Genomics Research and Involving People

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This thesis has been submitted in total fulfilment of the requirements for the
Doctor of Philosophy

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La Trobe University
Victoria, Australia

December 2021

Contents

Formal Acknowledgments

This thesis was written on Wurundjeri land, in the State of Victoria, Australia. I acknowledge and respect the traditional owners of this land. I also acknowledge that sovereignty was never ceded by Aboriginal peoples in Australia, and that this land was not peacefully settled. I also acknowledge the United Nations 'Declaration on the Rights of Indigenous Peoples' and acknowledge and respect the knowledge and cultures of all Indigenous peoples, leaders and Elders, past, present and future.

This doctoral research was supported by an Australian Research Training Program Scholarship and a La Trobe University Postgraduate Research Scholarship, which commenced on the 5th September 2016. Further personal and professional acknowledgements can be found in the section 'Full Acknowledgements', along with a statement acknowledging any real or perceived competing and conflicting interests.

Abstract

Public involvement in research occurs when the public, patients, or research participants are actively contributing to the research process. Involving people in research improves the quality, relevance and equity of research for everyone. Public involvement has been acknowledged as a key priority for prominent human genomics research initiatives in many different countries. However, to date, there has been no detailed analysis or review of the features, methods, and impacts of public involvement occurring in human genomics research projects worldwide, and evidence about the best methods to involve people remains limited.

This thesis explores how we can strengthen the principles of equity and human rights in genomics research, using evidence-informed methods. This doctoral research initially explored how people have been involved in human genomics research. I published a global review of nearly 100 current international genomics research projects and discovered that only one-third of them reported involving people. Applying learning from this, I co-designed research with four different groups to explore and evaluate practical ways of involving people. This represented a program of participatory action research across four different projects and domains of human genomics, each of which had different implications and challenges with regard to involving people the research cycle. The participatory action research projects included a large cohort study of >15,000 healthy, elderly research participants, a group of patients and families affected by a rare immunological disorder, an extended family of donor-siblings who share the same sperm-donor father, and the Australian Indigenous Precision Medicine project.

Involving each group had different challenges with regard to involving people in the research cycle. To describe methods and assess impact, I created a standardised way to report involvement across the four studies. I led an international team of more than 40 people to develop 'Standardised Data on Initiatives' (STARDIT), which is now being used by multiple organisations including Australian Genomics and the Wikimedia Foundation open-access journals.

Learning from this research indicates that people want to be involved and want to make decisions about genomics research and their own data, but they need support to get involved. In addition, researchers need support to involve multiple stakeholders in designing evidence-informed ways of involving people. Data from STARDIT can be used to support evidence-informed methods of involvement and strengthen equity in genomics research, helping to make sure the benefits of genomics research are for everyone.

Statement of Authorship

Except where reference is made in the text of the thesis, this thesis contains no material published elsewhere or extracted in whole or in part from a thesis accepted for the award of any other degree or diploma. No other person's work has been used without due acknowledgment in the main text of the thesis. This thesis has not been submitted for the award of any degree or diploma in any other tertiary institution.

Jack Stephen Nunn, 22nd December 2021

Plain English Summary

All life is made from code, from DNA. Human genomics research is the study of all the DNA in humans (the genome), together with the technologies that allow it to be analysed. When learning from genomics research is applied to human health, it is called genomic medicine, and it can improve lives.

In the next five years, it is estimated that nearly two billion people worldwide will have had their DNA sequenced. How can we ensure that future genomic research benefits everyone? How can we avoid a future where only those who can afford it are able to access those benefits? How can we involve people in genomic research, to maximise public health benefit?

Involving people in genomics research means sharing power, giving the public, patients and health technology consumers more control around decision making. Involving people is the best way to ensure genomics initiatives reflect the diversity of priorities in populations. It has been widely demonstrated that involving people in research improves the relevance and equity of research. Involving people increases public trust, acceptability and participation in genomics. It can also improve recruitment, ethical oversight and ultimately the quality of research outcomes. If genomics initiatives do not align with people's values, there is a risk that entire populations (in particular those people who are at greater risk of exploitation or under-represented, such as Indigenous peoples) will not participate in future initiatives, affecting the potential positive impact of genomics for decades. While funding bodies are increasingly requesting evidence of how people have been involved, data for evidence-informed ways of involving people in genomics research is lacking.

I am a public health researcher looking at genomic research and involving people in it. My PhD thesis explores how we can strengthen the principles of human rights in genomics, including using participatory action research. I have explored how can we do that practically, and what evidence is there about the best ways to involve people in other types of research, which can be applied to human genomics? For example, more people are discovering that a variation in their DNA might contribute to their risk of a disease, or that they or a loved one might pass on that risk to their children. Some people are told that the disease they are at risk of is so rare that it has not been well-researched, that there's no treatment or that treatment is too expensive. Some people with very rare DNA variations feel that finding a community of other people with the same variation is their only hope of surviving the disease, by working together to improve research and treatments. However, the best way for people to get involved in shaping the future of genomics research is not always clear.

As part of this doctoral research, I have published a global review of more than 100 current international genomics research projects and discovered that only one-third of them report involving people. Applying learning from this, I co-designed research with four different groups to explore and evaluate practical ways of involving people. This represented a program of research across four different projects and domains of human genomics, each of which had different implications and challenges with regard to involving people the research cycle.

I worked with participants in one of the largest clinical trials in Australia, to co-design a multi-generational study that explored the preferences of multiple stakeholders, including how they'd like to be involved in the trial. I also worked with a group of people affected by a rare disease, helping explore how they would like to be involved in future genomics research using online discussions. After starting my PhD, I decided to have my own DNA tested and discovered that my biological grandfather was a prolific sperm donor who fathered up to 1,000 people. After consultation with ethical experts and a co-design process with the siblings, I worked with 20 biological relatives from this group to co-design online discussions about future genomic research. Finally, I also worked with remote Aboriginal communities to co-design genomics research protocols.

Learning from this doctoral research has shown that people want to be involved and want to make decisions about their own data, but they need support to get involved. It's also demonstrated that researchers need to involve people in designing involvement plans, using evidence-informed methods to do this.

While I've learned that involving people has positive impacts, evidence about the best methods remains limited. For that reason, I created a standardised way to report involvement and I'm now leading an international team of more than 40 people to develop 'Standardised Data on Initiatives' (STARDIT). The working Beta version of STARDIT can report on research initiatives around the world in multiple languages. It has already been used by Australian Genomics to report planned work exploring how to involve people in genomics research. STARDIT can help us answer the question, 'What is the best way of involving everyone in shaping future research?' Once we can answer this question, we can all be involved in making sure the benefits of genomics research are for everyone.

Visual abstract

Executive summary

Background

Human genomics research is the study of all the DNA in humans (the genome), together with the technologies that allow it to be analysed. When learning from genomics research is applied to human health, it is called genomic medicine, and it can improve lives. Public involvement in research occurs when the public, patients, people affected by genomic variations of known or significance or research participants (hereafter 'people') actively contribute to the research process.

There is evidence that involving people in research assists with recruitment, improves its quality, relevance and acceptability, and promotes equity of research. Involving people in research means sharing power by giving the public, patients and participants control of aspects of the research – such as data access. In many countries, public involvement in human genomics research initiatives has been acknowledged as a key priority. However, to date, there has been no detailed analysis of the features, methods and impacts of public involvement in human genomics research, and no examination of practical, evidence-informed ways to involve people.

Involving people in genomics research (and in population-wide genomic initiatives) is essential for public trust, support, funding, acceptability and participation. Involving people is also the best way to ensure that genomics initiatives reflect the diverse priorities of populations. In many parts of the world including Australia, demonstrating evidence of how people will be involved in genomics initiatives is now part of the requirements for funding applications. If genomics initiatives do not align with people's values, there is a risk that entire populations will not participate in future genomic research. This would undermine the potential positive impacts of genomics for improving human health for decades to come, especially in the case of under-represented populations, or populations at greater risk of exploitation, such as Indigenous peoples. Although people are involved in genomics to a variable extent around the world, there is currently no standardised way to plan, report or evaluate how they are involved. To ensure that power is shared in culturally appropriate, culturally safe, transparent and cost-effective ways, those planning and funding future initiatives require new evidence-informed methodologies.

Research aims

The research aims for this doctoral research are to:

1. understand when and how people have been involved in human genomics research to date, and identify gaps that need to be addressed with new approaches and methods for involvement
2. apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research
3. develop a standardised way of planning, reporting and evaluating involvement in order to improve future genomics research.

Research methods

This doctoral study was conducted in a public health research setting, as distinct from an ethical, social or psychological research setting. The work is intended to be pragmatic, while also developing and applying theoretical frameworks. Several reviews were undertaken, including a systematic scoping review of public involvement in genomics research. These reviews have informed the case study methodology used on the subsequent four case studies. The case studies used participatory action research methods to explore the views and perspectives of four different groups of people associated with different human genomics research studies. The thesis describes the participatory action research process used during the involvement activities, and the subsequent co-design and implementation of the studies, as well as their associated impacts.

The case studies used different methods to involve people in every stage of the research cycle, supported by learning and development resources shared in different formats. Two used an online text-based discussion platform, two used face to face meetings, and one also used telephone interviews. The different methods used, and any associated impacts were reported in a standardised way.

Case studies

1. Involving research participants from Australia's largest clinical trial and cohort study of more than 15,000 healthy, elderly research participants, to co-design a future multi-generational research study (*ASPirin in Reducing Events in the Elderly study – ASPREE*)
2. Involving people affected by a rare condition in shaping future genomic research through working in partnership with a rare disease charity (*ausEE study*)

3. Co-designing genomics research with one of the largest known groups of donor-conceived siblings in the world (*Shared Ancestry study*)
4. Involving Australian Indigenous peoples in co-designing a research protocol for a precision medicine project (*Indigenous Precision Medicine project)*

Standardised Data on Initiatives (STARDIT)

This thesis also describes the co-creation of Standardised Data on Initiatives (STARDIT), a standardised way of reporting on the planning, execution and evaluation of involving people in human genomics research and other initiatives. I conceived STARDIT and led an international co-design process to co-create this new framework. During the co-creation process I applied it to the four case studies. I used the STARDIT framework for reporting on the participatory action research process used in the four case studies from different prominent genomics research projects in Australia. The framework was also used to create standardised data for a cross-case analysis of preferences for involvement, methods of involvement, and the impacts of involvement across the four case studies.

Results

This thesis has developed and used a standardised way of planning, reporting on and evaluating stakeholder involvement in genomics research and other related initiatives. Learning from this research will help advance the field of involving people in genomics research.

Results from reviews

The narrative review demonstrated there is not currently enough data to complete a meta-analysis of quantitative or qualitative data about involvement in genomics research. It informed the decision that a systematic scoping review was the most appropriate method to search for relevant data. Findings from this review also suggested that methods of involving people guided by the paradigm of participatory action research were most likely to have impacts.

The systematic scoping review provided a useful 'snapshot' of current international genomics research projects, by using a database provided by the Global Alliance for Genomics and Health (GA4GH). While a third of initiatives reported involving people, only 10% of initiatives reported impacts. The limited reporting of involvement suggested there would be intrinsic value in developing a more systematic method of both reporting and evaluating how people are involved in human genomics research, as data from such reporting could provide the evidence required to inform

future policy around involvement of the public in genomics research. The recommendations from this review informed the co-creation of STARDIT.

Results from Standardised Data on Initiatives (STARDIT)

STARDIT provides a standardised ways of planning, reporting and evaluating genomics research, including involvement. During this doctoral research, I co-created Standardised Data on Initiatives (STARDIT), a standardised way of reporting on involvement in genomic research and initiatives. The STARDIT system enables reporting in multiple languages and is applicable beyond public health genomics. It has already been used by projects beyond those described here, including by the Australian Genomics working group 'Involving Australia', and has been suggested for use in describing involvement in biobanks. In addition the STARDIT (Alpha version) was cited as 'useful' as a way of 'evaluating engagement' in an article supported by the Global Alliance for Genomics and Health (GA4GH). The STARDIT Alpha version is also cited in the GA4GH 'Framework For Involving And Engaging Participants, Patients and Publics In Genomics Research And Health Implementation' as a useful way of 'conducting evaluations of engagement'.

Results from case studies

As part of this doctoral research, STARDIT was also demonstrated as a way to map preferences (using the preference mapping tool STARDIT-PM), plan involvement and report the impacts of involving stakeholders across the four case studies (both research participants and study team members). In each case study, the process of involving people in the research led to positive impacts and outcomes, with no negative impacts or outcomes reported. Using STARDIT allowed these to be reported in a standardised way, while using STARDIT for the analysis made it possible to combine all the public domain data from the case studies and to categorise the data. This allowed comparison of STARDIT-PM data from 83 stakeholders in the three case studies in which preferences were mapped (by the facilitators of the online discussions in the ausEE and Shared Ancestry case studies, and by study team members from the ASPREE case study). By combining data about preferences for who should be involved in research from the Shared Ancestry and ausEE case studies, it was possible to show that 45 per cent of participants' responses 'widened' to include a preference for more people to be involved in the research (N=43/95). For 43 per cent, preferences stayed the same (N=41/95), and in 12 per cent they 'narrowed' (N=11/95).

Overall, my results suggest there is inherent value in planning and reporting involvement in a standardised way, which allows the creation of data from which to make evidence informed decisions about effective ways of involving people. In this thesis, I have led the co-creation process to build a way of doing this that anyone can use, and demonstrated how it can be applied in multiple

research settings as case studies. By reporting different case studies in a standardised way, I have shown how it is possible to combine the data in order to better understand data from multiple sources. In this thesis, I used this data to show that through the process of involving people in the genomic research, most participants' preferences for who should be involved in genomics research 'widened' to include more people.

Discussion

Learning from these case studies can be applied to various research settings, but is particularly valuable for research involving populations at greater risk of exploitation– such as people affected by rare diseases or Indigenous populations, where stakeholder involvement is critical. The published outputs from this thesis have collectively received more than 43 citations, demonstrating the interest in the issues explored in this thesis. During this thesis STARDIT has been used by projects outside the scope of this doctoral research, including by a working group led by Australian Genomics, to report prospectively on how people will be involved.

In order to ensure that planning, reporting and evaluation of involvement in research increases equity in public health genomics and other fields, further ongoing co-development of the methods described here (including STARDIT) is required.

Conclusion

Greater involvement of stakeholders in global human genomics research has intrinsic value worldwide. In this thesis, I reviewed contemporary practice and applied theoretical constructs in the context of real-world genomic research to develop novel ways of reporting impacts, described in peer-reviewed case studies. My thesis has demonstrated that STARDIT can be used as a standardised way of planning, reporting and evaluating involvement in genomics research, which works across multiple human languages. STARDIT has been demonstrated as a systematic, practical and effective way to co-design, report on and evaluate public involvement in genomics research. The data created by STARDIT has potential to inform best practice in future genomics research, and other disciplines. Further work to co-develop more systematic ways of reporting and evaluating such involvement, and supporting ways of embedding this practice into research would be highly beneficial.

List of thesis publications and outputs

Title	Status	Year	Contribution	Link	Cited	STARDIT report
Jack Nunn 2018 Visualise your thesis entry	Published (La Trobe University)	2018	100%. Sole author	Yes	N/A	No
Public involvement activities in 96 global genomics projects *The Human Genetics Society of Australasia Conference*	Conference poster (HGSA)	2018	70%. First author, main contributor	Yes	N/A	
Eosinophilic gastrointestinal disorders and genomics.	Published by registered charity	2019	70%. First author, main contributor (checked by qualified medical experts)	Yes	N/A	
Involving people in genomics research	Conference poster (American Society of Human Genetics Conference)	2019	70%.First author, main contributor	Yes	N/A	No
Public involvement in global genomics research: A scoping review[1]	Published in Frontiers in Public Health (peer reviewed)	2019	70%. First author, main contributor	Yes	31	No
Reducing health inequalities by involving Indigenous people in genomics research.[2]	Published in Health Voices - Journal of the Consumers Health Forum of Australia (Editorial board review)	2019	100%. Sole author	Yes	3	No
Standardised Data on Initiatives (STARDIT) Alpha Version	Pre-print (for public review)	2019	70%. First Author, main contributor	Yes	5	No
A Pathway to Precision Medicine for Aboriginal Australians: A Study Protocol[3]	Published in Methods and Protocols (peer reviewed)	2021	45%. Second of 10 authors. Contributed sections on participatory research.	Yes	1	Yes
Co-designing genomics research with donor conceived siblings[4]	Published in Research Involvement and Engagement (peer reviewed)	2021	70%, First author, main contributor	Yes	2	Yes
Equitable expanded carrier screening needs Indigenous clinical and population genomic data. [5]	Published in The American Journal of Human Genetics (peer reviewed)	2020	5%. One of 40 authors, contributed sections about participatory research	Yes	11	No
Involving elderly research participants in the co-design of a future multi-generational cohort study[6]	Published in Research Involvement and Engagement (peer reviewed)	2021	70%. First author, main contributor	Yes	1	Yes
Participating in a discussion on the UK Genetic Society's podcast	Published (Genetics Society Podcast)	2020	30%. Guest, featured interview. Spoke about PhD	Yes	N/A	No
Guidance for planning, reporting and evaluating initiatives: A multidisciplinary scoping review	Pre-print, Wiki Journal of Science	2021	70%. First author, main contributor	Yes	0	No

Title	Status	Year	Contribution	Link	Cited	STARDIT report
Involving people affected by a rare condition in shaping future genomic research[7]	Published in Research Involvement and Engagement (peer reviewed)	2021	70%. First author, main contributor	Yes	0	Yes
Participating in a discussion on an Australian Broadcasting Corporation radio interview [8]	Published on ABC website	2022	100%, interviewee	Yes	0	
Standardised Data on Initiatives - STARDIT: Beta Version[9]	Published in Research Involvement and Engagement (peer reviewed)	2022	70%. First author, main contributor	Yes	0	No

This table is correct as of 16th December 2021, citation data from Google Scholar.

List of publications which informed this thesis

This is a list of publications was created in parallel with this thesis during my PhD candidature. While they are not directly relevant to the thesis question, learning from these informed my thinking and understanding in certain areas.

Publication title	Status	Year	Contribution	Link
Research priorities in health communication and participation: International survey of consumers and other stakeholders[10]	Published (peer reviewed)	2017	10%. Co-author	https://doi.org/10.1136/bmjopen-2017-019481
Stakeholder involvement in systematic reviews: A scoping review[11]	Published (peer reviewed)	2018	10%. Co-author	https://doi.org/10.1186/s13643-018-0852-0
Development of the ACTIVE framework to describe stakeholder involvement in systematic reviews[12]	Published (peer reviewed)	2019	10%. Co-author	https://doi.org/10.1177/1355819619841647
Stakeholder involvement in systematic reviews: A protocol for a systematic review of methods, outcomes and effects[13]	Published (peer reviewed)	2017	5%. Co-author	https://doi.org/10.1186/s40900-017-0060-4
Involving the public in rare cancer care and research, textbook of uncommon cancer[14]	Published (peer reviewed)	2017	90%. First author, main contributor	https://doi.org/10.1002/9781119196235.ch3
Selecting, refining and identifying priority Cochrane reviews in health communication and participation in partnership with consumers and other stakeholders[15]	Published (peer reviewed)	2019	10%. Co-author	https://doi.org/10.1186/s12961-019-0444-z
STARDIT public consultation report – September to December 2019[16]	Published by registered charity	2019	100%. Sole author	https://doi.org/10.26181/5e8c0dd2976b8

What are systematic reviews?[17]	Published (peer-reviewed)	2020	90%. First author	https://doi.org/10.15347/WJM/2020.005

List of projects and work which informed this thesis

This thesis has used a participatory action research methodology. Accordingly, during my candidature I have led or been involved in a number of projects and other work which has informed this doctoral research.

Health

- Member of the Medical Services Advisory Committee Evaluation Subcommittee (MSAC-ESC), Australian Government Department of Health) – including involvement in creating recommendations for funding genetic testing (2019–present)
- Health Technology Assessment – Consumer Consultative Committee (Australian Government Department of Health) – involved in discussions about public involvement in health technology assessment decision making and processes (2019–present).
- Member of team at ASPREE, helping with co-design involvement for a future multi-generational study (2016–present).
- Member of the research team at the Centre for Health Communication and Participation, La Trobe University (2014–2018).
- Member of research team at Poche Centre for Indigenous Health (2018–present).
- Member of the Cochrane Council, Cochrane Consumer Executive Committee and the Cochrane Advocacy Advisory Board, international (2020–present).
- Member of the Australian Genomics working group 'Involving Australia' (2021-present)
- Director of an organisations which partners with the Industry Genomics Network Alliance (InGeNA), Australia, (2021-present)
- Relevant consultancy work including work with Australian Government Department of Health and World Health Organisation, and work with the Victorian Comprehensive Cancer Centre, to scope how people affected by cancer and the wider public can be involved in personalised cancer care.

Publishing

Strategy Liaison for the WikiJournals – involved in developing strategies for the not-for-profit open access publisher, including integrating STARDIT on the platform, international (2018–present).

Other

Director and founder of the charity 'Science for All', leading co-design of projects to involve the public in DNA research using participatory research methods and reporting using STARDIT (2018–present).

Chapter 1 – Introduction

In order to improve evidence-informed methodology, this doctoral thesis explores why people should be involved in genomics research, how they can be involved, and demonstrates ways of planning, reporting and evaluating involvement in a standardised way.

The research aims for this doctoral research are: to (1) understand when and how people have been involved in human genomics research to date; (2) apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research; (3) develop a standardised way of planning, reporting and evaluating involvement in order to improve future genomics research. The research aims and activity are summarised in more detail in Chapter 2 'Research aims and activity'. This chapter aligns with research aim 1 "understand when and how people have been involved in human genomics research to date, and identify gaps that need to be addressed with new approaches and methods for involvement".

Introduction to involvement in health research

What is public involvement?

In 1969 Arnstein wrote that, "Participation of the governed in their government is, in theory, the corner-stone of democracy".[18] Less than a decade later, this sentiment was enshrined in the World Health Organization's (WHO) 'Declaration of Alma-Ata', which stated that "the people have the right and duty to participate individually and collectively in the planning and implementation of their health care."[19] The Alma-Ata declaration was followed by this statement from the WHO in 1986:

> "Health promotion works through concrete and effective community action in setting priorities, making decisions, planning strategies and implementing them to achieve better health. At the heart of this process is the empowerment of communities – their ownership and control of their own endeavours and destinies. Community development draws on existing human and material resources in the community to enhance self-help and social support, and to develop flexible systems for strengthening public participation in and direction of health matters. This requires full and continuous access to information, learning opportunities for health, as well as funding support."[20]

Involvement should attempt to create a balance between individual autonomy and collective values in order to achieve equitable access to the things which improve health and wellbeing.[21] Variation in people's priorities means that it is important to try to articulate the differences and decision-making processes in a transparent way.[21–23] The process of involvement must be as inclusive as possible. For example, people must feel that they have been supported to influence the creation of health policy or services to ensure that they are acceptable.[21,24]

While there is no international consensus on terminology to describe involvement in health research,[25] the United Kingdom's National Institute of Health Research (NIHR) provides one of the most succinct and helpful definitions of involvement:

> "By public involvement we mean research being carried out 'with' or 'by' members of the public rather than 'to', 'about' or 'for' them,"[26]

As part of an international review in 2017, eight government research funders produced guidance that endorses patient and public involvement.[25] However, while international support for involvement is growing, definitions of 'involvement' are varied and inconsistent. Even between and within Anglophone countries, words such as 'engagement', 'involvement' and 'participation' are used interchangeably.[27–29]

Similarly, to be meaningful, words such as 'patient', 'public', 'community', 'consumer' and 'payer' need careful articulation. While terms such as 'people' or 'the public' exclude fewer people than the terms above, they also lose some meaning in the process of generalising. What is meant by 'the public' is defined more clearly by the UK Health Research Authority which states:

> "… the term public means patients, potential patients or members of the public including those with known genetic dispositions, carers and people who use health and social care services as well as people from organisations that represent people who use health and social care services."[300]

Noting that 'potential patients' includes everyone, the word 'stakeholders' can be a more useful term, if the usage of this term is defined carefully. In this thesis, the term 'stakeholder' means anyone who has a 'stake' in health research, in particular those with important knowledge, experiences, expertise or views that should be taken into account.[21,31,32] It can include, as sub-categories; researchers; research funders; policy makers, people affected by the research; people

with specific health conditions, people with specific genomics variations; patients and the general public (who may also be categorised as 'tax-payers' for publicly funded research). The term 'people' and 'the public' will be used to refer specifically to patients, potential patients, carers, payers, consumers of health technology and the general public, excluding professional researchers, research funders, policy makers and anyone else with a professional connection to research.

However, current variations in terminology and inconsistency in definitions in all human languages make it challenging to compare how different stakeholders are involved in different tasks in genomics, and why.[28,33] This creates significant problems in public health genomics, impedes comparison and the ability to share standardised data to support both ethical and evidence-informed methodologies. Ways of exploring the 'why' of involvement are explored in detail in the section 'Research Paradigms'.

Why involve people in research?

Involving people in shaping future genomics research can be important for a range of reasons. There is evidence that involving multiple stakeholders (including the public and patients) in research improves research and outcomes. Research prioritisation processes that do not involve people can result in a mismatch between the research and the needs of people the research is intended to help.[34] Involving people in the research cycle helps improves trust in research and public influence over research.[21,35,36] It can help ensure that research is conducted in an ethical, accessible, responsible and transparent manner.[37] It can also help ensure that research reflects the balance and diversity of priorities within populations.[19,38] Involving people in research can therefore be considered both a moral and a scientific imperative.[39–41]

Increasing the involvement of the public in human genomics research (hereafter 'genomics research') and genomics policy development has been identified as a crucial aspect of responsible research practice.[21,42] For genomics research to be successful, public support, in terms of funding decisions and willingness to participate, is necessary.[32] This is particularly important with respect to populations at greater risk of exploitation, such as those with histories of experiencing medical and research abuse.[43]

The principles and best practices of research involvement outlined above have not yet been fully integrated into the emerging field of human genomic research. Using an evidence informed methodology, my thesis seeks to help improve the translation of these principles into the field of human genomics, where they are highly relevant and urgently required.[44,45]

What is human genomics research?

Human genomics research is the study of all the DNA in humans (the genome), together with the technologies that allow it to be analysed. This analysis includes interactions of the DNA with other molecules and with the environment. When learning from genomics research is applied to human health, it is called genomic medicine, and it can improve and save lives.

Genomics research has the potential to improve disease prevention and to inform more targeted, more effective interventions. Genomics research works best when it is combined with rich data about a person's medical history, lifestyle and other information. With more data, we can make more accurate predictions. Around the world, more people are participating in human genomics research and it is impacting on more people's lives in multiple ways. The number of people involved in genomics research is predicted to grow substantially in coming years.[46,47] By 2025, it is estimated that nearly two billion people worldwide will have had their DNA sequenced. Responsible and effective public involvement in human genomics research is thus a global imperative.[10]

According to the Global Alliance for Genomics and Health, (as of March 2021)[48] there are currently 221 active genomics initiatives globally, noting this is not an exhaustive list. These initiatives include biobanks, databases and other data sharing initiatives. Nine of these are government-led health initiatives. As the cost of sequencing, analysis and data storage declines, the number of global initiatives is predicted to grow exponentially, with large segments or even entire populations participating in such research.

In addition, increasing numbers of people are purchasing online DNA tests from commercial companies and gaining information about their own DNA. Sometimes called 'direct-to-consumer' (DTC) or personal genomic tests (PGT), these can be broadly grouped into 'health-related' and 'recreational' tests,[49] with the latter including tests for wellbeing and ancestry purposes. Popular providers such as 23andMe and Ancestry DNA charge a one-off fee for the DNA test and result but claim the rights over the produced DNA data. DNA data is then used to improve their services and are often sold at a profit to other companies, which results in significant amounts of data for research.[50] Private 'direct-to-consumer' companies hold some of the largest DNA databases in the world, with 23andMe and UK drug maker GlaxoSmithKline recently entering a partnership to identify potential candidates for clinical drug trials.[51] The issue of informed consent in this process is explored in the Discussion section 'Implications for ethics and participatory action research'.

Crucially, in addition to receiving data on their own DNA, many of these services offer people the choice of access to their own data, sharing their data with other projects or 'donating' data for research. Communities of people researching themselves are also redefining the relationship between 'researcher' and the researched. Open access and 'community-led' projects – such as GedMatch,[52] OpenSNP[53] and Open Humans[54] – have potential to make access to interpretation of data and control over personal data more inclusive. However, these projects remain at risk of unauthorised uses of data and buy-outs – for example, despite being a not-for-profit volunteer initiated project, GedMatch has been used for high-profile criminal justice applications, and was recently sold to a venture capital firm.[52,55]

It is important to note that while this thesis is concerned with human genomics research in the context of 'public health', human genomics research is not limited to this domain. Genomics research and the applications of genomic data are not limited to public health and medical applications, and the 'blurring' of the boundary between medical and non-medical uses has been noted in a recent report by the UN.[56]

For many members of the public, such distinctions are academic, and they primarily associate the practical application of genomics research to domains such as ancestry and criminal justice.[57] Participants in two of the four cases studies in this thesis raised valid concerns about having data they provided for health research used by the criminal justice system. This issue is explored in the results section 'Different interests'.

In this thesis, the term 'genomics research' is used to encompass domains beyond health, government and academia. The term 'genomic medicine' or 'public health genomics' is used to provide specificity, when required.

Research or care?

Even within the domain of genomic research for 'medicine' and 'health', the conceptual and practical division between 'healthcare' and 'health research' is blurring, in particular, in the treatment of rarer cancers.[58] For some people with rarer cancers, participating in research is their only care option, so that from the patient perspective health research is also a 'service'. There is also often a temporal gap between the discovery of effective new treatments in research settings and their availability in clinical practice.[58] This interval can create pressure on researchers to provide clinical care that is not available to patients elsewhere.[58] Thus, the distinction between genomics

research and genomics medicine is increasing unhelpful. This is best illustrated by the comprehensive cancer centre model.

The USA's comprehensive cancer centre model (also used in Australia) aims to facilitate the translation of research (including genomics) into routine clinical care.[59] Using learning from genomics research to carry out genomic profiling (of people's DNA and tumour DNA) in order to inform medical treatment is known as 'precision medicine'– the aim of which is "giving the right treatment to the right patient at the right time".[60] Similarly, 'precision oncology' resists definition as 'one thing', and is more accurately an interconnected concept which is "erasing boundaries between the clinic, laboratory and healthcare industry".[61] Contemporary cancer centres blur distinction between research and health care as they must integrate data from clinical observations into laboratory research, develop clinical and public health intervention strategies from basic scientific discoveries, and provide high-quality treatment and diagnosis. In addition, they must develop educational resources about cancer prevention for the public.

Precise terminology in 'precision medicine' thus remains a challenge, with some patients reporting that such terms are not 'meaningful'.[62] Accordingly, in this thesis, involvement in human genomics research can include involvement in healthcare, health services and health research (including industry) where learning from genomics research is directly applied, or data from those activities inform genomics research.

Genomics research and involvement

> "No sensible decision can be made any longer without taking into account not only the world as it is, but the world as it will be ..." (Isaac Asimov, 1978).[63]

The data generated by genomics research is literally astronomical in scale.[64] Projects such as the 100,000 Genomes Project create hundreds of millions of data points, with global sequencing capacity expected to exceed 35 petabases per year.[64]

To be useful, such human genomic data require combination and comparison with similar sized datasets, including medical records and other phenotype data. As no single organisation worldwide has enough resources to fund research at the scale required for clinical genomics research, national genomics programs must collaborate of necessity.[65] These collaborations require ethical standards and protocols for data sharing and provide an important opportunity for the public to be involved in shaping decisions about research. Memoranda of Understanding signed by Genomics England with British Columbia in Canada and the Garvan Institute in Australia are examples of such

collaboration.[66] The Chief Scientist for Genomics England stated that this kind of data sharing and collaboration provides an opportunity to "showcase their achievements to patients and the public in order to promote further involvement from those communities".[65]

There is a growing need to increase public involvement in human genomics research and policy development and this has been identified as a crucial aspect of responsible research practice.[21,42] As successful genomics research relies on people choosing to share their data, public support is essential.[67] Many high-profile genomics research initiatives have already made public statements about the importance of involving people, with some governments positioning public involvement as a democratic right.[32,36,68] For example, in the 2017 report 'Generation Genome', the UK's Chief Medical Officer suggested that shaping the future of genomics research requires the "active involvement of many stakeholders including patients, health professionals, researchers, policymakers, and wider society," with a "key role for public engagement and involvement".[32]

Examples of public involvement in more recent projects are summarised in detail in the global systematic scoping review of public involvement in genomics (Chapter 4).[28] Initiatives which feature multiple ways for people to be involved include the UK Biobank, Genomics England and the USA's Precision Medicine initiative 'All of Us'. In Australia, the 'Genomics Health Futures Mission' (part of the Medical Research Future Fund) requires that the public should be "involved in setting the priorities" of the Mission.[69] In 2019, it became compulsory that grant applications demonstrate how people would be involved in proposed projects.[69] Australia's Medical Services Advisory Committee also seeks feedback on the health technology assessment (HTA) process from the public and people affected by genomic variations.[70]

Compared with other kinds of research, genomics research poses unique challenges in relation to involvement. Technological advances in genomics and the clear need for data sharing have seen a shift from autonomy towards concepts of reciprocity, solidarity and universality.[71(p2),72(p546)] The need for data from so many people, who share approximately 99.9 per cent of their genome with all other humans, challenges concepts of individualism and promotes concepts of 'solidarity'.[73] The inherent commonality in the DNA we all share gives rise to an expectation that any benefits from knowledge generated by genomics research will be shared by all humans. In a 2015 report, the United Nations International Bioethics Committee wrote of concepts of justice and solidarity, stating that:

> "… genetics promises to offer an unprecedented contribution to improve health care. These advancements should be shared with society as a whole and with the international community; any discrimination has to be avoided."[56]

Concepts of individualism and solidarity are explored further in the Methods (Chapter 3) in the section 'Individual rights or collective solidarity?''.

Genomics and other biomedical research present specific social, ethical and legal challenges, including those relating to communication and commercialisation of research results, balancing individual rights against the collective good, potential for genetic discrimination, and data protection. Emerging biotechnology should be subject to public debate about ethics and decision making, and orientated to the pursuit of the public good.[45(p22)] International cooperation is required to achieve real and effective public involvement in genomics research and innovation.[45(p2)] By involving the public in shaping future genomic research, we can create research that is more likely to benefit more people.[74(p6)]

However, those participating in either government or privately funded research often have limited information on which to base informed decisions about participation. This extends to information about what will happen with their data, control over their data once they are entered into databases, and opportunities to be involved in data-governance decisions.[28] While innovations in giving research participants control over their data are being made,[75] the public still has limited control over research questions, priorities or methods, and limited ways to hold researchers who breach law or agreed ethical standards accountable.[28] In addition, there is limited evidence on ways to carry out collective decision making at various levels of genomics research governance – including in international law, government policy and other research initiatives. Evidence is also limited in relation to the effectiveness of education to improve genomics literacy (with both the public and health professionals) and to support people to critically appraise information sources about genomics research.[28]

The social movements in biotechnology such as 'Do-it-yourself Biology' (DIY-Bio), warrant consideration in this thesis. Projects such as the Open Insulin project have highlighted the fact that access to medicine (including genomic medicine) is not universal or affordable for many people.[76] A University of Sydney project called 'Breaking Good', which worked with school children to create malaria drugs, indicates the growing acceptance of community-controlled research processes.[77] My own work with the charity 'Science for All' and the DIY-Bio charity 'BioQuisitve' on the 'Wild DNA' project showed how cheaper access to genomics sequencing and analysis is providing new frontiers for more people to get involved in doing research themselves. As more people are beginning to take direct action to solve issues of access to genomic medicine, the DIY-Bio movement is thus further

blurring boundaries between 'researchers' and 'the public'. This phenomenon is explored further in the Discussion section 'DIY-Bio and health technology'.

The need for research into public involvement in genomics research

Despite increasing interest worldwide in large-scale human genomics initiatives, limited research has been done on how the public are currently being involved, and no research has been conducted on the impacts and benefits of genomics initiatives involving the public.

While systematic reviews on involving the public in other types of health and medical research have been published,[37,78,79] there are no comparable reviews in human genomics. In other areas of medical research, many reviews have concluded that public involvement activities are inconsistently reported or under-reported[29,37,80–82] – including the specific ways in which people are involved in medical research and any impacts from involvement.[35,79,80]

Genomics research is a global enterprise and will need to continue to involve populations who speak diverse human languages. Accordingly, best-practice methodologies for involvement are needed and must work across these same boundaries. How such urgent challenges can be met is the subject of this thesis.

These issues are explored further in the Methods section 'Involvement in genomics: Mapping what is known and unknown', and in Chapter 4 in the global systematic scoping review of involvement in genomics research that I conducted as part of this thesis.

Research conducted as part of this thesis

This thesis describes how I formed partnerships with two prominent genomics projects in Australia and two communities of people affected by genomic research in order to conduct participatory action research in these four settings. This thesis reports the processes as separate case studies, using a method of standardised reporting in order to report and compare the case studies consistently. The results and learning from these case studies is summarised in Chapter 9 'Results: Comparison of all case studies' and Chapter 10 'Discussion'. This thesis concludes by summarising

how the observations, learnings and outputs from this thesis have advanced the field of evidence informed involvement in genomics research.

About this thesis

While this thesis focusses on involving people in the context of genomics research, it is a thesis about participatory action research, involvement in research and ways of planning, reporting and evaluating it in a standard way. Public health genomics was chosen as the area to apply these paradigms as it was identified as a complex area requiring urgent attention. Doctoral research for this thesis has been included in 8 reviewed (or under review) publications and 2 pre-prints. The published papers are included in this thesis in full or in part. To aid the examiners, peer-reviewed content is indicated by *italic text* before and after the peer-reviewed content. The section 'Publications and author contributions' summarises all publications associated with this thesis that I have authored or co-authored. This section outlines my contribution to each publication and shows how each is incorporated into, or has informed, this doctoral research.

Text-based additional files from the published peer-reviewed articles can be found in the appendices, with the data files hosted by the journals, accessible via the Digital Object Identifier (DOI) links.

In order to explain the iterative participatory action research processes used, the section 'Thesis Timeline' presents a visual timeline of the doctoral research and related activities. Visual summaries (infographics) of the research are also included, where possible, to make the content matter more accessible to those with different learning or processing preferences. The structure of this thesis is described in more detail below in the section 'Thesis Structure'. To facilitate non-linear reading of this thesis on electronic devices, hyperlinks (denoted by an underscore) are included in chapters and subsections, along with a detailed table of contents which can be found in the section 'Detailed contents'.

Formatting and referencing styles are consistent throughout, including the peer-reviewed articles, meaning all referencing is consistent throughout this document. The text and graphical content of the peer-reviewed articles in this thesis are otherwise identical to the published versions, although numbering of figures and tables within peer-reviewed articles is aligned with the published versions.

Formatted versions of articles created by the publishers can be found by following the hyperlinks in the references. This thesis uses the American Medical Association referencing style which uses superscript numbers.[0]

As this thesis has been written by one person (Jack Nunn), as the author, I will use a first-person active voice to describe things I have done. However, when they have been done as part of a team (for example, all the case studies), I will use the third person passive voice to describe what was done by the study team.

Audience

The audience for this thesis is anyone who is interested in learning more about evidence-informed ways of involving people in health research. Where possible, it has been written in plain English, with any specialist terms defined (see section 'Definitions'). The thesis is thus written for a general audience with no specialist knowledge required, noting that the methods used in this thesis (including paradigms, data collection and analysis) are explained within the thesis, with references provided where further reading may be helpful for the reader.

While this PhD focusses on involving people in genomics research, it is a thesis with relevant and transferable applications to participatory action research, and involvement in other kinds of research and ways of planning, reporting and evaluating such participatory research in a standard way. Genomics was chosen as the area of application for these methods as it was identified as a complex area requiring urgent attention. While a basic understanding of genomics is helpful, expertise in genomics is not required to understand this thesis.

Definitions as structured data

Words and related concepts used throughout this thesis are defined in Table 1.1. WikiData categories are used in order to standardise terminology, where possible. In some cases, I have created, amended or improved the Wikidata item.[1] These definitions therefore also exist as structured data, machine readable as part of both taxonomies and ontologies. This thesis (and STARDIT) both use Wikidata. Created in October 2012, "Wikidata is a large-scale, human-readable, machine-readable, multilingual, multidisciplinary, centralized, editable, structured, and linked knowledge-base".[83] It has been identified as a useful resource for biomedical data integration and semantic interoperability between biomedical computer systems, including allowing the database to be automatically processed by users as well as by machine learning.[83] In addition, it is highly relevant to the field of human genomics, as it has been used "in the fields of genomics, proteomics, genetic variants, pathways, chemical compounds, and diseases".[84] Wikidata also adheres to the FAIR principles of findability, accessibility, interoperability and reusability. [85]

[1] The Wikidata categories have been formatted as hyperlinks to the Wikidata item, as they are not in themselves references but links to structured data, which in some cases, I have edited the Wikidata online entry. For transparency, I have used my Wikimedia username 'JackNunn' for all such edits.

Table 1.1: Defining words and concepts

Word (Wikidata entry)	Definition
Barriers (Q109580927)	Barriers refers to things that might prevent something from happening.
Enablers (facilitators) and barriers (Q109580938)	Enablers refers to things that help enable something to happen. For example, paying people's travel expenses might enable them to get involved in research. *Note:* The word 'facilitators' is used in the published scoping review of public involvement in genomics, whereas 'enablers' is used throughout the rest of this thesis.
Exploitation (Q859884)	The word 'exploitation' in this thesis means the act or result of forcibly depriving someone of something to which they have a right, such as autonomy over the use of their own data. *Note:* The word 'vulnerable' was initially used in some published manuscripts of this thesis, however as a result of the co-design process of STARDIT and thesis review, it was decided that this word was problematic, as it is imprecise and vague about who is deciding who is vulnerable and why. [86] Accordingly, the word 'vulnerable' was subsequently changed to 'people/populations at greater risk of exploitation' in this thesis and later publications.
Genome (Q7020)	A genome is an organism's complete set of DNA, including all of its genes.
Genomics (Q222046)	Genomics is an interdisciplinary field of biology which explores the structure, function, evolution, mapping and editing of genomes.
Genomics research	The term 'genomics research' is used in this thesis to describe research into genomics domains beyond health and beyond government and academia. The term 'genomic medicine' or 'public health genomics' is used to provide specificity, when required.
Involvement in research (Q109581008)	Involvement in research refers to research being carried out 'with' or 'by' members of the public, rather than 'to', 'about' or 'for' them. This can include working to prioritise research topics, helping design research, helping manage it, and helping evaluate it.[87]
Participatory action research (Q7140444)	Participatory action research is an umbrella term which describes a number of related approaches, including forms of action research which embrace a participatory philosophy and include 'co-design' and 'co-production' of research.[88] It is a process whereby researchers, the public and other relevant stakeholders "work together, sharing power and responsibility from the start to the end of the project",[89] including knowledge generation and translation.[89]
People and 'the public' Public	The term 'people' and 'the public' will be used to refer specifically to patients, potential patients, carers, payers, consumers of health technology and the general public, excluding

(Q2388316)	professional researchers, research funders, policy makers and anyone else with a professional connection to research.
Stakeholder (Q109581040)	The term 'stakeholder' means anyone who has a 'stake' in health research or service, in particular those with important knowledge, experiences, expertise or views that should be taken into account. It can include: researchers; research funders; policy makers; people affected by the research; people with specific health conditions; people with specific genomics variations; patients; and the general public (including 'tax-payers' for publicly funded research).

Thesis structure

This thesis documents doctoral research which included multiple reviews and four case studies and was also informed by professional experience in parallel to the research.

A timeline showing thesis development is provided to aid reader understanding of its distinct parts and how they interrelate (Figure 1.1). The results chapters of this thesis are presented in the chronological order in which the research was conducted.

Chapter 1 introduces the concept of involvement and how it applies to genomics research. The current context of genomics research is summarised in order to show the importance and urgency of addressing the research questions examined in this thesis.

Chapter 2 summarises the research aims of the thesis.

Chapter 3 provides a detailed overview of the methods at each stage of the research process. It explores the perspectives, research paradigms and methods used and provides a summary of the research methods. The section 'Involvement in genomics: Mapping what is known and unknown' shows how the narrative review informed the choice of research paradigms, and how the scoping review informed the development of the 'Standardised Data on Initiatives' (STARDIT). This section concludes with a description of the co-creation process I led to produce STARDIT, the framework used throughout this thesis to describe the data from the case studies in a standardised way. The Beta version of STARDIT is included to explain the system which is central to the methods used in this doctoral research. The methods described in this section are further appraised in the Discussion Chapter, in section 'Methodological evaluation'

Chapter 4 (results chapter) contains the published, peer-reviewed systematic scoping review of involvement in global genomics research projects.

Chapters 5, 6 and 7 (results chapters) contain the three published, peer-reviewed case studies I led as part of this doctoral research.

Chapter 8 (results chapter) is a reflective case study I authored, about my work with organisations involving Indigenous peoples in genomics research. The chapter describes the co-creation of a research protocol, and explores the co-design process from the very inception of a research project, as well as how to report intended involvement in a standardised way using STARDIT.

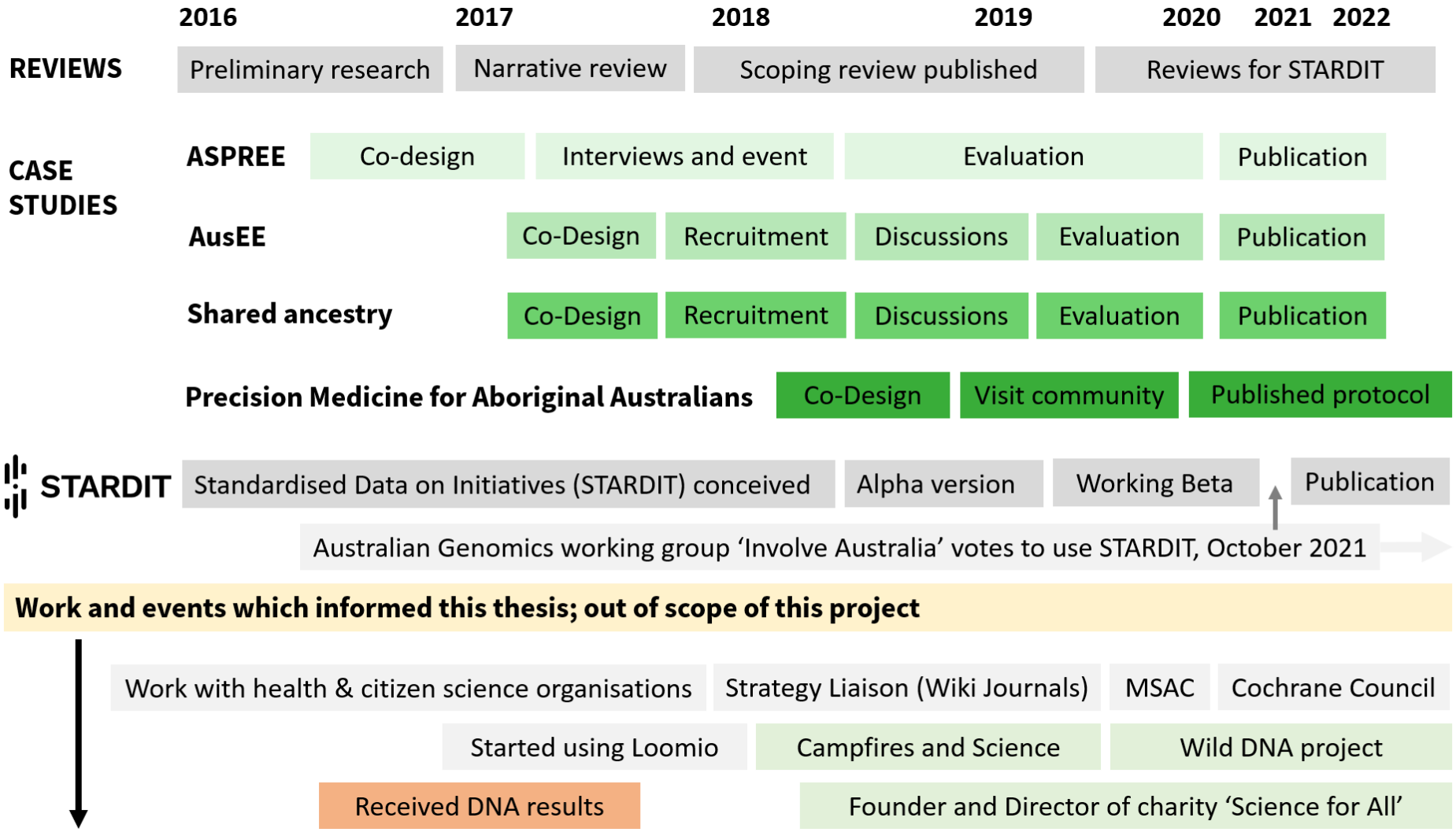
Chapter 9 summarises the results from all case studies and compares the findings from each. This cross-case analysis includes an exploration of the themes, generalisations, similarities and differences between these case studies. It uses STARDIT to compare the data in a standardised way using a combination of qualitative and quantitative data.

Chapter 10 discusses the data presented in the previous chapters and summarises the main findings, outputs and dissemination of findings. It synthesises the main ideas explored in the thesis and summarises the new knowledge generated by this doctoral research. It summarises the implications of the doctoral research in the context of human genomics research and wider contexts. The chapter concludes with a series of recommendations informed by the learning from this doctoral research.

Chapter 11 summarises this doctoral research and its conclusions.

All research procedures reported in the thesis were approved by the relevant Ethics Committee, with relevant ethics information included in each chapter.

Figure 1.1: Thesis timeline



Chapter 2 – Research aims and activity

The research aims for this doctoral research are to:

1. understand when and how people have been involved in human genomics research to date, and identify gaps that need to be addressed with new approaches and methods for involvement
2. apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research
3. develop a standardised way of planning, reporting and evaluating involvement in order to improve future genomics research.

This thesis will explore ways of co-defining problems, co-creating solutions and co-evaluating methods of involving people. It will demonstrate practical ways of involving people in different stages of genomics research across four different communities of shared interest. The research activity undertaken to achieve these aims was as follows.

1. Review of the literature to assess methods of public involvement that may be applicable for human genomics research (or those that are already being used).
2. Review of current human genomics projects around the world, to understand the degree of involvement currently occurring, and the gaps.
3. Application of participatory action research paradigm to studies of four different groups of people associated with human genomics research, either underway or in planning.
4. Planning and conduct of the involvement activities, and where appropriate, sharing of learning experiences and description of how involving people affected or informed future genomic research.
5. Co-creation and publication of a peer-reviewed standardised framework to compare the views and perspectives of different stakeholders about genomics research across the four case studies, including views about involvement, methods of involvement, and any impacts and outcomes of the involvement.

Chapter 3 – Methodological overview

Methodological summary

This thesis used multiple paradigms and methods, including mixed methods for data analysis. This chapter is therefore divided into sections which explain the background, justification and application of each paradigm and method at the various stages of my doctoral research. While it is necessary to summarise theoretical perspectives and provide a relevant philosophical discussion of research methodology, in this thesis this discussion is embedded in the context of how it was applied and the paradigms and methods used, rather than in abstract. An evaluation of the paradigms and methods used in this doctoral research can be found in the Discussion chapter, in the section 'Evaluation of research design and limitations'.

This chapter also outlines how learning from the different stages of research influenced the methodology used in the case studies, including research design and data analysis. A chronology of the different stages of the doctoral research is summarised in the 'Thesis timeline'. The timeline shows that this doctoral research was guided by an iterative process, and improved by applying the principles of participatory research at multiple stages, including during co-design and co-analysis. Qualitative and quantitative data were gathered in order to find gaps in knowledge and patterns in data. Learning from these findings was used to inform the next stages of the research.[90] For example, findings from the narrative and scoping reviews influenced my decision to use a participatory action research paradigm to guide the case studies. While this chapter outlines the methods used in each case study, to avoid repetition more detail on the methods used for each study is contained in other chapters.

This doctoral research was informed by a 'rights-based' paradigm and used the participatory action research paradigm to guide the research process for the case studies and for STARDIT. Participatory action research (PAR) and related methodologies such as 'co-design' are explored further in the next section.

Case study methodology was used to analyse the different projects, and a cross-case analysis examined commonalities and variations in the learning from each case study. Other frameworks for reporting and assessing public involvement were used to guide data collection, which included the Public Involvement Impact Assessment Framework (PiiAF) and the GRIPP2 reporting checklist.[80,91]

This doctoral research was conducted in a public health research setting. It is intended to be practical in nature, demonstrating ways of applying and developing theoretical frameworks and evaluating them.

This research is underpinned by the World Health Organization's 'Declaration of Alma-Ata' of 1978, which states that "the people have the right and duty to participate individually and collectively in the planning and implementation of their health care".[19] It is also informed by the United Nations statements on Human Genetics and the 'Report of the IBC on updating its reflection on the Human Genome and Human Rights'.[56]

Research paradigms

Health research requires a pragmatic approach to understanding problems and creating solutions and, accordingly, a pragmatic approach has been taken in this thesis.[90] Multiple research paradigms have been used to understand and solve the problems being examined. The word 'paradigm' has its roots in words which mean 'pattern' or 'model'. In plain English, 'paradigm' means an 'approach' or 'way of thinking'. For example, the terms 'co-design', 'co-production' and 'co-evaluation' describe a set of core values and principles applied at different stages of the research cycle. The terms describes multiple methodologies with similar components including knowledge and power sharing, involving people in every stage of a process, rather describing one single discrete methodology.[89]

In research, how a researcher uses a paradigm to understand the nature of knowledge (epistemology), ignorance (agnotology), or reality itself can affect why the research is conducted, how, and what it is possible to learn from it.[90] A pragmatic approach in health research attempts to unite qualitative and quantitative approaches, which are often considered as opposites. Quantitative research is associated with objective knowledge and positivist perspectives, while qualitative research draws on subjective knowledge and an interpretivist perspective. Both attempt to map reality and both have acknowledged limitations.[92(p2)] Researchers in applied fields such as evaluation research have moved towards pragmatism as a 'paradigm of choices' in research methods, rather than being confined to any particular narrow theoretical framework. The pragmatic method simply uses both qualitative and quantitative methods, whenever appropriate.[93(p257)]

The paradigms used in this thesis – such as participatory health research and critical pedagogy – actively dismantle 'labels' and linguistic separations between 'the researcher', 'the researched' and the research itself. Such an approach requires careful explanation and evaluation of these paradigms, justification for their use, and an account of how they were used. While they can be hard to define precisely, in this thesis

public statements about ‘paradigms of involvement’ are included as a measurable indicator of involvement in STARDIT.

It is worth noting that while this doctoral research is informed by multiple paradigms, it is being applied within the context of genomics research, where there is objective data in the form of the ‘hard’ code of DNA, which is itself objective, not subjective. While understanding, interpretation and technology are constantly evolving, concepts of power sharing in this highly standardised area of research need to be explored in parallel. The creation of a standardised way of discussing involvement was therefore required in order to work within the highly standardised processes required for genomics research and analysing DNA.[28]

Rights-based paradigm

This doctoral research has been influenced by a number of rights-based paradigms, primarily a human rights-based paradigm. While this section will not explore the entire history of human rights, it will explore how current codified rights are influencing involvement in genomics research. This doctoral research is grounded in such paradigms, seeking to both act within them, uphold and promote the ideals codified in these documents.

Human rights

The United Nations describes human rights as “inherent to all human beings”[94]. The United Nations (UN) 1948 Universal Declaration Human Rights states ”all human beings are born free and equal in dignity and rights”. Significantly for genomics research and data sharing, it also states all humans should be able to “receive and impart information and ideas”.[95] The UN also states that the concept of democracy is inherently interconnected with respect for all human rights and ”fundamental freedoms are interdependent and mutually reinforcing”.[96] The World Health Organisation’s 1978 ‘Declaration of Alma-Ata’ stated ”the people have the right and duty to participate individually and collectively in the planning and implementation of their health care”[19], further connecting concepts of democracy and self-government with universal rights in healthcare implementation. The UN’s 1997 ‘Universal Declaration on the Human Genome and Human Rights’ further codified how genomics interacts with concepts about individual and collective rights, and influenced international law and ethical norms in genomics research.[97]

The United Nations has provided much guidance on working with Indigenous peoples around the world [98], and the 'Declaration on the Rights of Indigenous Peoples United Nations' was a guiding paradigm

during this research[99], including the statement “Indigenous peoples have the right to be actively involved in developing and determining health, housing and other economic and social programmes affecting them”. Similarly, both FAIR[85] and CARE[100] data sharing principles were integrated into STARDIT, in order to ensure reporting aligned with international data sharing principles.

Informed by the United Nations ‘Universal Declaration Human Rights’ statement that all humans should be able to ”receive and impart information and ideas”[95], this doctoral research was also influenced by the Open Access movement, which can be considered part of this paradigm, in particular for those who cannot afford to access genomics research behind a paywall.[101] In 2020, the United Nations Secretary-General stated ”purposes that involve data and analytics permeate virtually all aspects of our work in development, peace and security, humanitarian, and human rights”, encouraging “everyone, everywhere” to ”nurture data as a strategic asset for insight, impact and integrity – to better deliver on our mandates for people and planet”.[102] All articles which are part of this thesis have been submitted for publication in peer-reviewed journals which are Open Access, free for anyone anywhere to access.

Within the paradigm of human rights are the rights of women and children, codified in the UN’s ‘Convention on the Rights of the Child’ and the UN’s statements on gender equity and equality.[103–105] The principles of self-autonomy and individual choice in health are monitored by the UN, in particular, women having the right to decide whether to terminate pregnancies.[106] In addition, the United Nations Convention on the Rights of Persons with Disabilities is also a guiding paradigm in this thesis. The specific implications of these rights paradigms on genomics research, including using data from DNA analysis to make informed decisions about parenthood, concepts of body sovereignty and concepts of ‘healthy births’ and eugenics are explored in the Discussion section ‘Implications for public discussion debate on the concepts of yōushēng (eugenics)’.

Consumer rights or human rights?

While the connection between human rights and democracy is significant, it is important to note that human rights and concepts of ‘social democracy’ can also be contrasted with ‘consumer rights’. From one perspective, the social democratic rights-based paradigm relies on collective action to create public health initiatives, codified by the World Health Organisation (WHO), which stated that health promotion is the process of ”enabling people to increase control over, and to improve their health” .[107] Similarly, Article 4 of the UN’s Universal Declaration on the Human Genome and Human Rights states ”the human genome in its natural state shall not give rise to financial gains”.[97] Genomic science will require a renegotiation of the social contract of public health systems and involving the public in this process is a priority [32(p27)]. This was summarised in the 2016 Annual Report of the UK’s Chief Medical Officer, who

wrote, ”the importance of ethical reflection and patient engagement in the development of the coordinated national and international developments in genomic medicine should be recognised”. [32(p27)] With the current and future genomics research and services likely to be a mixture of state and private partnerships, the involvement of the public is essential to balance individual rights against the public interest.[21] Genomics and other biomedical research present specific social, ethical and legal challenges. As the communication and commercialisation of research results continues, it is essential to balance individual rights against concepts of ‘the collective good’, and data protection. The Nuffield Council on Bioethics wrote in 2012:

> ”To address these challenges, leading international institutions stress the importance of public involvement in biomedical research and innovation”.[45(p2)]

Parallel to the human-rights based paradigms (but not independent of them) is the ‘consumer rights’ paradigm, where people are involved as ‘consumers’, ‘users’, ‘tax-payers’, ‘payers’ or ‘customers’. This model is grounded in free-market paradigms, based on the axiom that the market model will create services that are needed in response to the needs of the customers.[21] The ‘consumer’ paradigm can be seen as parallel but not identical to the ‘social democratic’ idea of public involvement. The history of the use of the word ‘consumer’ goes back as far as the Latin roots of the word, with roots in a word which means ‘under’, ‘below’ or ‘sub’.[108] The origins of the word are associated with a transactional merchant relationship where the ‘consumer’ takes goods or services, and to ‘consume’ can mean to use up or waste.[108] Yet in the last 30 years the word ‘consumer’ has commonly been associated with the ‘consumer rights’ movement, in particular in the context of health services and research. A recent assessment of the influence of public involvement on health research concluded that a ”consumerist approach is still predominant and that in reality the public voice has limited impact upon the research design or upon which research gets funded”.[109]

While more recently the UK, USA, Canada and other Anglophone countries have moved away from using the word ‘consumer’ to describe a member of the public, countries such as Australia continue to use the word in Government policy. [110] For example, the term ‘consumer’ is contentious in the UK [111], and has been actively rejected by some, being labelled as an ”overtly consumerist approach to involvement associated with internal-market legislation of the 1990s Conservative administration” which is in contrast to a citizens' rights based approach. [112] A recent UK report about the ethics of data sharing referred to “mere consumers, reduced to accepting or refusing an option presented to them, or choosing between several available ‘offerings’” [7] while the Labour party in England wrote that people should not be “mere

consumers of services but genuine and active partners in designing and shaping their care and support".[8] Similarly, commentators in the United States have argued the term 'consumer' even sits below 'customer' in terms of power relations.

> "It is a mistake to assume that patient involvement in the system is "consumerism" in either of its common meanings: 1) acquisition of goods and services in ever-increasing amounts, or 2) The protection or promotion of the interests of consumers". [113]

However, current Australian Government language describes consumers as 'potential users of healthcare services'. While the current 2016 National Health and Medical Research Council 'Statement on consumer and community involvement in health and medical research' states "Collectively, 'consumers' and 'community members' may be referred to as 'the public'", the word 'consumer' is used by government, charities and advocacy organisations as a shorthand for 'the community' and 'the public' .[110]

Similarly, DNA tests which people can buy, independent of healthcare systems, are often described as 'direct-to-consumer' (DTC) by genomic researchers and governments, who seek to differentiate them from other kinds of DNA testing for either healthcare, research or use in the criminal justice system.[114] The question of regulation of these services is central to this thesis. In other words, who decides who regulates whom? As one commentator wrote, "there is a sort of a paradox between the lagging implementation in health care of the few genetic tests with proven clinical utility, on the one hand, and the speedy DTC offer of tests, with or without clinical utility".[115]

With people no longer reliant on governments or healthcare providers for access to their own genomic data and any subsequent analysis, people have new power as 'consumers'. Tritter and McCallum argued in 2006 that this shift from "patient to customer, user, or consumer has been central to neo-liberal approaches to rationalising health services, facilitated by managerialism and marketization".[116] From this perspective, true 'user involvement' or 'consumer involvement' in genomics research is presented as a feedback mechanism for consumer views, which is an essential part of markets-driven services. [116] However, without real market choice, or the enabling conditions to make an informed decision, people must rely solely on giving 'feedback' in the hope of influencing service provision. For example, if a for-profit DNA testing company informs customers they will sell customer DNA data to those doing genomics research (as is indeed the case with both Ancestry DNA and 23andMe[117]), customers have limited power in this situation, and have options which include not using the service or providing feedback. Accordingly, both the social democratic and consumerist paradigms are significant in public health genomics and

genomics research. Ensuring that research is "civically responsible"[37] requires navigating how the public can be involved in both social democratic and consumerist paradigms.

Individual rights or collective solidarity?

Concepts of 'individual rights' and 'collective solidarity' need to be explored when applying rights-based paradigms, as the very nature of genomics research transcends the individual in order to understand individuals' variations in the context of whole populations, and sometimes other species.[118]

A report from the UK's Human Genetics Commission in 2002 articulated a way of considering data sharing in genomics in a fair and ethical way. By acknowledging that individuals who choose to share data may indirectly benefit others, the public are empowered to make a balanced and informed decision:

> "nobody should feel pushed into taking part in genetic research, when they make this decision people should be aware that by taking part they might help those suffering from disease". [119(p8)]

The report articulates that genetic knowledge may bring people into a "special moral relationship with one another", and proposes the concept of "genetic solidarity and altruism, which promotes the common good". [119(p8)] Increasing 'public awareness' of genomic technology was a key recommendation of a report by the Human Genomic Strategy Group in 2012 that also looked beyond national boundaries in solidarity, citing how genomics data sharing can be used to "benefit the care of patients across the NHS and indeed the world".[120(p7)] Similarly, Genomics England stated in 2015 that the comparison of specific rare disease sets across the world could add "immeasurably to knowledge" and potentially have "world markets". [121(p11)] These concepts are simultaneously being "eroded" by language such as 'personalised medicine', which ignores the enormous collective data-sets required to allow these interventions to arise.[122] Thus, careful attention needs to be given to both the language used to describe genomics research and related medical interventions and the ethical arguments in support of involvement and participatory decision making. Any participatory action research in this area needs to include an awareness of this language and attempt to involve people in mapping preferences and existing power structures.

A 2015 systematic review of 'stakeholders' perspectives on biobank-based genomics research emphasized the importance participants of biobanks felt of being informed about data-sharing practices, measures for privacy protection and applicable sanctions if the data were misused.[123]Participants regarded reidentification as a negligible risk as the benefit of data sharing for science and society outweighed the potential risk and they were generally very supportive of data sharing. [123] Interestingly, the same review suggests that unless there is experience of or evidence of serious data breaches, overly

rigid data protection could constitute 'paternalism' over patient preferences and be described as an "unjustified obstruction to research". [123] Risks such as perceived data breaches could be reduced by involving the public, who can be involved in scrutinising data storage and access, and accessing if it aligns with perceived values of what defines a 'breach' of trust in data storage and access. Involving the public in shaping genomics research can create research which is more likely to benefit more people. [74(p6)] Emerging biotechnology should be subject to 'public ethics' and decision making which is orientated by the pursuit of the public good.[45(p22)]

Issues such as 'shared ancestry', and exactly how many generations back one means when one uses the term 'shared ancestry' are central to all of the case studies in this thesis. People from all of the populations I worked with will be affected by decisions made by others in those communities, either in communities defined by shared ancestry, cultural or other shared interests. For example, people in either the donor conceived group, or the Aboriginal community, will share significantly more DNA variations with others in that community than with people from populations such as ones defined by national borders. Similarly, potential participants of the ASPREE multi-generational research study and those from the ausEE study may discover things about their genome which may have relevance to close relatives, including offspring. It is therefore possible that people who have not given consent to participate in genomics research may still discover information about how genomic variations they might have will affect risk of disease, or that such data obtained from close relatives may be used by the criminal justice system. In summary, decisions of close relatives to participate in research can directly affect people who have not given consent, and thus concepts of individual consent needed to be re-examined in this doctoral research.

Accordingly, when applying a participatory action research paradigm, the way in which data is shared must therefore be co-designed with participants and other stakeholders from the outset of any study which collects genomic data, in order to ensure individual and collective rights are balanced, and people at greater risk of exploitation are not exploited or exposed to harm. By ensuring those being asked to participate are also invited to be involved and supported to make informed decisions, it is more likely the research will be ethically acceptable and meet the needs of those participating.[21,34,36,124]

If genomics research does this poorly, and if it makes it difficult or overly complex for people to give informed consent to participate in genomics research, those people not participating may mean humanity's understanding of genomics and disease will be poorer, and more people will likely suffer from disease. An indirect outcome from improved public involvement in genomics research may be more

people volunteering to share their data, and thus more useful data which can be used to help more people.

How the rights-based paradigm shaped this doctoral research

Concepts such as 'involvement', 'ethics' and 'democracy' cannot always be expressed fully in linguistic constructs, as they carry resonances of concepts such as equality, justice and human rights.[125] Further still, translating such descriptions between cultures and languages presents difficulties. For example, in the context of research, at the root of the word 'ethics' is not one single definition, but an entirely subjective concept, expressed in English by the words 'fairness' or 'justice'.[126] Similarly, the concept of 'involvement' in research (and associated concepts) is connected to these concepts and thus there is an array of subjective feelings about these concepts, seeking expression in a limited English lexicon. Terms such as 'consumer' and 'community member' also raise questions of 'who is involved in deciding who decides who is in each of these categories' or groupings. The need for clarity in such terminology, and ways of working across human languages to provide such standardised terminology was an objective of this doctoral research. These issues are explored in more detail, along with proposed solutions, in the section 'Standardised Data on Initiatives – STARDIT: Beta Version'.

Participatory action research

This section will describe the participatory action research paradigm, and how it was used to involve four different groups, in varying formats. As the methods used for each group varied, here the rationale for selecting various participatory action research methods will be provided.

Informed by the reviews conducted as part of my doctoral research (including the narrative review), I chose a participatory action research paradigm to guide the process with co-design and reporting informed by guidance from a number of sources.[80,91,127] Participatory action research is an umbrella term which describes several related approaches, including forms of action research which embrace a participatory philosophy and include 'co-design' and 'co-production' of research. [88] These approaches share a process whereby researchers, the public and other relevant stakeholders "work together, sharing power and responsibility from the start to the end of the project",[89] including knowledge generation and translation.[89]

An important contextual consideration in participatory action research is the concept of individuality, and the extensive laws, rights and moral codes which enshrine the protection of 'self-rule', with the biomedical model subordinate to the rights of those considered competent to make informed decisions.

[128(p58)] However, 'self-rule' or autonomy is only part of the context in which genomics research operates, and does not fully articulate concepts such as the trust one may place on experts to make decisions on our behalf [129(p9)], in what might be described as consensual heteronomy.

As discussed in the human rights paradigm section, the tension between 'private interest' and the ideal of 'public good' must be explored as part of the participatory action research process. Concepts such as 'public good' in the context of genomics research cannot be defined by an empirical collecting and collating of multiple self-interests (such as public surveys), so participatory action research provides a guiding paradigm to help navigate the complex ethical issues which must be fully explored in order to balance and prioritise the sometimes competing rights of individuals. [129(p9)] In other words, something other than individual preference is required to explore value conflicts. [129(p10)] How this is actualised in participatory action research and in the emerging and complex field of genomics research is what is being investigated as part of this doctoral research.

Guided by the participatory action research paradigm, I worked in partnership with a number of organisations and communities in order to involve people in discussions about the future of genomic research. Where possible, potential participants were included in the design of each project. The main groups I interacted with, and the results of involving them in the research process, are briefly introduced below and will be presented as case studies in the format of their submission for publication.

Defining participatory action research

Participatory research is an umbrella term which describes a number of related approaches, including [88(p1)]:

1. community-based participatory research
2. participatory action research (including critical participatory action research)
3. participatory health research
4. community-partnered participatory research
5. cooperative inquiry
6. other forms of action research embracing a participatory philosophy which may include 'co-design', 'co-production' and 'co-evaluation' of research.

The term 'participatory action research' will be used to refer to all variations of this approach, unless explicitly stated. At the core of participatory action research is critical reflexivity, a process which asks people involved to reflect on the causes of problems, any solutions and the actions that people can take to improve the current situation. [130(p11)] It is a form of collective self-reflective enquiry undertaken by

participants in order to understand their situation from a number of perspectives, including rationality and a sense of justice. [131(p153)]

In a health context, participatory action research attempts to reduce health inequalities by supporting people to be involved in data collection, reflection and, ultimately, actions to improve their own health [132]. It is an interactive process, seeking to understand and improve things through change. [132] Participatory action research integrates knowledge translation into the research process, by involving those who can inform future actions as partners in the research.

While participatory action research can be a challenge to define, the quotation below from Orlando Fals Borda summarises both the underlying philosophy and ways of implementing the participatory action research paradigm, including participatory knowledge translation[133]:

> Do not monopolise your knowledge nor impose arrogantly your techniques, but respect and combine your skills with the knowledge of the researched or grassroots communities, taking them as full partners and co-researchers. Do not trust elitist versions of history and science which respond to dominant interests, but be receptive to counter-narratives and try to recapture them. Do not depend solely on your culture to interpret facts, but recover local values, traits, beliefs, and arts for action by and with the research organisations. Do not impose your own ponderous scientific style for communicating results, but diffuse and share what you have learned together with the people, in a manner that is wholly understandable and even literary and pleasant, for science should not be necessarily a mystery nor a monopoly of experts and intellectuals.

The concept of 'dominant interests' is especially important in the context of research with Indigenous peoples around the world and the UN's recognition that their culture can be threatened by 'dominant' cultures[98], which was explored in the previous section 'Human rights'. Methods of mapping such 'interests' in a standardised way is discussed further in this chapter in the section 'Standardised Data on Initiatives – STARDIT: Beta Version'.

Defining who is participating

Defining the people and communities involved in participatory action research can be difficult. Using a definition whereby anyone who is not a professional researcher is a member of the community can blur lines which are already ill-defined. [134(p6)]

Other traditional labels can be unhelpful in the participatory model as the labels project power structures onto people. For example, 'service user' may include anyone affected directly or indirectly by a service, 'consumer' uses an economic market model to classify health and research, 'patient' places it within a medical model and 'survivor' provides are more psychiatric perspective of those 'affected'. [134(p7)]

While co-creation of a term that people would like to use to describe themselves would be best-practice, practically a definition must be arrived at from the outset of the research.

Origins and parallels

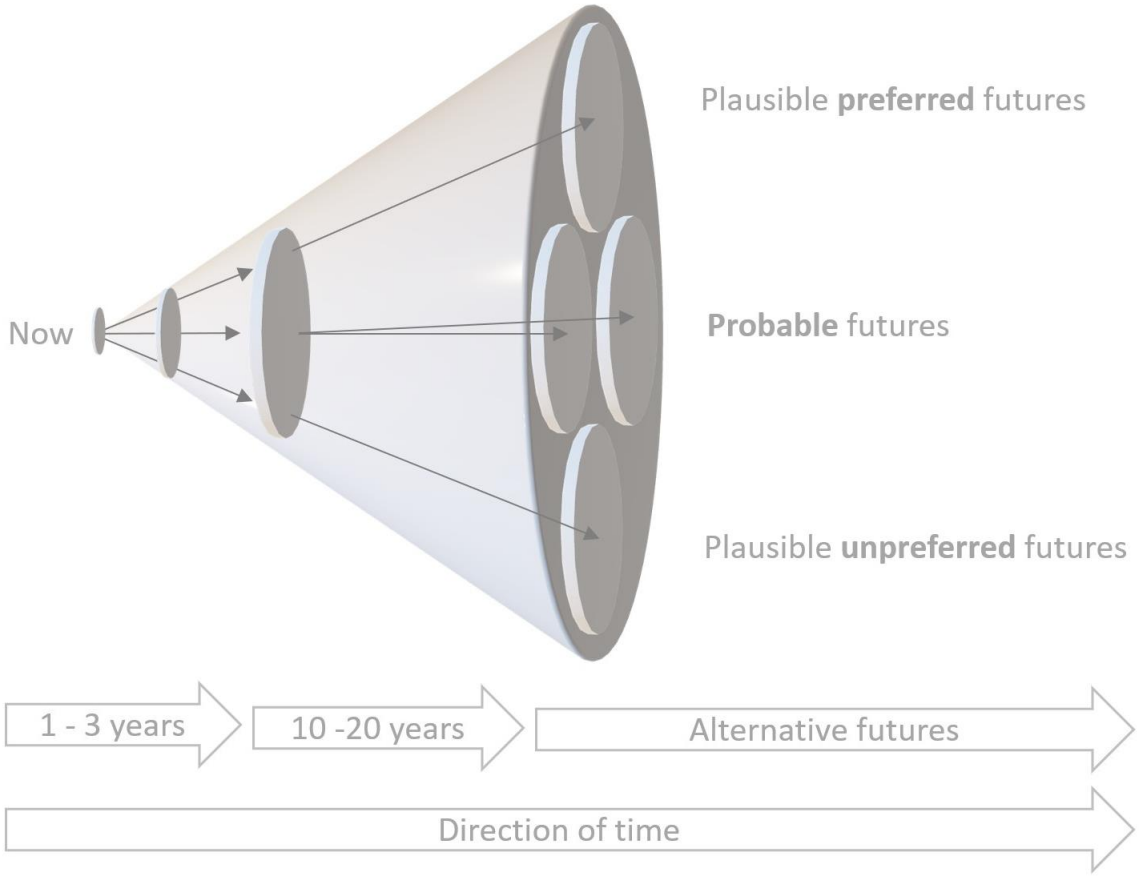
The origins of participatory action research have roots in methodological critiques of conventional research, including positivist research, which did not acknowledge power structures inherent in research and other settings. In particular, this kind of research did not acknowledge the position of relative power of the researcher in comparison to the research participant.[135] At the core of participatory action research is Paulo Friere's concept that reflection about the world requires action in order to transform it[136(p12)]:

> "to see the world not as a static reality, but as a reality in process, in transformation".

In this paradigm, reflection and action are interconnected in 'conscientisation', where critical thinking is developed through dialogue and involvement.[109] This approach is also connected to the concept of 'critical pedagogy'. Freire summarised this by stating that "reflection without action is sheer verbalism" and "action without reflection is pure activism, or action for action's sake".[132]

This perspective requires viewing the world as something changing and changeable. This was articulated in a 1993 World Health Organisation report which articulates alternative futures, dividing them into possible, plausible, probable and preferable. [137(p5)] See Figure 3.1 for a visualisation of this concept.

Figure 3.1: Alternative futures



Adapted from Bezold C, Hancock T. 'An Overview Of the Health Futures Field' for the WHO Health Futures Consultation. 1993. https://apps.who.int/iris/bitstream/handle/10665/61479/WHO_HST_93.4_eng.pdf

Such a view of preferable alternative futures immediately raises the question 'preferable for whom', challenging health researchers and policy makers to balance the varying and sometimes competing or conflicting needs and preferences of multiple stakeholders. Participatory action research offers an opportunity for multiple stakeholders to be involved, including people affected by certain health inequalities to take part in shaping this future, collectively attempting to articulate and then achieve what is preferable. This process involves learning from the past, examining potential realities, likely realities and preferred realities through an interactive process of involving communities of shared interest in appraising, shaping and affecting these potential realities. [138(p24)]

Participatory approaches in research share a number of significant connections to existing qualitative methodologies and methods, yet the constructivism at the core of participatory action research ensures that this research method reflects that there is no 'one truth' or objective answer, but that there are multiple ways to ask a question and try to improve health inequalities. [134(p2)] Involving people in health and research in this way recognises that some improvements in health can only be achieved by actively

involving people. [129(p4)] Effective public health, and thus, public health genomics, requires a range of methods beyond epidemiological methods in order to reflect the diversity of the issues it is trying to solve [139(p174)]. The question of which or whose values should direct decisions in health and research, while complex, can be navigated by participatory methods, as long as they are transparent, accountable and with clear boundaries for decision making agreed in advance. [130(p15)]

Epistemology is the study of what is known and how we know it is, yet a more recent but equally important discipline is 'agnotology', which is the study of what we don't know, and why.[140] This includes crucial concepts such as culturally induced ignorance, which are central to applied critical pedagogy and the participatory action research paradigm.

Contextual considerations

There are a number of interrelated areas which require explanation in order to give participatory action research a meaningful context in relation to this thesis. The following sections explore these important perspectives.

Positivism and scientism

The positivist forms of knowledge which can be applied to social sciences (and other natural sciences) require the researcher to aspire towards an objectivity from that which is being researched. At the core of this thinking is a reductionist philosophy that if all component parts of a system (including society) are understood, reliable predictions can be made. Critics of this reject this as 'scientism', which is the belief that true knowledge can only be gained using empiricism. [141(p29)] While this system can be helpful in some contexts and has led to many scientific advances, it reaches difficulties when trying to articulate and codify concepts such as values, emotions, lived experience of disease and ethics.

Positivism often requires people aspiring towards objectivity to then interpret, codify and analyse both quantitative and qualitative data. This often includes attempting to collect and analyse data in relation to the subjective aspects of people's lives, such as ethical values. It also requires subjective judgement on the part of the researcher in relation to research design (methods, analysis and reporting) and requires a consideration of how best to aspire towards objectivity. [130(p15)] Participatory action research differs from more traditional forms of research as the conceptual division between the researcher and the researched is blurred, removing the 'object' from research, making all stakeholders partners in the process.[132] By removing 'subjects' or 'respondents', the reductive methods of more traditional kinds of research are avoided, with the 'data' remaining in context as a record of subjective experience. The participatory action research paradigm accepts that humans cannot separate conscious experience from the concept of

external reality in order to describe an object. Human experience is where subject and object meet, with any subsequent resulting action affecting future experiences of subject and object. [132]The study of genomics adds yet another layer of complexity to this, with our individual DNA code (which exists as an objective object) in some cases affecting our individual instances of subjective, conscious experience (qualia). For example, our DNA can affect our colour perception, with some people being labelled as 'colour blind'. Subsequent interactions of our inherited DNA with our environment (including culture), or 'nature or nuture' questions, require careful articulation of who is deciding what is described as subjective or objective, and why.

The aspiration towards objectivity and detachment when applying such positivist paradigms is particularly difficult for exploring people's views and preferences about genomics research, as no one person is detached from being affected by this research, with genomics research having a quality of 'solidarity' or 'shared-ownership'.[119(p5)] In contrast, participatory action research requires no separation between subject and object, recognising that the two are combined in the experience of the individual, and enhanced by communication with others. A person's experience of group dynamics, inter-personal relationships and a sense of community all affect the relational knowledge of the individual. [130(p15)] This kind of critical participatory action research requires a 'self-critical' perspective, as the research process is not simply one of collecting data, but attempting to nurture self-reflection, recognising that the experiences of the researchers and participants are interwoven and must be acknowledged. This is particularly true for research involving autoethnographic methods, which is explored further later in this chapter in the section 'Research with family members and close relatives'.

Freire's influence on the participatory action research paradigm can also be applied to qualitative research, challenging 'interpretivist' methods, which place confidence in the value of subjective accounts of lived experience as being authentic and reliable.[142] Freire's epistemology rejects that consciousness creates a reliable and infallible copy of reality. Participatory action research would recognise that accounts of lived experience are constructed from certain power dynamics and cultural traditions. Such dynamics themselves require critical appraisal, including asking all stakeholders to actively reflect on power relations, and where appropriate, challenging or improving them. [142(p154)]

The combined relational and reflective knowledge which is created by the participatory action research process is an 'embodied knowledge' which creates an important link between knowledge and action, with the translation stage of the research cycle effectively taking place within the individual. [130(p16)] By extension, this research method also transcends the research process and 'the researched' people and

can extend into changed individual behaviours, thus into communities and the wider public. Methodological theories explaining the importance of measuring such transformations and impacts are explored in the section 'Theoretical approaches to case studies'.

Democracy

In 1969, Arnstein wrote that the 'participation of the governed in their government is, in theory, the corner-stone of democracy'[18]. Fewer than ten years later this sentiment was enshrined within the World Health Organisation's 'Declaration of Alma-Ata' which stated 'the people have the right and duty to participate individually and collectively in the planning and implementation of their health care.'[19] This was followed up by the following WHO statement in 1986[20]:

> Health promotion works through concrete and effective community action in setting priorities, making decisions, planning strategies and implementing them to achieve better health. At the heart of this process is the empowerment of communities - their ownership and control of their own endeavours and destinies. Community development draws on existing human and material resources in the community to enhance self-help and social support, and to develop flexible systems for strengthening public participation in and direction of health matters. This requires full and continuous access to information, learning opportunities for health, as well as funding support.

Participatory action research is highly dependant on the social and political context, with democracy regarded as an important enabling condition.[134(p5)] With complex (and often international) communities evolving online, an important emergent enabling condition is the free exchange of information regardless of borders[95], which in practical terms means access to technology and an uncensored internet connection. These democratic principles, when applied to internet access mean that the location of the person involved is often secondary in consideration to their ability to connect to communities of interest uninhibited by technological restrictions or censorship. For example, traditional ideas of 'citizenship' and nationality are less important to people affected by rarer diseases caused by a genomic variant than being able to connect online and form communities of interest with similar people, regardless of location. As Steven Pinker wrote, "people who have grown up with the democratization of information will not tolerate paternalistic regulations that keep them from their own genomes, and early adopters will explore how this new information can best be used to manage our health".[143] People affected by rare diseases are already creating online communities which span continents and involve those people affected in helping decide research priorities.[144,145] As participatory action research can be international, with people sharing local and national knowledge at an international level[130(p9)], this paradigm can guide

how such communities interact with more traditional research stakeholders such as government and industry, with democracy as an underlying principle guiding how participatory action research is used. However, as George Orwell noted[146]:

> In the case of a word like democracy, not only is there no agreed upon definition but the attempt to make one is resisted from all sides ... the defenders of any kind of regime claim that it is a democracy and fear that they might have to stop using the word if it were tied down to any one meaning.

Thus the task of constantly co-defining what democracy means, what it looks like, and how it works in relation to genomics research is an ongoing task, with participatory action research at the core of how the meaning of such concepts can be collectively agreed and acted upon.

Research with family members and close relatives

Participatory action research is one of the ways of over-coming potential ethical issues of close family research by attempting to address power imbalances by making sure everyone is involved in decision making at every stage of the research. The decision to work with my own family members (including my own mother) as described in the Shared Ancestry case study was a decision made after consulting with both methodological and ethical advisors.

As the concept of 'researcher objectivity' is challenged by participatory action research, having a researcher involved who 'has a stake' in the issue or is an 'insider' and is also affected may improve trust in the process among those participating. [147] In addition, as a researcher with an 'insider' status, I was more likely to be able to offer a novel interpretation of the data than someone 'outside' or unconnected to the research. While having the perspective of being an 'insider' or a person comparably affected can aid a researcher in understanding and empathising with the other research participants, the status of being an 'insider' or 'outsider' is often blurred.[135] Similarly, participants might view themselves as 'outsiders' of the research system, able to offer creative input or challenge researchers' assumptions.[148] Participatory action research also challenges the binary dichotomy of 'insider' or 'outsider' status, which is particularly relevant to genomics research in relation to concepts such as 'shared ancestry', where there is no definitive articulation of when 'shared ancestry' begins or ends, and thus must be co-created with participants.[149] In other words, all life on earth has 'shared ancestry', so where exactly do we draw the linguistic line, and who is the 'we' that gets to decide?

As some of the data analysis in this thesis involved analysing data from my own close relatives, or people I am closely related to, the study was both ethically and methodologically complex. Having a researcher involved in the project who is considered an 'insider' by research participants may also improve trust in the process among those participating.[147] Informed by autoethnographic methods, I took a number of steps to improve both the quality of the data collected and the analysis.[150] As well as keeping a reflexive research diary, study team members and research participants were involved in reviewing the analysis and commenting, as a way to increase the number of subjective interpretations of the data.[151]

The requirement of transparency in the research process helps demonstrate a practical application of the theories of Karl Popper, regarding the method of science as 'critical rationalism', which rejects empiricism and 'observationalist-inductivist' forms of knowledge, instead, asking, 'does what is observed align with other observations', and thus can a theory be 'falsified' or not.[152] Thus, there is no one single 'objective' perspective to provide 'the truth'. For example, even the analysis of data by researchers is affected by their own lived experiences.

The social constructivist methods used in the Shared Ancestry case study here provided a way of exploring and using this subjectivity to deepen understanding, incorporating the valuable data this subjective perspective provides. This perspective aspires towards a shared 'truth' rather than an abstract concept of 'objectivity' which an 'outsider' has, and an 'insider' does not. For example, the process of analysing online discussion is complex and involving research participants in analysing the data is an 'interpretive' process, relying on a social constructivist approach.[153]

Popper wrote[152]:

> "the search for truth is only possible if we speak clearly and simply and avoid unnecessary technicalities and complications".

Using STARDIT (including standardised preference mapping) introduced some clarity and consistency into the complex case study, providing a standard framework for data analysis, and thus facilitating comparison with other case studies.

The process of participatory action research

As the name suggests, participatory action research requires an action, something which is changed and then evaluated collectively. [131(p153),135(p136)] A number of practical tools exist to support researchers starting a participatory action research process, enabling them to assess whether the research can be described as 'participatory'. For example, checklists exist which ask questions such as 'is the community of interest clearly described or defined?'.[154] Guidance suggests that participatory action research must be located in the reality of daily life. [130(p9)] For example, if people are members of online communities defined by a shared interest in a rare disease, the research must take place inside these structures, in an accessible way. Participatory action research can be carried out by various stakeholders including members of the public, health professionals and academic researchers, with all members of the group having equal influence or "equitable co-governing powers". [130(p9),155(p8)] The following sections explore the stages of participatory action research considered in the case studies described in this thesis.

1: The purpose and appropriateness of participatory action research

The purpose of participatory action research is to help assess what needs to be done, take action and then evaluate any actions that take place. It aims to help participants to improve their understanding of problems and to help elucidate any potential actions they can take. [135(p135)] Participatory action research is an appropriate method when an issue or problem is complex or may involve a number of ethical issues which require resolution. If conducted correctly, it should help participants understand their own circumstance and be able to make an informed choice about any future actions. [135(p138)]

2: Design of participatory action research

It is considered best-practice to include participatory action research elements in the research design. [134(p2)] A review of participatory action research suggests the method can improve how culturally appropriate the research is to the group involved and also ensures the method is logistically realistic. [155(p8)] We involved potential participants of all three case studies where we conducted research in giving feedback on both the research design itself, and also how we planned to involve participants in the research process.

With participatory action research there are two kinds of reflection that are relevant when designing research. Firstly, personal reflection, which examines the personal assumptions, values and experiences of all stakeholders involved. Secondly is epistemological reflection, which requires a recognition of the

limits of research methods, and ultimately, science itself. [134(p14)] Such exploration of both the strengths and limitations can help ensure that all stakeholders have realistic expectations about what can be achieved by incorporating participatory action research into research design. Multiple stakeholders were involved in this process for all of the case studies described in this thesis.

3: Ethical considerations before starting participatory action research

In addition to all the ethical considerations of any kind of research, the participatory action research process has additional ethical issues to consider. Researchers must be cautious of raising expectations at the start of the research process that might not be realistic. For example, making it explicit whether the process will examine the current context and potential actions, or actually carry out an action and evaluate it. [135(p143)] An exploration of attitudes about confidentiality must also take place at the start of the research to ensure that incorrect assumptions have not been made. For example, it may be incorrect to assume participants wish to remain anonymous, as they may wish to be co-authors. Different participants might have different preferences so these must be balanced with the research design and agreed in advance. [135(p144)] By exploring such attitudes during the co-design stage of the case studies, we hoped to ensure no such incorrect assumptions had been made.

Another ethical consideration is the potential for conflict arising during the participatory action research process, stemming from differing levels of perceived status and power between stakeholders. [130(p22)] Consideration also needs to be given to those who may be intentionally or unintentionally excluded from the research. [134(p8)] While this will always remain an unknown to some extent, certain actions can be taken to reduce exclusion and create the enabling conditions for people to be involved, for example, involving people in the design of recruitment strategies.[156]

Similarly, certain skills and knowledge are required for some people to participate, for example, using online tools to communicate. Skills and knowledge, such as an understanding of the research process and working as part of a group, can often be improved through learning interventions.[134(p13)] If there is no shared language between communities of shared interest, such linguistic competencies might be considered an unrealistic barrier to overcome in some contexts, although in some cases this presents an opportunity to partner with certain communities defined by a shared language, co-creating learning resources and paying members of the community to be involved in translation of other relevant information.

When representing findings, it is important that the research process preserves the multiple perspectives and voices found within the data.[134(p19)] With the online discussions used in two of the case studies in this

thesis, this meant careful documentation of online discussions (anonymising where appropriate) in order to directly quote participants, rather than generalise or paraphrase. Accordingly, where possible, we incorporated as many direct quotations as possible into the published case studies. As there is still a risk that the overall narrative might not represent what people involved in the research felt occurred, we invited everyone involved to review the data analysis and write up, in order to ensure they felt the findings represented what was said.

Ethics approval was obtained from respective organisations before starting the research processes for each case study. This process is described in more detail in each case study.

4: During participatory action research

During the process, everyone involved in the research must reflect on how a number of different factors may enable or hinder the research process. [134(p16)] These include personal values or experiences and potentially difficult or negative reactions. For example, might some discussions 'trigger' difficult emotions? Relationships within the group also need to be considered, including ensuring participants feel safe in participating in the research study. We co-created group agreements in both of the online discussions in order to ensure participants had an opportunity to shape decisions on any behavioural boundaries during discussions.

For people involved in participatory action research who are not health professionals, the process also involves critical health literacy. This is distinct from functional or interactive health literacy as it describes how people can act together with others to improve any factors which affect the health of the group as a whole. [130(p12)] As a result, initial questions in the group discussion of the communities of interest acknowledged power structures, such as universities involved in the research. Discussions then attempted to map who currently has the power why they do and people's views about how it might impact on research. For example, such discussions were used to guide the research design for the Shared Ancestry case study, ensuring that potential participants were happy with the choice of the facilitator who would be used to facilitate the discussions.

Facilitation is a vital role in the participatory action research process and the person who is an intermediary is an egalitarian, attempting to promote equal dialogue, while aspiring to achieve specific goals or outcomes.[130(p16)] Involving impartial or neutral facilitators for some discussions may reduce undue influence on discussions, as leadership requires facilitating shared-decision making at every stage of the research. Facilitation needs to create the enabling conditions for good communication, including respecting everyone's individual dignity and privacy. This ensures that people feel they can trust the people involved and the process. [134(p6)] The non-professional researchers involved in participatory action research may perceive the research and the process differently as the research progresses. Anxiety or

distrust may change to a feeling of self-confidence and of belonging, if the enabling conditions are created. [134(p13)]

As facilitation is also a process that requires self-critical reflection, we created a private online space for the facilitators from both case studies which used online discussion, in order to share experiences and reflections, and offer support to one another.

5: Participatory data analysis – no consensus required

The participatory action research process does not require consensus, it is the process of uncovering and examining different perspectives. Knowledge is created in the communicative spaces, created and facilitated by the participatory action research process. The concept of productive conflicts followed by useful negotiation is helpful, as it assumes there will not be homogenous perspectives yet also presumes the process for managing conflict will be sufficiently robust to result in useful negotiated outcomes [155(p2)]. Involving participants in co-creating boundaries and agreeing upon processes for such discussion and ensures that the discussion is aligned with their own expectations and values. If the process is well designed, it will engender trust through encouraging shared behaviours which are aligned with the universal human values of dignity and respect. [130(p16)]

This process also allows an exploration of what is known, what we know is unknown, agnotological exploration (a study of culturally induced ignorance) and an agnoiological exploration (that of which we will always be ignorant). By exploring the limits of both the positivist method and our own knowledge, this mapping can produce a helpful framework within which to focus discussion and action. With everyone in the role of 'co-researcher', collective learning transforms into a process whereby people can act based on research findings and have an impact beyond a traditional definition of the scientific community. [130(p17)] In this sense 'co-researchers' can then move from a stage of co-designing to co-implementing solutions. [157]

6: Sharing findings from participatory action research

The process of articulating and sharing findings should be participatory, with issues such as authorship discussed transparently at the start of the research. [135(p144)] Accordingly, revisions to the ethics application were made in order to incorporate feedback from potential participants as part of the co-design process. This included improving plain English summaries of how data from the planned research would be analysed, shared, and how participants could be involved in making sure it aligned with their expectations. Crucially, the process of involving participants in checking the information being shared provided an extra stage of reassurance that no data was being shared that participants did not want

shared. This was of particular importance to participants from the ausEE case study who might have insurance policies affected by any data breaches, or participants from the Shared Ancestry study who could be inadvertently identified in the public domain.

7: Defining and measuring impact in participatory action research

Defining and measuring impact from involving people in research is challenging, as outcomes can be both short term and long term, and the tasks people were involved in can vary from person to person. [130(p17)] A recent review of participatory action research suggested that positive outcomes of participatory action research can include ensuring culturally and logistically appropriate research, enhance recruitment capacity, generate professional capacity and competence in stakeholder groups, increase the quality of outputs and outcomes over time, increase the sustainability of project goals beyond funded time frames and during gaps in external funding, and create system changes and new unanticipated projects and activities.[155(p2)] The examples of negative outcomes identified by the review illustrated why these outcomes were not a guaranteed product of participatory action research, but rather were contingent on key aspects of context. The section 'Theoretical approaches to case studies' explores this in more detail, including why we attempted to co-define and measure impacts from the participatory action research process.

Research methods

Reviews

As part of this doctoral research, I conducted several reviews and published a summary of my research into various systematic review methods in the WikiJournal of Medicine under the title, 'What are systematic reviews?'.[17] The methods used for these reviews are summarised in Table 3.1. Further detail about each review, the method used and any learnings, can be found in the section 'Involvement in genomics: Mapping what is known and unknown'.

Table 3.1: Summary of review methods

Review title	Method description
How are the public involved in health research and what are the impacts? A narrative review.	This 'systematic narrative review' summarised five systematic reviews identified in the area of public involvement in research. Data extracted from the reviews included the kind of involvement taking place (the tasks), how it was done (the method), at which stage of the research cycle it occurred, and any impact on the research that might have occurred.
Public involvement in global genomics research.	Using a list of human genomics research projects from an existing database hosted by the Global Alliance for Genomics and Health, I conducted a scoping review (assisted by co-authors) in which I systematically searched public domain websites for information reported on involving the public in this research.
Guidance for planning, reporting and evaluating initiatives.	A multidisciplinary scoping review, the purpose of which was to show the considerable variation in guidance for reporting, planning and evaluating initiatives.

Case studies

This section summarises how the case study method was used to describe the four case studies in this thesis, including a cross-case analysis. It summarises the literature on case study theories and methodology, examines types of case study, and discusses how to ensure quality and rigour in case study method. It describes the principles underpinning the choice of the case study methods used in this doctoral research and, for each of the four case studies, details variations in the methods used. This section also explores case study selection criteria and summarises rejected case studies. The data sources for each project are described, including scope, exclusions and methods of analysis.

Case study methodologies

Introduction

Case studies can be used to describe or explain phenomena and the context in which they occur.[158] Case study methods are used in many disciplines, including anthropology, biography and ethnography.[153] Ethnography is a way of recording human events that combines deliberate observation, interpretation and analysis.[159] Case studies can be designed to suit each individual case and research question, allowing more flexibility than approaches such as grounded theory.[153,160]

What is the use and purpose of case studies?

Case studies are useful for generalisation or to challenge generalisations, that is, the purpose of a case study is not always to produce outcomes that are generalisable.[153] Studying exemplary cases increases understanding in any discipline.[161] While it is possible to generalise from one case, consideration needs to be given to the power of the example. If it is a 'black swan' – an exceptional case that challenges a whole theory (for example, 'there are no white swans') – a single case can have significant importance when generalising beyond the single case.[161] Using empirical falsification, it is possible to identify potential 'black swans' and develop a case study which falsifies the proposition that white swans do not exist.[162(p66)] However, falsification is limited as a method as it is not always practical or possible to provide evidence that some things are false.[162(p66)] Strategic selection of case studies may be required in order to advance knowledge in a certain area. Case studies are, therefore, helpful for context-dependant knowledge, rather than for creating predictive theories or universal generalisations.[161]

Types of case study

The diversity of case study methods is reflected in the multiple ways they are described and categorised. Conducting a case study employs multiple methods and theoretical frameworks that attempt to capture the complexity of the case being explored.[153,163(p65)] The explorative and, often, interdisciplinary nature of

case studies requires creativity and innovation, and this means that the multiple methods will constantly evolve.[153]

In 1995, Stake defined three types of case study: 'intrinsic', 'instrumental' and 'collective'.[163(p4)] The evolution in case study methods since then is indicated by the addition of several subcategories of case studies, defined more recently by Hyett in 2014. Hyett's types include 'illustrative', 'exploratory', 'cumulative', 'critical instance', 'explanatory', and 'reflective practice'.[153] These case study types are summarised in Table 3.2.

Table 3.2: Case study types

Case study type	Summary
Intrinsic	**An intrinsic case study** can be used to explore the unique phenomenon of a case as distinct from others. Some cases are chosen as case studies because the researcher recognises that they have an intrinsic interest that might not be generalisable.[163(p3)]
Instrumental	**An instrumental case study** uses a particular case to explore an issue or phenomenon in more detail by examining the wider context in greater depth. An unusual case often works better than one regarded as typical as it is more likely to provide novel data. The purpose of an instrumental study is to understand the particular case, but it can also attempt to provide data that could produce a valid modification of a generalisation by using inferences.[163(p109)] This method requires knowledge of the individual case which can then be applied to knowledge of others.
Collective	**A collective case study** involves exploring multiple cases in a sequence or at the same time. It provides more context than is possible for one case alone.
Illustrative	**An illustrative case study** provides an opportunity to elucidate a particular area of enquiry and seeks to provide a common language about that area.
Exploratory	**Exploratory case studies** can be used to help identify questions and outcome measures prior to a larger investigation.
Cumulative	**Cumulative case studies** attempt to combine data from several cases at different points in time. They provide an opportunity to generalise without needing to repeat studies.
Critical instance	**A critical instance study** examines a case which has unique interest, often with no intention of generalising any findings. It is a useful method for exploring causal relationships.
Explanatory	**Explanatory case studies** can use both qualitative and quantitative research methods to explore and describe phenomena, explain causal relationships and develop theory.[164] Findings from explanatory case studies can be compared for cross-case analyses.[165]
Reflective practice	**A reflective practice case study** (or 'reflexive case study') is a post-event analysis that reports experiences, insights, debates and discussion.[166] It can be used to inform others who are encountering similar situations. The method and structure of reflective case studies is informed by the 'reflection cycle'.[167]

Ensuring quality

As seeking methodological guidance is advised when developing case study methods and theoretical frameworks, several experts were consulted as part of the research design for this thesis.[153]

In general, a high-quality case study must clearly describe the study design and justify methodological decisions.[153] However, in a 2013 critical review, Hyett identified that few case studies described the study design in sufficient detail.[153]

Improving the scientific rigour of case studies can be achieved by:[153,158]

1. using theoretical sampling (using certain frameworks for certain types of analysis)
2. asking participants to validate data (involving them in checking analysis)
3. ensuring transparency throughout the process (describing steps in detail and justifying them)
4. reporting details of researchers' relationships with cases, and how these might influence the research process.

A well-conducted case study does not contain intrinsic bias. In fact, rigorous case study methodology usually ensures the falsification of more preconceived concepts than other methods available to researchers.[161]

Theoretical approaches to case studies

While both qualitative and quantitative data are both valid in case study methods,[161] in this thesis some epistemological approaches regarding how and why data were gathered for the case studies require rigorous justification.

A range of theoretical approaches are summarised in Table 3.3.[153,158] Some methods overlap, but this is not shown in the table. However, where possible, related approaches are shown adjacent to each other.

Owing to the often imprecise language used to describe these theoretical approaches, I have used the structured data Wikidata categories, which are associated with each approach. Wikidata is used in STARDIT reports. In some cases, I have created, amended, or improved the Wikidata item.[2]

[2] For transparency, I have used my Wikimedia username 'JackNunn' for all such edits. The Wikidata categories have been formatted as hyperlinks to the Wikidata item, as they are not in themselves references but links to structured data. In some cases, I have edited the Wikidata online entry.

Table 3.3: Summary of theoretical approaches

Theoretical approach	Summary	Wikidata entry
Interpretive	• Attempts to understand meaning from different perspectives. • Attempts to build theories based on these different perspectives.	Q2779065
Positivism	• Establishes variables in advance. • Attempts to spot if they fit with an expected pattern or theory. • Refines and tests the theory in relation to data. **Limitation:** Meets difficulties when trying to articulate and codify concepts such as values, emotions or lived experience of disease and ethics.	Q131015
Critical	• Questions any assumptions the researcher might have. • Includes context such as political or social context. • Interprets how power and control potentially affect behaviour. **Limitation**: Focusing only on power relations may mean other factors are not fully explored.	Q301751
Post-positivism	• Holds that a researcher's knowledge, experience, values, theories and hypotheses may influence what is observed and how it is observed.[168] • Considers both quantitative and qualitive data are useful, if critically appraised for bias. • Attempts to build theory from case studies.[169(p533)]	Q2371887
Constructivist	Considers that: • in the context of learning (or 'education'), learners construct new understandings and knowledge by integrating them with existing knowledge and experience. • a well-run discussion facilitated by a neutral facilitator (using a number of methods and modes) is central to exploring knowledge and values and co-creating new knowledge or preferences.	Q207103
Cognitive constructivists	• Views the process of knowledge or experience as being influenced by social transaction – as something shared, which allows participants to actively reorganise elements of their knowledge (for example, as part of an online interaction).	

Theoretical approach	Summary	Wikidata entry
Social constructivism	• Views the data in a way in which meaning is negotiated through the co-construction of knowledge in the discourse. • The researcher has personal interaction with the case.[170]	Q1135710
Online research methods (ORMs)	• Describes multiple ways that researchers can collect data via the internet, including online focus groups, online interviews, online qualitative research and cyber-ethnography (explained below).	Q6035951
Online-ethnography (or cyber-ethnography)	• Adapts ethnographic methods to the study of the communities and cultures created through computer-mediated social interactions.	Q1816310

Theoretical approach used for case studies

Using more than one theoretical approach is preferable in health research.[158] I adopted a combination of critical, interpretive and post-positivist approaches to analyse the case studies in order to provide richer and more diverse data than using any single method alone. Accordingly, I used two theoretical approaches when planning and conducting the case studies: social constructivist and post-positivist. These are summarised and appraised in more detail in this section.

The post-positivist approach outlined by Eisenhardt is an attempt to build theory from case studies.[169(p533)] In recognition of how a researcher may affect the case study (the observer paradox), I worked with case study teams and participants to develop clear protocols aimed at reducing observer paradox, that is, how a researcher may affect the case study. This post-positivist approach involved triangulation of multiple data sources from both the study team and participants, who were also involved in analysing and comparing data.[153] To measure the outcomes of involvement, we included in the data collection researchers' subjective accounts of what they learned throughout the process of involving people as this represents a valid way of measuring outcomes from involvement.[171]

Combining the methods described above into a single case study method bridged the theoretical paradigms of post-positivism and constructivism.[153] The philosophy underlying this 'bridge' asserts that, if reality is assumed to be objective, it must still be interpreted by people (subjects) who are subjective beings. [139,172,173] Constructivism describes how people sometimes called 'research subjects' work together with researchers to collaboratively construct meaning from data.

As our own direct experience of reality is inescapable, tools such as language (including that used in data analysis) offer ways to attempt to escape subjective experience and arrive at a collective experience.[174(p139)] Through acknowledging the subjective nature of all enquiry, and attempting to justify methods and overcome or label bias where it might occur, the subjectivity of the case-study method used in this doctoral thesis is defensible.[161]

In addition, in large organisations or other complex power structures (as described in the ASPREE study), critical realism offers a useful framework for analysing the interactions between people's internal and external realities and the observer of those realities. Critical realism can be summarised as a way to explore causally relevant levels of reality, using multiple perspective to explore interactions case by case.[175] A combination of both an interpretive and post-positivist approach therefore provided richer and more diverse data than using one of those methods alone. Using online research methods enabled me to adapt traditional discussion methods and co-create the methods described in this thesis to suit case study participants. Working with potential participants to co-design the study enabled a melding of objective and subjective data and using rigorous, transparent methods improved both the validity and richness of the data.[153] Use of similar coding methods (including the STARDIT-PM categories) in the two online studies also improved the replicability and validity of the data.[176]

Use of asynchronous text-based discussions in the online discussions also aligned with the participatory action research (PAR) paradigm, as it is more inclusive than other approaches. It also supports people's different cognitive needs or preferences by providing access to information and discussion in a more accessible format.[177] This theoretical approach also creates the enabling conditions for everyone to be heard equally, in contrast to synchronous or face-to-face discussion that might be dominated by certain people. It is more inclusive because it is flexible and does not exclude people who, for example, might work full time or have caring responsibilities.

The reflective practice case study of Chapter 8 'Involving Australian Indigenous peoples in precision medicine' is distinct from the other three case studies, in that it is a post-event analysis that reports experiences, insights, debates and discussion from my individual perspective (checked by another researcher involved for validity).[166] The method and structure of reflective case studies is informed by the 'reflection cycle', and is explained in more detail in the methods section of Chapter 8.[167]

Case selection

Selection of cases for this thesis was informed by a number of factors, which were appraised using the following questions.[178]

1. Was it a population affected by genomics research, as distinct from the general public?
2. Was it pragmatic, that is, was it possible to establish a mutually trusting and effective relationship within the time and resources available to the research project?
3. Was the power dynamic equal and not exploitative (would the research offer participants something rather than just being passive subjects)?
4. Were there conflicting or competing interests which could negatively affect the research?
5. Was the proposed case study ethical, (including would the research be a burden on the populations' or partner organisations' capacities)?

Cases studies can be defined as 'typical' or 'deviant', 'influential' or 'extreme', but these are one-dimensional labels that rely on the perspective of a single labeller and thus may change according to an individual's perspective.[178] To facilitate an understanding of cross-case characteristics, Table 3.4 explores and labels the characteristics of the case studies in this thesis from multiple perspectives. Using data available to the study team, each column explores a different feature of each case study. To make comparison with other cases more useful, the 'population label' groups the cases into a larger population grouping. For example, a population affected by a rare disease may be grouped with other rare diseases.

The 'representative features' column explores features of the case that may be *similar* to others in the same population label, for example, with respect to equality of access to treatment. As deviance may affect data utility or generalisability, 'deviant features' explores how a case study may *differ* from others within the same population label. 'Methods of involvement used' outlines the participatory methods used to involve people during the case study. 'Generalisable data' explores what data from the case study may be able to be applied to other populations within the defined population label. Lastly, the 'Usage' column refers to how all data from the case study may be used.

Case study	Population label	Representative features	Deviant features	Methods of involvement used	Size	Generalisable data	Usage
a. ASPREE	'Typical' cohort study with associated biobank.	Large government funded cohort study, with biobanking and international data sharing practices.	Only healthy people over 70 years. Study team using co-design methods to explore acceptability of future studies.	Representative on study team, telephone interviews, face-to-face events, co-authors on paper.	59	Learning from methods of involvement and reported impacts.	Confirm or disconfirm theories about methods of involvement. Explore likelihood of any causal mechanisms
b. People affected by Eosinophilic Gastrointest-inal Disorders (ausEE)	'Typical' community of shared interest defined by being affected by a rare disease.	Charity started by person affected, offering information and support with an established online community.	Charity had attempted to involve people in prioritising research decisions prior to participatory action research.	Representatives involved in co-design, online surveys and online discussions, commented on paper.	26		
c. Shared Ancestry	'Typical' group of people who are donor conceived half-siblings.	Most people in the group made the discovery after buying direct-to-consumer genetic tests and making contact via online communities.	Group is exceptionally large. A member of the study team discovered that their mother was part of the group while researching potential case studies.	Representatives involved in co-design, online surveys and online discussions, commented on paper.	18		

d. Indigenous Precision Medicine Initiative	'Typical' group of people from a remote Aboriginal community.	The Aboriginal community is remote and experiences multiple forms of inequity and poor health outcomes.	Members of the Aboriginal community have positive experiences of genetic medicine and have previously been involved in another co-designed project.	Representatives involved in co-design, face-to-face events, surveys.	N/A*		

Table 3.4: Selected case study characteristics

*the co-design process is ongoing and therefore the numbers of people involved are not quantifiable at this stage of the research

Unselected case studies

Several case studies were considered for this doctoral research project. Appraisal of potential partnerships can be considered a proto-stage of co-design. Following such appraisal, it was decided not to partner with certain organisations in the co-design of research projects. So that candid and accurate explanation for these decisions can be provided, these organisations are not identified. The complexities of establishing a partnership relationship with an organisation, before discussion of project co-design can begin, are illustrated below.

An Australian-based cystic fibrosis charity

Exploratory meetings considered hosting online discussions for community members, particularly regarding prioritising drugs for commissioning. To inform the process, the study team also consulted an independent person affected by cystic fibrosis outside of the formal meetings with the charity. Internal reorganisation within the charity meant that the timing of the partnership would not be mutually suitable. In addition, pharmaceutical company funding of the organisation placed study team members with links to Cochrane in a potentially difficult position. Due to these problems in timing and ethics, this case study was rejected.

An Australian-based comprehensive cancer centre

Comprehensive cancer centres work to integrate genomics into care pathways for patients. Following an invitation to present at the Centre, a member of staff at the Centre approached me and suggested a potential partnership. After several meetings, the study team decided that it was too early in the project to involve people in the way discussed. However, as an independent consultant, I developed a training session for the Centre to help people (including cancer patients affected by rare cancers) understand how they could get involved with the work of the Centre. Project staff continued to share information about the project and relevant learning with the Centre. Staff changes at the organisation delayed further discussion about partnerships.

At the end of this doctoral research project (September 2020), I was approached by the Victorian Comprehensive Cancer Centre and worked with them (in my capacity as Director of Science for All) on a project to support public involvement in precision oncology and scope how people affected by cancer and the wider public can be involved in personalised cancer care.[179] While not a part of my doctoral research, learning from the doctoral research was applied to this work, and a STARDIT report in the form of a protocol was produced describing proposed plans for involving the public in the work of the cancer centre. While permission was given to share an outline of the work conducted on the Science for All website, owing to a confidentiality agreement, the majority of this work remains unpublished at the time of writing (November 2021).

An Australian-based cancer centre research cohort

Meetings were held with staff working on a cancer research project with a focus on clinical application of research. The project had links with several other Australian-based genomics research organisations. The meetings explored ways of working together. Accountability for involving patients and other stakeholders in the research was emerging, but is as yet unclear. Staff changes during the project set up phase complicated communications and the case study was rejected on the grounds of timing.

A rare cancer charity with organisations in Australia, UK and USA

Face-to-face meetings held in Australia and the UK explored potential ways of working together. People affected by the rare cancer had formed three aligned patient organisations in Australia, USA and the UK and were funding their own researcher. The study team explored ways of working with the group, but concluded that the organisation was seeking a way to manage communications and decision making about, but not limited to, research within the organisation. Any discussion hosted by the study team would have had to encompass the full scope of the organisations' concerns, which would have been out of scope of this research project. While it was a promising project, it was rejected owing to complexity and timing. To inform the design of other projects, the study team stayed in touch with staff from these organisations and shared information and learning about best-practice.

Selected case studies

Table 3.5 provides an overview of the case studies selected for this research.

Table 3.5: Overview of selected case studies

Case study cohort	Summary	Method	Data sources	
a. Healthy people over 70 (ASPREE)	The ASPirin in Reducing Events in the Elderly (ASPREE) study is a prominent longitudinal primary prevention trial involving a large research cohort in Australia (>16,000 elderly Australians).	Working in partnership with staff and participant advisors, we involved people in the planning of a possible future multiple-generational cohort study, encompassing genomics and other types of research. The work culminated in a face-to-face event, where I facilitated a session exploring people's views and perspectives about involvement in genomic research.	1. Diary 2. ASPREE Newsletter 3. Meeting records 4. Email discussions 5. Reports on progress 6. Interview participant initial feedback 7. Meeting about interviews 8. Interview data 9. Interview summary 10. Meeting about event.	11. Interview 12. Event planning feedback 13. Pre and post event information and questions 14. Facilitation plan and relevant reflections 15. Event recording 16. Participant feedback about event 17. Email summaries of event 18. Notes from event 19. Meeting notes.
b. People affected by Eosinophilic Gastrointestinal Disorders (ausEE)	The charity ausEE works to improve the lives of people affected by rare eosinophilic disorders.	Working in partnership with representatives from the organisation, an online discussion forum was created to explore views and perspectives about genomic research of people affected by rare immunological eosinophilic gastrointestinal	1. Diary 2. Emails and meeting notes 3. Online pre-discussion survey 4. Learning resources for participants and facilitators 5. Online discussion with participants	

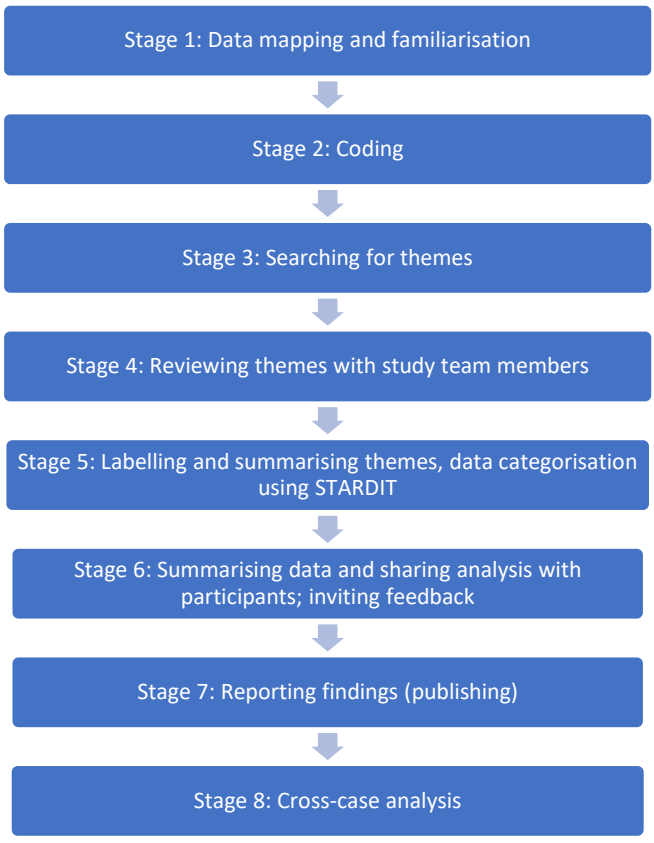
Case study cohort	Summary	Method	Data sources
		disorders. Online discussions were co-facilitated and moderated by Jack Nunn and a representative from the organisation, Kylie Gwynne, who is also a co-investigator.	6. Online discussion with facilitators 7. Online post-discussion survey 8. Follow up survey for facilitators.
c. Donor-conceived siblings (Shared Ancestry)	A group of related family members who share the same ancestor (a common sperm donor) were invited to participate in the project.	As a group of (an estimated) >500 half-siblings around the world who share a common father, the group presents a unique opportunity for human genomic research. Potential participants were involved in designing the study and an expert facilitator (not a relative of any member of the group) led and moderated discussions.	1. Diary 2. Emails and meeting notes 3. Online pre-discussion survey 4. Learning resources for participants and facilitators 5. Online discussion with participants 6. Online discussion with facilitators 7. Online post-discussion survey 8. Follow up survey for facilitators.
d. Indigenous Precision Medicine Initiative	The Poche Centre for Indigenous Health is leading the 'Indigenous Precision Medicine Initiative'. The Centre plans to work with communities to co-create research protocols for creating reference genomes to improve clinical translation of genomics research.	This project will work in partnership by observing and assessing the process and standards set for the other case studies that form part of this doctoral research – including reporting planned activity as a STARDIT report.	1. Meeting notes (including notes from face-to-face meetings with community members, with identifying information not recorded) 2. Emails with study team 3. Comments on shared documents.

Data summary and analysis

The case study design used in this thesis was informed by international best practice, including use of standardised case study reporting to improve both quality and rigour.[180,181]

In addition to quantitative analysis, qualitative data from each case study were analysed thematically in a number of stages including data mapping and familiarisation; transcription; manual coding; searching for themes; reviewing themes with team members; labelling and summarising themes; and reporting the findings.[182] An alpha version of the STARDIT tool was developed and used in parallel with the thematic analysis to organise data into pre-defined 'super-categories'. Using the STARDIT reporting tool, this allowed consistent comparisons with other case studies and data to be made. Several other frameworks for reporting involvement in research also informed data collection and analysis.[21,91,183–185]. Figure 3.2 summarises the stages of data analysis.

Figure 3.2: Stages of data analysis



Quantitative cross-case analysis

During this thesis, I developed a standardised way of planning, reporting and evaluating stakeholder involvement in order to improve future genomics research using standardised data (STARDIT). This included mapping the preferences of different stakeholders using standardised preference mapping (STARDIT-PM), applied to multiple data sources including online discussions, survey responses and meeting notes. The thematic analysis of the 'super-categories' mentioned above allowed the generation of quantitative data, for example, how many participants from each study shared views about a specific data category. This data is summarised in Table 9.2.

Baseline and follow-up preference mapping

In addition to the 'super-categories', baseline and follow-up data was collected from some case studies (AusEE and Shared Ancestry) about specific preferences relating to the research cycle, and who should be involved in which tasks. In order to map changes in preferences about involvement in research, participants were presented with an identical question at the start and the end of the research process. Data came from the question '**Which aspects of any future research genomics research should be influenced by the following?'**. Participants answered using a tick-box grid to indicate which aspects of research they felt should be influenced by which stakeholder groups. Figure 3.2.0 shows this grid. This grid allowed the creation of quantitative data and analysis of participants preferences, and if and how they changed between the start and the end of the studies.

Figure 3.2.0: Preference mapping grid

	Everyone (any member of the public who is interested)	Anyone who might be indirectly affected by the research	Only people who are directly affected by the research	Only people who are participating in the research	Only people with a professional role in research
0. All aspects mentioned below (leave others blank if ticking this)	○	○	○	○	○
1. Finding questions to ask (identifying research topics)	○	○	○	○	○
2. Deciding which questions to prioritize and fund	○	○	○	○	○
3. Deciding how to try and answer the question (the research method)	○	○	○	○	○
4. Attempting to answer the question (carrying out the research, including collecting information)	○	○	○	○	○
5. Trying to understand if it is possible to answer the question (analyzing the information)	○	○	○	○	○
6. Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)	○	○	○	○	○
7. Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)	○	○	○	○	○
8. Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)	○	○	○	○	○
9. Designing how people are involved in the research	○	○	○	○	○

Involvement in genomics: Mapping what is known and unknown

This section summarises the reviews which have been conducted as part of this doctoral research, as summarised in Table 3.6. In the following sections, I summarise a number of mixed-methods reviews that I conducted in an attempt to map both the knowledge and gaps in knowledge which exist around the questions of this thesis. Abstracts for the narrative review of public involvement and the scoping review of reporting frameworks and guidelines are included in the body of the thesis. The full reviews are presented in the Appendices. This allows for the scoping review of involvement in genomics research to be included in full as 'Chapter 4 – Scoping review of involvement in human genomics research'. Table 3.1 above provides a summary of all the review methods, while Table 3.6 below summarises the learning from each review.

Who knows?

Mapping both what is known and unknown is important for policymaking and science evaluation.[186] Angiology is the study of ignorance of that which we can never know (what is unknowable), while agnotology is the study of what we do not know, and why in the realm of what is knowable.[140] The latter can be thought of as the social construction of ignorance. The best documented example is of people working for the tobacco industry who attempted to confuse a correlation between smoking and cancer by funding research into alternative causes, paying experts to promote smoking and producing pro-smoking 'propaganda'.[187] Combined with systemic under-funding of research into causation, the World Health Organization projects that deaths from smoking will reach 10 million annually by 2030,[188] partly caused by deliberate attempts to induce cultural ignorance about the risks.

The study of culturally-induced ignorance is absolutely central to genomics research. For example, some observers have commented that "the world has placed excessive emphasis on the study of numerically and economically dominant European and Asian populations", [189] with these populations over-represented in current databases of human genomes. This makes genomic medicine less useful for people from population groupings not represented in current databases, as any variations of known or unknown significance in these populations are not as well understood. The impact of this on Indigenous peoples is explored in Chapter 8, and in the paper I co-authored titled 'Equitable expanded carrier screening needs Indigenous clinical and population genomic data'.[5]

The methods used for these reviews are informed by the 'Routledge International Handbook of Ignorance Studies'.[140] They attempt to map what is known about involvement in genomics research, what is not known, and why. Connected to these concepts is the 'right to ignorance', whereby some people choose not to know certain things about their own genome. Such preferences can influence the design of genomics research, including data sharing protocols. This is explored further in the case studies in Chapters 5 and 7.

What do we know about people's attitudes to genomics research?

Extensive research about people's attitudes towards participation in genomics research has been conducted. However, it remains the case that limited research has been done on people's involvement in genomics research and any impacts of such involvement.

A 2015 systematic review of stakeholders' perspectives on biobank-based genomic research emphasised the importance to biobank participants of being informed about data-sharing practices, measures for privacy protection, and applicable sanctions if the data were misused.[123] Participants were generally very supportive of data sharing and considered that the benefits for science and society outweighed the potential risk of re-identification.[123] This suggests that, unless there is evidence of serious data breaches, overly rigid data protection could constitute "paternalism" over patient preferences and be described as an "unjustified obstruction to research".[123]

The UK's 100,000 Genomes Project recognised that many people who joined as project participants did so because their participation would "benefit generations to come".[121(p3)] A 2014 study noted that this project can be viewed as potentially benefiting "anyone, anywhere". Participants do not expect to benefit personally, and there is no sense that they are trading-off donation of data in exchange for direct benefits for themselves or their families.[190(p6)]

In a 2016 study, 80 per cent of people who had shared their genomic data said they did so in order to contribute to the advancement of medical research.[191(p1)] In a 2020 global survey, most people viewed DNA data as different from other medical data. Notwithstanding, people were most willing to donate DNA data when the recipient was specified as a 'medical doctor'. They were least willing when the recipient was a 'for-profit researcher'.[67] Similar beliefs about participating in research in order to benefit society as a whole have been reported in a number of other countries, including the USA and Australia.[192–194]

While many surveys and reviews have focused on people's perspectives about participating in genomics research, there is very limited information about their preferences with respect to how

they would like to be involved, that is, how they would prefer to share power in decision making, planning and analysis. For example, little is known about how people would like to influence decisions on data access or how they would prefer this to be achieved in practical terms. Such gaps in our knowledge have serious implications, in particular, for populations at greater risk of exploitation, such as Indigenous peoples whose culturally-specific preferences for involvement may not have been integrated into the planning of large genomics research projects. These populations are thus less likely to be involved in critical decisions about data sharing.

Table 3.6: Summary of learning from the reviews

Review title	Learning points
How are the public involved in health research, and what are the impacts? A narrative review (see Appendices)	1. Language to describe involvement is varied and sometimes inconsistent. Standardised reporting would be advantageous. 2. Involving people in health research generally has beneficial outcomes for research and for health service planning. 3. Involving people as early as possible is considered best practice. 4. A number of high-impact actions can be taken that do not necessarily require considerable budgets or time. 5. Face-to-face involvement appears to involve greater cost in relation to impact, compared with other forms of involvement, including online methods. 6. Involving people in planning how the public will be involved is crucial. 7. Other than generic positive impacts, the most often reported impacts were improving the relevance of the research, improving dissemination of findings, and improving data collection.
Public Involvement in Global Genomics Research: A Scoping Review[28]	8. The methods to involve people reported by most initiatives were formal groups (63 per cent) and public events (41 per cent). 9. People performed diverse tasks when involved, including identifying research priorities, being a project co-investigator, participating in research design and management, sharing views about the research, and overseeing data access. 10. Initiatives reported various impacts from involving people, including a mobile outreach bus and improvements to governance frameworks. 11. Specific genomics research tasks that people can get involved in included articulating phenotypes. 12. Methods of involving people and tasks performed are varied. Evidence on effective ways of involving people is not clear, with data likely under-reported. Consistent reporting on and evaluation of involving people is required.
Guidance for planning, reporting and evaluating initiatives: A	13. Guidance on reporting, planning and evaluating initiatives varies across disciplines. Many disciplines which share common methodologies use inconsistent language to describe similar methods, tasks and communication modes.

Review title	Learning points
multidisciplinary scoping review[195]	14. There is an urgent need to create standardised reporting of data about initiatives, including consistency of terminology to describe planning and reporting (including reporting impacts and outcomes), and consistent ways of sharing data to evaluate initiatives across disciplines. 15. To facilitate machine learning, terminology needs to be translated consistently between human languages. The findings of this review support the continued co-creation of the reporting system 'Standardised Data on Initiatives (STARDIT).[196]

Narrative review: How are the public involved in health research more broadly, and what are the impacts? (Abstract)

A more detailed summary of this narrative review is included in the Appendices.

Before beginning research specifically into genomics research and involvement, it was important to explore existing literature and terminology on involvement in health research in general. The methods from this review informed the design of later reviews, as well as helping inform methodologies by exploring impacts from different methods, and identifying the need for something like STARDIT to be created. The main learnings from this review which informed this doctoral research were that language and terminology is inconsistent in the area of public involvement in research; systematic reviews called for improved reporting and consistency; online involvement methods had a lower cost and more impacts than face to face involvement; and participatory action research (PAR) was the method with the most reported outcomes. Thus participatory action research was chosen as the guiding paradigm of the doctoral research, and online methods of involvement were integrated into the research plan in 2017.

Abstract

Aim

We wanted to understand how the public are involved in shaping health research and summarize the established methods of involving the public that might be applicable to genomics research. By looking at any impacts that this involvement might have had, we wanted to identify possible methods and approaches that may inform our future plans for involvement and impact assessment.

Method

A systematic search of systematic reviews relating to public involvement in research was conducted using boolean[3] operators in CINAL, Medline and Google Scholar. Data extracted from the reviews included what kind of involvement was taking place (the type), how it was done (the method), which stage of the research cycle it occurred and any impact on the research that might have occurred.

[3] Boolean operators are used when searching to connect words together. The three basic boolean operators are: AND, OR, and NOT. They can help focus a search by connecting information

Results

This systematic narrative review summarises five systematic reviews identified in this area. Involvement was reported at each stage of research with mostly positive impacts reported at each stage. Involvement in data collection and analysis were the most reported stages of involvement. Agenda setting was the most frequently reported stage of the research cycle for involvement. Most of the methods of involvement described specified a method involving people in a group structure, rather than individually. A total of 27 different methods of involvement were extracted, with community based participatory research having the most impacts. Other than generic positive impacts, the most impacts reported were improving the relevance of the research, improving dissemination and improving data collection. The stages of research with the most reported impacts were 'agenda setting' and 'data collection', with dissemination the next highest.

Conclusion

Involving people in health research generally has advantageous outcomes for research and health service planning. Involving people as early as possible is considered best-practice. There are a number of high impact actions which can be taken which do not necessarily require considerable budgets or time. Face to face involvement appears to have an increased cost in relation to impact, compared to other forms of involvement, including involving people using online methods. Involving people in planning how the public will be involved is crucial. While there are limitations in the data available, it is clear that involving the people at any stage of research has value, with impact at all stages being possible if people are involved early in the research cycle. More work is needed to better document the impacts of involvement in research.

Scoping reviews background

A systematic scoping review was chosen as the method for exploring how the public are currently involved in global genomics research. In addition, a second review was conducted, informed by the co-design process for STARDIT.[195]

The short section below is an extract from a peer-reviewed article I wrote with a co-author titled 'What are Systematic Reviews?' for the WikiJournal of Medicine[17]*. The extract focuses on scoping reviews, and formatting and references have been incorporated into this document. The STARDIT report for this article is here: https://www.wikidata.org/wiki/Q101116128*

What are scoping reviews?

Scoping reviews are distinct from systematic reviews in several important ways. A scoping review is an attempt to search for concepts by mapping the language and data which surrounds those concepts and adjusting the search method iteratively to synthesize evidence and assess the scope of an area of inquiry.[197,198] This can mean that the concept search and method (including data extraction, organisation and analysis) are refined throughout the process, sometimes requiring deviations from any protocol or original research plan.[199,200]

A scoping review may often be a preliminary stage before a systematic review, which 'scopes' out an area of inquiry and maps the language and key concepts to determine if a systematic review is possible or appropriate, or to lay the groundwork for a full systematic review. The goal can be to assess how much data or evidence is available regarding a certain area of interest.[199,201] This process is further complicated if it is mapping concepts across multiple languages or cultures.

As a scoping review should be systematically conducted and reported (with a transparent and repeatable method), some academic publishers categorize them as a kind of 'systematic review', which may cause confusion. Scoping reviews are helpful when it is not possible to carry out a systematic synthesis of research findings, for example, when there are no published clinical trials in the area of inquiry. Scoping reviews are helpful when determining if it is possible or appropriate to carry out a systematic review, and are a useful method when an area of inquiry is very broad[28], for example, exploring how the public are involved in all stages systematic reviews.[13]

There is still a lack of clarity when defining the exact method of a scoping review as it is both an iterative process and is still relatively new[202]. There have been several attempts to improve the standardisation of the method[197,199,201,203], for example via a PRISMA guideline extension for scoping reviews (PRISMA-ScR)[198]. The International Prospective Register of Systematic Reviews (PROSPERO) does not permit the submission of protocols of scoping reviews[204], although some journals will publish protocols for scoping reviews.[13]

This is the end of the extract from 'What are Systematic Reviews?' published in the WikiJournal of Medicine.[17]

Why were scoping reviews used in this thesis?

The main finding from the narrative review was that language and terminology to describe involvement is inconsistent in the area of public involvement in research. Multiple systematic reviews concluded that improved reporting and consistency was required. Accordingly, and on the advice from colleagues at the Cochrane Consumers and Communication review group, it was decided that a systematic review (meta-analysis) would not be useful impossible when terminology is imprecise and existing systematic reviews conclude there is systemic under reporting.[199,201]

For the review of public involvement in genomics research, after a number of preliminary test-searches, it was concluded that the limited amount of both peer-reviewed literature and grey-literature meant that systematic website searches were likely to generate more data. After reviewing a number of different methods, an existing international database managed by the Global Alliance for Genomics and Health (GA4GH) was selected as the data source for the scoping review. Further information about the selection of data sources is available in Chapter 4. The database was curated by GA4GH staff, and verified by them, ensuring the quality of the data. The alternative method was to use a non-verified database, created by the authors for the specific purpose of the scoping review, using a sampling method which relied on using commercial search engines which have opaque search algorithms. While a systematic site search was conducted using commercial search engine on the number of websites from the GA4GH database, this was in parallel with manual searching.

The second scoping (summarised below) as part of the STARDIT co-design process was an attempt to understand the variation in reporting of initiatives and any involvement in them, in order to inform both future systematic reviews and proposed standardised ways of reporting data on initiatives.

Guidance for planning, reporting and evaluating initiatives: A multidisciplinary scoping review

In order to move towards standardised reporting of initiatives, I led this scoping review in order to show the current variation in guidance on planning, reporting and evaluating initiatives. The need for this review was identified by the co-design process for the STARDIT Alpha version, and informed by the learning from 'Public involvement in global genomics research: A scoping review'.[28]

It is important to note that this review is not limited to one discipline and includes information from disciplines including health research, health technology assessment, environmental research, basic research, community based participatory research and educational research health, international development and community arts projects. The objective of this review is to display the considerable variation in this area in order to inform both future systematic reviews and proposed standardised ways of reporting data on initiatives.[196]

This review is not intended to summarise, appraise or find consistencies in ways of planning, reporting or evaluating initiatives, nor is it an attempt to systematically map and compare all data. This review has been designed as a first step towards future systematic reviews which could explore the question more thoroughly. It is also designed to help appraise the need and feasibility of a standardised way of reporting initiatives across disciplines.[196]

The following extract is from the abstract for the article 'Guidance for planning, reporting and evaluating initiatives: A multidisciplinary scoping review', which is currently a pre-print awaiting submission in 2022, where a full version of this review can be found.[195]

Authors

Jack S. Nunn, Steven Chang, Sue Gilbert, Saloni Swaminathan, Aidan Levy

Abstract

Background

The principles of involvement in research and other interventions (and associated methodologies such as participatory action research and citizen science) are the same across all fields (health research, health technology assessment, environmental research, basic research, community based participatory research and educational research), information about initiatives and their impacts is not consistently reported across disciplines. Additionally, the linguistic variation between terms such

as ‘involvement’, ‘engagement’ and ‘participation’ makes it difficult to conduct systematic comparisons of how people have been involved in initiatives, and any impacts. Standardised Data on Initiatives has been identified as one solution to standardised data about initiatives. However, before attempting to create a multi-disciplinary solution, it is important to map the current variation across disciplines for planning, reporting and evaluating initiatives.

Methods

The design of this scoping review was informed by the guidance on Preferred Reporting Items for Systematic reviews and Meta-Analyses extension for Scoping Reviews (PRISMA-ScR) Checklist for this scoping review. Multiple information sources were included in the search. These included, EBSCOhost Research Databases, Academic Search Complete, Business Source Complete, CINAHL, Education Research Complete, ERIC, MEDLINE, MEDLINE (with Full Text). Any information source which included information about how an initiative was planned, reported or evaluated was initially included as ‘guidance’, before being screened for relevance. Initiatives were included if they described an action, intervention, project or other kind of participatory process.
In order to answer the review question, the authors agreed that in addition to searching databases, the search would be improved by consulting subject area experts and citation searching (also known as ‘citation mining’ or ‘pearl growing’)[198]. As part of the expert consultation and citation searching, the search also included government websites (including reports and other documentation), non-government organisation websites (including charities and not-for-profits), and United Nations websites. We scanned the references of all information sources selected for inclusion from experts. The most recent search was conducted on 3rd April 2020.

Results and discussion

An initial 20,868 results were refined and screened by two authors (SS, JN), with further screening completed by a third author (AL). The initial database search found 529 relevant articles. After further screening, more articles were removed which were not relevant, leaving 316 relevant articles. From the database search, a total of 83 unique reporting tools or well-described methods (including standardised reporting tools or validated surveys) were found, with the ‘health and outcomes’ category and ‘health research’ categories both having 16 results using reporting tools. 228 articles provided guidance on planning initiatives, 122 provided guidance or ways of reporting initiatives or methodology, 225 provided guidance on or reported methods of evaluating initiatives, and 100 reported involvement or described ways stakeholders (including patients and the public) were involved.

From the expert and citation search (including 'pearl growing'), a total of 407 relevant resources were found, including 158 different reporting standards, guidelines and taxonomies were found across disciplines. This included 7 different biodiversity reporting standards, and 15 different reporting standards for health research.

Guidance on planning, reporting and evaluating initiatives exists across disciplines, with most being relevant to health research and some being environmental research. For initiatives which span disciplines, it is often not clear which methods of planning, reporting and evaluation are most appropriate, for example, an initiative to reduce air pollution to improve public health would span the disciplines of public health, environmental studies, policy and potentially education. The variation in language and associated concepts makes cross discipline comparison of methods extremely difficult and almost impossible to do in a systematic way.

Conclusions

Guidance on reporting, planning and evaluating initiatives is varied across disciplines. Many disciplines which share common methodologies use inconsistent language to describe similar methods, tasks and communication modes. There is an urgent need to create standardised reporting of data about initiatives, including consistent terminology to describe planning, consistent terminology for reporting (including reporting impacts and outcomes) and consistent ways of sharing data in order to evaluate initiatives across disciplines. Additionally, terminology needs to be consistently translated between human languages to facilitate machine learning. The findings of this review support the continued co-creation of the proposed reporting system 'Standardised Data on Initiatives (STARDIT).

This is the end of the extract from the article 'Guidance for planning, reporting and evaluating initiatives: A multidisciplinary scoping review', which is currently under review.[195]

Mapping the gaps in knowledge

How the reviews informed this doctoral research

The results from reviews conducted for this doctoral thesis provided an important context for this research. This short section summarises the main learning points from the reviews regarding current gaps in knowledge and practice. Learning from the narrative review informed development of the scoping review and, subsequently, the initiation of STARDIT. Development of STARDIT was informed by methods from the scoping review, with other interested people and organisations invited to be

part of the co-design process. The Alpha version of STARDIT, created to elicit feedback on the proposed framework, identified the need for guidance on planning, reporting on and evaluating initiatives, including genomics research initiatives. The literature review informed the development of the STARDIT Beta version. The interdisciplinary nature of public health genomics showed that STARDIT needed to work outside of the specific 'medical' context, and the co-design process highlighted the need for an interdisciplinary solution to reporting involvement in research and other initiatives.

How do we know how people are involved?

There is currently no standardised way to describe, report on and evaluate the impacts of involvement, making it difficult to establish best practices.[28,171,205–209] According to a 2018 systematic review, existing tools for reporting public involvement in health research such as GRIPP2 do not appear to have been widely adopted,[210] especially in low and middle-income countries.[211] Furthermore, tools like GRIPP2 are only appropriate for reporting on English language health research and offer no structured or publicly searchable databases for analysing report data.[212]

No existing tools for reporting involvement include indicators of involvement that are consistently described or reported, and the terminology used varies around the world. Subjectivity is evident in definitions of indicators of involvement. To consistently assess and report on involvement, objective measures need to be developed. Some concepts related to involvement cannot be reduced to linguistic labels such as 'ethics' and other values, so objective measures (or indicators) of who was involved and how in co-defining such concepts is important.

What evidence is there about the most effective ways of involving people?

Lack of consistent reporting about involvement in genomics research mean that it is not currently understood which methods, techniques and approaches are optimal, most acceptable, and preferred by the public, researchers and other stakeholders.[28] Evidence is therefore lacking on the best ways to integrate involving people as a routine part of the research cycle. In addition, many researchers are unfamiliar with the notion of involving people in their research, or are reluctant or ill-equipped to begin. Standard tools and guidance on effective ways of involving people are rarely available to them.

What are people's preferences for involvement?

While there are multiple ways to plan and report on people's participation in designing involvement in initiatives, [91,212–216] there is no standardised way to plan, report and evaluate this data. Plans for involving people must meet the needs of researchers (be realistic within time, budget and other resources) and the people they are trying to involve. Involving people at the outset in designing how the public and other stakeholders will be involved in an initiative can help ensure these needs are met.

Why Standardised Data on Initiatives (STARDIT) was created

The creation of Standardised Data on Initiatives (STARDIT) and the STARDIT Preference Mapping tool (STARDIT-PM) were developed in response to the gaps identified above. The next section summarises how STARDIT was co-designed, co-created and resulted in a Beta version, in which it is now possible to report data about initiatives (including genomics research) in a standardised way across multiple human languages. This includes being able to describe how people were involved, in which tasks, and any impacts of the involvement.

Standardised Data on Initiatives (STARDIT)

Learning from the reviews described in this section inspired me to initiate and design 'Standardised Data on Initiatives', and begin a co-design process to refine it. This section aligns with research aim 3 "develop a standardised way of planning, reporting and evaluating involvement in order to improve future genomics research".

Context for STARDIT co-creation

The need for the creation of a system such as STARDIT was identified during both the narrative review and the scoping review of public involvement in genomics research. Results from both reviews showed that reporting of involvement was inconsistent, with varied terminology even within Anglophone countries, as well as between them.

Retrospective context for STARDIT co-creation

This short section was added post-examination in August 2022, in order to further elucidate the context for why STARDIT was created, rather than using existing data reporting tools such as GRIPP. Please note some of the references in this section had not been written at the time of STARDIT co-creation, they are included to indicate the issues that were anticipated by STARDIT, rather than in response to existing literature which emerged during the writing of this thesis.

My work with Cochrane colleagues on the project 'Development of the ACTIVE framework to describe stakeholder involvement in systematic reviews'[12] required my input in suggesting data

categories to describe stakeholder involvement in systematic reviews. Combined with my awareness of this work, projects such as GRIPP,[183] and my attendance at early co-creation workshops for projects such as the 'Public Involvement Impact Assessment Framework' (PiiAF),[91] I became increasingly convinced that the pattern for reporting frameworks that were being developed to describe public involvement in English seemed to be in the direction of specialising towards specific fields, and creating further variation in terminology, rather than towards standardised terminology. In addition many frameworks were being developed in the English language only, with no attempt made to map concepts and terminology across other languages.

In addition, my work on the 'Wild DNA'[217,218] project made me aware of how there were similar issues reported with inconsistent reporting of both the people doing 'citizen science' and the methods.[210] Further, the known issues of inconsistent data sharing and reporting in both human genomics[219] and environmental DNA[220] practices convinced me that a system which could be used across these disciplines would have potential benefits.

I also noted a limitation of many reporting frameworks (including the many standards of COPE and EQUATOR) was that they required data to be shared in the format of a PDF attachment or additional file for a peer-reviewed article. The data was not shared in a structured way, or easily machine readable. This limited the usefulness of the data owing to the format it was being reported. Through my voluntary work at the Wiki Journals, I was aware of Wikidata, and conceived of a system where people could create reports which were structured using Wikidata and created using an online form to create a 'living report', rather than requiring a PDF attachment which would go out of date and was unable to share any additional impacts or outcomes.

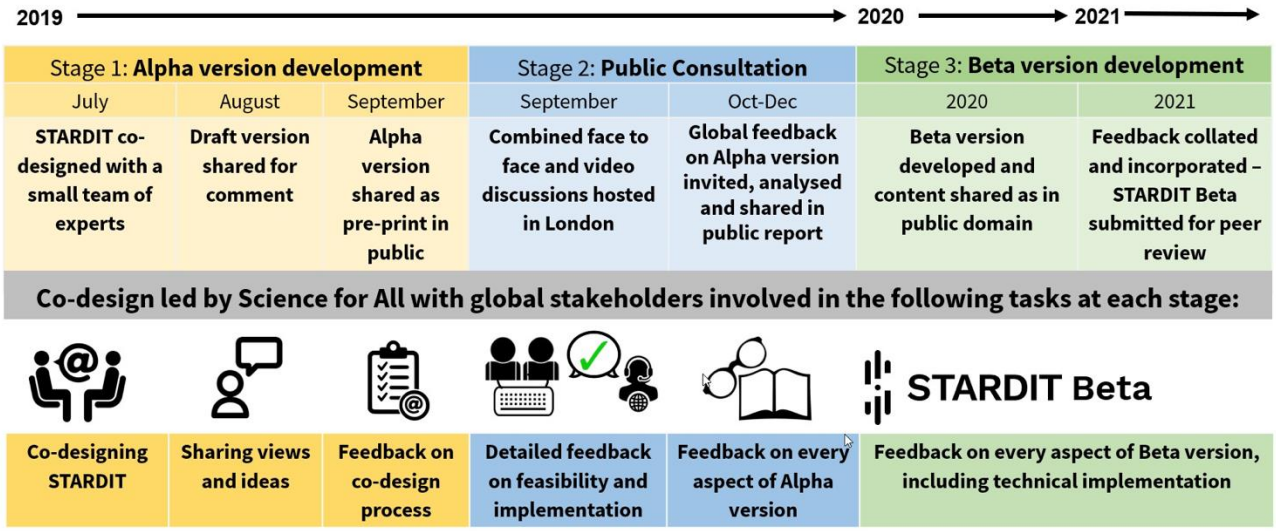
In addition, while designing the research projects for this thesis it became apparent that the relatively small numbers of people would be participating in two of the case studies (the Shared Ancestry study and the ausEE study). This meant that extrapolations from the data would be improved if data reporting was standardised across the multiple case studies in order to consistently combine the results with other studies for comparison[221], facilitating a cross-case analysis from standardised data.

STARDIT co-creation

After I had the initial idea for STARDIT, I decided to proceed a co-design process. The narrative review (and other literature) identified a participatory method as the most appropriate for the development of a system such as STARDIT.

I consulted experts in the field, including the then Chief Operating Officer of INVOLVE (then part of the UK's National Institute for Health Research), a Chief Editor of the journal *Research Involvement and Engagement* and the Chief Editor of the *WikiJournal of Science*. One suggestion included renaming it from '**Sta**ndardised **R**eporting on **D**issemination, **I**nvolvement and **T**ranslation' to '**Sta**ndar**r**dised **D**ata on In**it**iatives'. I designed the co-design process, and used this process to co-create the Alpha version, with more detail provided in the report 'STARDIT Public Consultation Report – September To December 2019', which is included in the Appendices[16]. Detailed information about the subsequent co-creation process is included in the STARDIT Beta version. A visual summary of the co-design process can be seen in Figure 3.3.

Figure 3.3: Visual Summary of STARDIT co-design process



How STARDIT was used in this doctoral research

STARDIT was used throughout this doctoral research, with learning from using the Alpha version (version 0.1) used to inform the development of the Beta version (version 0.2). STARDIT 0.1 was used to map preferences for involvement, plan involvement and case study, report case study method and data, compare data from each case study and evaluate impacts and outcomes. In addition a public domain STARDIT Beta version 0.2 report with structured machine readable linked-data is stored in the public domain and included in each published case study.

Standardised Data on Initiatives – STARDIT: Beta Version

The remainder of this chapter has been published in a peer-reviewed journal. The article and the additional files for this article can be found at the link can be found at this link: [221]

https://doi.org/10.1186/s40900-022-00363-9

Figure and table numbers in the remainder of this chapter have been changed to align with this thesis. Please note, as the additional files for the STARDIT Beta version are over 25,000 words long, they have not been included in this thesis to reduce the file size. They can be accessed via the link above.

Authors

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Please note, affiliations can be viewed at the above link.

Plain English summary

All major problems, including complex global problems such as air pollution and pandemics, require reliable data sharing between disciplines in order to respond effectively. Such problems require evidence-informed collaborative methods, multidisciplinary research and interventions in which the people who are affected are involved in every stage. However, there is currently no standardised way to share information about initiatives and problem-solving across and between fields such as health, environment, basic science, manufacturing, education, media and international development.

A multi-disciplinary international team of over 100 citizens, experts and data-users has been involved in co-creating STARDIT to help everyone in the world share, find and understand information about collective human actions, which are referred to as ‘initiatives’.

STARDIT is an open access data-sharing system to standardise the way that information about initiatives is reported, including information about which tasks were done by different people.

Reports can be updated at all stages, from planning to evaluation, and can report impacts in many languages, using Wikidata. STARDIT is free to use, and data can be submitted by anyone. Report authors can be verified to improve trust and transparency, and data checked for quality.

STARDIT can help create high-quality standardised information on initiatives trying to solve complex multidisciplinary global problems. Among its main benefits, STARDIT offers those carrying out research and interventions access to standardised information which enables well-founded comparisons of the effectiveness of different methods. This article outlines progress to date; current usage; information about submitting reports; planned next steps and how anyone can become involved.

Abstract

Background and objective

There is currently no standardised way to share information across disciplines about initiatives, including fields such as health, environment, basic science, manufacturing, media and international development. All problems, including complex global problems such as air pollution and pandemics require reliable data sharing between disciplines in order to respond effectively. Current reporting methods also lack information about the ways in which different people and organisations are involved in initiatives, making it difficult to collate and appraise data about the most effective ways to involve different people.

The objective of STARDIT (Standardised Data on Initiatives) is to address current limitations and inconsistencies in sharing data about initiatives. The STARDIT system features standardised data reporting about initiatives, including who has been involved, what tasks they did, and any impacts observed. STARDIT was created to help everyone in the world find and understand information about collective human actions, which are referred to as 'initiatives'. STARDIT enables multiple categories of data to be reported in a standardised way across disciplines, facilitating appraisal of initiatives and aiding synthesis of evidence for the most effective ways for people to be involved in initiatives.

This article outlines progress to date on STARDIT; current usage; information about submitting reports; planned next steps and how anyone can become involved.

Method

STARDIT development is guided by participatory action research paradigms, and has been co-created with people from multiple disciplines and countries. Co-authors include cancer patients, people affected by rare diseases, health researchers, environmental researchers, economists, librarians and academic publishers. The co-authors also worked with Indigenous peoples from multiple countries and in partnership with an organisation working with Indigenous Australians.

Results and discussion

Over 100 people from multiple disciplines and countries have been involved in co-designing STARDIT since 2019. STARDIT is the first open access web-based data-sharing system which standardises the way that information about initiatives is reported across diverse fields and disciplines, including information about which tasks were done by which stakeholders. STARDIT is designed to work with existing data standards. STARDIT data will be released into the public domain (CC0) and integrated into Wikidata; it works across multiple languages and is both human and machine readable. Reports can be updated throughout the lifetime of an initiative, from planning to evaluation, allowing anyone to be involved in reporting impacts and outcomes.

STARDIT is the first system that enables sharing of standardised data about initiatives across disciplines. A working Beta version was publicly released in February 2021 (ScienceforAll.World/STARDIT). Subsequently, STARDIT reports have been created for peer-reviewed research in multiple journals and multiple research projects, demonstrating the usability. In addition, organisations including Cochrane and Australian Genomics have created prospective reports outlining planned initiatives.

Conclusions

STARDIT can help create high-quality standardised information on initiatives trying to solve complex multidisciplinary global problems.

Introduction

Background

Many problems facing life on earth transcend the capacity of any single discipline to address. For example, problems such as pandemics, air pollution and biodiversity destruction cannot be characterised solely as 'public health', 'environment' or 'education' problems.[222,223] Solving such problems calls for holistic approaches[224] and will require governments, industry, research organisations and people around the world to work in partnership.

People need access to valid and reliable information to make informed decisions[225], which typically requires evidence. Depending on the context, this evidence-informed approach is called 'research', 'evaluation'[226], 'international development', 'education' or an 'initiative'. Hereafter all of the above will be referred to as 'initiatives'. For example, when deciding a response to a pandemic, standardised data can improve retrieval of relevant information which can be used to inform which affected individuals or organisations could be involved in the design of the response and which outcomes are most important[227]. This can include deciding which stakeholders should be involved in which tasks, such as prioritising outcomes.

In this article we explain how Standardised Data on Initiatives (STARDIT) builds on work to date by standardising a wide variety of data in a format applicable across multiple sectors, disciplines and languages. It is hoped that the creation of this evidence base will add to understanding and evaluating what works, for whom, why, and in what circumstances.[28,35,79,228]. Hereafter, data generated by an initiative (including raw data), information about the data (meta-data) and information about the initiative will all be referred to as 'data' unless otherwise specified.

In 2020, the United Nations Secretary-General stated that 'purposes that involve data and analytics permeate virtually all aspects of our work in development, peace and security, humanitarian, and human rights', encouraging 'everyone, everywhere' to 'nurture data as a strategic asset for insight, impact and integrity – to better deliver on our mandates for people and planet'[102]. Similarly, the United Nation's Paris Agreement highlighted the critical role of 'sharing information, good practices, experiences and lessons' in response to preventing irreversible climate change[229]. While organisations such as Cochrane (health) and The Campbell Collaboration (social sciences) are working to create high-quality systematic reviews of medical, social and economic initiatives, there remain limitations to the data available for such reviews. After a recommendation from the

Organisation for Economic Co-operation and Development (OECD), successful data sharing initiatives in biodiversity exist, such as the Global Biodiversity Information Facility (GBIF)[230], however there also remain limitations and accessibility issues in sharing and standardising biodiversity data[231,232].

It is often essential to include those affected by initiatives in the design and delivery of those initiatives[34]. For example, with an initiative to respond to a pandemic, those creating and delivering an initiative, and those affected by the outcome may be the same people. Forms of participatory action research where anyone can be involved in any aspect of research[233] (including amorphous terms such as 'citizen science'[234]) are increasingly recognised as crucial paradigms for solving such global problems such, as they can help ensure that initiatives are aligned with the priorities of those affected[235–237]. However, while the importance of involving people is clear[35], evidence-informed methods of doing so are limited.[28,171,205–208]

A recent statement defined a role for the public in 'data intensive' health research.[238] While in the health research disciplines there are over 60 different tools or frameworks for reporting or supporting public involvement, most published tools or frameworks are not used beyond the groups that developed them, and none work across multiple disciplines or languages.[209] Current reporting methods also lack information about the ways in which different people are involved in initiatives, making it difficult to collate and appraise data about the most effective ways to involve different people. In addition, 'citizen science' and 'participatory action research' are blurring the lines between concepts such as 'researcher', 'public', 'patient' and 'citizen'. [28,134,239–242]

The STARDIT system features standardised data reporting about initiatives, including who has been involved, what tasks they did, and any impacts observed. STARDIT was created to help everyone in the world find and understand information about collective human actions, which are referred to as 'initiatives'. In addition to providing new standardised data categories for describing who was involved in which tasks of an initiative, STARDIT can also incorporate the many existing data standards (see the supplementary resources 'Using Standardised Data on Initiatives (STARDIT): Beta Version Manual'), thus creating a unifying system for data hosting, linking and analysis. STARDIT can also report any different 'interests' of stakeholders and the ways power is shared between different stakeholders. The word 'stakeholders' here includes the public, those who have important knowledge, expertise or views that should be taken into account and others with a 'stake' in an initiative.[21,31]

Stakeholders can also include people who have financial, professional, social or personal 'interests'. An 'interest' can include a kind of commitment, goal, obligation, duty or sense of connection which relates to a particular social role, practice, profession, experience, medical diagnosis or genomic variation [243]. These can include financial or other interests which may compete or conflict with 'public interest'.[140] For example, a systematic review found that industry funded research is more likely to have outcomes favouring those with financial interests who are sponsoring the research[140,244]. Other examples include people from certain sub-populations (including those from populations more likely to be exploited[86]), Indigenous peoples, or people affected by rare diseases may have a personal interest in initiatives relevant to those specific populations, separate to the 'general public'.[4,7,28,58] For example a person with a rare disease may have a personal 'interest' in research into a treatment for that disease[7]. STARDIT allows standardised reporting of stakeholders and any interests.

Sharing data in a consistent way may help ensure that benefits of initiatives are shared more equitably (for example, by improving accountability).[28] In addition sharing information about who 'owns' or controls access to data and how such data access decisions are made can help people make informed decisions about participating in research[7]. By reporting involvement in initiatives, STARDIT also allows acknowledgement of people otherwise excluded from the public record – such as patients, people donating personal data, medical writers, laboratory assistants, citizen scientists collecting or analysing data, custodians of traditional or Indigenous knowledge, translators, interviewers, coders and code reviewers.

Objective

The objective of STARDIT is to address current limitations and inconsistencies in sharing data about initiatives. The STARDIT system features standardised data reporting about initiatives, including who has been involved, what tasks they did, and any impacts observed. STARDIT is designed to support a culture of partnership across disciplines and beyond, and is, wherever possible, aligned and interoperable with existing reporting models and frameworks such as those used in health, environment, manufacturing, publishing, government policy, education, arts and international development (see Table 1). In addition, the STARDIT Preference Mapping (STARDIT-PM) tool provides a standardised way to report information about different stakeholders' preferences, including preferences for power-sharing and methods of involving people during an initiative (see section 'Mapping preferences for involvement').

In alignment with the UNESCO Recommendation on Open Science,[245] the co-created values of the STARDIT project state that designs and code should always be open access and relevant licenses should always be those which allow others to build on and improve the project, while maintaining central control over quality (such as the Creative Commons Attribution-ShareAlike 4.0 International license (CC BY-SA 4.0) and the GNU General Public License (GPL) 3.0 for code. STARDIT data will released into the public domain (CC0) and integrated into Wikidata, which is a free and open knowledge base for collaboratively editing structured data[246]. The working Beta Version of STARDIT uses Wikidata to enable definitions to be co-created by contributors anywhere in the world, and therefore works across human languages, with interoperability with other platforms planned for future versions.

Potential applications

STARDIT's potential applications are summarised in Table 1. Among the principal applications, STARDIT offers public access to standardised information which enables the comparison of methods with the most impacts, such as ways of involving stakeholders in initiatives. The United Nations defines assessing impact as 'establishing cause and effect chains to show if an intervention has worked and, if so, how'.[247] With more data being shared, STARDIT could support decision making when planning stakeholder involvement in initiatives, and enable more people to assess the rigour of impact assessments.[247] This will be achieved by structuring the data in a way to allow such comparisons between different outcomes and methods of involving people, including using machine learning algorithms (including artificial intelligence).

In addition, STARDIT could be used to share information which makes research more reproducible[248,249], improving accessibility to the information required to critically appraise research and evidence and thus improving trust in processes such as the scientific method[250,251], and facilitate an appraisal of different knowledge systems, including Indigenous knowledge systems[252]. Such data sharing could also improve the translation of trusted, quality research and data, by empowering people to both access and appraise relevant data. For example, improved access to more standardised information (in multiple languages) about data and outcomes, could help to facilitate more informed collaborations between researchers and those monitoring and protecting critically endangered species, particularly where there is no common language.[253–255]

For example, many industries use self-regulatory processes to govern industry practices, with examples including the Forest Stewardship Council (FSC), Marine Stewardship Council (MSC)[256], Certified B Corporations,[257] and multiple Good Manufacturing Practice (GMP) guidelines. STARDIT

could be used to improve public awareness of, and access to the data already reported by such self-regulatory standards. Increased transparency could, for example, support people to make informed decisions when investing or buying products; automate analysis of data to facilitate such decisions, and improve accountability overall.

Defining 'initiative' and 'involvement'

As STARDIT is designed to report data across disciplines, distinctions between concepts such as 'intervention', 'research', 'project', 'policy', 'initiative' (and similar terms) are of secondary importance compared with communicating 'the aims or purposes of specified actions'; 'who did which tasks or actions'; 'are there competing or conflicting interests', and the 'outcomes from a specific action'. In this way, STARDIT can be used to report on any kind of collective action, which can include interventions, projects or initiatives – including a clinical study, education interventions or any kind of evaluation.[226,258,259] In this article, we use the word 'initiative' to describe any intervention, research or planned project which is a kind of collective human action. We define 'involving' people as the process of carrying out research, initiatives or interventions with people, rather than on them.[260] Involvement occurs when power is shared by researchers, research participants, and other relevant stakeholders (such as the public, industry representatives and experts). While meanings of these terms are often imprecise and can be used interchangeably, 'involvement' here is distinct from 'engagement'. We consciously use 'involvement' rather than 'engagement' to emphasise active participation that goes beyond simply receiving information about initiatives. We use 'engagement' here to mean where information and knowledge about initiatives is shared, for example, with study participants who remain passive recipients of interventions.[261–263]

Using and developing data standards

The current Beta Version of STARDIT maps terms and concepts using the Wikidata initiative (part of the Wikimedia Foundation)[36], which includes definitions (taxonomy), a way of describing relationships between concepts (ontology)[37], and a system to translate definitions and ontology between many languages. Examples of existing taxonomies include the National Library of Medicine's Medical Subject Headings (MeSH), which are used extensively in multiple kinds of literature reviews.[38]

How to involve people in combining or merging overlapping taxonomies for different subsets of data has been identified as an important question in the process of taxonomy.[264,265] By using Wikidata, STARDIT can be used by anyone to store both publicly accessible data and meta data (data about

data), and link to hosted structured linked data. While STARDIT is a novel element set, where possible it will also incorporate element sets from established data standards and map them where possible (see Table 5 in Additional file for examples of data standards which could be incorporated). This includes standard elements and value sets and controlled vocabularies.[266] The terms used in this paper are working terms, which will be progressively standardised over the lifetime of the project.

Structured Wikidata can help define terms and concepts clearly and unambiguously, in a transparent and open way. For example, colours in the spectrum are described by a standard numerical code in Wikidata, whereas the names of colours change according to different languages. Also, people with different DNA variations will also experience some colours differently. Similarly, the Wikidata entry for 'patient' has the human-readable definition of 'person who takes a medical treatment or is subject of a case study' (translated into 54 other languages) and a machine-readable definition consisting of dozens of semantic links to and from other Wikidata entries.[39] The terms 'participant' and 'research participant' are similarly coded, defined and translated. For terms that do not currently exist in Wikidata (for example, 'biobank participant'), a definition can be contributed by anyone in any language, refined by other users, then coded and translated into multiple languages by Wikidata. Developing taxonomies and ontologies will be an ongoing process facilitated by the current Wikidata infrastructure, and may require creating additional tools to create more inclusive ways of involving people in developing taxonomies.[40]

Methods and paradigms

Participatory action research

STARDIT development is guided by participatory action research (PAR) paradigms, which guides initiatives by aiming to involve all stakeholders in every aspect of the development and evaluation of an initiative[130,267]. Participatory research is a form of collective, self-reflective enquiry undertaken by people in order to understand their situation from different perspectives.[131] Development has also been influenced by existing work in health research, including the multidisciplinary area of public health, which incorporates social, environmental and economic research. In a health context, participatory research attempts to reduce health inequalities by supporting people to be involved in addressing health issues that are important to them, data collection, reflection and ultimately in action to improve their own health[132]. At the core of participatory research is 'critical reflexivity'. The process asks people involved to reflect on the causes of problems, possible solutions, take any actions required which might improve the current situation, and evaluate the actions[130].

Rights-based paradigm

The United Nations (UN) Universal Declaration Human Rights states everyone should be able to 'receive and impart information and ideas'.[95] The UN also states that democracy, development and respect for all human rights and fundamental freedoms are interdependent and mutually reinforcing'.[96] To uphold human rights and 'environmental rights',[268] and for 'the maintenance of peace', people require 'media freedom' in order to 'seek, receive and impart information',[96] free of unaccountable censorship. STARDIT has been created in order to help anyone uphold these universal rights, by providing a way to share open access information in a structured way with a transparent process for quality checking.

Cultural neutrality

Values, assumptions, ways of thinking and knowing are not shared universally. The participatory process used for developing STARDIT required and will continue to require that it attempts to map cultural variations, in an attempt to avoid unconsciously reinforcing particular (often 'dominant')[98] values. Transparent acknowledgement of differing values and perspectives is critically important, in particular when mapping if different stakeholders' values are complimentary or opposing. A participatory process requires mapping all of these perspectives and, where possible, involving people in labelling different perspectives and values. For example, STARDIT has already been used to map the varying perspectives of multiple stakeholders when planning a multi-generational cohort study.[6]

Many problems facing humans are shared by non-human life forms and ecosystems, including rapid climate change, air pollution and sea-level rise. If initiatives are to operate in inclusive, culturally-neutral ways, reconsideration of the language used to describe relationships between humans, non-human life and the environment is essential.[269] Environmental and social sciences are challenging and redefining colonial-era concepts of what can be 'owned' as property or who 'owns'. [269,270] As a result, ecosystems such as rivers and non-human animals, are being assigned 'personhood'.[271–273]. For example, a public consultation by a 'dominant' group might ask, 'who owns the rights to the water in a river system?'[98] This question imposes the dominant group's values on people who may not share the same concept of 'ownership'. In this way, Western European legal and economic traditions are frequently incompatible with those of some Indigenous peoples'.[269,274,275]

The participatory process used for developing STARDIT has attempted to be transparent about how different stakeholders have been involved in shaping it in order to improve how the system can be used to map values and provide more culturally neutral guidance for planning and evaluating involvement in initiatives. However, it is acknowledged that it will be a challenging process to 'de-

colonialise' and 'de-anthropocise' language and action,[276,277] as this may be perceived as a challenge to some people's cultural attitudes which may not align with the United Nation's universally enshrined principles of democracy, human rights and environmental rights. In addition, ongoing co-design will be required to ensure STARDIT is as accessible and inclusive as possible.

Development phases and methods

Both the STARDIT Alpha version (0.1) and the Beta version (0.2) have already involved people from diverse disciplines and backgrounds in the development, as this is integral to its effectiveness (Figure 3.3). It has been co-created using methodologies informed by PAR and other health research reporting guidelines.[278] PAR describes related approaches which involve experts (such as researchers), the public and other stakeholders "working together, sharing power and responsibility from the start to the end of the project".[88,89]

The Alpha version of STARDIT (version 0.1) followed the recommendations of a 2019 scoping review led by Nunn et al, which mapped public involvement in global genomics research.[28] This review stated that 'without a standardized framework to report and transparently evaluate ways people are involved, it will be difficult to create an evidence base to inform best-practice'.[28] This review was followed by an additional review (conducted in 2020 led Nunn et al, and to be submitted for publication in 2022), which mapped international guidance for planning, reporting and evaluating initiatives across multiple disciplines, and found 158 different reporting standards and reporting guidelines across disciplines (see the preliminary results in Table 7 of Supplementary resource 1)[195]. This included 7 different biodiversity reporting standards, and 15 different reporting standards for health research. STARDIT was also informed by a number of PAR projects, [4,279,280] and a report for the Wikimedia Foundation by the charity Science for All.[281]

The charity Science for All has hosted the co-creation process since 2019. Science for All is a charity based in Australia which support everyone to get involved in shaping the future of human knowledge, with co-created values guiding their work[282]. Development was informed by a number of literature reviews and guidelines, with methods of involving people in the development of STARDIT guided by the Enhancing the Quality and Transparency of Health Research (EQUATOR) network's approach to developing reporting guidelines.[278,283]. Methods of involving people included public events, online discussions and a consultation process. Owing to there being no formal budget for this project, the ability to actively involve people who can't afford to volunteer their time for free was restricted. Details about how inclusive ways of involving people were used are included in the publication consultation report.[284] This includes information about working with people from lower, middle and high-income countries, Indigenous peoples from Australia and Indonesia, people affected by cancer and rare diseases from Europe and the Americas, and people with expert knowledge of protecting endangered animals and eco-systems. The STARDIT project is actively

seeking funding from organisations which align with our values, in order to ensure the project is as inclusive as possible.

The co-creation process is currently being supported pro-bono by Science for All, and has also received in-kind support from individuals and organisations worldwide. A modified Delphi technique was used at some stages, with this method to be reviewed when co-creating future versions.[285,286] Many people were invited to provide feedback on all aspects of STARDIT, including its feasibility, design and implementation. They could comment anonymously using online forms and shared documents, in online discussion forums, via email or during face-to-face or video meetings.

After the feedback from the Alpha version was collated, work began on the Beta version. Between January 2020 and August 2021 multiple meetings and presentations took place to inform the Beta version, with some planned face to face involvement cancelled owing to the COVID-19 pandemic. Online activities where feedback on STARDIT was invited and given included interactive presentations by JN to the WikiCite 2020 Virtual conference[287], Poche Centre for Indigenous Health[288], Ludwig Boltzmann Gesellschaft[289], La Trobe University[290], Australian Citizen Science Association[291] and Rare Voices Australia. In addition, between February 2021 and May 2021, a total of 27 people provided feedback on the Beta version via the online form and collaborative document. Over 7000 words of feedback and comments were provided via the online form with 144 separate points, comments or corrections[284]. More detailed information about the consultation process for the Alpha and Beta versions up to May 2021 can be found in the 2020 and 2021 public consultation reports [16,284]. Further information about who was involved in the Beta Version development and proposed future development phases can be found in the supplementary information.

Science for All also hosts an online working group which continues to guide the development of STARDIT according to the terms of reference[292]. Anyone is welcome to join the working group, contribute to discussions and vote on decisions and ensure alignment with other initiatives. STARDIT and all associated work and co-designed logos (see Figure 3.2.1 and Figure 3.2.2) are currently published under the Creative Commons Attribution-ShareAlike 4.0 International license (CC BY-SA 4.0)[293], with the quality of any future iterations being the responsibility of not-for-profit host organisations and future licensing decisions to be made transparent, with anyone invited to be involved. The co-design process so far is summarised in Figure 3.3, with further information about the process available in Additional File 1. Further information about the STARDIT logo is available in Additional File 1.

Visual language and logo development

A culturally neutral logo was required for STARDIT in order for it to be recognised, including one which does not contain Latin alphabet letters so it could work across other scripts. Working with a professional graphic designer, the logos were co-designed, with anyone invited to give feedback and rank their favourite options. Inspiration for the logo was drawn from multiple traditions, with both tally sticks and DNA codons being integrated into the design of the logo, both systems of knowledge transfer which require a complementary half, representing the checking or tallying of the STARDIT data.

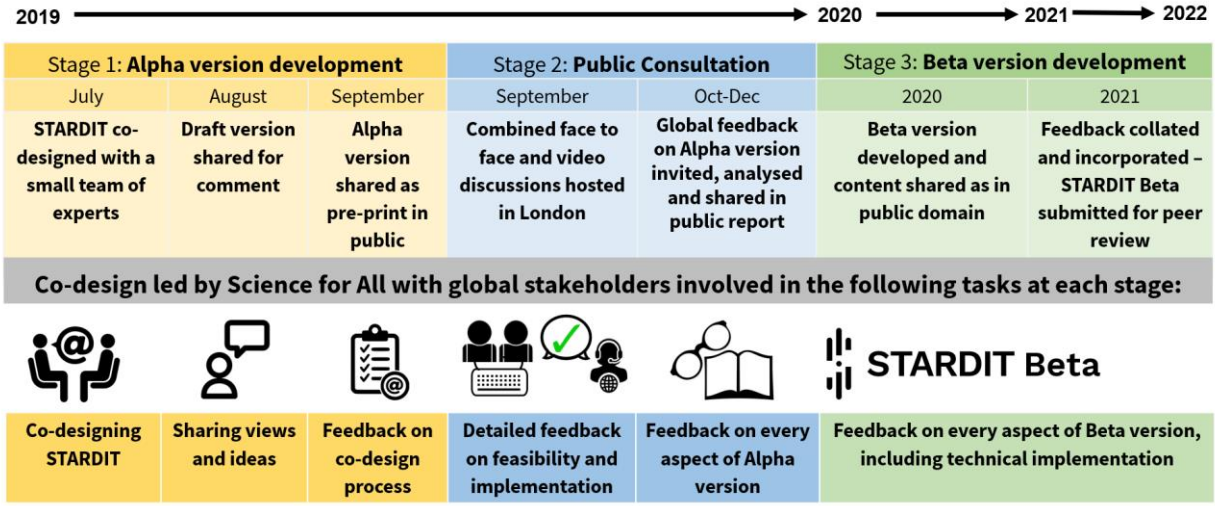
The novel and pronounceable word ‘STARDIT’ was purposely created, and it is proposed that the name for STARDIT in other languages be a phonetic way to spell the purposely invented word, with standardized spelling or signs to follow future co-design processes.

Future versions of the logo could contain Quick Response (QR) codes around the logo which link to report DOIs, to allow people to scan them easily on portable internet connected devices with cameras. Figure 4 is a mock up of such a QR code, linking to the first ever STARDIT report. Additional information about the data quality of each report could also be embedded in the logo by with colours which can be discriminated by those who have a decreased ability to see colour or differences in colour.

Figure 3.2.1: STARDIT logo

Figure 3.2.2: STARDIT small icon

Figure 3.3: Visual Summary of STARDIT co-design process



Version One implementation

Once STARDIT Beta (version 0.2) has been submitted for publication, a Beta version implementation article will be published, demonstrating the use of machine learning to generate STARDIT reports using mapped data from a number of international partner organisations. Work will then begin on the next version (version 1.0). Those involved with STARDIT development will disseminate information, gather feedback and recruit more people and organisations to participate as project partners and potentially funders. This stage is estimated to take between 2 to 3 years, at which point a working group will formally invite other appropriate partner organisations (such as the UN and WHO) to adopt the STARDIT framework. A Steering Group will be established to oversee and continually improve the STARDIT system. STARDIT will require continued working with publishers, research funders and governments to encourage adoption of the reporting tool. More detail on the proposed next stages can be found in the Additional File 1, in the section 'Development phases'.[9]

Results

This section summarises the results from the process of co-designing STARDIT. Since the start of the project in 2019, over 100 people from multiple disciplines and countries have been involved in co-

designing STARDIT. A working Beta version was publicly released in February 2021 (ScienceforAll.World/STARDIT). Subsequently, STARDIT reports have been created for peer-reviewed research in multiple journals and multiple research projects.[3,4,7,279,280,294,295] In addition, organisations including Cochrane[296,297] and Australian Genomics[298] have created prospective STARDIT reports outlining planned initiatives that will use STARDIT to report them. The Cochrane Council voted to use STARDIT to report planned work on creating a values statement[296,297], while the Australian Genomics working group 'Involve Australia' voted to use STARDIT to report their planned work.[298]

Beta Version interface

A link to the working Beta version can be found at: ScienceforAll.World/STARDIT/Beta.[299] The data fields in the STARDIT system co-created during the process described in this article are summarised in Tables 4. Table 3.10: presents the full version of the data fields. The 'Minimum Contribution Reporting Form' (MICRO) specifies the minimum information required to make a STARDIT report and these fields are highlighted in the table and marked with an asterisk (*).

Authorship

Acknowledging those involved in reporting ensures accountability for accuracy and increases trust in report content. STARDIT reports must be completed by named people who are accountable for the data being reported. Ideally, a public persistent digital identifier (for example, an ORCID number)[300] or an institutional email address will be linked to authors' names using Wikidata.

Reports cannot be completed anonymously, but STARDIT editors can redact author details from publicly accessible reports for ethical reasons (such as privacy or risks to safety).

Report authorship can be led by any stakeholder, including people associated with, or affected by, the initiative such as employees, researchers, participants, or members of the public. The affiliations of people formally associated with the initiative can be shared in a report.

Submission and Editorial process

Reports can currently be submitted to STARDIT via a simple online form or emailed as a document file. At present, only data which is already publicly accessible can be included in a STARDIT report. It is a way of collaboratively structuring data, not a primary repository for data. Once a report is submitted, editors can review content for quality control (for example, checking that publicly accessible URLs and URIs align with the data in the report), but will not critically appraise the initiatives or methods. The Editorial process is currently parallel to the WikiJournal process, involving selected Editors from these journals. While Editors will not approve the ethics of the initiative, a transparent process for considering ethical issues will be considered before publishing a report. The Editors may consider questions such as, 'Does data need to be redacted in order to prevent harm and protect or preserve life?' or, 'Is personal information being shared without consent?' For more

information about the Editorial process for reviewing data quality and ethical considerations, see the section 'Editorial and peer review of STARDIT reports' of the supplementary resource 'STARDIT Manual Beta Version'.
Once approved by the Editors, the STARDIT data will be entered into the database in a machine-readable format using structured data, based on the widely used Resource Description Framework (RDF) developed by the World Wide Web Consortium (W3C), which is used by Wikidata.[301] Each STARDIT report is assigned a unique Wikidata item number and all previous versions are navigable in a transparent history.

In future versions, it is proposed that stakeholders will be able to submit reports directly via an application programming interface (API), which will facilitate machine automation of STARDIT report creation. In addition, machine learning algorithms could be programmed to generate STARDIT reports from existing databases. As humans and machines submit reports, categories or meta-tags will be suggested (such as 'patient', 'member of the public'), with the option of adding, or co-defining, new categories using the Wikidata system for structured data.[302]

The database will generate a unique version number for the report with a Digital Object Identifier (DOI). To create an immutable version, the report will also be using the Internet Archive (a charity which allows archives of the World Wide Web to be created, searched and accessed for free).[303] Finally, the report will be assigned a status, with the data quality checking being described as:

- manually added, no human review (low quality checking – no DOI assigned)
- machine added, no human review (low quality checking – no DOI assigned)
- human review (medium quality checking – DOI assigned pending Editorial decision)
- peer or expert reviewed, with publicly accessible sources for indicators and references checked (higher quality checking – DOI assigned pending peer or expert review)

Processes for data checking and assigning report status need to be further developed and agreed by the STARDIT working group. For example, developing a transparent process if a report has been created about an initiative with no involvement from anyone associated with the project, or only one subset of stakeholders. In such cases, the Editorial team might give a short period of time for any other stakeholders to be involved in checking and editing any information.

Updating reports

STARDIT will enable reports to be updated as initiatives progress over time. Updates will be reviewed by the STARDIT Editors. Once an update is approved, the system generates a new version number, while also preserving the original report. Updates might include, for example, information about involvement in the initiative, or about dissemination, translation, co-creation of new metrics to assess impacts, or longer-term outcomes.[214]

Table 3.10: Summary of STARDIT Beta Version data fields

A minimum dataset is required for a STARDIT report. This is called the Minimum Contribution Report (MICRO) and the required categories are highlighted and marked with an asterisk (*). Relevant Wikidata items and qualifiers for these fields are provided in Additional File 1, in the section 'Developing taxonomies and ontologies',[9] and on the Science for All STARDIT Beta webpage.[299]

Section	Data category	Data field
Core: Initiative context - This information locates the initiative within a clear context.	Identifying information	Initiative name*
		Geographic location(s)*
		Purpose of the initiative (aims, objectives, goals)*
		Organisations or other initiatives involved (list all if multi-centre)*
		Relevant publicly accessible URLs/URIs
		Other identifiers (e.g RAiD[304], clinical trial ID[305,306])
		Keywords or metatags – including relevant search headings (e.g. MeSH[307])
		Other relevant information (free text)
	Status of initiative	What is the current state of the initiative?* Select from: 1. **Prospective**– this report is prospective or describes planned activity

Section	Data category	Data field
		2. **Ongoing** – the initiative is still taking place 3. **Completed** – the initiative has finished (*evaluation and impact assessment may be ongoing*)
		Date range (start and end dates of initiative)
	Methods and paradigms	Methods of the initiative (what is planned to be done, or is being reported as done). Include information about any populations or eco-systems being studied, any 'interventions', comparators and outcome measures (qualitative or quantitative)* *If appropriate, include a link to a publicly accessible document (such as a research protocol or project plan)*
		Include any information about theoretical or conceptual models or relevant 'values' of people involved with this initiative, including any rationale for why certain methods were chosen
Report authorship – Information about who completed the report and how *Please note this section can be completed multiple times if there are multiple authors*	Identifying information for each author *(authors can be anonymised in the public report but at least one verified identity will need to be sent to STARDIT Editors to attempt to prevent falsified reports)*	Name*
		Publicly accessible profiles, institutional pages*
		Open Researcher and Contributor ID (orcid.org) *
		Tasks in report completion
		Other information
	Accountability	Key contact at initiative for confirming report content (include institutional email address)*
	Date	Date of report submission *(automatically generated)*

Section	Data category	Data field
Input: Ethics assessment	Ethics approval information (if applicable)	Assessing organisation or group*
		Approval date and approval ID - *include any relevant URL*
Input: Human involvement in initiative Who is involved in this initiative and how? Editors assessing involvement may need to use the STARDIT 'Indicators of involvement' tool	Details about how each group or individual was involved in the initiative	Who was involved or how would you label those involved (select from group labels or submit new group label name in free-text)* *You can name individuals or use 'labels' to describe groups of people such as 'professional researchers', 'service users' or 'research participants'. Additional 'labels' or 'meta-tags' to describe people may be added if appropriate.*
		How many people were in each grouping label?
		Tasks of this person or group (list as many as possible)* – *including any information about why certain people were included or excluded in certain tasks (such as data analysis)*
		Method of doing task? How did these people complete these tasks? (what methods were used) – *for example 'group discussion' or 'reviewing documents'*
		Communication modes? What modes of communication were used – *for example, 'group video calls', 'telephone interviews' or 'postal survey'*
		How were people recruited, contacted or informed about these tasks?
	Involvement appraisal	Methods of appraising and analysing involvement (assessing rigour, deciding outcome measures, data collection and analysis)

Section	Data category	Data field
		Enablers of involvement (what do you expect will help these people get involved – or what helped them get involved) *Examples of enablers*
		Barriers of involvement (what do you expect will inhibit these people from getting involved – or what inhibited them from getting involved). Are there any known equity issues which may contribute? *Examples of barriers, and any attempts to overcome them*
		How did the initiative change as a result of involving people? *For example, did the initiative design or evaluation plan change?* *Note: this can be answered separately for different individuals or groupings of people*
	Involvement outcomes, impacts or outputs	Were there any outcomes, impacts or outputs from people being involved?* *When describing these, attempt to label which groupings were affected and how. These can include impacts on people, organisations, processes or other kinds of impacts.*
	Learning points from involving people	What worked well, what could have been improved? Was anything learned from the process of involving these people?
	Stage	Which stage of the initiative were these people involved? *(please provide information about any distinct stages of this initiative, noting some may overlap)*

Section	Data category		Data field
	Financial or other interests (including personal or professional interests)		Describe any interests (financial or otherwise), conflicting or competing interests, or how anyone involved may be personally, financially or professionally affected by the outcome of the initiative* *Including any relevant information about authors of this report.*
Input: Material involvement in initiative Mapping financial or other 'interests'	Financial		What was the estimated financial cost for the initiative.
			Funding information (link to publicly accessible URL if possible) - *this may include the project funder, funding agreements, grants, donations, public ledgers, transaction data or relevant block(s) in a blockchain*
	Time		How much time was spent on this project *Note: this can be answered separately for different individuals or groupings of people*
	Other		Describe any costs or resources that cannot be measured financially or quantitatively - *this may include expertise, traditional or Indigenous knowledge, volunteer time or donated resources*
Outputs: Data including code, hardware designs or other relevant information	Sensitive data	Secure criteria	Data adheres to relevant industry/discipline data security requirements
		Repository	How is data entered, changed or removed within a repository?
		Usage	Who is the data from this initiative shared with?
			Who has access to sensitive data and how is this decided?

Section	Data category		Data field
		Safety	Is data encrypted? Is it anonymised or de-identified? What methods are used for re-identification? What is the risk of unauthorised re-identification?
	Open data	FAIR criteria	Data adheres to FAIR criteria[85]
		Findable	Describe relevant metadata, how the data is machine readable and other relevant information
		Accessible	How can data be accessed – *include any information about authentication and authorisation*
		Interoperable	How is data interoperable or integrated with other data? *Include information about applications or workflows for analysis, storage, and processing, and resulting file formats or other outputs*
		Reusable	How can data be replicated and/or combined?
	Indigenous data	CARE principles	Data adheres to CARE principles[100,308]
		Collective Benefit	How will Indigenous Peoples derive benefit from the data
		Authority to Control	How will Indigenous Peoples and their governing bodies determine how relevant data are represented and identified
		Responsibility	How will those using the data provide evidence of these efforts and the benefits accruing to Indigenous Peoples
		Ethics	How have Indigenous Peoples' rights and wellbeing been centred during the data life cycle
	All data	Hosting	Where is it data stored and hosted -s*hare any location data if appropriate*

Section	Data category		Data field
		Owner	Who 'owns' the data or claims any kind of copyright, patent(s), or other specific types of intellectual property - *include relevant open licensing information*
		Analysis methods	Describe methods used to analyse the data (including a link to any relevant code and information about validity)
		Usage	How can data be used? *Include information about license and attribution*
		Dissemination	How is information about this data disseminated? *For example, how are results from analysis shared?*
		Impact	impact/effect of the output
		Data control	Who controls access to the data? How are decisions about data access made? Is data anonymised or de-identified? What methods are used for re-identification? What is the risk of unauthorised re-identification? How is this risk managed?
		Management and quality	Which person (or organisation) is responsible for managing (or 'curating') the data?
			Who is accountable for ensuring the quality and integrity of the data? (this may be an individual or organisation)
Impacts and outputs: Publications, events, changes, learning items etc.	What was learned		What new knowledge has been generated? (if appropriate, include effect size, relevant statistics and level or evidence)*
	Knowledge translation		Describe how the learning or knowledge generated from this initiative has or will be used

Section	Data category	Data field
	Impacts	Have there been any outcomes, or has anything changed or happened as a result of this initiative that isn't captured in previous answers?*
	Measurement and evaluation	How has or how will this be measured or evaluated?
		Who is involved in measuring or evaluating this?
		Who was or is involved in deciding on the outcomes used to evaluate any impacts or outcomes? How were they involved?
Information completed by Editors		
STARDIT report version number (assigned)		Report number assigned to distinguish it from any future updated reports
Indicators completed by Editors and/or peer reviewers Editors and peer reviewers assessing the report will need to look for indicators in the following categories on publicly accessible URLs*	Indicators of involvement	Use the STARDIT 'Indicators of involvement' tool
	Indicators of data practice compliance	Use the relevant criteria
	Indicators of translation and impact	
	Other indicators	

Scope and applications

STARDIT is the first and only data-sharing system that enables standardised sharing of data about how people are involved in any type of initiative, across any discipline, including involvement in the planning, evaluation and reporting of initiatives. In addition it allows comparison of both evaluation

methods and any impacts or outcomes in relation to standardised terminology. Table 3.7 summarises the proposed scope and potential applications of the STARDIT.

Current usage

STARDIT provides a way to report data about who did which tasks in an initiative. STARDIT reports have already been used to describe a number of research projects, including data about who did which tasks, ethics approval, funding, methods and outcomes.[4,279,280]

In health and medicine, STARDIT is already being used by an Australian Genomics working group to have describe planned work to improve guidance on involving the public in genomic research[298]. The Cochrane Council voted to use STARDIT to outline a proposed process for co-creating a Cochrane values statement[297,309]. Other projects which have used STARDIT reports include participatory action research projects involving a large cohort study of >15,000 healthy, elderly research participants[294], a protocol for precision medicine for Aboriginal Australians[3], and a group of patients and families affected by a rare immunological disorder[7], and a project involving extended family of donor-siblings who share the same sperm-donor father.[4,295]

The Wikipedia-integrated open access peer reviewed WikiJournals are also using STARDIT, which has articles which are integrated into Wikidpedia.[17] For example, a STARDIT report has been created to share information about a Wiki Journal of Medicine article about systematic reviews (with an associated integrated Wikipedia page)[17], including information about authors, editors and peer-reviewers.[310] This allows readers to critically appraise the source before deciding whether to use or share it.

An environmental research project has also used STARDIT to report the initiative, which works with citizen scientists to locate critically endangered species using eDNA[217,218]. Currently, the Standardised Data, which makes up the STARDIT reports, is structured in WikiData, and hosted in the STARDIT report format using WikiSpore, which is hosted on Wikimedia Cloud Services, and is used as an experimental and supplementary space to develop potential Wikimedia projects.[311] Figure 3.3.1 summarises how Standardised Data is organised.

Figure 3.3.1: STARDIT Technical Information Summary

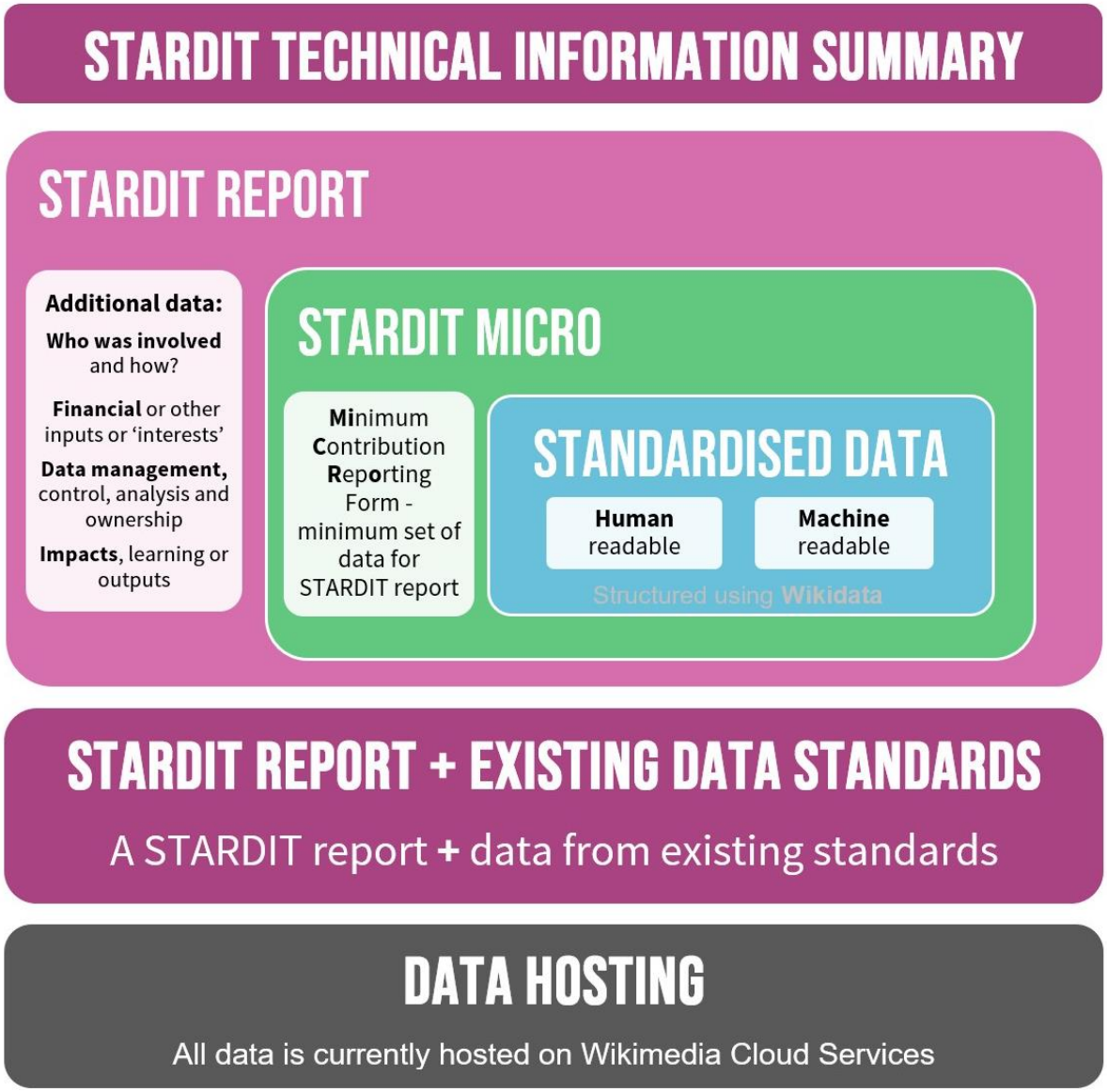


Table 3.7: Example applications of STARDIT

Further examples of how STARDIT can be used are provided in the supplementary information, including; using STARDIT in genomic research for mapping phenotypes and reporting who was involved in helping define and describe them; providing data to critically appraise information sources (including public videos); report data about case studies consistently; creating 'living systematic reviews' and training machine learning from STARDIT data.

Area	Sub-Area	Relevant data categories
Research	Health research	**Reporting:** Funding, conflicting or competing interests, co-design, experts involved, people affected involved, methods, process for deciding and measuring outcomes, protocols, who is accountable for ensuring protocol is followed, information about data storage, sharing, ownership and custodianship, information about data security practices and standards, information about consent and withdrawal processes evaluation of entire research process, ethical review, information about data analysis and data validation
	Social research	
	Genomics research	
	Environmental research	
Policy	Health and social policy	**Reporting:** Values of people involved, sources of data and evidence, data on past and current initiatives and spending[312], process for policy (or proposed policy) creation, process for deciding and measuring outcomes, experts involved, people affected involved, policy or manifesto writers, conflicting or competing interests of people involved, purpose of policy (what needs have been identified, how and by who), outcomes from policy (including outcomes measured by those affected by policy), policy evaluation (reporting if it achieved what was intended)
	Other government policy (transport, arts, education, environment etc)	
	Foreign policy	
	Proposed policy (including draft policy and manifestoes)	
	International development	
Education and learning	Educational initiatives	**Reporting:** Sources of data and evidence for intervention, purpose of intervention, process for educational intervention creation, funding, conflicting or competing interests, experts involved, people affected involved,

Area	Sub-Area	Relevant data categories
		conflicting or competing interests, process for deciding and measuring outcomes, outcomes from intervention, evaluation of intervention, ethical review
Arts	Community arts projects	**Reporting:** Purpose of project, process for project design and implementation, experts involved, people from communities intended to benefit involved, funding, conflicting or competing interests, process for evaluating project, project evaluation, project outcomes.
	Arts funding	**Reporting:** People involved in deciding funding process, purpose of funding, people allocating funding (funding sources), funding amount, conflicting or competing interests, process for deciding outcomes of funding, evaluating the funding allocation process
Information, media and cultural heritage	Health and medical information	**Reporting:** People involved in researching, writing (including medical writers), creating, reviewing (including peer reviewers), disseminating and funding, information about any potential risks (to human health or lifeforms, natural or cultural heritage), information about who assessed those risks and how (for example, medical information standards[313]), information about consent to appear in images and verified appearances of public figures, information about ownership of data or knowledge (including concepts of intellectual property, copyright and license information, relevant blockchains and non-fungible tokens), evaluating knowledge translation, reporting impacts and outcomes.[281]
	Disaster and emergency communication	
	Public interest, factual information commentary, documentaries and other informative media	
	Intangible cultural heritage (including folklore, traditions, language), traditional, local and	**Reporting:** who created any content containing the Indigenous or traditional knowledge, what tasks they had, how this knowledge was shared and any relevant concepts of 'owning' or 'property'; reporting who knows certain things (for example, people who are recognised as

Area	Sub-Area	Relevant data categories
	Indigenous knowledge and wisdom	'Preservers of Important Intangible Cultural Properties'[314]); reporting who is recognised as an Elder, community leader, Indigenous elders or leaders (and by who); reporting who does or does not have permission to verify, share, redact or edit content (including stories, beliefs, cultural practices and medicine)[315]; information about data custodianship[252], information about any potential risks (to human health or lifeforms, natural or cultural heritage); information about who assessed those risks and how, information about informed consent process, information about any cultural sensitivities or restrictions (including relevant information about gender, clan, tribe or other culturally constructed groupings)[316–318], information about relevant laws and lore[252], ethics processes (including who was involved and how), reporting impacts and outcomes from dissemination.[281]
	Tangible cultural heritage (including cultural property[319])	**Reporting:** Who was involved in creating the property, any concepts of ownership or guardianship in relation to the property, data about ongoing management (including monitoring, exhibiting, restoring or moving), data about cultural significance and stakeholders involved in defining this
	Hardware designs (including hardware architecture, device designs or other abstract representations)	**Reporting:** Who was involved in creating the designs and how, who reviewed them and how (including relevant safety, regulation or standards information), what formats are the designs shared as and in what medium, information on license(s), outcomes and impact of the hardware
	Code and algorithms	**Reporting:** who created code (including algorithms), who is involved in reviewing and scrutinising code (including who is involved in which ethical review processes), what code is part of which distinct projects or forks, what language the

Area	Sub-Area	Relevant data categories
		code is in, what medium (for example, machine or DNA), information about ownership of data or knowledge (including concepts of intellectual property and copyright), information on license(s), purpose of code, outcomes and impact of the code
Management and monitoring	Environmental and natural heritage, natural resource management	**Reporting:** data about who was involved in service design, monitoring and management processes, data about funding for monitoring or management (for example, funding for pollution monitoring), data about how information will be stored and shared (including what will be redacted and data security), data about who decides what data will be redacted and how this decision is made, information about how data will be analysed (including relevant code and algorithms) and how learning from data will be shared, information about relevant data privacy legislation and regulation
	Public and private essential services management (health, infrastructure, waste and recycling, water and sewage, electricity)	
	Data management and monitoring	
Evaluation	Process evaluation	**Reporting**: data about processes (industrial, public health, organisational)[226], people involved, outcomes
	Evaluation of participatory methods (co-design)	**Reporting:** data about participatory research methods and compare outcomes.
	Transparent rating	**Reporting:** Processes of transparency rating (or 'scoring') data quality about initiatives based on how much information about the initiatives is shared in a publicly accessible way (or reasons for redaction, including Indigenous knowledge).
Production, consumerism and business	Industry standards	**Reporting:** Internal processes and data sharing practices of self-regulating industry standards (for example, the Forest Stewardship Council, Marine Stewardship Council[256] and Certified B Corporations[257]), data sharing principles, process evaluation (including by those affected).

Area	Sub-Area	Relevant data categories
	'Green' industries and eco-tourism	**Reporting:** Transparent process for defining 'green' and 'eco', experts involved, people affected involved, process for deciding and measuring outcomes, outcome measures, evaluation of process.
	Infrastructure, construction and interiors	**Reporting:** Transparent reporting of sources of building and furniture materials, such as wood (including relevant DNA information to verify sources of timber), metals and other materials (including information verifying the supply chain is slavery free), data from building and structural assessments
	Finance and financial services	**Reporting:** who is involved in decision making (including investment and divestment), who scrutinises decision making, who is involved in holding individuals to account and who scrutinises this process, competing or conflicting interests of people involved in decision making, data about how concepts such as 'ethical investments' are defined, impacts or outcomes from investments or donations, data sharing practices and security practices, data about who scrutinises security practices.
	Donation and philanthropy	**Reporting:** Any stated purposes or caveats for donation, organisations or individuals donating, how money was spent, who was involved in deciding how it was spent, what was the method for deciding this, who is accountable for overseeing this, any outcomes or impacts.
	Other products (medical devices, electronics)	**Reporting:** Experts involved in production, other people involved in production process, resources involved in production process (including relevant DNA information to verify products from plants, animals and fungi), ingredients, funding for resources (for example demonstrating it is 'slavery free'), process reporting
Products for human use or ingestion	Food	
	Medicines	

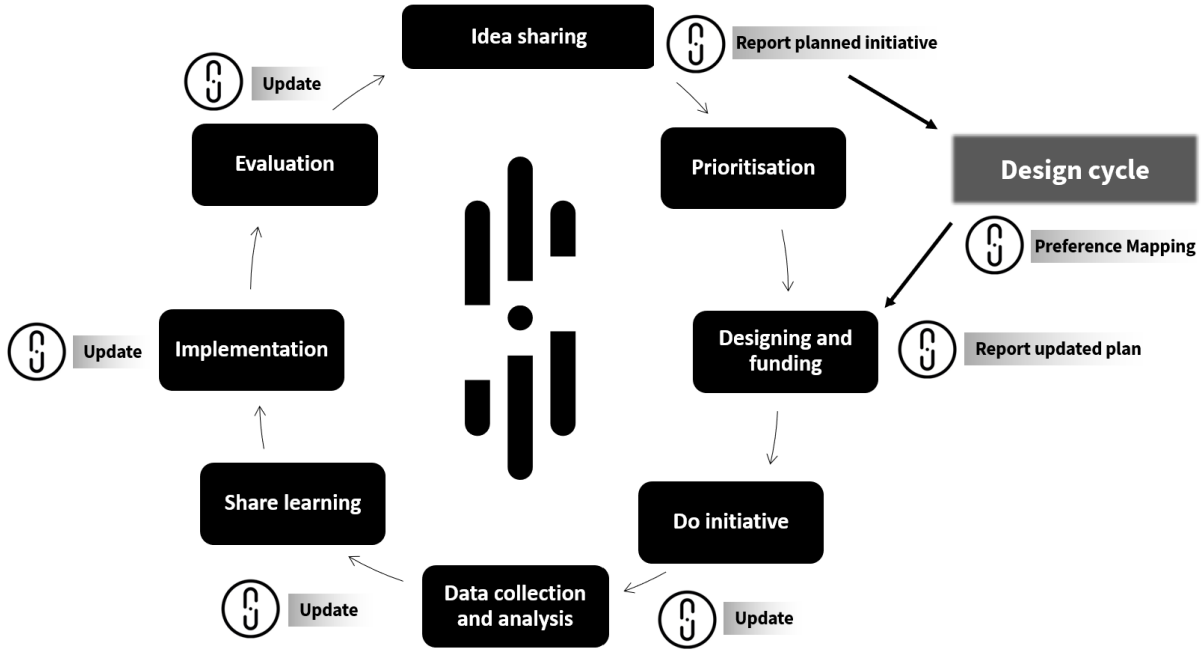
Area	Sub-Area	Relevant data categories
Products for non-human lifeforms	Food	(including Good Manufacturing Practice), regulation and authorisation processes (for example medicines and medical devices), code and algorithm checking (for example, autonomous vehicles) process for designing impact assessment, impact assessment (including human and environmental), experts involved in dismantling process (including recycling), other people involved in dismantling process and disposal, evaluation of product according to transparently decided outcome measures
	Medicines	
	Other products	
Health Technology assessment	Assessment process for pharmaceuticals, devices, procedures and organisational systems used in health care	**Reporting:** Process for deciding health technology assessment (oversight and scrutiny), sources of data and evidence, process for deciding and measuring outcomes, experts involved, people affected involved, conflicting or competing interests, outcomes from assessment decisions (including outcomes measured by those affected by assessment decisions), collation of adverse event reports from Governments and reputable sources, assessment evaluation (did it achieve what was intended?), results of economic evaluations
Health and social care and services	Health care and services	**Reporting:** Process for assessing needs (including who was involved, the method and budget), process for prioritisation of services (including budgets and 'rationing' decisions), process for designing and implementing service or care (including who was involved, the method and the budget), process for evaluating service or care (including impacts), patterns for evaluating service improvement initiatives, process for reporting adverse events and malpractice (including the overview and scrutiny of this process), process for identifying patterns of sub-optimal service, process for responding to malpractice or other identified issues, process for identifying impact indicators (including geolocation data)
	Social care and services	
Other services		

Further examples of how STARDIT can be used are provided in the supplementary information, including; using STARDIT in genomic research for mapping phenotypes and reporting who was involved in helping define and describe them; providing data to critically appraise information sources (including public videos); report data about case studies consistently; creating 'living systematic reviews' and training machine learning from STARDIT data.

Using STARDIT

Across all disciplines, 'plan', 'do' and 'evaluate' are recognised as distinct stages of initiatives[320]. While there are many ways to involve different people in these stages, standardised reporting and thus evidence-informed methods of doing so are lacking.[19,28,35] Figure 3.4 describes how STARDIT can be used to map how people might be involved in designing, doing, reporting and evaluating initiatives, starting with 'idea sharing'.

Figure 3.4: Planning and evaluating initiatives using STARDIT



Reporting initiative design in STARDIT

Questions such as, 'Who decides how people are involved?' and, 'Who is involving whom?' and 'what are people's preferences for ways of working' can be difficult to answer and is an active area of research[7,321]. For example, planning a healthcare initiative requires input from experts as well as from the people the initiative is intended to help[19]. Figure 3.5 summarises a way of using STARDIT to report the design process of initiatives, with Table 3.8 providing details about how involvement from different stakeholders can be reported at different stages. The section 'Detailed reporting of design

using STARDIT' in the supplementary resource 'STARDIT Manual Beta Version' provides more comprehensive information.

Figure 3.5: Reporting initiative design in STARDIT

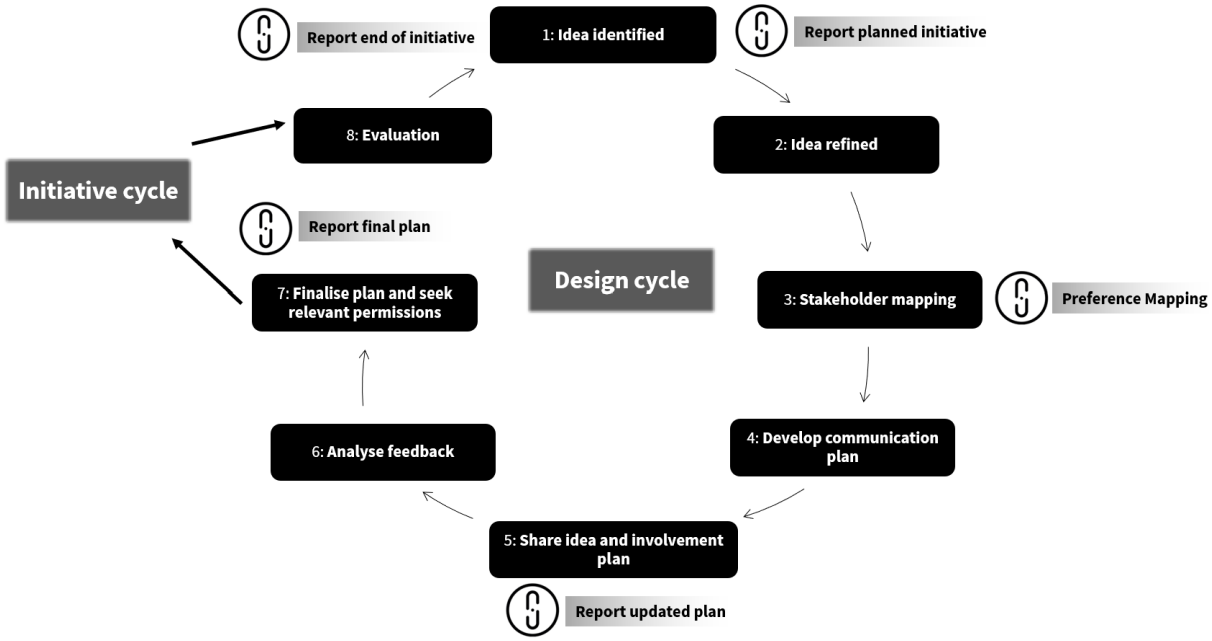


Table 3.8: Summary of reporting initiative design in STARDIT

Initiative stage	Data reported
Stage 1: Idea identified: An idea for an intervention, project or research is identified and articulated	Report planned initiative
Stage 2: Idea refined The idea is refined with a small group of stakeholders[35,91,134,138,322–325]	
Stage 3: Stakeholder mapping: Existing stakeholders attempt to map who is included and who might currently be excluded from the process[134,185]	Preference Mapping
Stage 4: Co-create communication plan Develop a communication plan to invite people to co-create involvement[134,322,326]	
Stage 5: Share plan: Share the idea (according to the communication plan) and ask for feedback on it (including the involvement plan) [138,156,327]	Report updated plan
Stage 6: Analyse feedback: Collect and analyse feedback, share results. [326(p1)]	
Stage 7: Finalise idea and involvement plan: Co-create the plan (including the plan for involving people), seek relevant permissions and ethics [328,329]	Report final plan
Do initiative (see 'Planning and reporting initiatives using STARDIT')	
Stage 8: Evaluate involvement: Evaluate the process and the impact of both the initiative and involving people in the initiative	Report end of initiative

Mapping preferences for involvement

Involving multiple stakeholders in designing how people should be involved in initiatives is considered best practice, as it may facilitate power sharing and improve the process overall.[28,330] Current explanations of participatory research methods, and the language used to describe them, vary considerably. There is no agreed, consistent way to describe how people have been involved in a research initiative, or to report the impacts of their involvement.

The STARDIT Preference Mapping (STARDIT-PM) tool provides a standardised way to report the preference of multiple stakeholders. Anyone can be involved in creating a STARDIT report, which means that data on the impacts and outcomes of participation can be contributed by diverse

stakeholders. Such reports will help researchers make informed decisions when planning participation in research.

For example, a recent study showed how a charity for people affected by a rare disease involved a small number of people affected by the rare disease in discussing preferences for how best to involve the wider community of people affected in future research prioritisation and planning.[7] Those involved had a good understanding of any specific needs or preferences for involvement, and shared preferences for the tasks (such as overseeing data access), method (facilitated discussions) and mode of involvement (online text-based discussion). The STARDIT-PM data about this processes showed a preference for being involved using online discussions, and the STARDIT report stated that involving people influenced the way the charity planned to involve people prioritising research in the future.[279]

Examples of completed STARDIT-PM can be found in the additional files of a number of research projects.[4,279,294] Table 3.9 summarises questions which can be asked to map stakeholder preferences with respect to involvement in initiatives.

The first stage of preference mapping requires individuals to self-identify as belonging to a specific grouping of people. People from that grouping then share views on how people from other groupings could be involved (or which groupings should not be involved). For example, labels for such groupings could include:

- only people with a professional role in the initiative
- everyone (any member of the public who is interested)
- anyone who might be indirectly affected by the initiative
- only people who are directly affected by the initiative
- only people who are participating in the initiative
- only people with a financial interest in the initiative.

As a consistent mapping tool for use across all initiatives, STARDIT would allow both comparison of diverse stakeholder views and exploration of similarities and variations in relation to preferences for involvement. Used alongside other planning tools, this information could help align initiatives with stakeholders' preferences. In this way, how stakeholders are involved throughout an initiative could be co-designed from the outset. Analysis of the data about preferences should involve stakeholders from multiple groupings to ensure that a diversity of perspectives are involved in assigning meaning to any data.

Table 3.9: Questions for mapping preferences for involvement

Question	Rationale for question
Which stakeholder group does this person align with?	To establish which grouping(s) the person identifies as being part of – for example 'researcher' or 'participant' (noting any groupings should be co-defined)
Describe any financial relationship or other interest this person has to this project	To provide a public record of any potential conflicting or competing financial interest
Views on the purpose and values of the research	To establish the purpose of the research, and the motivations and values of the initiative from multiple perspectives
Describe how you think the learning from this initiative could be used	To establish views about knowledge translation and application of learning
Views on who data from this project should be shared with and how	To establish that person's view about data sharing and ownership
Views on who should be involved (which 'groups' of people) – including who should not be involved – *following answers may be categorised depending on the stakeholder group*	To establish that person's views on which 'groups' of people they think should be 'involved' in research – that is, having a role in shaping the research design, direction and outcomes *Note: Answers may require sub-categories if there are multiple categories for who should be involved*
Views on specific tasks of this person or group	To establish that person's views on the tasks of the specific stakeholders who they think should be involved.
Preferred modes of communication	To establish that person's preferences on communication modes with stakeholder groups
Views on what methods should be used	To establish that person's views on which methods should be used to involve people – for example 'online survey'

Question	Rationale for question
Views on facilitators of involvement	To explore that person's perceptions of what might facilitate involving specified groups of people and help inform the design of involvement
Views on barriers of involvement	To explore that person's perceptions of what might be a barrier to involving specified groups of people and help inform the design of involvement
Views on what the outcome or output of the involvement could be	To ascertain the expectations of that person about what involving the specified groups of people might achieve
Views on which stage of the research this group should be involved?	To establish that person's views on which stage of the research the specified groups of people should be involved in

Values

The STARDIT co-design process included co-defining shared values. It was agreed that the STARDIT project must be implemented in a way which encourages those involved to acknowledge cultural values and assumptions in a transparent way. For example, some people can be labelled as having human-centred (anthropocentric) values, which values natural resources in relation to benefits they can provide for humans. In contrast, some people who think the value of nature should be measured using non-human outcomes can be labelled ecocentric[331]. A participatory process requires mapping all of these perspectives and, where possible, labelling them.

The values for STARDIT were adapted from an existing values statement co-created by the charity Science for All,[332] with values specific to the STARDIT project summarised in Table 4. Further information about the values are provided in the supplementary resource 'STARDIT Manual Beta Version'.

Table 3.9.1: Values of the STARDIT project

Value	Summary
System and language agnostic	STARDIT is system and language agnostic, it should always be designed to work across and with as many systems as possible, in as many countries and languages as possible
Designs and code should always be open access	In alignment with the UNESCO Recommendation on Open Science,[245] STARDIT designs and code should always be open access and relevant licenses should always be those which allow others to build on and improve the project, while maintaining central control over quality (such as the Creative Commons Attribution-ShareAlike 4.0 International license (CC BY-SA 4.0) and the GNU General Public License (GPL) 3.0 for code)
Participatory paradigm	STARDIT development will be guided by the participatory action research (PAR) paradigm[130]. PAR is an umbrella term which describes a number of related approaches, including [88(p1)], community-based participatory research, participatory action research (including critical participatory action research), participatory health research, community-partnered participatory research, cooperative inquiry. It may also include other forms of action research embracing a participatory philosophy which may include 'co-design' of research and other kinds of research which might include forms of 'public involvement' (or sometimes 'engagement'). The plain English definition of the paradigm is that power to control the project with be shared in a transparent, inclusive and equitable way
United Nations rights-based paradigm	STARDIT will be guided by the United Nations rights-based paradigm, including human rights, environmental rights and other emerging rights

Discussion and future versions

Since the inception of this project in 2019, subsequent world events have included; the worst bushfires in Australian history[333] in parallel with misinformation campaigns funded by industries whose actions increase the severity and frequency of such fires;[334,335] the COVID-19 pandemic and associated "infodemic" of misinformation;[336] continued violence inspired by misinformation;[337–339] and "infowars" of information control which continue to take place alongside wars fought with

physical weapons.[340] The need for tools which can provide a way for all global citizens (and their machines) to share, asses, verify, edit, and link data has never been greater or more urgent. STARDIT is one such tool, which, by using Wikidata, will make use of existing and trusted infrastructure, and allows people to co-define types of data in multiple languages.[313,341,342]

STARDIT is the first system that enables sharing of standardised data about initiatives across disciplines. It enables reporting of who was involved, any impacts of stakeholders' involvement, and outcomes of initiatives over time. This functionality addresses a serious limitation of the current peer-reviewed publication process in which articles are not easily updated. However, there is no single process for making decisions that would improve and refine the processes, language and taxonomies associated with reporting initiatives, including who was involved in which tasks.[343] Similarly, based on feedback from Indigenous community leaders, patient representatives and others, it is essential to ensure access to learning and development opportunities is available to support people to both access and create STARDIT reports. The STARDIT project therefore needs to continually appraise the inclusiveness and effectiveness of its multidisciplinary, multilingual system, including accessibility of interfaces. To achieve this, the project will continue to work with its partner organisations, including the Wikimedia Foundation, a global leader in this field.

The co-design process for STARDIT (hosted by the charity Science for All) ensured people from multiple organisations and countries were involved in both creating and refining STARDIT, ensuring it is usable and relevant in multiple disciplines. Consultation with experts, and source materials from around the world, have informed the design of STARDIT. Co-authors come from disciplines including health research and services, environmental research and management, economics, publishing with over 20 different institutions represented. Future versions should be informed by a regular, systematic search, review and appraisal processes, using the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) data set,[344] used for reporting in systematic reviews and meta-analyses.

While there are multiple methods for mapping values,[345,346] there is currently no agreed, standardised way to map the values (beliefs and personal ethics) of those involved in initiatives and those creating reports in STARDIT. Further research is needed to facilitate mapping of values and detect whether certain perspectives are being consciously or unconsciously excluded.

STARDIT seeks to be an easy-to-use way for people from multiple disciplines to share data about initiatives. However, amassing sufficient reports to create a useful database is estimated to take at least 5 years, and will likely require machine learning. For example, adversarial machine learning may be used in parallel with humans (for verifying data) to generate STARDIT reports from existing publicly accessible data at a scale and speed otherwise impossible for humans alone to achieve.[347] Similarly, the process of creating 'living systematic reviews' from STARDIT reports is currently theoretical and would require significant development and rigorous testing to realise.

It is important to note that access to Wikidata is actively blocked by governments or internet service providers in some countries. While such censorship limits people's ability to contribute or critically appraise data, STARDIT has been designed to be both interoperable with existing standards, and 'future proofed' by being system and language agnostic, to allow interoperability with existing and emerging data systems beyond Wikidata.

Science for All will continue to host the co-creation process and to monitor and evaluate the project. However, an open, transparent governance process that enables anyone to be involved in decision making and ongoing co-design of STARDIT will need to be established, and is proposed in more detail in the supplementary resources.

Ensuring that the STARDIT development process is inclusive and ethical, and that the database is quality assured, is paramount to ensuring that STARDIT is credible, useful and trustworthy. STARDIT currently relies on volunteers and pro-bono services from not-for-profit organisations. In the future, people should be paid for certain tasks, especially if the project is to avoid excluding the involvement of those from lower socio-economic backgrounds who may not be able to afford to volunteer their time. For the success and longevity of this project, a sustainable, transparently decided funding model needs to be established, which ensures both the independence of the data, the hosting process and the governance.

Conclusion

This article summarises work to date on developing Standardised Data on Initiatives (STARDIT), an open access web-based data-sharing system for standardising the way that information about initiatives is reported across diverse fields and disciplines. It provides a way to collate and appraise data about how different people have been involved in different tasks of multiple types of initiatives. The current usage by multiple initiatives demonstrates to usability of STARDIT, and will inform the next stages of development. In accordance with the principles of transparent participatory action

research, the authors invite the involvement of any interested persons in developing and improving the next version of STARDIT, Version 1.0. Detailed and up-to-date information about STARDIT is available on the Science for All website (ScienceforAll.World/STARDIT).[348]

This is the end of this chapter, and the content from the peer reviewed article at this link:[9]

https://doi.org/10.1186/s40900-022-00363-9

Chapter 4 – Public involvement in global genomics research: A scoping review

This chapter is adapted from the published, peer-reviewed 2019 review titled 'Public involvement in global genomics research: A scoping review'.[28] *Please note:*

- *Figure numbers have been changed to align with the format of this thesis, and some formatting has been adapted, including integrating references into this thesis.*
- *This paper uses the term 'facilitators', where later chapters use the term 'enablers' to avoid confusion with those facilitating discussions.*
- *The published version of this article can be found here, along with the supplementary material: https://doi.org/10.3389/fpubh.2019.00079*

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Abstract

Public involvement in research occurs when the public, patients or research participants are actively contributing to the research process. Public involvement has been acknowledged as a key priority for many prominent human genomics research initiatives worldwide. However, to date, there has been no detailed analysis or review of the features, methods and impacts of public involvement occurring in human genomics research projects worldwide. Here, we review the reported public involvement in 96 human genomics projects (initiatives), based on a database of initiatives hosted by the Global Alliance for Genomics and Health, according to information reported on public domain websites. To conduct the scoping review, we applied a structured categorization of criteria to all information extracted from the search. We found that only a third of all initiatives reported public involvement in any capacity (32/96, 33%). In those reporting public involvement, we found considerable variation in both the methods and tasks of involvement. Some noteworthy initiatives reported diverse and comprehensive ways of involving the public, occurring through different stages of the research project cycle. Three notable initiatives reported a total of eight distinct impacts as a result of involving people. Our findings suggest there would be intrinsic value in having more public

involvement occur in human genomics research worldwide. We also suggest more systematic ways of reporting and evaluating involvement would be highly beneficial, to help develop best practices.

Introduction

In human genomics, there is a growing need to increase the involvement of the public in research and policy development, as this has been identified as a crucial aspect of responsible research practice. [21,42] The concept of 'public involvement' in research is defined as research that is carried out 'with' people rather than 'on' them.[260] Public involvement can also be defined as when the public, patients or research participants actively contribute to the research or policy development process.[349]

The number of people involved in genomics research is predicted to grow substantially in coming years.[46,47] By 2025, it is estimated that nearly 2 billion people worldwide will have had their DNA sequenced, creating a global imperative for responsible and effective public involvement in research.[10] Many high-profile genomics research initiatives have already made public statements about the importance of involving people, with some governments positioning public involvement as a democratic right.[32,36,68].

The benefits of involving the public in research are wide-ranging. They include improving trust and public influence over research[21,35,36]; ensuring that research is conducted in an ethical, accessible and transparent manner; and ensuring that research reflects the balance and diversity of priorities within populations.[19,38] However, with the growing interest and importance of large-scale human genomics initiatives worldwide, there has been limited research into how the public are currently being involved. There has also been no assessment of the resulting impacts and benefits, including genomics initiatives that have involved the public.

While involving the public in other types of health and medical research has been the subject of previous systematic reviews[37,78,79], comparable reviews have not been published in human genomics. Many of the existing reviews on other areas of medical research conclude that reports of involvement activities are inconsistent or under-reported[29,37,80–82] and that the precise ways in which people are involved in medical research are not well reported, including any impacts from involving people.[35,79,80]

Our review provides a summary of currently reported public involvement in 96 global human genomics projects, listed on a database managed by the Global Alliance for Genomics and Health (GA4GH), a recently formed international organisation seeking to enable responsible genomics data

sharing within a human rights framework.[350] The list provides a representation of the current landscape of human genomics research worldwide, and a snapshot of contemporary practice with regards to public involvement in genomics research.

This scoping review provides a new perspective by exploring how these genomics initiatives have conducted and reported public involvement to date, including any impacts, facilitators and barriers of involvement. The intention is that resulting data will help inform future directions for integrating public involvement into genomics research and policy development, and inform the development of ways of routinely reporting and evaluating any involvement.

Methods

Source

Using a list of human genomics research projects ("initiatives") from a database hosted by the GA4GH (see Supplementary Materials 'Table 1'), we systematically searched public domain websites for information reported on involving the public in research. The database was curated by GA4GH staff, last verified August 2016, and contains information about the type of the genomics research initiative (i.e. consortium, data-sharing initiative, organisation(s), repository or research project), the type of data gathered (i.e. whole-genome or whole-exome sequencing), the geographical scope of the initiative, number of participants (cohort size), relevant disease areas, and the public domain URL of the website for the organisation or initiative (as some 'initiatives' involve a number of organisations). The scoping review methodology can be summarized in three stages (see 'Figure 4.1: Scoping review study overview and results summary' for overview):

Stage 1 – Defining "involvement" and the search strategy

We developed a criteria to define 'involvement' based on the International Association for Public Participation's participation spectrum and other studies.[36,184,349,351] This included reports of 'consultation', 'involvement', 'collaboration' and 'empowerment'.[352] Involving people in genomics research was defined as the 'active involvement' in shaping and guiding research, rather than only providing data.[81,352,353] We defined specific tasks related to involvement at different stages of the research cycle[260], such as the sharing of views to influence research, or co-creating the research.[29,354,355] 'Consequential' involvement meant involvement contributing to the research process, as distinct from involvement which is ignored or not incorporated.[356–358] We could not

always determine whether involvement was consequential based on the available information, so an assumption was made that all methods reported resulted were 'consequential'.

Stage 2 – Searching websites (data extraction)

Public domain websites of all the initiatives in the GA4GH database were searched for reports of involvement and associated impacts. The date range for website searching and data extraction was 16th August to 28th November 2017. The exact text from the URLs where data was extracted from was collected to allow reanalysis, with all relevant URLs archived using an online archive service to preserve the content and the date of extraction.[303]

We used search engine operators to systematically scan the text of each public domain website for relevant phrases, including all grammatical variations of the words used (for example, deriving 'involvement', 'involves', 'involved' and 'involving' from the root word 'involve'). Grammatical variations of specific phrases (denoted by inverted commas) were generated using tables to systematically create a series of search strings for each domain. For example, this search string returned 4 results:

> *site:www.ukbiobank.ac.uk/* "public involvement" OR "involves public" OR "public involved" OR "involving the public" OR "involve public"

Reports of involvement were assessed by a member of the research team (JN), then independently assessed by an additional member of the research team, with a random sample assessed by a third investigator (PL). Any disagreements between the team on the data included was discussed until a consensus was reached. Informed by previous reviews, the search terms for the concept of involvement were; "engagement", "involvement" and "partnership".[351,359–362] The search terms to describe the people involved were; 'citizen(s)', 'community', 'consumer(s)', 'lay', 'patient(s)', 'public', 'stakeholder(s)' and 'user(s)'.

In addition to using a standard list of terms, adaptive (context dependent) search terms were sometimes required when searching domains where terms were specific to the region or initiative. Adaptive search terms were; 'advocate(s)', 'carer(s)', 'civil society', 'client(s)[363]', 'customer(s)[363]', 'group(s)', 'participant(s)', 'payer(s)', 'population(s)[358]', 'PPI' (an acronym commonly used in the UK which stands for 'patient and public involvement'), 'residents' (geographical grouping)[91],

'representative(s)', 'taxpayer(s)' and 'volunteer(s)'. For more details on search method, see Supplementary Materials 'Table 2'.

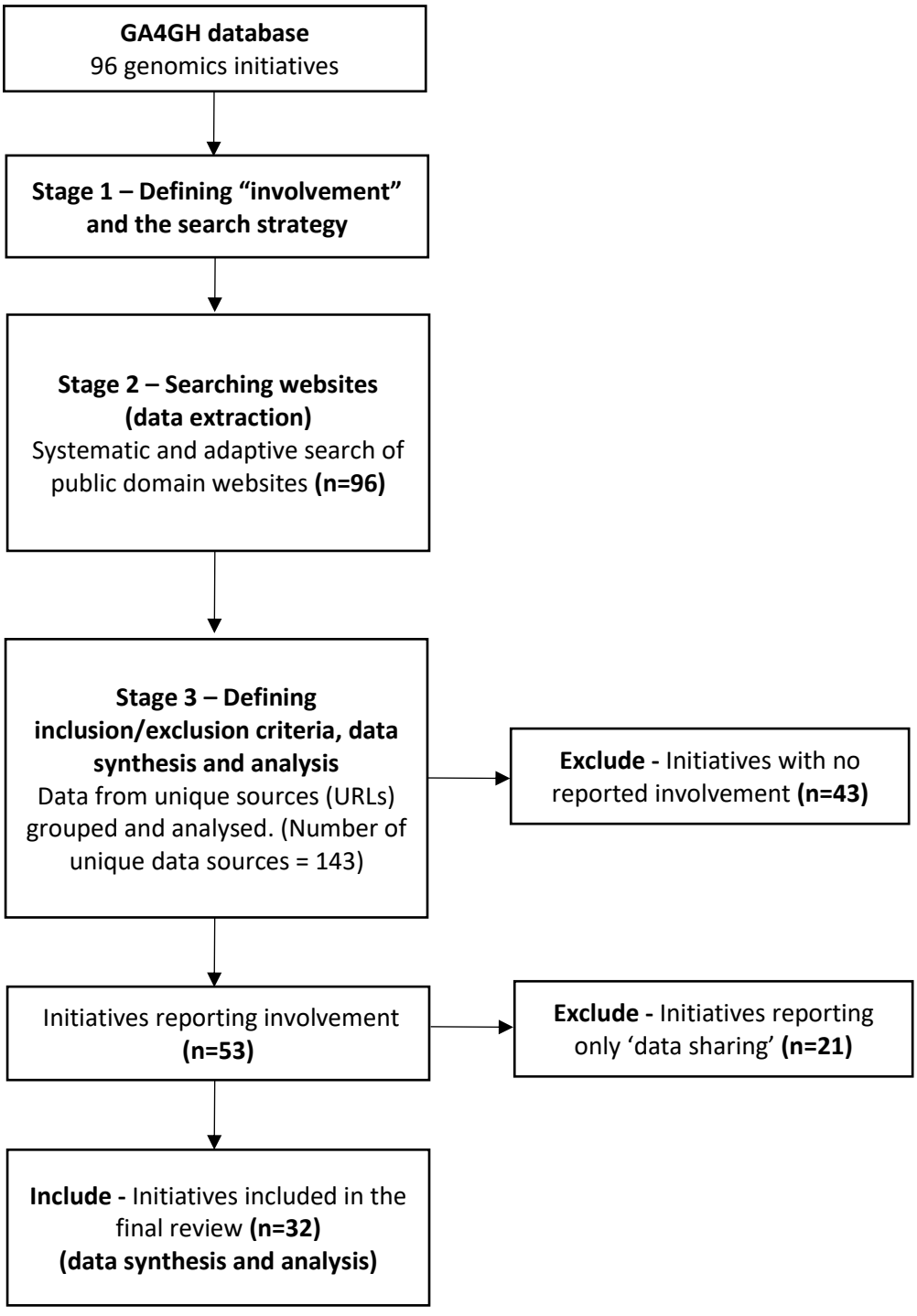
Stage 3 – Defining inclusion/exclusion criteria, data synthesis and analysis

Defining the inclusion and exclusion criteria was an iterative process informed by published scoping review methodologies.[364,365] Initiatives reporting no involvement were excluded from further analysis. Initiatives were categorised as 'no involvement' if the context of words such as 'participation' were used to describe 'research participants' (research subjects) only, rather than aligning with the concept of involvement already articulated.[349,366] Reported impacts were excluded if they were phrased as anticipated future impacts (using terms such as 'we expect'), rather than reporting real results. Initiatives reporting 'data sharing' as the only type of involvement were also excluded. Initiatives reporting any other type of involvement, according to our definition, were included and proceeded to data extraction (structured categorization of extracted search data).

Extracted data was categorized (data synthesis) based on the following types of information; **a**) the *method* of involvement (*how* people were involved)[353]; **b**) the *tasks* they were involved in (what people *did*)[353]; **c**) the *stage* of the research (using an expanded version of an existing framework[37], informed by INVOLVE)[367]; **d**) *who* was involved, for example 'research participants', 'patients' and 'public' (informed by the Concannon '7Ps Framework' taxonomy)[80]; **e**) *reported facilitators or barriers* of involvement; and **f**) publicly-reported *impacts* (informed by section 7 and 8 of the GRIPP2 framework).[80,353]

As there is currently no standardised way to report and group methods of involving people or descriptions of people involved[353,358], grouping was informed by methods of previous reviews (for example, grouping similar methods of involving people[353]) and by using previously established nomenclature.[355,361,366] The initial grouping (JN) was reviewed by other authors (PL). While previous reviews have used frameworks to label the 'roles', 'degrees' or 'levels' of involvement or 'control'[29,353], we did not use these frameworks as they require subjective judgements to be made, often with insufficient data.[355,368–370]

Figure 4.1: Scoping review study overview and results summary



Results

Of the 96 initiatives searched, based on our criteria, only a third reported involving people in some capacity (32/96, 33%) (Table 4.1). These 32 initiatives were included in the final analysis (data synthesis).

Reported methods of involvement

The reported methods of involving people were organised into categories, shown below in **bold**, with the number of total initiatives reporting each method shown in brackets:

- **Citizen science** (n=2) **-** people involved beyond data collection, research design or data analysis, towards co-creation across all aspects of the scientific process[326];
- **Consultation** (n=4) - an organised consultation or dialogue process;
- **Formal discussion** (n=8) - formalised 'focus groups', forums or interview structures;
- **Formal groups** (n=20) - a working group or committee (including ethics and data access committees, 'scientific advisory groups' and 'steering groups');
- **Generic involvement** (n=11) - informal, such as meetings, 'partnership', or an unspecified method;
- **Newsletters** (n=2) **-** or mailing lists;
- **Online tools** (n=7) - websites, social media, or online community hosting;
- **Public events** (n=13) - with discussion - including initiatives hosting public debates, workshops, discussion spaces or conferences;
- **Surveys** (n=10) - including questionnaires; and
- **Other** (n=7) - methods not described by other categories.

Some initiatives reported using multiple methods to involve people. Reports of involving people also showed that some methods, for example 'formal discussion', can use different modes of communication, including face to face, online (for example, 'massive open online courses'), or a combination of the two.

Table 4.1: Summary of G44GH initiatives reporting public involvement

Initiatives from a database provided by the Global Alliance for Genomics and Health were searched for reports of public involvement (based on public domain websites). Each initiative has been assigned an ID number. The type (method) of involvement was categorized using specific criteria.

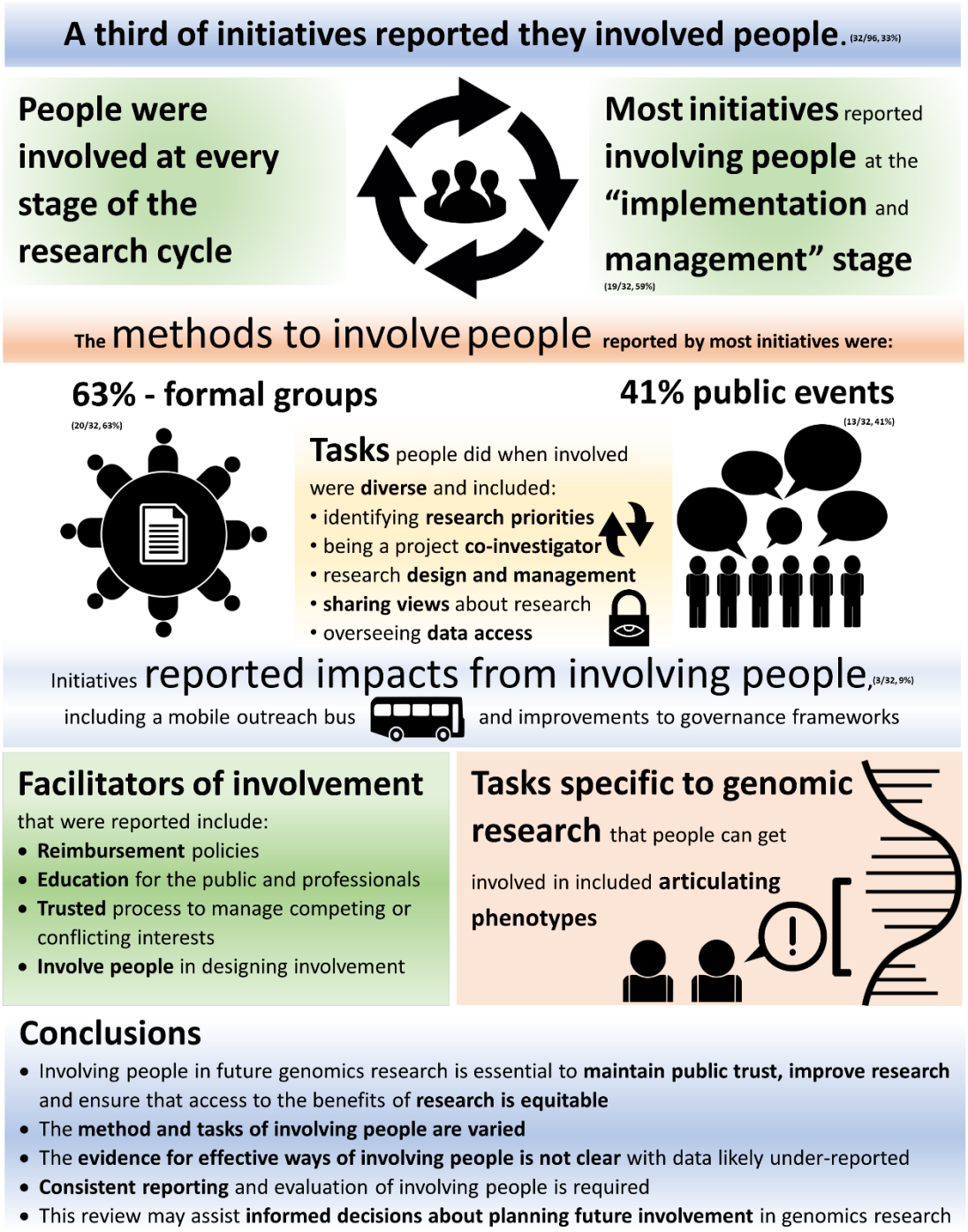
Name of Initiative/Organization	ID	Geographic Region (cohort size)	Reported methods of involving people
100k Wellness Project	**1**	North America (100000)	*Online tools, Other*
Australian Genomics Health Alliance (AGHA)	**8**	Australia (1800)	*Formal groups, Other, Public events*
Biobanking and Biomolecular resources Research Infrastructure (BBMRI)	**11**	Europe (N/A)	*Formal discussion formats, Public events*
Cancer MoonShot 2020	**16**	North America (20000)	*Generic involvement*
Clinical Sequencing Exploratory Research (CSER)	**21**	North America (6000)	*Generic involvement*
DECIPHER	**24**	International (21475)	*Formal groups*
East London Genes & Health	**26**	Europe (100000)	*Formal groups, Generic involvement*
Electronic Medical Records and Genomics (eMERGE)	**27**	North America (55028)	*Surveys*
ELIXIR	**28**	Europe (N/A)	*Consultation, Formal groups, Public events*
France Genomic Medicine 2025	**33**	Europe (N/A)	*Consultation, Generic involvement*
Genome in a Bottle	**35**	International (N/A)	*Generic involvement, Public events*
Genomics England	**37**	Europe (100000)	*Consultation, Formal discussion formats, Formal groups, Generic involvement, Other, Public events, Surveys*

H3Africa	**41**	Africa (60000)	*Formal discussion formats, Generic involvement*
Implementing Genomics in Practice (IGNITE)	**44**	North America (73000)	*Formal groups, Public events*
International Rare Diseases Research Consortium (IRDiRC)	**50**	International (N/A)	*Formal groups, Generic involvement, Other, Public events*
Kaiser Permanente Research Program on Genes, Environment, and Health (RPGEH)	**52**	North America (500000)	*Formal groups*
Kaviar	**53**	North America (N/A)	*Formal groups*
Matchmaker Exchange	**57**	International (N/A)	*Formal groups, Online tools*
MSSNG	**60**	North America (10000)	*Formal groups*
MyCode Community Health Initiative	**62**	North America (250000)	*Formal groups*
MyGene2	**63**	International (500)	*Online tools*
openSNP	**65**	Europe (2500)	*Citizen science, Online tools, Surveys*
Precision Medicine Initiative / 'All of Us'	**69**	North America (1000000)	*Citizen science, Formal groups, Formal discussion formats, Generic involvement, Online tools, Other, Public events, Surveys*
Public Population Project in Genomics and Society (P3G)	**72**	International (N/A)	*Formal groups, Online tools, Public events, Surveys*
Qatar Genome Project	**73**	Asia (1161)	*Surveys*
RD-Connect	**74**	Europe (2500)	*Formal discussion formats, Formal groups, Generic involvement, Newsletters, Online tools, Surveys*

The Clinical Genome Resource (ClinGen)	**84**	North America (N/A)	*Formal groups, Other*
Tohoku Medical Megabank Project	**86**	Asia (150000)	*Formal discussion formats, Public events, Surveys*
Transforming Genetic Medicine Initiative (TGMI)	**88**	Europe (N/A)	*Public events*
UK Biobank	**92**	Europe (500000)	*Consultation, Formal discussion formats, Formal groups, Generic involvement, Newsletters, Other, Public events, Surveys*
Undiagnosed Diseases Network (UDN)	**94**	North America (8000)	*Formal groups*
Vanderbilt's BioVU	**96**	North America (215000)	*Formal groups, Public events*

Figure 4.2 summarizes overall findings from data synthesis. There was variability in the methods and tasks of involvement reported. This supports previous findings that involvement in biomedical research is diverse, varied and described using different language.[371]

Figure 4.2: Summary of global genomics review results



Reported tasks of involvement

The tasks people were involved in (what people did when they were involved) were diverse. Tasks included; identifying research priorities related to people with specific diseases; communicating priorities to scientists, clinicians and health policy makers, [IDs 11, 37, 50, 74]; designing or improving how people will be involved in the research [IDs 41, 50]; educating professionals involved in the research [ID 8]; developing workshops and conferences [IDs 44, 94], offering culturally appropriate information about research to people in community groups [ID 37]; providing feedback on the cultural and linguistic appropriateness of public domain research documents [ID 96]; and translating information into 'lay' language [ID 92]. Tasks also involved sharing views and perspectives about multiple aspects of research projects [ID 37, 92, 96]; articulating phenotypes [ID 65]; and being a project co-investigator [ID 21].

Some initiatives reported involving people in the task of giving feedback and sharing views and perspectives about the 'acceptability' of specific aspects of the research design. For example, research management, governance [IDs 27, 41, 92, 72], accountability, planning, policy, protocols, data access and data use [IDs 37, 74, 84, 92], consent, re-contact, withdrawal, confidentiality, benefit sharing, project closure and recruitment [IDs 37, 62, 74, 92]. A number of initiatives also involved people in the task of sharing views and perspectives on issues of perceived social and ethical importance (including being told about potentially serious incidental findings) [IDs 37, 74, 96], or to scrutinise a project to ensure it aligned with public interest [ID 11].

Reported stages of involvement

Most reports of involvement were at the 'implementation and management' stage of research (19/32, 59%), followed by 'dissemination' (12/32, 38%), 'evaluation' and 'study design' (both 9/32, 28%) and 'data analysis' (8/32, 25%). The stage with the lowest number of initiatives reporting involvement was 'funding' (1/32, 3%) with the next lowest being 'identifying topics' and 'prioritisation' (both 4/32, 13%). Four initiatives reported involving people at every stage of research [IDs 21, 50, 69, 74].

Reported impacts of involvement

Nearly 10% of the initiatives reporting involvement also reported impacts of the involvement (3/32, 9.4%). The method with the most reported impacts was 'public events' (4/8, 50%), followed jointly by 'formal discussion formats' and 'surveys' (2/8, 25%). Actions taken as a result of involving people included the creation of a mobile outreach bus [ID 37]; improvements to ethical and governance frameworks [ID 92]; and improved participant information and consent documents [ID 37]. Three

initiatives reported a total of eight distinct impacts as a direct result of involving people. Some impacts were reported as being a result of using a combination of methods.

Reported facilitators and barriers to involvement

A number of specific facilitators of involvement were reported, including: reimbursement policies [ID 21], with people involved paid for their time [IDs 92, 94], travel [IDs 74, 94] accommodation [ID 74] and expenses [IDs 74, 92]; education and learning opportunities for the general public [IDs 1, 11, 41], ensuring people involved are informed and can make informed decisions [ID 11]; education for health professionals [IDs 41, 50], providing opportunities to learn about how to involve people [IDs 41, 50], and governance which is trusted by all stakeholders to be able to manage real or perceived competing or conflicting interests [ID 50]. The only barrier reported was limited venue size, which inhibited the number of people involved [ID 92]. This also implies a limited budget, which is an important but likely under-reported implicit limitation on all involvement methods.

Discussion

This review provides an overview of reported public involvement occurring in prominent human genomics projects worldwide, during a period of rapid growth for genomics research. We identified significant variability in the way in which involvement is reported. This variation of reported involvement suggests diversity in both the ways people are being involved in genomics, and in the varied and emergent language used to report and describe involvement, consistent with other areas of biomedical research. [36,372] While there are similarities with the principles of involvement in other kinds of biomedical research, this review has identified three different tasks not found in other reviews.[371]

Because the results from this review suggest there is currently no standardized way of reporting and therefore evaluating how people are involved, there is a risk that best-practice will be hard to define or even absent in future evidence reviews. This has implications, as the number of people involved in genomic research is predicted to grow exponentially. Without a standardized framework to report and transparently evaluate ways people are involved, it will be difficult to create an evidence base to inform best-practice.

While a third of initiatives reported involvement, a majority of projects did not (64/96, 66%). Some prominent initiatives involving the genetic analysis of thousands of people did not refer to public involvement in any way. This is somewhat concerning given that involving the public has been identified as a crucial aspect of responsible research practice in genomics.[21] Whilst we acknowledge

the probable under-reporting of involvement activities on public-domain websites, we argue public involvement in human genomics needs to increase.

Findings from this review also suggest it is best-practice to involve multiple stakeholders (including the public) in designing how people will be involved in research (co-design of involvement plan), to involve the public throughout the lifetime of a project in certain tasks (such as overseeing data access) and to evaluate the involvement with both qualitive and quantitative data.

Involving people in planning involvement may improve how appropriate, effective, efficient and equitable it is. Involving people in the design of planned methods of involvement by identifying what is considered 'good practice' was reported by H3Africa [ID 41] and the International Rare Diseases Research Consortium (IRDiRC), and reported as a facilitator of involvement by the IRDiRC [ID 50]. The IRDiRC [ID 50] also reported both qualitative and quantitative data should be used to evaluate involvement, although there is currently no way to systematically collect and analyse such activity.[130]

If involvement is more effective when the public are invited to help plan it, standardised reporting and evaluation will help make informed decisions at every stage of involvement from co-design through to evaluation.

Implications for policy and practice

With the impact of some genomics research data likely to be measured in decades, some of the initiatives offer a useful insight into planning and funding sustainable (long-term) involvement for the entirety of an initiative's lifetime.[68] For example, Genomics England [ID 37] and the UK Biobank [ID92], as exemplars, both reported multiple ways of involving people, at different stages of the research cycle, conducted over a number of years. Other initiatives, such as the International Rare Diseases Research Consortium (IRDiRC) [ID 50] and the Public Population Project in Genomics and Society (P3G) [ID 72], also publicly stated the importance of planning sustainable involvement over the duration of a project. These initiatives demonstrate a standard of involving people which could eventually be used to inform international best practice.

The IRDiRC also reported that involving people throughout an entire project helped maintain trust by scrutinising and managing competing or conflicting interests [ID 50]. Similarly, the UK Biobank reported that involving people in the ethics and governance should not be one-off and must be

ongoing [ID 92]. The method of using 'formal groups' was more common for more complex or ongoing tasks such as overseeing data access, policy development, research management and improving research protocols.
Some initiatives, such as openSNP, reported tasks that were specific to genomics research, such as articulating phenotypes [ID 65]. Involvement in this kind of task might have important implications when working to usefully describe people's subjective lived-experiences across multiple languages, for example, rare diseases and mental health.[373]

Public involvement in articulating phenotypes also suggests that the traditional boundaries between terms such as 'research', 'healthcare', 'patients', 'research participants' and 'the public' may be increasingly challenged by the methodology of future genomic research.[374] Findings from this review show that both 'the public' and 'patients' are already involved in every stage of research, including collecting and analysing data.[374] Any future standardised reporting of involvement will need to keep pace with the continually evolving language to describe not only what research is, but who is involved and how.

Limitations

While the database hosted by GA4GH includes many of the most prominent human genomics research initiatives worldwide, the database is not exhaustive. There are several known genomics initiatives which involved people that were not part of the database. Therefore, the GA4GH selection cannot be considered entirely systematic or representative. However, it does provide a reasonable indication and snapshot of the current global landscape in human genomics research.

Our data collection was limited to self-reported information reported on English language websites only. This likely under-reports the total amount of public involvement occurring. For example, some initiatives may have conducted involvement, and not reported it publicly. This indicates a current lack of standardization or best-practice in reporting involvement activities in human genomics research, which we feel could be improved.

Of the public involvement activities reported, we did not systematically follow up reports to confirm they had taken place, or if involvement was 'consequential'.[356–358] While this is a limitation of the review, it also reflects the inconsistent and often incomplete ways genomics research initiatives report impacts of involving people. For example, the impact of how involvement influenced research

was only reported by three projects - Genomics England [ID 37], the Qatar Genome Project [ID 73] and the UK Biobank [ID 92].

A number of reported methods did not provide sufficient information to make a clear decision about how to group a method. For example, many reports of involvement simply referred to a 'workshop', 'meeting', or other 'public events', where people were able to get involved by sharing views and perspectives. As a result there is potentially significant overlap between some methods, which could have been articulated more clearly if more data were available.

Reports of 'data sharing' were excluded, as they were not considered as public involvement. While sharing data may enable people to be involved in some capacities (for example, in analysing data), data sharing is not necessarily an indicator that people were involved in the analysis of data. The complexity within the term 'data sharing' in genomics, and how people can be involved in the analysis and interpretation of data also requires further consideration.[123,375,376]

Conclusion

Involving people in the future of genomics research is essential to maintain public trust, improve research outcomes, and to ensure that access to the benefits of research is equitable.[21,79,374] While a third of initiatives reported involving people, only 10% of initiatives reported impacts. The limited reporting of involvement suggests there would be intrinsic value in developing a more systematic method of both reporting and evaluating how people are involved in human genomics research. Data from such reporting could provide the evidence required to inform future policy around involvement of the public, as human genomics research continues to grow.

Supplementary Material

1. '**GA4GH database and results table'**: This table combines a database of human genomics research projects hosted by the 'Global Alliance for Genomics and Health' with a summary of results from a scoping review of currently reported public involvement reported on public domain websites. The data is organised alphabetically, with organisations reporting involvement listed first. A direct link to Additional File 1 can be found here: https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_1_xlsx/octet-stream/Table%201.XLSX/1/446268

2. **'Systematic search method':** This document describes the search method, including how standard and adaptive were used to search the public domain websites of all the included initiatives in the GA4GH database for reports of involvement and any impacts. A direct link to Additional File 2 can be found here: https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_2_docx/octet-stream/Table%202.docx/4/446268

This file is also included in the Appendices.

This is the end of the chapter adapted from the published, peer-reviewed 2019 review titled 'Public involvement in global genomics research: A scoping review' at this link: https://doi.org/10.3389/fpubh.2019.00079

Chapter 5 – Involving elderly research participants in the co-design of a future multi-generational cohort study

This chapter aligns with research aim 2: "apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research"

*This chapter is adapted from the published, peer-reviewed 2021 article titled '*Involving elderly research participants in the co-design of a future multi-generational cohort study'.[6]

Please note:

- *Figure numbers have been changed to align with this thesis, and some formatting has been adapted, including integrating references into this thesis.*
- *This artcile uses the term 'enablers', to avoid confusion with those facilitating discussions, where previous chapters used the term 'facilitators'.*
- *The published version can be found here, along with the additional files:*[6] https://doi.org/10.1186/s40900-021-00271-4

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Plain English Summary

Research which follows a population of people over time is a valuable way to learn about what contributes to both wellness and disease. Some studies of this kind look at multiple generations and such studies have had significant positive impacts on public health. However, such studies are challenging to establish and expensive to maintain.

It has been proposed by Paul Lacaze, an investigator on the ASPirin in Reducing Events in the Elderly Extension study (ASPREE-XT), that the study would be a good basis for a future multigenerational research study (MGRS). There is evidence that involving potential participants in co-designing research can improve the quality of the study design, recruitment and acceptability of the research.

A team of people including a current ASPREE-XT participant were involved in planning how potential participants would be involved in the co-design process. An advert was sent to 14,268 participants of the ASPREE-XT observational cohort study to invite them to be involved in the process of planning a future multi-generational research study.

Fifty-nine ASPREE-XT participants were interviewed by telephone and 18 attended a face-to-face workshop event. We used a newly developed standardised format to plan and report how participant involvement activities positively impacted the study design (Standardised Data on Initiatives - Alpha Version 0.1).

Involving participants positively impacted the proposed study design by improving the research objectives, developing protocols, influencing funding decisions and improving ethics applications. This case study provides evidence that including participants in the design of a research study positively impacted the study design, participants and researchers.

Abstract

Objectives: To report as a case study the process of involving study participants and how this impacted the co-design of a proposed multi-generational research study, using a novel standardised reporting tool.

Background: An advert was sent to 14,268 participants of the ASPREE-XT observational cohort study to invite them to be involved in co-design and planning of a future multi-generational research study. Participants were aged 74 years and older from Australia, located in both metropolitan and rural locations.

Interventions: We used participatory action research to involve elderly research participants in the co-design of a proposed multi-generational cohort study between 2017-2019 using newsletters, telephone interviews and an in-person workshop. Primary and secondary outcome measures: We used the 'Standardised Data on Initiatives (STARDIT)' Alpha Version to plan and report how participant involvement activities positively impacted the study design.

Results: Fifty-nine ASPREE participants were interviewed by telephone and 18 participants attended a face-to-face workshop event. Involving participants positively impacted the proposed study design by improving the research objectives, developing protocols, influencing funding decisions and

improving ethics applications. Learning points included the importance of maintaining the ideals of the ASPREE study (respect, quality and transparency); research participants' preference for the option of receiving results (including genetic results); participants' need for involvement in decisions about recruitment, data access, governance and other ethical issues; and the preference for different communication methods, including both face-to-face and online methods. The process was highly valued by all stakeholders, including research participants, study staff and lead investigators. Involvement of participants was described by the lead study investigator as "enormously helpful".

Conclusions: This case study provides evidence that including participants in the design of a research study positively impacted the study design, participants and researchers. Using a standardised reporting tool to describe the methods and impacts provides a way for learning from this case study to inform future research studies planning to involve people.

Introduction

The ASPirin in Reducing Events in the Elderly (ASPREE) trial (2010-2018), a randomised controlled trial for aspirin in healthy older people, recruited 19,114 participants from Australia and the USA[377–380]. The median age at recruitment was 74 years. The ASPREE Healthy Ageing Biobank is a sub-study which has collected biospecimens from over 15,000 ASPREE participants, alongside detailed medical records, lifestyle, cognitive function and physical testing data. [381] ASPREE researchers are conducting various types of genomics and biomarker research. ASPREE had a remarkably high retention rate, with only 1.2% of participants withdrawing from the study and 90% still attending annual visits after an average of 4.5 years follow-up.[382,383]

ASPREE XT is a follow up observational study that was established in 2018 to continue to collect data from ASPREE participants for another 5 years. ASPREE-XT participants are over the age of 74 (defined as elderly[384]) and Australian participants are located in both metropolitan and rural locations. Multiple stakeholders, including a participant advisor, proposed a new multi-generational research study (MGRS) which could recruit direct descendants of ASPREE-XT participants, as part of a longitudinal observational study alongside their relatives for two generations or more. MGRS are challenging to establish and expensive to maintain, yet their value to medical and epidemiological research is significant. ASPREE-XT has been proposed as a platform for a multi-generational study. Recruiting ASPREE-XT participants and their descendants to a MGRS would provide a large starting population by international standards.

Previous multi-generational, longitudinal cohort studies have had significant positive impacts on public health. Examples include the Framingham Heart Study,[385] Lothian Birth Cohort[386] and Dubbo Osteoporosis Epidemiology Study,[387] all of which had substantially lower starting populations than ASPREE-XT. [388] ASPREE-XT provides a rare opportunity for such a study in Australia, already combining high-quality medical record data with genomic data on a large number of elderly Australians. [388] Additionally, the cohort is already well engaged, with all surveyed participants supportive of a proposed MGRS.

The aim of this study was to report as a case study the processes of involving ASPREE-XT participants in the co-design of a proposed multi-generational research study, and how this impacted study design. The research objectives were to involve potential participants in the co-design of a new MGRS; plan the process in a standardised way; identify themes and preferences; and evaluate then report the process using a standardised reporting tool.[196]

ASPREE-XT's unique focus on healthy ageing adds substantial value to public health, medical, epidemiological and geriatric research. Previously collected genomic (including epigenetic) data, combined with detailed medical data and ongoing cognitive assessment, allow that a MGRS with the ASPREE-XT cohort would be of considerable value to science for decades to come, for example in dementia research.[389] The proposed study would examine health outcomes in a large, well-monitored cohort; and provide data to help inform our understanding about the genetic and environmental determinants of health and disease, across multiple generations. However, this is only possible if people choose to participate.

Many clinical research studies are underpowered due to poor recruitment and retention[390]. Involving participants and the public in research design has been shown to improve the recruitment [207,391], quality and relevance of research. [28,392] The concept of involvement is research being done 'with' people rather than 'on' them. [260] Involving the public, patients, research participants and other stakeholders in actively contributing to the research process can lead to a range of positive impacts and outcomes. [349]

In human genomics research, the need to involve the public and other stakeholders is a crucial aspect of responsible research practice. [21,28,42] The term 'stakeholder' here means anyone who has a 'stake' in the research, in particular those with important knowledge, expertise or views that should be taken into account. [21,31] This includes ASPREE-XT participants, study staff and academic research

investigators and the wider public. At the earliest stage of the research cycle (the conceptual stage), some current ASPREE-XT participants were invited to be involved in the co-design of a new MGRS. In this paper we aim to outline how people were involved in the co-design process in order to appraise the methodology and inform future best-practice.

Terminology

We have used consistent language in this paper to describe concepts such as 'involvement'. To aid readers, Table 5.1 provides definitions of important terms used consistently throughout this paper.

Table 5.1: Definitions of terms

Involvement – The words **'involvement'** or 'being **involved'** describe the concept of people being 'involved' in research. This is when research is carried out 'with' people rather than 'on' them. [260] 'Involvement' can also be defined as when other people aside from the research team, such as the public, patients, research participants and other stakeholders, actively contribute to the research process. [349] It is the 'active involvement' in shaping and guiding research, rather than only providing data. [81,352,353]
Participant – a person who participated in the process of sharing views and perspectives about the proposed MGRS, including sharing views about preferences for any future involvement. The term 'ASPREE-XT participants' will be used when specifically referring to participants from the existing study.
Participant advisor – before inviting people to become participants, it was necessary to involve a small number of participants to help advise and plan the process. These participants were chosen from an existing reference group of ASPREE-XT participants.
Stakeholder – this term includes anyone who has a 'stake' in the research, in particular those who have important knowledge, views or perspectives that should be taken into account. [21,31] In this paper it refers to participants, participant advisors and ASPREE-XT study team members (including researchers, ASPREE-XT participant assessors and lead investigators).
Study team – this process was guided by the study team, who consisted of academic researchers, ASPREE-XT participant assessors, a participant advisor and a lead investigator.
The process – this term will be used to describe the process of involving ASPREE-XT participants by inviting them to share views and perspectives about a potential future MGRS. This process includes the co-creation of this case study with participant advisors.
Participatory action research (PAR) - this term describes a number of related approaches, including forms of action research which embrace a participatory philosophy and include 'co-design' and 'co-production' of research. [88(p1)]

Materials and Methods

Study design

A participatory action research (PAR) method was chosen to guide the process with co-design and reporting guided by a number of frameworks. [80,91,127,393,394] Participatory action research is an umbrella term which describes a number of related approaches, including forms of action research which embrace a participatory philosophy and include 'co-design' and 'co-production' of research. [88(p1)] During the study design, we applied this co-design process where researchers and other relevant stakeholders (including research participants) "work together, sharing power and responsibility from the start to the end of the project",[89] including knowledge generation and translation. [89,330] Figure 5.1 summarises the process we used (Fig 1). The ASPREE participant advisor was an integral member of the study team, through each stage.

Guiding Frameworks

The process was guided by a number of international participatory action research methodology frameworks[89,185,395], including the International Collaboration for Participatory Health Research.[130] The participatory action research method was also informed by an international review of involvement in genomics research carried out by some members of the study team[28]. Learning from this review informed the subsequent development and application of an Alpha version of 'Standardised Data on Initiatives' (STARDIT).[196] STARDIT includes a tool to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. STARDIT was then used to guide co-design of the process, and to subsequently report how people were involved, using standardised data. An Alpha version of the STARDIT framework was also used in parallel with the thematic analysis to organise data into pre-defined 'super-categories' which allow consistent comparison with other data using this reporting framework.[196]

We used a case study research methodology to record and describe the process of involving participants in the co-design of a proposed MGRS. Case study research methodology is a form of empirical inquiry and this study is presented as an instrumental case study, where the purpose is to understand the particular case and to provide data that could produce useful generalisations by using inferences from the data. [163(p109)]

The selection of the population for this case study was informed by a number of factors which were appraised by the study team, including ethical, pragmatic and population considerations.[178,396] One

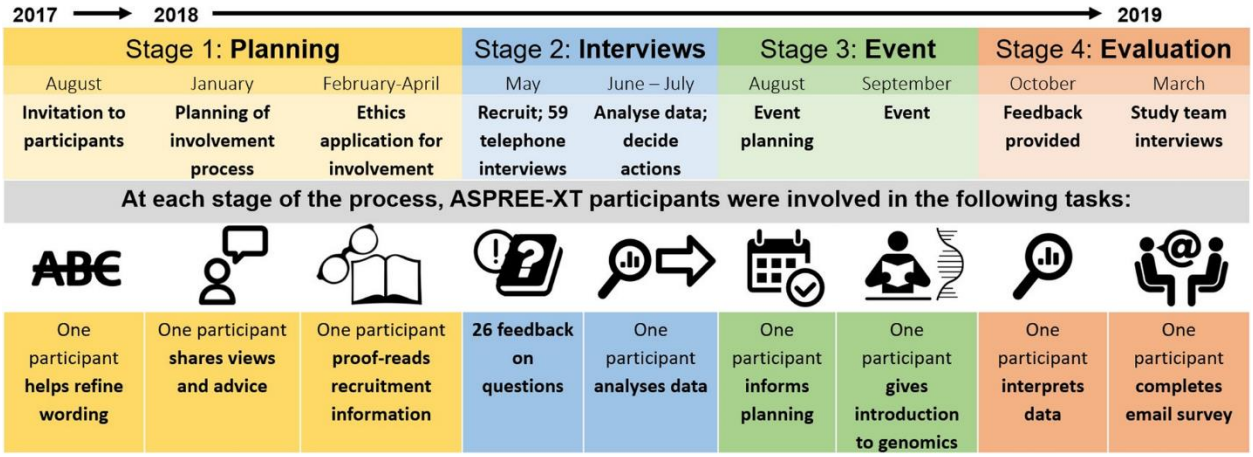
of the study team members (PL) is an investigator with the ASPREE-XT study, which was used as a starting point by the study team to explore the appropriateness of this case study.

Data collection and reporting

The data collection and reporting was informed by a number of frameworks for reporting involvement in research.[21,80,91,183–185,196,397] Qualitative and quantitative data was collected from multiple sources, including meeting notes, audio recordings, documents, emails and surveys. data sources were analysed using a number of methods including thematic analysis. Further information about how data sources were analysed is in Table 5.2.

Stages of research

Figure 5.1: Process timeline of the participant involvement



Stage 1: Planning

The study team held four meetings to co-design the involvement activities. One participant advisor was involved in a number of tasks including reviewing and improving the written information, telephone interview questions, and the facilitation plan for the event.

Stage 2: Recruitment and telephone interviews

An advert was placed in a newsletter to 14,268 ASPREE participants. The process we used for recruiting and involving ASPREE-XT participants has been divided into four chronological stages, summarised above in Figure 5.1.

After the recruitment and consent process, participants were interviewed by telephone. A summary of the questions and responses from the telephone interviews is shown in Table 5.3. For a list of all questions asked, see Additional File 2 in the Appendices. Participants were asked about their

willingness to provide feedback throughout the study, and to be involved in study design, as well as preferences for modes of communication. The definition of involvement below used in the script was co-designed with participants for subsequent interviewees, with versions 1 through 1.3 piloted with 15 participants.

Pre-question script: 'Traditionally, research studies have been designed and conducted only by researchers, and people invited to participate. We are challenging this idea of researchers being the only experts.'
Question: Would you be willing to provide feedback and share your views and perspectives by commenting throughout the research process?'

Twenty relevant interviews were transcribed, coded and categorised (JN), with relevant interviews identified by two investigators independently (JN, PL). [182] To reduce any unconscious selection bias, a sample of over 10% of the interviews was selected at random.

Stage 3: Event

Eighteen participants attended a four-hour workshop event in central Melbourne, led by JN, who is an experienced facilitator. The event was co-designed by the study team, and was informed by interview data and international best-practices for involvement events. [89,398]

The event included an introduction to the proposed MGRS by the lead ASPREE-XT genomics researcher (PL); a plain-English introduction to genomics by an expert in genomics who is also an ASPREE-XT participant (BH); a summary of the telephone interview results by the interviewer (MS); and an interactive session which included open questions about the types of information participants would like returned and recruitment of family members (JN).

The final session included a presentation and interactive discussion about involvement in research, led by the event facilitator (JN). This session explored preferences about how people would like to be involved, with open and closed questions. Questions included preferences about tasks and modes of communication.

Throughout the event, participants shared their views on a range of issues through interactive discussions, voting (by show of hands) and anonymous written feedback.

Stage 4: Evaluation and analysis

Table 5.2. Summary of data collection and analysis

Data source description	Analysis method
Meetings – including meeting notes, recordings and relevant documents	**Qualitative** (content analysis)
Telephone interviews – including recordings, notes made by interviewer, summary documents and related emails	**Qualitative** (thematic analysis, content analysis) **Quantitative** (number of responses)
Event – including audio and video recordings, interviews, written notes and feedback forms	**Qualitative** (content analysis) **Quantitative** (number of responses)
Study team surveys – responses to open ended questions by email	**Qualitative** (content analysis)
Other data – this included reflexive research diaries, relevant emails, financial and other relevant documents	**Qualitative** (thematic analysis) **Quantitative** (analysis of cost)

Evaluation and reflection

All members of the study team (except JN) were surveyed six months after the face-to-face event in order to integrate the valuable views and perspectives of those involved in co-designing and delivering the process. Design of surveys was informed by frameworks for planning and reporting public involvement.[80,91] The study team were asked 11 questions (available in Additional File 1 in the Appendices) and the data from the four interviews was coded and categorised (JN) using STARDIT.[196]

Data analysis

The stages of qualitative data analysis included data mapping and familiarisation; transcription; coding; searching for themes; reviewing themes with study team members; labelling and summarising themes; and reporting the findings.[182] In order to enhance validity of the analysis, two authors (JN, MS) independently analysed the data thematically, which was then checked (triangulated) by a third author (PL).[153,182,399] Standardised categories (STARDIT) were used during content analysis of the data in order to facilitate comparison with other research projects.[196] More information about the data sources and a STARDIT report available is in Additional file 1 in the Appendices.

Results

The results are presented in three sections. Section one provides results from the stages outlined in Figure 5.1, section two shares the perspectives of ASPREE-XT participants and other stakeholders, and in section three we describe how participant involvement impacted study design in seven ways.

We quote participants directly, assigning each a unique number. Figure 5.2 summarises the entire process, results and impacts. For quantitative data from the telephone interviews, see Additional File 3 in the Appendices.

Section 1: Results from stages 1-4

Results from Stage 1: Planning

The input of the participant advisor during the early co-design stage had clear benefits, in particular in identifying the best approaches for engaging the broader ASPREE population; improving the wording of participant information resources and improving question design for the interviews.

Results from Stage 2: Recruitment and telephone interviews

After reading the advert in the newsletter, 76 ASPREE-XT participants agreed to participate in the process. We interviewed 59 people by telephone, with the remaining 17 not responding to a number of follow-up calls.

Interview participants expressed a strong interest in receiving results from genetic research. The strongest preference was for genetic results of medical significance (rather than ancestry or drug-response).

All telephone interview participants expressed willingness to provide feedback throughout the process of planning the proposed MGRS. While most participants understood the concept of involvement, 9 of the 20 analysed expressed self-doubt about how they could be involved. Of 20 analysed telephone interviews, five participants seemed unclear about what tasks they could be involved in, suggesting the concept of participant involvement was new to them. Six participants sought clarity about what was expected of them when being involved, or discussed the skills and knowledge required. Further, participants stated that the goals and processes of involvement needed to be clear in order to avoid “spending too much time” and were willing to be involved as long as the task had purpose and was not “just for the sake of chatting” (P1).

During the telephone interview stage, the interviewer initially “struggled to explain” the concept of involvement (as distinct from participation in an active study) (MS). Co-designing and making changes to the language used in the telephone interviews appeared to improve participant understanding of the concept of involvement. Changes included providing a clear definition and pre-question script and including actual examples of involvement (such as overseeing ethical decisions about data access), thereby avoiding jargonistic abstract concepts such as ‘involvement’.

Table 5.3: Summary of data from telephone interviews

Questions (closed)	Results	% of interview participants
1. Do you think participants like yourself should be involved in helping design research projects, or just researchers?	Just researchers	39
	Participants involved	46
	Unsure	11
2. Would you be willing to provide feedback and share your views and perspectives by commenting throughout the research process?	Yes	100
	No	0
3. Would you like to be more involved in study design rather than just participating?	Yes	65
	No	12
	Unsure	23
4. Would you be more or less likely to participate in research if participants were involved in design	More likely	25
	Less likely	0
	Unsure	13
	Wouldn't influence participation	13
5. What is your preferred mode of communication for being involved	Face to face event	15
	Post	31
	Online discussion	3
	Online questionnaire	15
6. If genetic research results were made available, which types of genetic testing would you be interested in? *	Medical	97
	Ancestry	97
	Drug response	94
7. If only one option for genetic testing was available, which one would you prefer?	Medical	65
	Ancestry	12
	Drug response	6
	No preference	18

*Note that percentages indicate the number of participants who responded with that response. On question six, participants could select multiple answers, so totals do not add up to 100%.

Results from Stage 3: Event

Eighteen of the invited 76 participants attended the face-to-face event. When specific tasks were discussed at the event, 10 participants expressed interest in being involved in recruitment and communication tasks, 7 in data access decisions, and 11 in ethical decisions. When surveyed after the event, the study team reported that involving participants positively impacted the proposed study design by improving the research objectives, developing protocols, influencing funding decisions and improving ethics applications. Videos of the event discussions and interviews with participants will be shared in the public domain.

Results from Stage 4: Evaluation and reflection

Participant feedback from the workshop event was analysed, with participants reporting it as a positive experience, one describing it as "brilliant" (P7). When surveyed, all members of the study team reported that the event achieved its intended aims and was a positive experience. No negative impacts were reported from any participants or study team members at any stage of the process.

One study team member described that through involving people, their perceptions "significantly changed" as they learned how participants could have "valid, interesting, and often novel ideas that researchers may not have considered". The lead investigator (PL) noted:

> "What I learnt is that sometimes the researchers' assumptions about certain things can be off or even incorrect, and that researchers can miss critical points that are important to participants and the study".

During the process, both participants and study team members reported changed views about the value of involvement in research, demonstrating 'transformative learning' and co-construction of knowledge. [130,131,267]

A number of significant learning points were identified by the study team when responding to the question "do you have any advice to other researchers planning involvement for their research". Significant learning points are summarised in Table 5.4.

Table 5.4: Summary of learning points from study team

Summary of learning points from study team
1. Fund and prioritise involvement, make it a requirement
2. Ethics processes take time, but can improve plans
3. Know your audience – don't make assumptions
4. Value diversity in experience and knowledge
5. A supportive team improves the experience for all

Cost and value

The entire process of involving people was estimated to cost $10,000 AUD, including staff time, catering and event venue hire. The value of the process was summarised by the lead investigator who stated "I learnt a lot from the process and am very glad we made the effort".

Section 2: Perspectives of ASPREE participants and other stakeholders

Recurring themes were identified by the study team from the data sources analysed, and seven specific impacts were reported as a result of involving participants (summarised below). The analysis is divided into two sections: (1) participant views about the proposed study, (2) stakeholder views about involvement in the proposed study.

1: Participant views about the proposed study

Participants were very positive about the proposed MGRS, with one stating 'I think it sounds very good'[P20]. Altruism was a primary motivator for participation, with participants suggesting that outcomes of a MGRS could include benefits to themselves (personally and for their families); improving healthcare more generally; and the potential for saving lives, preventing diseases, improving quality of life, and improving future research.

Participant views on types of information they would like returned from genomics research and how the study should be funded (commercial funding versus public funding) were diverse and did not always align with the study team's prior expectations. For example, two thirds of event participants wanted access to their own genomic data, which was described by the lead investigator as 'very at odds with the current system' (PL).

2: Stakeholder views about participant involvement in the proposed study

Participants were supportive about being involved, with all participants supportive of being involved by providing feedback throughout the research process (100%, 32/32), with a typical participant

response being ‘I’d be happy to be involved’ [P3]. Views about enablers were shared in three of the 20 interviews coded, by all 18 of the event participants and all study team members surveyed. Views about barriers were shared in eight of the interviews coded and by half of the study team surveys. Further data is categorised in Table 5.5. Additional mapping of preferences for involvement using the STARDIT-PM tool can be found in Additional File 1. [196]

Table 5.5: Enablers and barriers for involving participants

Enablers	Quotations	Barriers	Quotations
Financial remuneration for people's time; financial support for travel and accommodation	Running a business 'limits me and my time' [P15]	Living in rural areas and travel logistics a barrier to participation in face-to-face events	'I'd like to be involved online rather than face to face because of travel difficulties' [P6]
Learning and development opportunities for participants and researchers	'if you tell me what would be useful, I could do it' [P13]	Poor explanation of abstract concepts such as 'involvement', which can be jargonistic	'there seemed to be confusion, or a lack of understanding of what this 'involvement' would actually look like' (MS)
Small groups at events gave more people a chance to share perspectives	'Small group' discussions at the event 'worked well' (Study team member)	Self-doubt about their skills or knowledge mean they don't think they could be involved	'I'd probably ask a stupid question' [P2]
Early notice of events	'give me enough notice' [P10]	Some people not comfortable being part of a face-to-face group	'I get very uncomfortable in a group of people' [P12]
Clear information about timings and time commitments, frequency of involvement and available support	Ensure 'people are advised what's going to happen at the workshop' [P8]	Lack of clarity about expected time commitments	Being involved 'depends on what's involved and time' [P1]
Clear information about purpose and expectations of involvement, feeling their involvement has consequences	'What's the endpoint – what's the goal?' [P11]	Unclear about what tasks they could be involved in	'I don't know how I could but willing to help' [P13]

Enablers	Quotations	Barriers	Quotations
Independent facilitator when working in groups (either face-to-face or online)	I'd feel more comfortable if I had someone who was facilitating [P14]	Face to face discussions 'dominated' by more confident or knowledgeable people (Study team member)	'Participants from professional backgrounds to some extent dominated some of the discussions' (Study team member)
Short events ensure people do not get fatigued	'any longer and I think fatigue would have dampened the enthusiasm' (Study team member)	People feel they have limited time, are busy with work or social commitments	'I don't have a lot of time left in life' [P1]
Having access and literacy in using computers and online tools	'if I could negotiate [online discussions] I'd be happy to do that' [P15]	Lacking access, literacy or trust in using computers, smartphones or online tools (including social media)	'I'm hopeless with computers' [P16], 'I don't have internet' [P12]
A selection of flexible communication modes (such as face to face and facilitated online discussion forums)	'I'd be happy to be involved – more online but if there was an occasional need to come into the city I'd be happy to do that' [P3]	Only one mode of communication, such as expecting people to travel to events	'online is often easier', face to face only 'as long as it's not too far' [P22], 'travel distance is an issue' [P20]
Involving people in research ethics and governance	'ethics is the difference between right and wrong – you know what's right and you don't do what's wrong' [P18]	A 'researchers know best' attitude that doesn't value the process of involving people (Study team member)	Researchers 'don't see the forest for the trees' [P19]

The lead investigator stated “the feedback has been so valuable” and that it will be “built into the design” of future research. However, the study team also identified barriers to involving people which exist for researchers. One study team member reported that at the start of the project they were “unsure how people without a science or health background” could be involved. They reported a personal shift during the involvement process from not understanding how participants could be involved and being concerned about “asking too much” to believing that, “with adequate resources (financial, training, time...), people can be involved in all aspects of genomics research”.

Other barriers identified by the study team included delays in obtaining ethical approvals for involving people and the cost of involvement in both time and money. One study team member reflected in the follow-up survey that they had “worried too much” about the time-burden of involving participants. The concern of “not putting further pressure” on participants was a theme in email communication between the study team when making decisions to limit contact with participants throughout the process of involving them.

Survey data from the study team also suggests that adequate funding, a supportive team, involving participants in the very earliest stage of research planning and co-designing inclusive methods of involvement all contributed to the impacts reported.

Section 3: Impacts on study design from stakeholder involvement

Involving stakeholders in the co-design process impacted the study in seven specific impacts ways. By asking for participants’ views on aspects of the proposed study design, the study team gained insight into participant preferences and opinions. While there was diversity in views, the process allowed the study team to improve aspects of the study design. These impacts are summarised in Table 5.6.

Table 5.6 ‘Summary of impacts on study design’

Impact on planned research	Summary of impact
1: Recruitment and sample collection	Recruitment and consent for the MGRS will occur online wherever possible, and saliva samples (rather than blood) will sent by post to be used as biospecimens for DNA analysis.
2: Participant communication	A short video and ‘information pack’, which will explain the MGRS study, will be created to assist with recruiting family members.

3: Participant involvement in governance	Participants will be invited to be involved in overseeing governance, including funding decisions.
4: Data access	Study participants should be involved in controlling data access decisions and policies.
5: Communication and ways of involving participants	Participants will be included on study advisory groups, including for study recruitment and communication, data access and ethical oversight using multiple communication modes.
6: Provide feedback to participants about the research	Participants will be informed about the impact of the research, and how their involvement has affected the design and management of the study.
7: Create learning and development opportunities	Learning and development opportunities will be created for potential participants, researchers and other stakeholders.

Impact 1: Recruitment and sample collection approach

A discussion about participants' adult children being 'time-poor' highlighted the importance of a study design which minimised the time burden on younger generations. Only a third of participants thought their children would be willing to do a blood test. This allowed the research team to make a more informed decision about 'trade-offs' between the data that can be collected via blood or saliva, versus the potential effect on recruitment.

Impact 2: Participant communication

Most participants reported willingness to be involved in recruiting family members to a new study, if given appropriate information and supporting documents. Relatively inexpensive information resources, such explanatory videos, could have a significant impact on the success of the recruitment and the study as a whole. One participant also advised that information produced for participants by researchers can be confusing, and that laypeople can help simplifying it.

Impact 3: Participant involvement in governance

Event participants unanimously agreed that they should be involved in all aspects of the research, whereas 11 of participants from the 20 analysed interviews thought that they should be involved in study design. One interview participant felt only researchers should be involved as they are "the qualified people" (P8). Five of the interview participants expressed the view that non-researchers are required in research as they provide an important alternative perspective.

Participants had mixed views about commercial organisations funding research. Four event participants were against it, some were ambivalent, and the majority indicated no objections. One participant was concerned about the risks posed by involvement of commercial organisations with opaque "vested interests", asking "what is in it for them?" (P2), referring to individuals and organisations with real or perceived conflicting, competing or commercial interests. Participants suggested that involving people in governance (including funding and ethical oversight) was a way of mitigating this risk.

Study team members reflected that public funding would be preferable to commercial funding, as the responses at the event suggested that a commercially-funded study might negatively impact recruitment. As a result, the study team altered the proposed design to involve participants in governance, oversight and funding decisions.

Impact 4: Data access

Participants shared the view that they would like different kinds of genetic information returned from the research (see Table 5.3), including personal medical, ancestry and pharmacogenomic results. Two-thirds of event participants wanted access to their own genomic data, and had mixed views about who else should have access. All event participants stated they were comfortable with their data being held by academics, and none were comfortable with data being held by a commercial company. However, one participant suggested not "ruling private companies out completely" from research (P9).

General practitioners (GPs) were generally trusted to access and interpret genomic data, but participants felt GPs should not have access to data that they did not. All but two event participants agreed they should exclusively control access to their own data, with those disagreeing mentioning cognitive decline as a reason for a co-managed access model.

Some participants had concerns about themselves or their biological relatives (especially offspring) finding out information they "might not want to know" (P6), or having it imposed on them.

Impact 5: Communication and ways of involving participants

Preferences for communication mode differed between interview participants and event participants. Interview participants preferred questionnaires via post (30%, 18/50), and face-to-face events (25%, 15/59), over online questionnaires (17%, 10/59) and online discussions (8%, 5/59). Event participants suggested face-to-face meetings were helpful but only when there was an "occasional need" (P3). Participant responses also suggested that limiting face-to-face events where possible (in favour of telecommunication) may mean involvement is more inclusive.

Event participants felt certain tasks (such as reviewing information) could be done "more online" (P3). Participants spontaneously suggested using online, moderated forums and suggested that these should be hosted by trusted institutions (such as universities) rather than commercial organisations, as some 'don't trust' social media companies [P17].

Impact 6: Provide feedback to participants about the research

Event participants stated that keeping people informed about what has been learned from the study is a good way of keeping people engaged in the study and improving retention. For example, ASPREE-XT sends a regular newsletter to participants. Participants also stated that they would like to be informed about when their involvement has made a difference.

Impact 7: Create learning and development opportunities

Participants often stated their willingness but also their uncertainty about how they could be involved. Learning and development opportunities were identified as an e of involvement by both a participant advisor.

Figure 5.2. Co-designing multi-generational genomics research

Discussion

The participatory action research method gave insights into participants' preferences that measurably impacted on the proposed study design. The improvement of the interview design using the co-design process illustrates the value of a flexible and iterative approach to involvement in a study.

Participants' preference for being informed about both the study outcomes and the outcomes of their involvement is supported by other studies which suggest that communicating about the research regularly and sharing results may improve retention. [216,400,401] This is particularly relevant for those planning involvement in genomics research, which may span decades. [28]

Such regular communication should also be combined with learning and development opportunities, which could also help facilitate participant involvement by ensuring that people understand the values which motivate the tasks they are being asked to be involved in. [28,345]

Participant views about data storage and access aligns with a 2015 survey by Genetic Alliance Australia, which indicated people mostly trusted universities and research institutes to use personal genetic information for research, with commercial companies least trusted. [402] Event participants' unanimous preference for having access to their own data and a general trust for GPs to access and interpret genomic data also aligns with findings from other studies. [403–405]

Participants' concerns about unintentional disclosure of data to biological relatives who 'might not want to know' certain information highlights the challenges of asking participants about information preferences, and recognises that this ethical decision extends beyond individuals [P6]. This issue is common to almost all ongoing and proposed genomics research studies and should be urgently addressed by contemporary individualist ethical frameworks. [404]

Study strengths

By asking participants their preferences, the study team gained useful insights to inform the design of the proposed study. Participants preference for being involved in decision making about funding sources, data management and ownership, and what information to share with participants will help ensure any future study design aligns with participants' values, ensuring the design is culturally safe and culturally competent. [406] Similarly, the participants preference for being involved in reviewing

participant information aligns with other research which suggests that involving potential participants in reviewing information can help improve recruitment. [369]

The effective involvement of 'stakeholders' also includes involving all relevant staff and health professionals at all levels of an initiative, who may have important knowledge or perspectives that senior research staff do not.

The 'transformative learning' during the process reported from both study participants and the study team was an important impact captured by the participatory action research (PAR) method.[130,131] The process showed that it was valuable to create regular involvement opportunities for each stakeholder. Reporting this process in a standardised way using 'Standardised Data on Initiatives' (STARDIT) meant that impacts such as transformative learning could be reported and that this case study can be compared to similar studies in the future [49].

Study limitations

Because the 76 participants who responded to the original advertisement were self-selected volunteers, our findings may not necessarily be accurate for the whole ASPREE-XT cohort. Our sample may reflect a sub-set of individuals who feel more strongly supportive of a proposed MGRS than the cohort average. However, the data from this process is still useful and valid.[57]

The number of interviews which were transcribed (20) and analysed was high for a case study. [407] As there is no agreement in case study literature on whether to code all or some of the data, our methodology balanced exhaustive examination of the data with the capacity bias imposed by time constraints. [182,408] Therefore, data transcribed and coded from the interviews does not include all the data collected in the interviews.

While the mixture of written and verbal feedback at the event ensured a range of ways to give feedback, the voting process (which involved people raising their hands in front of everyone) may have given different results if it was an anonymous ballot.[59] Future research should explore and compare preferences for different methods of voting.

While the process of involving people described here did not exclude people based on language,[409] the original ASPREE study required a certain level of English language skills in order for people to participate in some of the cognitive assessments and public events. The process described here may

therefore have excluded people who cannot read English or do not feel confident speaking in English, such as people who speak English as a second language.

Conclusions

This case study provides evidence that including participants in the design of a research study positively impacted the study design. As many research studies are negatively impacted owing to poor recruitment and retention, such evidence is increasingly important for informing involvement in future studies. The process of involving ASPREE-XT participants in the design of a new MGRS was highly valued by stakeholders, and was positively impactful for both participants and the study team. The lead investigator stated "the feedback has been so valuable" and that it will be "built into the design" of future research. Learning from the case study suggests that adequate funding, a supportive team, involving participants in the very earliest stage of research planning and co-designing inclusive methods of involvement all contributed to the impacts reported.

The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participant and study team members' views about the value of involvement. For example, one a study team member stated they 'learned that participants have valid, interesting, and often novel ideas that researchers may not have considered' and their 'perceptions of involving participants in planning medical research have significantly changed', which can be viewed as an impact of 'transformative learning'. [130,131] Using a standardised reporting tool to describe the methods and impacts provides a way for learning from this case study to inform future research studies planning to involve people, including studies beyond the discipline of public health genomics.

This is the end of the extract from the publication 'Involving elderly research participants in the co-design of a future multi-generational cohort study': https://doi.org/10.1186/s40900-021-00271-4

STARDIT data: Involving elderly research participants in the co-design of a future multi-generational cohort study

This is an extract from a report which uses the Standardised Data on Initiatives Alpha version (STARDIT).[196] A full version of this report can be found here[294]: https://doi.org/10.1186/s40900-021-00271-4

A 'living' version of this report can be found here: [410]

https://wikispore.wmflabs.org/wiki/STARDIT/Involving_ASPREE-XT_participants_in_co-design_of_a_future_multi-generational_cohort_study

Involvement	
Who was involved	1. 3 academic research investigators 2. An ASPREE participant assessor 3. An ASPREE-XT participant
Specific tasks of this person or group (list as many as possible)	Everyone listed above was involved in co-designing every stage of the process. This included refining wording of participant information, sharing views and advice about the process, proof-reading documents, providing feedback on questionnaires, analysing data, informing planning, presenting information to participants, interpreting data, and participating in email surveys.
What was the outcome or output of the involvement of these people? What changed as a result of involving people?	Improved participant information resources, improved wording that is culturally appropriate, improved question design for interviews, improved learning resources for participants, improved co-design process. By piloting different versions of the questionnaire, we were able to get feedback from participants that the wording of the question about involvement was difficult to understand. In partnership with participants and the study team, the wording was changed to include a short statement explaining what 'involvement' meant and the perceived benefits (see Additional file 2).
Mapping financial or other 'interests'	

Describe any financial relationship or other interest this person has to this project	Three members of the study team were employed by Monash University during this process.
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	It will be published open access
How is it stored and hosted?	It will be shared on a public domain repository.
Who is analysing the data?	The study team described above
How is information about this data disseminated?	1. It will be published in an open access journal 2. It will be shared as an item in a newsletter to participants of the ASPREE-XT study 3. Learning from this process will be presented at conferences, shared on social media and through other channels (such as podcasts).
How is the data FAIR[85]?	Data will be shared in the public domain under a Creative Commons.
Impacts and outcomes	
What new knowledge has been generated? (if appropriate, include effect size, relevant statistics and level or evidence)	4. Involving participants in co-designing a proposed study resulted in changes to the design of the proposed study 5. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participant and study team members' views about the value of involvement, which can be viewed as an impact of 'transformative learning'.
Outcomes - Describe how the learning or knowledge generated from this initiative has or will be used	1. Knowledge from this process will inform the design of a future multi-generational study 2. Learning from this process can inform future involvement activities

Ethics approval and consent to participate

The Ethics Committee of the Alfred Hospital (Melbourne, Australia) granted approval for this project, which was considered Low Risk Review and approved on 16/04/2018.

Project No: 180/18.

Project Title: ASPREE Genomics Engagement: Planning of Future Multi-Generational and Family Studies.

Principal Researcher: Dr. Paul Lacaze.

Participants gave informed consent to participate, with further consent being granted by participants for sharing of recordings from the process.

Chapter 6 – Involving people affected by a rare condition in shaping future genomic research

This chapter aligns with research aim 2: "apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research"

*This chapter is adapted from the published, peer-reviewed 2021 article titled '*Involving people affected by a rare condition in shaping future genomic research'.[7] *Please note:*

- *Figure numbers have been changed to align with this thesis, and some formatting has been adapted, including integrating references into this thesis.*
- *This paper uses the term 'enablers', to avoid confusion with those facilitating discussions, where previous chapter used the term 'facilitators'.*
- *The published version can be found here, along with the additional files: https://doi.org/10.1186/s40900-021-00256-3*

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Plain English Summary

There is evidence that involving potential participants and the public in co-designing research can improve the quality of the study design, recruitment and acceptability of the research, but appropriate methodologies for doing this are not always clear. This mixed methods study examined ways of involving people affected by a rare disease in shaping genomic research.

In this article we describe how people were involved in the co-design process, and ways to plan, report and evaluate involvement in research, including impacts. We demonstrate using a novel way of sharing data in a standardised way to plan, report and evaluate how participant involvement activities positively impacted the study design (Standardised Data on Initiatives - Alpha Version 0.1 -

STARDIT). STARDIT is an open access data-sharing platform being developed to standardise the way that information about initiatives is reported.

We conducted pre and post surveys and facilitated online discussions. There were six outcomes from the process, which included participants reporting an improved understanding about how to get involved in research and that learning resources were useful. Participants reported changed views about involvement, with most participants 'widening' their perception of who should be involved in research to include more people. Participants enjoyed online discussions and asked to stay involved in the research process. The partner organisation reported that similar online discussions will be used in future research prioritisation processes. Standardised reporting of this study will help inform the future involvement of participants and the public in the design and conduct of genomic research.

Abstract

Background

There is evidence that involving potential participants and the public in co-designing research can improve the quality of the study design, recruitment and acceptability of the research, but appropriate methodologies for doing this are not always clear. In this study we co-designed a way of involving people affected by a rare genomic disease in shaping future genomic research about the condition. The aim was to report the process, experiences and outcomes of involving people in genomic research in a standardised way, in order to inform future methods of involvement in research co-production.

Method

Participants were recruited from an online community hosted by an Australian-based rare disease charity and were over the age 18 years. Once people gave consent, we shared learning resources with participants and invited them to complete an online survey before joining a two-week facilitated online discussion, followed by a second online survey. We used the novel tool 'Standardised Data on Initiatives - Alpha Version 0.1' (STARDIT) to map preferences, plan involvement and report any outcomes from the process, with quantitative data analysed descriptively and qualitative data thematically analysed.

Results

Of the 26 people who gave consent and completed the initial survey, 15 participated in the online discussion and 12 completed the follow-up survey. STARDIT was used to report six outcomes from the process, including 60% of participants' responses showing a change towards 'widening' their view of who should be involved in research to include more people. Outcomes also included an improved understanding of research and how to be involved. Participants enjoyed online discussions, found learning resources useful and asked to stay involved in the research process. The partner organisation reported that a similar online discussion will be used in future research prioritisation processes.

Conclusion

Involving people in co-designing the process improved the study design, ensuring it met the needs of participants. Whilst the study includes participants from only one disease group, using STARDIT allowed us to map people's preferences and report the methods and outcomes from involving people, providing a way for learning from this case study to inform future research studies beyond the discipline of public health genomics.

Introduction

Genomic research may lead to better understanding and management of the Eosinophilic Gastrointestinal Disorders (EGID), including Eosinophilic Oesophagitis (EoE). EGIDs are long-term (chronic) inflammatory conditions that affect the lining of the throat, stomach and gut (epithelium). EoE is the most common type of EGID and is most likely caused by exposure to food antigens.[411] EoE affects people of all ages, gender and ancestral backgrounds.[412] Genomic research may lead to better understanding and management of the disease.[413] EoE is a rare disease and involving people affected by EoE in shaping future research could help ensure that the research is relevant, well-designed and aligned with patient priorities.[34,58] However, currently there is no standardized way of planning and reporting how people are involved in shaping future genomics research.[28]

This illustrative case study documents a participatory action research process with the charity ausEE, to co-design a way of partnering with people affected by EoE (including their carers), to help shape future research. The study aimed to examine the processes, experiences, barriers and enablers of participant involvement in genomic research about one condition EoE to inform methods of genomic research co-production. In this article we aim to outline how people were involved in the co-design process, and how Standardised Data on Initiatives (STARDIT) can be used to plan, report and evaluate involvement in research, including impacts.[414]

We created a prototype online discussion forum, guided by the principles of participatory action research (PAR) from the International Collaboration for Participatory Health Research, and guidance on co-design.[89,130] This allowed us to explore and report the practicalities, enablers and barriers of using an online discussion as a way of involving people in genomic research.

Involving the public, patients, research participants and other stakeholders in actively contributing to the research process can lead to a range of positive outcomes. These can include improving the recruitment[207], quality and relevance of research. [28,392] Involvement is when research is carried out 'with' people rather than 'on' them. [260] In human genomics research, the need to involve the public and other stakeholders is a crucial aspect of responsible research practice, which can help ensure outcomes of importance to all stakeholders are included in decision making processes.[21,28,34,415] The term 'stakeholder' means anyone who has a 'stake' in the research, in particular those with important knowledge, experiences, expertise or views that should be taken into account. [21,31] This can include people affected by Eosinophilic Oesophagitis (EoE), the study team, and the wider public, although in this process we did not involve the wider public. At the earliest stage of the research cycle (the conceptual stage), people affected by EoE were involved in the co-design of this study.

Methods

In order to co-design the study from an early stage, a representative from ausEE was contacted, inviting the organisation to partner with the research team, with a representative from ausEE invited to be part of the study team and another invited to give feedback on the proposed study design. The representatives were involved in a number of tasks including reviewing and improving the written information, online survey questions, and the facilitation plan for the online discussion.
In order to facilitate comparison with other studies, we used the novel system 'Standardised Data on Initiatives - Alpha Version 0.1' (STARDIT) to map preferences, plan involvement and report any outcomes from the process.[196] This included reporting co-design positively impacted the study. STARDIT is an open access data-sharing platform being developed to standardise the way that information about initiatives is reported across diverse fields and disciplines, including information about which tasks were done by who. Quantitative data was analysed descriptively and STARDIT was also used in parallel with a thematic analysis, which organised data into pre-defined 'super-categories' which allow consistent comparison with other data using STARDIT.

Learning resources were both co-created and selected by the investigator team, working in partnership with the Australian Genomics Health Alliance and co-refining the selection with potential participants, including working in partnership with potential participants and the Australian Genomics Health Alliance, using the outcomes of a landscape analysis of educational materials as a starting point for selecting resources.[416] Final resources were checked by a medical professional specialising in EoE.

Study Design

A participatory action research (PAR) paradigm was chosen to guide the process with co-design and reporting informed by guidance from a number of sources.[80,91,127] Participatory action research is an umbrella term which describes a number of related approaches, including forms of action research which embrace a participatory philosophy and include 'co-design' and 'co-production' of research. [88]. It is a process whereby researchers, the public and other relevant stakeholders "work together, sharing power and responsibility from the start to the end of the project",[89] including knowledge generation and translation.[89] Elements of this study were co-designed in parallel with another similar study, therefore some aspects were inflexible and thus 'co-refined' rather than 'co-designed'.

We used case study methodology to record and describe our experience involving participants in an online discussion about genomics research. The selection of the population for this case study was informed by a number of factors which were appraised by the study team, including ethical, pragmatic and population considerations.[178,396] One of the investigators (PL) had a professional relationship with the charity ausEE, which was used as a starting point by the study team to explore the appropriateness of the case study.

The case study is presented as an instrumental case study, where the purpose is to understand the particular case and can attempt to provide data that could produce useful generalisations by using inferences from the data.[163(p109)] We collected and analysed both qualitative and quantitative data during the involvement activities, informed by a number of frameworks and standards.[393,394]

In addition, we analysed other data from participant survey responses, online discussions, meeting notes, emails, surveys of the study team and reflexive diary entries of one member of study team (JN). Coding and thematic analysis of qualitative data was carried out by two authors independently (JN, KG) and checked by another author (PL), following best practices for enhancing validity in qualitative methods.[182] Two authors of this paper also shared personal comments in the online

discussion (KG, SG), which have been anonymised using participant numbers. In order to aid analysis and comparison with other case studies, we used a novel way of sharing data in a standardised way (Standardised Data on Initiatives - Alpha Version 0.1) to map preferences for involvement, plan involvement, report and evaluate how people were involved in the participatory action research process, and how this positively impacted the study design.[196]

STARDIT is an open access data-sharing platform being developed to standardise the way that information about initiatives is reported across diverse fields and disciplines, including information about which tasks were done by who. It also offers a way to add updates throughout the lifetime of an initiative, from planning to evaluation and reporting any impacts. Authors from this paper were involved in co-creating STARDIT, and learning from this process informed the development of the reporting tool.[16]

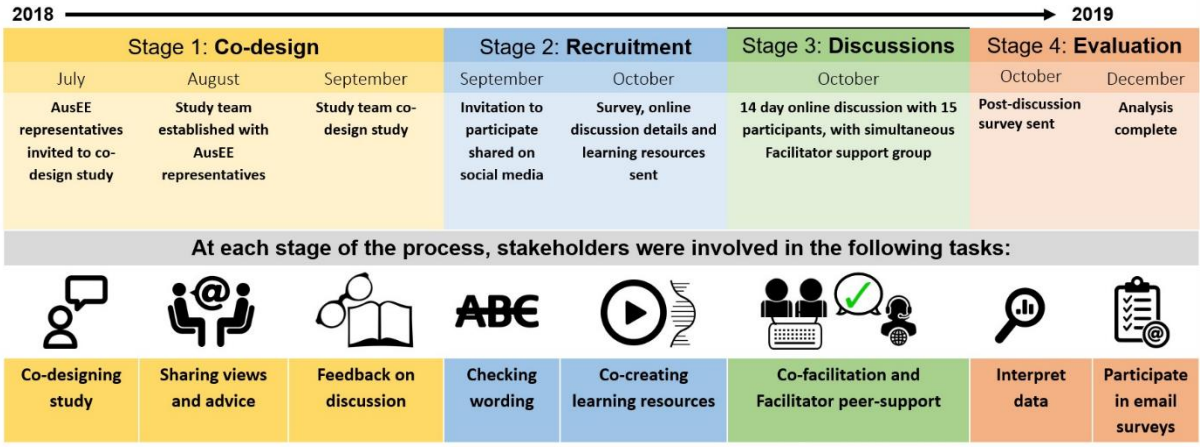
Participants and recruitment

This study recruited participants from an existing online community managed by the Australian-based charity ausEE. A link to the informed consent form and learning resources was shared by ausEE on a closed Facebook group. We recruited people if they were either a parent, a carer, a partner, family member or loved one of someone with EGID who is under 18; or someone who was over 18 with EGID. If people gave consent, they were invited to complete an online pre-discussion survey and sent instructions for joining the online discussion.

Stages of research

There were four stages of the research process: 1. Co-design; 2. Recruitment and surveys; 3. Online discussions and 4. Evaluation and reflection. The multiple stages of the co-designed research are summarised in Figure 6.1.

Figure 6.1: Stages of research

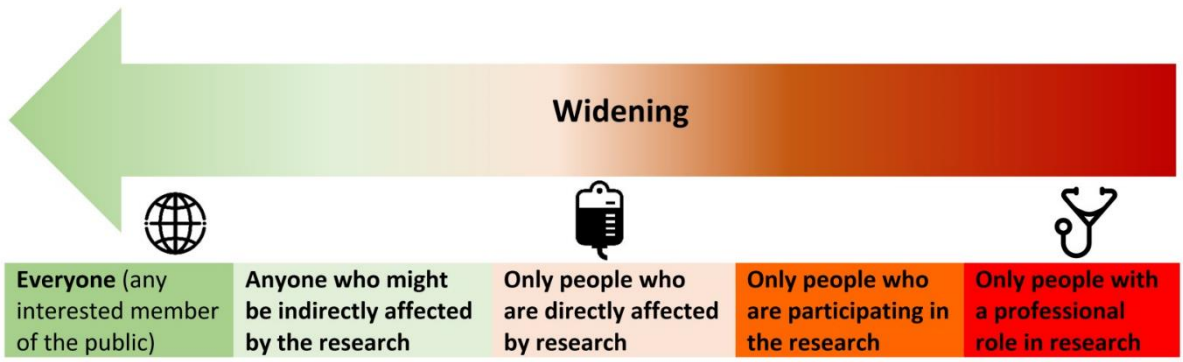


Initial contact was made with ausEE in July 2018 and two investigators from ausEE were invited to join the study team (SG, KG). After ethics approval was obtained, an invitation to participate in the study was shared in the ausEE Facebook group and member newsletter in September 2018. This group has members who live in both Australia and New Zealand. Before being asked to give consent, the study was summarised in plain English and two learning resources were shared with participants in order to provide context to the study (see Additional File 1 'Data and analysis').

The invitation to participate contained a link to the participant information document and the informed consent form. If people gave consent to participate, they were invited to complete an online pre-discussion survey, which had demographic data categories informed by other similar studies to allow comparison.[49]

Participants who gave consent were then contacted by email, with information about joining the discussion shared alongside relevant learning resources. Participants were also sent a follow-up survey after participating in the online discussion. Questions relating to 'Who should be involved in research' were the same as in the pre-discussion survey to allow comparison. Participants could choose from the categories outlined in Figure 6.2, with a change in direction towards more people being involved labelled as 'widening', the inverse as 'narrowing'.

Figure 6.2: Who should be involved in research?



Online discussion methodology

We conducted an online discussion to maximise flexibility about when participants could contribute (for example, people in different time-zones or those with different caring responsibilities). A significant advantage of online discussion platforms is that they are accessible to a greater number of people. This creates a more equitable platform for people to participate, compared to a synchronous (simultaneous 'real-time') discussion.
We offered anonymity in online discussions to avoid people inadvertently disclosing sensitive or personal information, which may have serious implications, for example, impacting on eligibility for future health insurance. Participants could choose to use their real name and email address, or remain anonymous by using pseudonyms or temporary email accounts.

Before participants joined the online discussion, seven learning resources were shared with them. This included a short 60 second online video about the study context and purpose[417], a one-page infographic summary of a scoping review about genomics research[418], and a short two-page summary of genomics and contemporary research relating to EoE was co-created with ausEE, the study team and experts in genomics (see Additional File 1 'Data and analysis').[419]

An open-source software web application 'Loomio'[420] was installed on virtual machines hosted by an Australian Government initiative called 'Nectar Cloud'.[421] Participants could securely log-in from anywhere in world and participants' data was stored securely on servers physically located inside Australia.

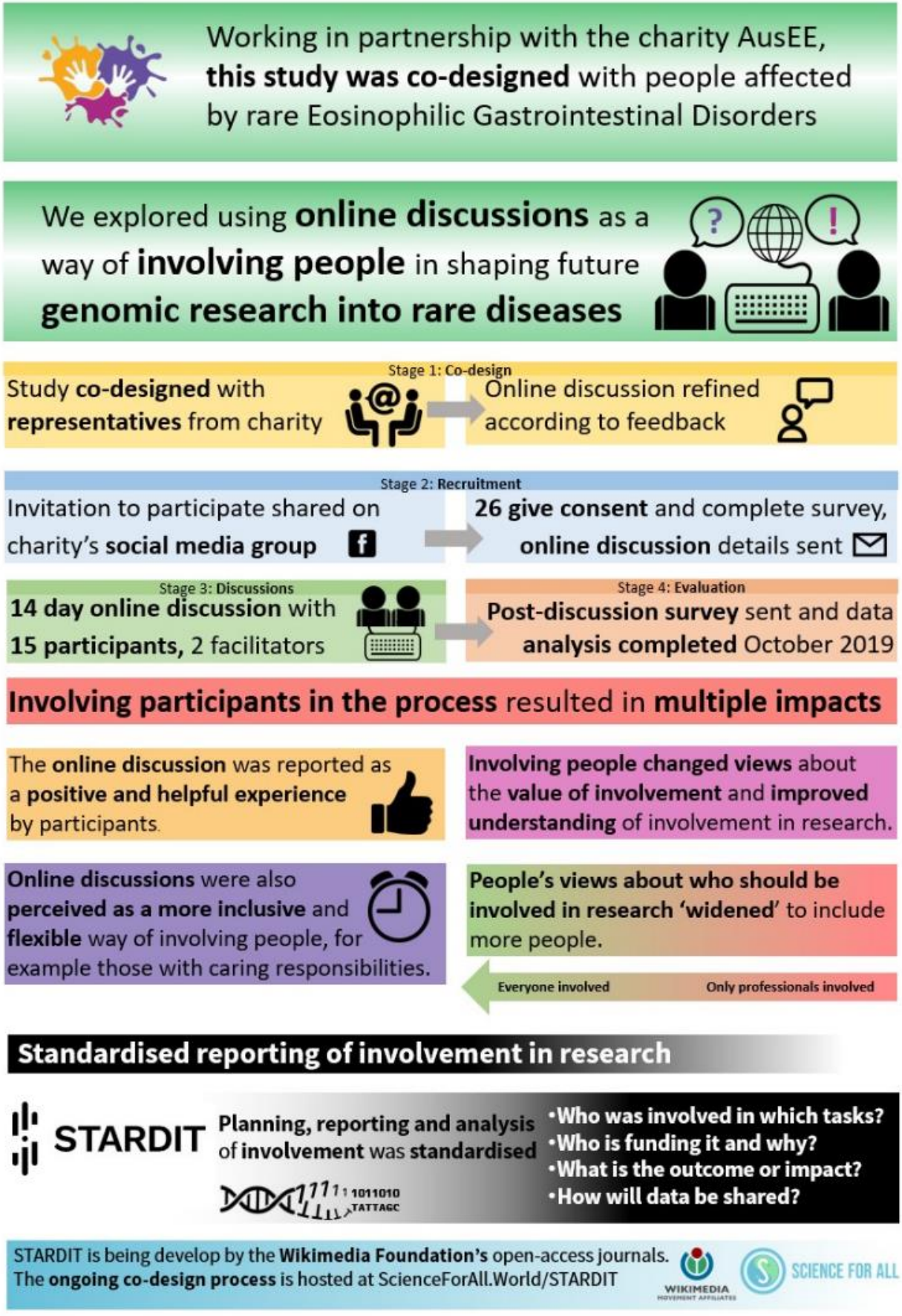
Two people facilitated and moderated the discussion for 14 days (JN, KG), one of whom was a parent of a person affected by EoE (KG). Participants were invited to co-create their own boundaries for the group discussion at the start by reviewing provided statements and suggesting amendments. The facilitators judged when to introduce new topics (depending on the engagement with each topic) with the recommended schedule below in Table 6.1 used as a template.

Table 6.1: Online discussion overview

Question	Day
What do you understand by the word 'research'?	Day 1
What do you understand by the phrase 'genomic research'?	Day 1
Why do we do research?	Day 2
Which aspects of any future genomic research should be influenced by the following groups of people?	Day 4
What methods do you think could be used to involve those people in future genomics research?	Day 6
Do you have any ideas, thoughts or reflections that have not been shared yet?	Day 7
Discussion closed	Day 14

Results

Figure 6.3: Process summary



Stage 1: Co-design

The input of the representatives during the planning and co-design stage had clear positive impacts, particularly in improving educational resources and ensuring the online discussion was advertised using wording appropriate to the existing online community. For example, representatives from ausEE helped change the study design to include explicit opportunities for participants to learn more about genomics and EoE, avoiding participation being perceived as having a one-way benefit. During the co-design process it was also decided to exclude people who were under 18 and people who stated they were representing someone who was over 18, as people who were 18 and over had the choice to represent themselves.

Stage 2: Recruitment and surveys

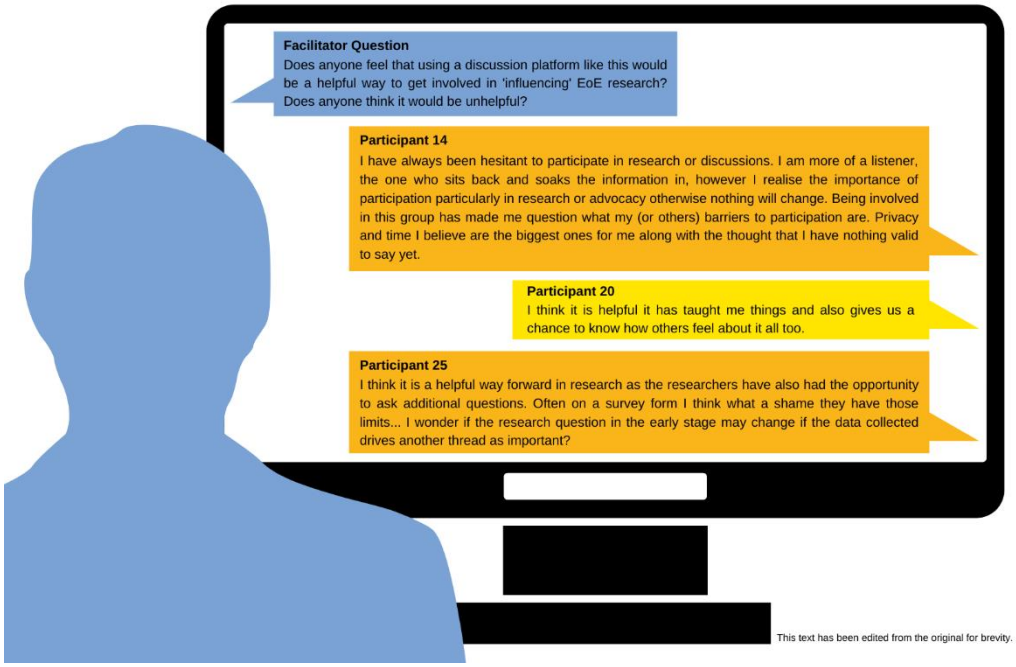
In total 26 participants completed the pre-discussion survey, 12 completed the follow-up survey. These responses are summarised in Table 6.2. All but one of the participants were female, with most reporting they were parents of a person with an EGID. Self-reported educational attainment was mostly 'degree (bachelors), diploma or post-graduate', with one participant reporting they had professional experience in genomics. Most participants were between 30-45 years old and participants all lived in Australia, except for one who lived in New Zealand.

A total of 41 responses were given by 12 participants who shared their identity at both the baseline and follow-up, including ten questions about which aspects of genomic research should be influenced by different stakeholders which identical at each stage. Of the 41 responses to the ten questions, 60% showed a change towards 'widening' involvement (N=25/41), 36% of responses stayed the same (N=15/41) and 7% 'narrowed' (N=3/41). Recurring themes were identified by the study team, and six specific outcomes were reported as a result of involving participants in this process.

Stage 3: Online discussions

A total of 15 people participated in the online discussion. The President of the charity ausEE also identified herself by name in the discussion (SG). All but two participants chose to use their real name in the online discussion, which they had provided when registering and giving consent. A visual representation of the online discussion is illustrated in Figure 6.4.

Figure 6.4: Online discussion visualisation



Stage 4: Evaluation and reflection

Participant feedback about the online discussion was sought via survey and was positive, with one participant describing the process as ‘very good’ [P25]. Participants reported that they enjoyed being involved and that participating in the discussion changed their views about involvement. The visual summary of the scoping review about involvement in genomics that was shared at the start of the discussion was described as a ‘great summary document’ [P21]. One facilitator noted the group were ‘well informed’ about research [P16], with the discussion indicating a high level of general understanding about research, genomics and the associated ethical, legal and social implications. For example, one participant stated ‘I wonder about the limitations of genomic sequencing but believe knowing the result is an adult human right’ while adding they were unsure ‘how much’ they would want to know themselves [P25]. Another participant mentioned the ‘moral minefield’ of pre-implantation genomic screening, noting that “not all that is ‘marked’ will come to be” [sic] [P3].

Table 6.2: Summary of pre and post survey responses

Question	Results
What made you decide to respond to our invitation to participate in this project? 22 responses (pre-discussion)	Fourteen participants stated they decided to participate as they wanted to help improve knowledge of the disease and help find a cure. Two people were specifically interested in genomic research [P6, P9], and one person reported they were 'researching themselves' and their sons' genomic variations [P5].
What do you hope to get out of participating in this discussion? Do you have any specific expectations? 20 responses (pre-discussion)	Four participants wanted to 'learn more' [P16]. Five participants stated they wanted better outcomes for patient care and treatment protocols, with three participants stating they wanted to be actively involved in helping research to improve outcomes. Two participants wanted to hear the perspectives of others. Five participants stated they had no expectations.
Do you have any ideas about how the different people could influence future research? 19 responses (pre-discussion)	One participant stated that people with a rare disease and their families are 'likely to have different priorities from scientists' [P16]. Another suggested that 'sharing patient experiences, priorities of research areas' and involving people in co-defining 'ultimate patient outcomes' were ways people could influence research [P21].
Is there anything in particular you liked or thought was helpful about how the discussion was conducted? 8 responses (post-discussion)	One participant said they 'enjoyed the interaction, helpful links with information about genomics and the topic threads' [P21]. Another added they liked 'being able to read others thought processes on each topic' [P3]. Others said 'the responses to mine were timely and provoked further questions that made me think in new directions' [P25]. Two participants said that 'having different topic questions/threads was helpful', as was the 'information provided to start' of some threads [P29]. One participant added that 'it was great because even though busy with my son' they could travel and 'still catch up and learn things and do my input too'[P30].
Is there anything you didn't like, thought was unhelpful or could	Only two participants provided answer for this question other than 'no'. One participant stated 'I didn't like the platform it was conducted on' as it was not 'user-friendly' [P15]. Another added

have been improved about how the discussion was conducted? 4 responses (post-discussion)	that sometimes facilitators 'added another question too quickly before a number of people had a chance to answer the first one' which risked leaving 'some people behind' [P3].
Did you have any expectations from participating in this research that were met or not met? 8 responses (post-discussion)	One participant stated that they wanted to 'learn more about the difference of the gene and genomics and that was met' [P30], and three others stated their expectations had been met, with none reporting they had not.

Outcomes from the process

There were six outcomes reported from this process, which are summarised in Table 6.3.

Table 6.3: Summary of outcomes from the process

Outcome	Summary
1. Learning resources useful	Participants stated that the learning resources were useful and helped improve their understanding of genomics, research and the associated ethical, legal and social implications.
2. Changed views	Participants reported that the process of being part of an online discussion gave them an opportunity to 'learn a lot' about the views and experiences of others and reflect [P25]. Participants reported that this 'challenged' them into 'rethinking' or changing or their own views on certain issues [P25].
3. Enjoyed online discussions	Four participants said they enjoyed being part of the process and preferred it when compared with other methods such as face to face discussion or interviews.
4. Online discussions to be used in future research prioritisation	The President of ausEE stated that the discussion had 'given her an idea' and that she would combine face to face and online discussions into a process to involve people in deciding how the medical research fund is spent.
5. Participants asked to stay involved in the research	Participants requested to stay involved in the research process, including in analysing data and being co-authors on the paper.
6. Improved understanding of how to get involved in research	Participants reported improved understanding of genomics research, including how they could be involved.

Participant views about involvement genomic research

While the focus of the online discussion was involvement in genomics research, the themes of the discussion reflected the interconnected nature of the subject, including ethical, legal and social implications of genomics research. Issues discussed included research prioritisation and funding; data sharing; health technology assessment, and health and life insurance. Fourteen participants shared the view that they wanted to be involved in improving knowledge of EGID and helping find a cure 'so other parents don't go through what we went through' [P25]. Similarly, participants wanted to help genomic research in the area improve the lives of people affected by an EGID.

Ten participants identified specific enablers of involvement in genomic research, including 'equity'. Open-source data and information was identified as an enabler of involvement [P3]. Emotional connection to an issue was identified as being either an enabler or a barrier to involvement, with one participant stating 'there needs to be a balance' [P21]. Other enablers included participants being 'able to contribute without putting their personal situation at risk' with data 'well protected so as not to be used against an individual or group of people' [P3].

The theme of the 'intention' and 'purpose' of research emerged, with a discussion on vested interests. One participant identified that people affected by EGID 'certainly do' have a vested interest [P3]. Participants were concerned that research priorities were driven by money. Four participants mentioned 'insurance' as an area of concern which requires public involvement and scrutiny with regards to data sharing, with one stating it is a 'valid community fear' that research data will be used 'for' insurance companies or other for-profit organisations [P3]. One participant stated that 'progressive watering down of privacy protections' meant they were concerned their children would be 'refused insurance because of a decision I have made'[P16], such as participating in research.

Stakeholder views about the online discussion

Participants identified specific things about the online discussion which they felt enabled participation or were barriers to participating. Participants 'enjoyed the interaction' of the online discussion and were 'supportive, positive and open' [P21] [P3]. Four participants found the links to learning resources helpful. Participants felt online discussions are 'good to help' people get involved, in particular those who are unable to travel or live in remote areas [P20]. The research method used in this online discussion was described as 'very good' as the time frame allowed for 'life responsibilities and also gave time to process new information and think about others' comments' [P25]. However, one participant and one facilitator felt the discussion was 'too quick' [P3]. Other participants reported the length and pace was good, highlighting that co-design is an important element in designing similar future discussions. One participant stated that 'this style of discussion here has been interesting and by learning from others I have found some of my initial thoughts have changed' [P25]. Other participants liked the idea of a multi-stage way of involving people using a combination of face to face and online discussion [P25], which aligns with other models used in priority setting.[15,422]

One participant mentioned that 'one of the things that I really love about this research is that the research topic is on involving people in research and this research itself does exactly that - involves people in the research'[P21]. The 'open discussion' at the end where participants could raise any subject or question was also highlighted as a 'helpful way forward in research' as it allows participants to identify and focus on important areas, and researchers an opportunity to ask clarifying questions, additional questions and re-focus larger research questions. Other participants noted they might be influenced by the answers of others and that as this is part of the research process, it would need to be a consideration in data collection methodology [P21]. For example, people might share different information in an anonymous survey compared to an online discussion, owing to concerns about privacy or because another participant has said something which has prompted a memory they might otherwise have forgotten.

Discussion

Involving participants in co-designing the research process resulted in changes to the study design, including improving language used in recruitment and learning resources. Involving people in online discussions about involvement in research changes people's views about who should be involved in research, including participants 'widening' their views about who should be involved in research to include more people.

Most participants did not seek anonymity during online discussions, and this may reflect that participants were recruited from a social media group where their identities were already known and a sense of community already developed. It may also suggest that participants wished to be known to other participants in order to give and receive support inside and outside of the discussion.

However, optional anonymity in online discussions can also be viewed as an advantage by some participants, by removing elements of identity which may be associated with power disparity, such as appearance, age or gender or future discrimination by health insurance providerr[423]. Anonymity for some may therefore lead to a disinhibition effect, with participants feeling more able to express views and perspectives that they may not feel confident sharing in other contexts.

Learning from the outcomes

The participants were highly engaged in the process, possibly as all self-reported a personal motivation for improving research in this area. The participants enjoyed being part of the process and none reported difficulties with the platform, which may be a result of familiarity with other online platforms, which may be correlated with self-reported higher degree level education. One

participant wrote ‘Thanks for this platform, process, everything really I've enjoyed being part of it’ [P21].

However, one participant said that while diagrams and visual aids were helpful, they wanted ‘more diagrams when reading through concepts or ideas’ [P3]. This suggests that online text-based discussions might not suit those with a preference for visual communication and highlights the importance of co-designing online discussions with potential participants who may have diverse learning or access needs.

The learning from ‘Outcome 2: Changed views’ aligns with other studies, showing that the act of being involved in a discussion or in research can have outcomes which are a form of ‘transformative learning’ for both participants and study team members.[171] At the core of the participatory action research method is ‘critical reflexivity’, a process which asks people involved to reflect on the causes of problems, any solutions and the actions that people can take to improve the current situation.[130] It is a form of collective self-reflective enquiry undertaken by people in order to understand their situation from multiple perspectives.[136] In this sense changed views of both researchers and participants involved can be viewed as an impact of ‘transformative learning’, with the reflexive re-examination of views and perspectives part of the participatory action research paradigm.

Strengths

While this study was planned and completed before the COVID-19 pandemic, the methods of involving people online described here now have an unexpected relevance to many disciplines, as research projects around the world seek to involve people online in novel ways, and evaluate such methods in a standard way.

Using STARDIT for standardised preference mapping, planning and reporting of involvement meant outcomes from the process could be mapped more effectively[130,196], including outcomes from this process beyond the date of this publication.

Measuring outcomes such as ‘transformative learning’ can be challenging. This process overcame such challenges by using baseline and follow-up surveys.

It is significant that some participants preferred the online discussion method over face to face discussion or interviews, as this highlights the importance of using STARDIT-PM to map the potential

participant's preferences when co-designing involvement, as this helped ensure research design meets the needs of those participating and those involved. Authors from this paper were involved in co-designing STARDIT, and learning from this process informed STARDIT Beta Version 0.2.

In order to share power effectively and ensure this article reflected everyone's experience of the process, the two people affected by EGID who were participant representatives were invited to be co-authors of this article. As participant representatives, they were involved from the very earliest stages of this research project. In addition there were opportunities for other participants to give feedback on the manuscript (including checking data analysis). In order to demonstrate the value of participant representatives' contributions, the study team ensured they were invited to propose their own flexible time frames to contribute effectively to the study. This flexibility, and flexible deadlines from the article publisher in response to COVID-19, helped ensure they could balance providing feedback on data analysis and article versions with any caring responsibilities.

Limitations

The study design allowed people to be anonymous at each stage, so there was limited data from people who shared identifying information at each stage, meaning that some anonymous participants might be counted twice. However, as this study is a relatively small sample, statistical extrapolation from this case study is limited. This study also recruited from an existing online community which used a social media group, so data may reflect the views of people who have more experience using online platforms than the general public and are thus more competent.

Some participants demonstrated passive behaviour in online discussions. For example, some participants logged-in multiple times, read comments but did not contribute comments. While the follow-up survey attempted to capture views of these participants, it was not completed by everyone, so it is hard to assess why certain people did not contribute more. However, one participant reported learning while reading and not contributing, and therefore there may be under-reporting of some learning outcomes from participants who were more passive .[424]

This study was designed in parallel with another similar study to allow comparison of data sets[4], so some aspects were inflexible (such as the choice of open-source discussion platform) and could only be 'co-refined' rather than 'co-designed'.

The decision to host the discussion on a separate platform (Loomio) from the one hosted by ausEE (on Facebook) was made for ethical reasons, as the study team could ensure control and ownership of the data. This may have been a barrier to some who decided not to participate, perhaps as the new platform was unfamiliar. However, data on this was not collected. While the gender balance of the participants was reflective of the wider online community from which participants were recruited at the time, data on educational background was not available so it is unclear if the educational background of participants was statistically representative. Future studies should seek to explore ways of appraising who might be excluded from research using more standardised processes.[425]

Conclusions

This study provides valuable insights to the involvement of participants in research co-design and its associated benefits for researchers and participants. The co-design process improved the design of the study and ensured it met the needs of participants. Through sharing and discussing views in an asynchronous online discussion, participants reported their views changed. This aligns with other studies and demonstrates that transformational learning occurs through the process of involving people. After participating in the online discussion, people's views about who should be involved 'widened' to include more people. Participants wanted to be involved in shaping future genomic research in order to improve it, especially being involved in ethical oversight and scrutiny to improve safeguards regarding data use and privacy.

Whilst the study includes participants from only one disease group, using a standardised reporting tool allowed us to map people's preferences and report the methods and outcomes from involving people. Such reporting also provided a way to report the benefits to both participants and researchers, providing a way for learning from this case study to inform future research studies beyond the discipline of public health genomics.

Contributions

JN wrote the manuscript and analysed the data, facilitated online discussions, designed and carried out the process, evaluation of the process and the reporting (including STARDIT and GRIPP2 reports). KG gave feedback on the process design, facilitated online discussions, advised on methodology and paradigms, edited the manuscript, analysed data and checked data analysis edited the manuscript. SG was a participant advisor during the process and gave feedback on the process design,

participated in the process and gave feedback on the manuscript. PL gave feedback on the process design, checked data analysis and edited the manuscript. All authors helped co-design the process, read and approved the final manuscript. All participants who opted to stay involved were invited to give feedback on this manuscript.

This is the end of the published peer-reviewed article 'Involving people affected by a rare condition in shaping future genomic research'.[7]

STARDIT MICRO Report: Involving people affected by a rare condition in shaping future genomic research

This is an extract of data from a report which uses the Standardised Data on Initiatives Alpha version (STARDIT).[196] A full version of this report can be found here[279]: https://doi.org/10.1186/s40900-021-00256-3 The WikiData version can be found here: https://www.wikidata.org/wiki/Q100403236

Involvement	
Who was involved	Group 1: Academic research investigators (Jack Nunn and Paul Lacaze) Group 2: People affected by the rare disease representing the charity ausEE with experience of academic research (Kylie Gwynne) Group 3: People affected by the rare disease representing the charity ausEE (Sarah Gray) Group 4: People affected by the rare disease who are members of the online community and participated in the study
Specific tasks of this person or group	Group 1 and 2: Involved in co-designing every stage of the process, analysing data and member checking during the thematic analysis Group 3: Involved in co-designing the recruitment and giving feedback on the proposed study design and as an author of the paper. Group 4: Invited to give feedback on the paper
Outcomes (from involvement)	Improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the group to describe themselves), improved online discussion, improved learning resources for participants, improved co-design process.
Mapping financial or other 'interests'	
Describe any financial relationship or other interest this person has to this project	Two investigators are affected by the rare disease, which is why they were invited to be part of the study

Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	It will be published open access in peer reviewed journals with identifying information removed in order to anonymise it as much as possible.
How is it stored and hosted?	It will be shared on a public domain repository.
Who is analysing the data?	Group 1-3: The study team described above Group 4: participants were invited to review the analysis and give feedback to ensure they felt it reflected their experience of the process
How is information about this data disseminated?	1. It will be published in an open access journal 2. It will be shared with participants of the research and also other members of the sibling group who have joined it since the study commenced 3. Learning from this process will be presented at conferences, shared on social media and through other channels (such as podcasts).
How is the data FAIR[85]?	Shared in the public domain licensed under a Creative Commons.
Impacts and outcomes	
New knowledge	1. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources 2. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'.
Outcomes	3. Learning from this process informed subsequent discussions in the charity ausEE about involvement in research, including proposed improved ways of involving people

Ethics approval and consent to participate

The La Trobe University Human Ethics Committee approved this study.

Project number: HEC18242

Project Title: Genomics Research and Involving People: ausEE

Chapter 7 – Co-designing genomics research with a large group of donor-conceived siblings

This chapter aligns with research aim 2: "apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research"

*This chapter is adapted from the published, peer-reviewed 2021 article titled '*Co-designing genomics research with a large group of donor-conceived siblings',[4]. *Please note:*

- *Figure numbers have been changed to align with this thesis, and some formatting has been adapted, including integrating references into this thesis.*
- *This paper uses the term 'enablers', to avoid confusion with those facilitating discussions, where previous chapters used the term 'facilitators'.*

https://doi.org/10.1186/s40900-021-00325-7

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Plain English Summary

Human genomics research is growing rapidly. There is evidence that involving potential participants and the public in co-designing research can improve the quality, recruitment and acceptability of the research. However, more evidence about effective methods for involving people is required, especially those in sub-populations which are inherently high interest to researchers and thus with a higher risk of being exploited by medical researchers.

In this study, we worked with a large group of donor-conceived siblings who were conceived from the same sperm donor. We sought their views regarding participation in possible future research. We co-designed a way of involving them in discussion about their own "terms of engagement" with research. Online discussions gave group members an opportunity to share their views, and take

initial steps towards developing their own research governance model. We used the 'Standardised Data on Initiatives (STARDIT)' Alpha Version to report involvement, allowing findings to be compared with other studies.

Group members who participated reported enjoying the experience and identified some advantages to online discussions over other methods, including time to reflect on answers and learn collectively. Most participants' preferences about who should be involved in research design 'widened'.

Participants' learning from the process also informed subsequent discussions in the sibling group about participation in research, including about how to make informed decisions about participating. Involving people in this way has the potential for a 'transformative learning' impact, where people's perspectives and attitudes change as a result of being involved. This is particularly important for people in populations at greater risk of being exploited including under-represented sub-populations which are of high interest to genomic researchers.

Abstract

Background: Human genomics research is growing rapidly. More effective methods are required for co-design and involving people, especially those from populations at greater risk of exploitation, such as those of inherently high interest to medical research. This case study documents how we worked with a large group of donor-conceived siblings who share the same sperm donor father, to explore how they might want to engage with and influence any future genomic research.

Method: A participatory action research process was used to explore the views of a group of 18 known donor-conceived siblings; who are part of a larger group of up to 1000 people who share the same biological father. The discussion explored views about how the group would like to be involved in future research. Five members participated in co-design; 12 completed a pre-discussion online survey; and six participated in an online discussion forum and evaluation survey. The online discussion was led by one facilitator, supported by the study team.

Results: Co-design informed the research process. Participants reported enjoying the overall experience of the surveys and discussion forum, which were perceived as inclusive and flexible. Most participants' views regarding the value of involvement in research changed during the process, and 'widened' about who should be involved. Participants were supportive of future research being done with the siblings group. All who completed the final survey requested to remain part of the co-design process. Other themes in the online discussion included concerns about conflicting interests and a desire for research participation to improve the situation for people affected by assisted conception. The process informed later discussions in the sibling group about participating in a self-managed biobank and informed decision making about participating in genomics research.

Conclusion: Findings from this study help inform how people from certain sub-populations should be involved in planning and defining their participation in genomic research; particularly those from populations at greater risk of exploitation or high-interest populations. This process provides a replicable and practical method of involving potential participants in co-designing genomics research using online discussions, with reported positive outcomes. Reporting this study using 'Standardised data on initiatives (STARDIT)' to report the process allows comparison with other studies.

Introduction

Human genomics research involves defining sub-populations, measuring DNA changes within them and linking them to traits or outcomes in order to understand how DNA variation can contribute to human health and disease. The more genetically similar people are, the more likely it is that they will share the same DNA variations that contribute to a given trait such as wellness or disease. Therefore, historically human genomic research has focused on restricted populations who share common biological ancestry, including large families or founder populations, where less genetic variation has led to more clear links between DNA and disease.[426]

Since the advent of affordable online ancestry DNA testing, increasing numbers of people are taking DNA tests to understand genetic ancestry (genealogy). However this DNA testing can sometimes result in returning unexpected information, including the revelation of being sperm donor-conceived.[427] These genetic results can also immediately link people to a broader group of biologically related people who also share the same biological father.[427]

Owing to the increasingly affordability of direct-to-consumer DNA testing[427], a growing number of large groups of donor-conceived siblings are now being discovered, some with over 100 half-siblings.[428,429] In some cases, people from such sibling groups were conceived before regulation or clear legal oversight of assisted conception services. For example, in the UK, a government register of donors was proposed as early as 1949[430], but such a register was not established until 1991[431]. By 1958 the total number of donor conceived people in the UK was estimated to be 7,500, and 100,000 in the United States.[430] Some countries, including developed economies such as Japan, still have no clear legal regulation or oversight of human sperm and egg donation.[430,432,433]

A historical and contemporary lack of regulation means that there may be many undiscovered large sub-populations of individuals who share a common sperm donor. Such sub-populations may provide new insights into medical, genetic, sociological and psychological studies, and therefore are of inherently high interest to research, including genomics research. However, such interest also makes them at greater risk of exploitation.[2,5,434] There are prior examples of other groups with shared biological ancestry having research conducted “on them” rather than in partnership “with them”.[1,435,436] The preferences of people within these sub-populations about how they would like to work with researchers conducting research with them and how they would like to be involved are established.[1]

Here, we involved people who know themselves to be members of a donor-conceived sibling group in co-designing this research. This case study documents a participatory action research process with members of one of the largest biologically-related (donor-conceived) family groups ever documented (hereafter 'the sibling group').[428,430,437–439]

Members of the sibling group are all offspring of one sperm donor, the scientist Bertold Wiesner, a consulting biologist at the Royal Northern Hospital in the 1940s.[440] He may have fathered up to 1,000 offspring between 1942 (or earlier) and 1967[430,438,439] through the medical practice of his wife Dr Mary Barton, despite a 1945 British Medical Journal paper where Barton and Wiesner stated they set an 'arbitrary limit of 100 children for each donor'.[441] This study represents the first of its kind done with this group of siblings. The 18 siblings known to each other at the time of this research project are the living members of the cohort who are both aware that they are part of a larger group of up to 1000 people who share the same biological father and have chosen to make ongoing contact with each other (hereafter 'known siblings').

A formal database of who is descended from Wiesner is not known to exist. It is therefore possible that some descendants have lived their whole lives and then died, without knowing their biological father was Wiesner. The purpose of this research was to involve known siblings in online discussions about how they would like to be involved in future research, to better understand preferences of the group members in order to inform any future genomics research. More information about this sibling group and relevant contextual and historical information can be found in Additional File 1.

We have also reported the participatory action research process and outcomes using the novel system 'Standardised Data on Initiatives (STARDIT)' , which is an open access data-sharing system to standardise the way that information about initiatives is reported.[196,295,442] More detailed information about STARDIT can be found in the section about the Beta version.[9]

Methods

Study Design

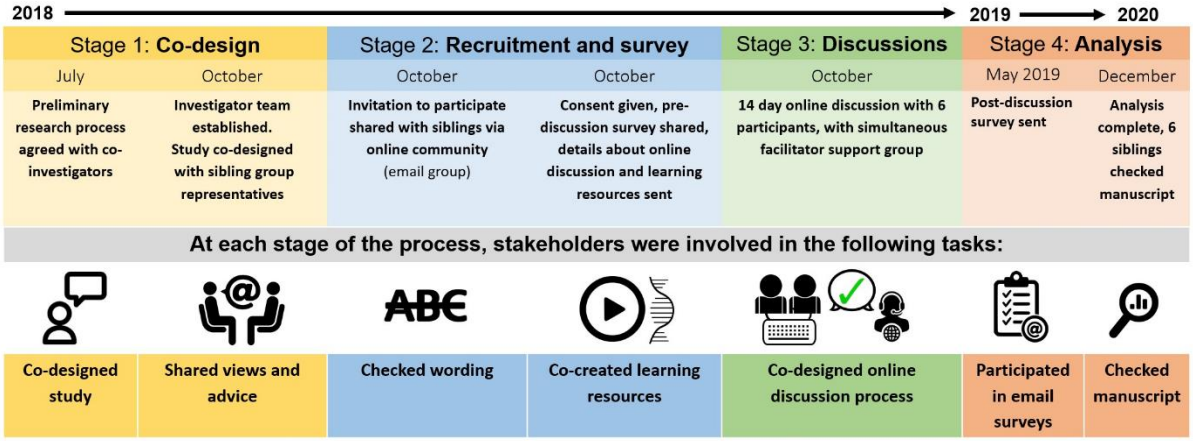
A participatory action research method was chosen for the study with co-design and reporting guided by a number of frameworks. [80,91,127] 'Participatory action research' is an umbrella term which describes a number of related approaches, including forms of action research which embrace a

participatory philosophy and include 'co-design' and 'co-production' of research.[88,233] It is a process where researchers, relevant stakeholders and sometimes the public "work together, sharing power and responsibility from the start to the end of the project"[89], including knowledge generation and translation.[89] The term 'stakeholder' means anyone who has a 'stake' in the research, in particular those with important knowledge, experiences, expertise or views that should be taken into account.[21,31]

After one of the investigators (JN) discovered his biological grandfather was Bertold Wiesner, the study team invited a researcher who had previously worked with members of the sibling group (MC) to join the study team. The biological relationship of the investigator JN was then used as a starting point by the study team to work closely with both potential participants and experts in human research ethics from the La Trobe University to ensure the method was acceptable and that no one (including the study team) would be exposed to avoidable risk. Guidance on ethical involvement of potential participants in research design remains unclear[127], in particular when it involves individuals conducting research with biological relatives.[443] Accordingly it was decided by the study team to commence co-design only after ethics approval was obtained. Feedback from the co-design process was later integrated into the study design through multiple subsequent modifications to the initial research plan, with each modification being reviewed and approved by the Human Ethics Committee.

The study had four stages: 1. Co-design; 2. Recruitment and pre survey completion; 3. Online discussion and post survey and 4. Analysis, with stakeholders involved in tasks at each stage (Figure 7.1).

Figure 7.1: Stages of research



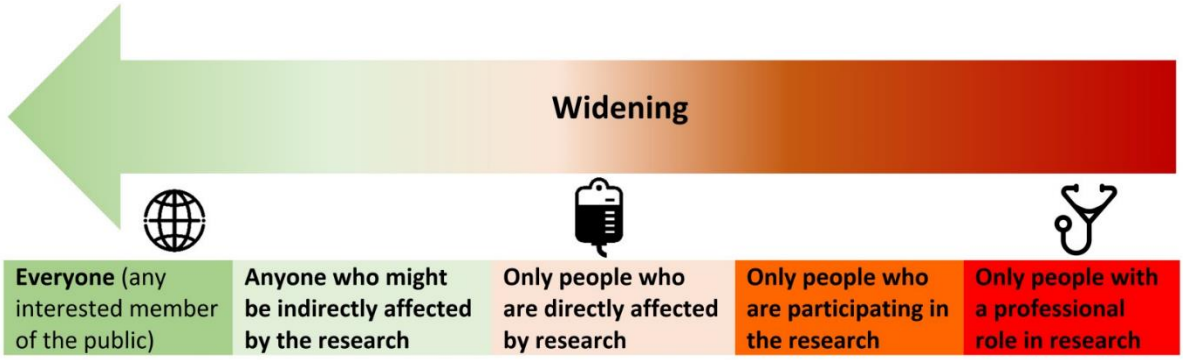
However, reporting of such involvement is often inconsistent, of variable quality[1,209], or the reporting frameworks themselves can have limitations.[444] STARDIT was used to report involvement, as it overcomes some of the identified limitations of other reporting tools by allowing people to self-identify with multiple labels such as 'researcher' and 'patient', and by transparently allowing multiple stakeholders to be involved in reporting which stakeholders have completed which tasks.[442] It has been proposed as a way of describing the 'who', 'how' and 'what' of research, and as a way to report two-way learning and 'transformational learning' amongst other impacts.[7,33]

As the studies were designed to facilitate comparison in data, accordingly, some aspects were inflexible and thus more accurately described as 'co-refined' rather than 'co-designed'. For example, questions in the pre- and post-surveys relating to 'who should be involved in research' and were designed to explore any changes in views on which stakeholders should be involved in which tasks during research, and were worded identically in each study in order to allow comparison with standardised data.[7,196] Participants could choose from the categories outlined in Figure 7.2, with a change in direction towards more people being involved labelled as 'widening', the inverse as 'narrowing'. The facilitator (MC) was also asked to complete a survey, which was informed by the Public Involvement Impact Assessment Framework Guidance (PiiAF)[91] and the questions were informed by the GRIPP2 reporting checklist.[80] The survey was identical to a survey used in other studies to facilitate data comparison.[6,7] All survey questions can be found in full in 'Additional File 1 - Data and analysis'.

Similarly questions about demographic information for the two studies were informed by the Genioz study to allow comparison with a wider dataset.[7,49] The choice of the software used in the online

discussion was also fixed owing to ethical constraints, including physical data location and ownership by participants.

Figure 7.2: Who should be involved in research?



Online methods of discussion are appropriate where a group of people is geographically dispersed and face to face discussion is impractical.[445] We therefore selected an online text-based discussion format, where people could log on at any time and contribute (asynchronous), including replying to other people's comments (threaded discussion). This platform allowed flexibility compared to 'real time' (synchronous) discussion, especially regarding participants contributing from different time-zones. An open source software web application called 'Loomio'[420] was installed on virtual machines hosted by an Australian Government initiative called 'Nectar Cloud'.[421] Participants could securely log-in from anywhere in world and participants' data was stored securely on servers physically located inside Australia.

The online discussions were facilitated for 14 days and led by one facilitator (MC). The facilitator was trained in advance to use the online discussion platform, given learning resources, invited to be part of a community of practice of other facilitators running similar discussions[7], and given support via telecommunications (by JN) to discuss any emerging issues. At the start of the online discussion, the facilitator asked participants to consider agreeing on boundaries relating to acceptable conduct, and invited members of the discussion to co-create a group agreement on conduct. The facilitator judged when to introduce new topics (depending on the engagement with each topic) with the recommend schedule below in Table 7.1 used as a template. The facilitated discussion period also included the provision of learning materials (short videos, infographics and information about research terms). These learning resources are included in 'Additional File 1 - Data and analysis'.

Table 7.1: Discussion overview

Question	Suggested staging point
Agreeing boundaries	Day one
What do you understand by the word 'research'?	Day one
What do you understand by the phrase 'genomic research'?	Day one
Why do we do research?	Day two
Which aspects of any future research genomic research should be influenced by the which groups of people?	Day four
What methods do you think could be used to involve those people in future genomics research?	Day six
Do you have any ideas, thoughts or reflections that have not been shared yet?	Day seven
Discussion closed	Day 14

The learning resources provided during data collection drew on the outcomes of an analysis of educational materials by the Australian Genomics Health Alliance. [416] Resources were selected by the investigator team in partnership with the Australian Genomics Health Alliance and refined following feedback from the volunteers. Detailed information about which learning resources were shared at which stage of the study can be found in Additional File 1.

Data analysis

We collected and analysed both qualitative and quantitative data during the participatory action research process. Data sources included pre- and post- surveys; online text-based discussions; meeting notes; emails between the study team members; surveys of the study team; comments shared by two of the study team (JN, MC) during the online text-based discussion; and reflexive research diary entries of one member of study team (JN) (Table 7.2). The data collection and analysis was informed by a number of frameworks and approaches for conducting and reporting qualitative research[393,394]; and conducting[153,158,446–449,160,161,163,178,180,182,397,399] and reporting case studies.[196,450] Participatory research processes, including stakeholder involvement, were informed by frameworks and best-practice for conducting[21,184,185], reporting[196,212,451], and assessing involvement in research.[91]

Coding and thematic analysis of qualitative data was carried out by two authors (JN, MC) independently; the analysis then checked (triangulated) for validity by a third author (PL), and

participants were invited to review the analysis ('member checked'), which is best practice for enhancing validity in qualitative methods.[182] In addition to quantitative analysis, each source was analysed using the method of thematic analysis, which involved stages including data mapping and familiarisation, transcription, coding, searching for themes, reviewing themes with study team members, labelling and summarising themes, and reporting the findings.[182] Participants were invited to review the analysis before publication in order to check whether they felt it reflected the research process accurately. They were also invited to share any further data via the STARDIT report.[295]

For the online discussion, we also used case study methodology to record and describe our experience of involving participants. This process is presented as an 'instrumental' case study, where the purpose is to understand the particular case, and attempt to provide data that could produce useful generalisations by using inferences from the data.[163] The co-design of the case study was informed by best practices for enhancing validity and rigour in the case study methodology.[153,160,452,180–182,397,399,447–449] In order to aid analysis and comparison with other case studies, we used 'Standardised Data on Initiatives' (STARDIT) Alpha Version to consistently map preferences for involvement and report ways people were involved in the participatory action research process.[196] Both the STARDIT report and the preference mapping can be found in the additional files. Further information about the case study method, data and both qualitative and quantitative analysis is provided in 'Additional File 1 - Data and analysis'.

Table 7.2: Summary of data analysis

Data source description	Analysis method
Meetings – including meeting notes and relevant documents	**Qualitative** (thematic analysis)
Online survey – text data from open and closed questions	**Qualitative** (thematic analysis, STARDIT-PM) **Quantitative** (number of responses)
Online discussion – text data from a facilitated and moderated online discussion of both participants and a separate one for the study team	**Qualitative** (thematic analysis, STARDIT-PM) **Quantitative** (number of responses)
Study team surveys – responses by email	**Qualitative** (thematic analysis, STARDIT-PM)
Other data – reflexive research diaries,	**Qualitative** (thematic analysis)

Participants and recruitment

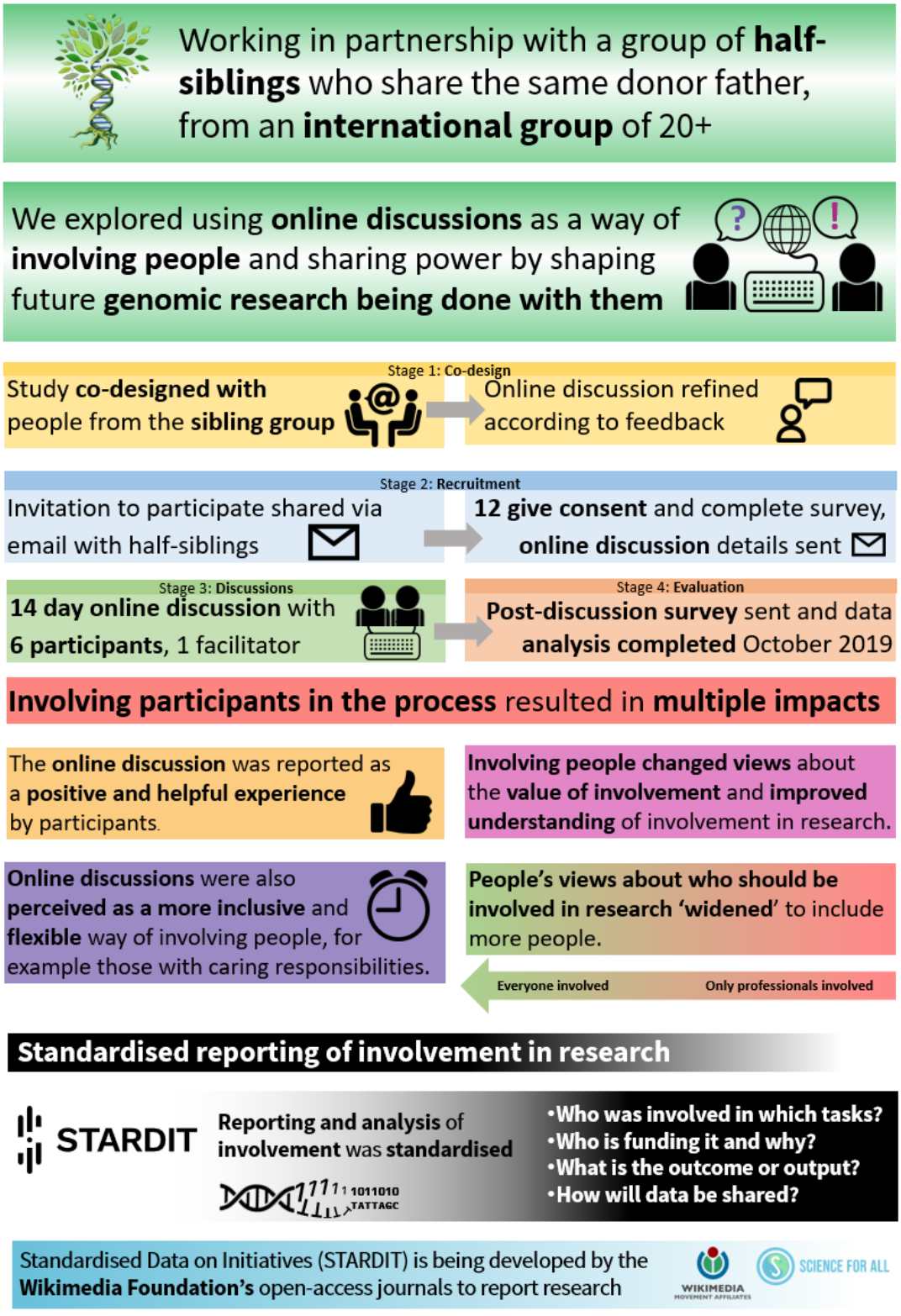
The selection of the particular shared ancestry population for this case study was informed by a number of factors which were appraised by the study team, including pragmatic considerations coupled with extensive consultation with professional ethics advisors and other experts.[178]

In October 2018 one member of the sibling group forwarded an email invitation to the other 17 known members to join the study. The invitation to participate contained a link to the participant information document; a plain English summary about the study; some learning resources about genomics research; and an informed consent form. We offered the choice of anonymity (through using pseudonyms or temporary email accounts) in survey completion and the online discussions to allow people to participate without disclosing sensitive or personal information.

Those who gave consent were invited to complete the online pre-discussion survey and were then contacted by email directly by a member of the study team (MC), who shared information about joining the online discussion alongside relevant learning resources. This included a short 60 second online video about the study, giving information about the context and purpose, and a one-page infographic summary of a scoping review about involvement in genomics research.

Results

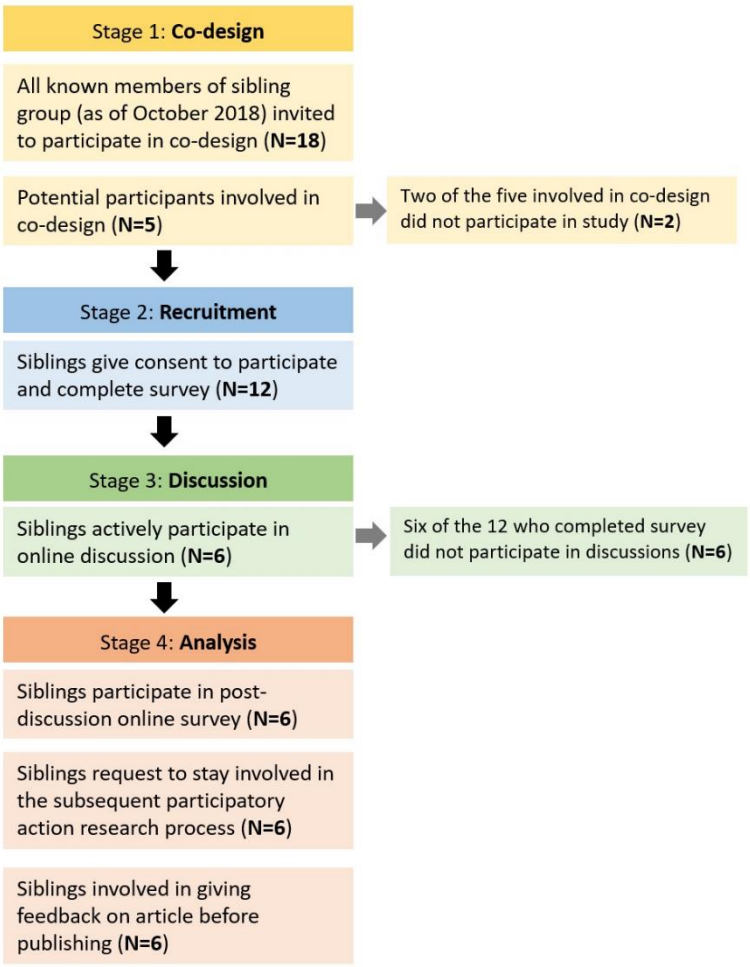
Figure 7.3: Summary of the research process



Stage 1: Co-design

Concepts such as ‘co-design’ and ‘co-creation’ are used here to describe involving people in the respective tasks such as helping design the research project, or providing input during the creation of a learning resource. These concepts can be considered as part of participatory action research.[88] Five members of the sibling group gave feedback during the co-design process (Stage 1), three of whom went on to participate in the study. Their input during the planning and co-design stage had clear positive impacts, particularly in improving educational resources and ensuring the pre-discussion survey used appropriate and acceptable wording to describe the sibling group, the members and the sperm-donor. During the co-design process it was agreed that the initial study should only involve the siblings, excluding any of their offspring. It was agreed during the co-design process that direct communication with the sibling group would be conducted by a biologically unrelated member of the study team (MC) once the study commenced, owing to the ethically novel situation of one of the investigators (JN). Figure 7.4 summarises how many people participated in the stages of the study. Of the 18 known living siblings in 2018, 14 participated in the co-design stages or the study.

Figure 7.4: Sibling group involvement and participation



Stage 2: Recruitment and surveys

Of the 18 members of the donor sibling group, 12 gave consent to participate and completed the pre-discussion survey of whom six participated in the online discussion. All six completed the follow-up survey in May 2019 and shared consistent identifying data at all stages, including the initial survey, the online discussion and the follow-up survey, allowing a comparison of results throughout the process.

Of the 12, seven were female and five were male. All stated that they were comfortable describing other descendants of their biological father as 'half-siblings'. Self-reported educational attainment was mostly 'degree (bachelors), diploma or post-graduate', with one participant having professional experience in medicine and genomics. All were aged between 50-74, with most living in the UK. Detailed demographic information can be found in 'Additional File 1 - Data and analysis'. Most agreed with the statement that they have a shared interest in discussing future research which

might affect them, including genomic research. More information about participant demographics can be found in Additional File 1.

From the six participants who completed both the pre-discussion and follow-up surveys, a total of 54 responses were given. Of these, 35% showed a change towards 'widening' involvement (N=19/54), 8% 'narrowed' (N=8/54) and half stayed the same (N=27/54). More detailed data is provided in Additional File 1.

Stage 3: Online discussions

Six people participated in the online discussion in October 2019. Only one chose to use a pseudonym in the online discussion, so the study team could identify them. Some of those who gave consent to participate did not make comments but logged-in multiple times and read comments. Table 7.3 shows how many comments were made by each participant in the online discussion. The themes where most participants shared views are summarised in Table 7.4. The most discussed themes included the unique nature of the sibling group, that anyone should be involved in research, questioning who decides ethical oversight in research, questioning research for profit and sharing concerns about genomics research being used for political purposes.

Table 7.3: Number of comments in online discussion

Participant ID	Number of comments
P2	12
P4	14
P5	42
P6	15
P7	9
P9	13
Facilitator (MC)	65

Table 7.4: Quantitative summary of themes

Theme	Number of participants
Anyone should be involved in research	**6**
Research with sibling group is unique and complex but important	**6**
Those affected by research should be involved	**6**
Research for profit and 'bullying' by 'big pharma'	**6**

Theme	Number of participants
Who decides who decides what is ethical?	**5**
Concerns about genomics research being used for political purposes	**5**
Finding out they are part of sibling group has been a positive experience	**4**
View on topics for research	**4**
Participants reported changed views and perspectives as a result of participating	**4**
Desire to improve situation for people affected by assisted conception	**3**
Interested in learning what other siblings think and discuss issues	**3**
Concerns about control of knowledge and data	**3**
Questioning which groups should have 'equal influence'?	**3**
Questioning eugenic attitudes to genomic variations	**3**
Views on participation in genomics research	**3**
Participants learned about genomics	**3**
Motivation for participation to help researchers and sibling group	**2**
Uncertainty about what they can offer but happy to help	**2**
What is the purpose of research?	**2**
Experts should be involved (over seen by ethics boards)	**2**
Developments in genomics have significant implications for society	**2**

Stage 4: Analysis

Participant feedback in the post-discussion survey was positive, including that the experience was 'interesting', and that they 'enjoyed thinking about the questions posed and reading the responses of others' and the 'perceptive comments' of the facilitator [P7] [P4]. Two reported experiencing some usability issues with the online platform. All who completed the survey opted to stay involved in the next stages of the participatory action research process.

Table 7.5: Summary of pre and post survey responses

Question	Results
What made you decide to respond to our invitation to participate in this project? 12 participants responded (pre-discussion)	Participants wanted an 'opportunity to be involved in research' [P3], wanted to 'learn more' [P7] and regarded involvement as a 'civic duty' [P10]. Others stated this study may be useful to 'future donor offspring' [P11] and one participant stated

	'curiosity' was a reason for participation. Four participants noted a familial connection to a member of the study team (JN).
What do you hope to get out of participating in this discussion? Do you have any specific expectations? 8 participants responded (pre-discussion)	Participants were 'interested in hearing what their half siblings think' [P4] and wanted an 'opportunity to discuss' and 'think through the issues involved' [P4] [P6]. One participant said they wanted 'the satisfaction of knowing that I may have contributed' to the study [P10]. Another wanted 'to be useful to the researchers' [P11]. One participant noted an expectation of anonymity while participating.
Do you have any ideas about how the different people could influence future research? 7 participants responded pre-discussion and 3 post-discussion	Five participants said 'everyone should be involved in research', with one adding 'not just scientists and researchers' [P5-pre]. One participant said 'anyone with an opinion' should be involved [P3-pre]. Another stated 'researchers and those who are affected by what is being researched' [P4-pre]. One participant stated 'People who know their subject but do not have hidden motives or agendas' should be involved [P2-pre]. One participant said the answer depended on 'what kind of research it is' [P7-pre] and one mentioned 'ongoing discussions' using online tools [P5-post]
Is there anything in particular you liked or thought was helpful about how the discussion was conducted? 5 participants responded (post-discussion)	One participant stated they 'liked and appreciated the opportunity to participate' [P6]. One participant stated the process 'seemed to work well' [P7]. One participant added 'I think it is commendable that there is a concern about participatory research' [P9]
Is there anything you didn't like, thought was unhelpful or could have been improved about how the discussion was conducted? 5 participants responded (post-discussion)	Two participants reported finding the platform 'complicated' and problematic [P5]. Two participants stated they would have liked 'more time' for the process [P4].
Did you have any expectations from participating in this research that were met or not met? 6 participants responded (post-discussion)	Four participants stated their expectations were met. One responded that they 'found some of the questions very complex and had difficulty answering them' [P5].

Outcomes from the process

There were 8 outcomes reported from this process, which are summarised in Table 7.6, with additional information available in the accompanying STARDIT report (Additional File 2).

Table 7.6: Summary of outcomes from the process

Outcome	Summary and learning point
1. Improved understanding of genomics informed participation in future research	Participants reported their understanding about genomics research increased as a result of participating in the study. Learning from this process informed subsequent discussions in the sibling group about participation in research, including a proposed self-managed biobank.
2: Learning resources useful	Participants reported finding the information resources and videos useful. **Learning point:** Creating learning resources in multiple formats (hyper-text, infographic summaries, videos with subtitles animations) will ensure that information is more useful for people with neuro-diverse learning needs.
3: Changed views and perspectives as a result of participation	Participants reported their views and perspectives changed as a result of participating. **Learning point:** Online discussion facilitated collaborative learning and the changed views of participants can be viewed as an impact of 'transformative learning'.[131,171,445]
4: Participants asked to stay involved in the research	All participants who completed the follow-up survey opted to stay involved in the research process. **Learning point:** Participants demonstrated 'critical reflexivity', a stage of participatory action research which asks people involved to reflect on their views and the causes of problems and to be involved in exploring any solutions and the actions that people can take to bring about improvements.[130,131,136]
5: Participants enjoyed the online discussions	Participants stated the experience of participating was 'interesting' and they 'enjoyed' it [P7] [P4], despite some usability barriers. **Learning point:** Some participants stated they preferred online discussions over face to face discussion or interviews, which highlights the importance of mapping potential participants' preferences when co-

	designing involvement, to ensure research methods meet the needs of participants.
6: Improved understanding of how to get involved in research	Participants reported improved understanding of how to get involved in research; this helped inform decision making for individuals when invitations were sent to members of the group to participate in genomics research after this study and unconnected to this study.[295] **Learning point**: Pre and post discussion surveys and standardised reporting (STARDIT-PM) are useful tools for mapping changes in understanding and views.[130,196]
7: Co-design changed study design	Feedback from participants resulted in changes to the study design including improving language used in recruitment and learning resources. **Learning point:** Involving participants in helping co-create learning resources can improve them.
8: Method for future research co-design established	Participants stated that the methods used in this process could be helpful when co-designing future stages of proposed genomic research with the sibling group. **Learning point:** STARDIT can be used to map preferences and impacts from future co-design processes.[196] Learning from this process is relevant to sub-populations where people share recent ancestry such as some Indigenous populations[2,3,5], and sub-populations of people affected by rare diseases.[58]

Participant views about genomic research and involvement

Participants demonstrated improved understanding of the difference between participation in research and involvement in research by the end of the discussion, although initially some were confused by the distinction. One stated 'I am a strong supporter of patient involvement in medical care' and that 'involving members of the public' in genomic research was important in order to 'have their views, reactions, interpretations, questions, concerns sought, interacted with, and considered' [P11].

All six participants of the online discussion thought that anyone should be involved in research, with one saying 'everyone should have a voice not just scientists and researchers' [P5]. Another added that it is a 'good idea to involve research subjects in formulating the research questions' [P10].

Methods of involving people were discussed in detail with a number of options explored. Participants explored ideas around the purpose of research and one stated that research participants should be involved in 'agreeing purpose, parameters and methods' [P7]. All expressed concern about research for profit and those with financial interests influencing research without the public being involved.

One participant noted that being 'highly educated' was an enabler for involvement as was having a 'bit of time on their hands' [P4]. Another noted they didn't feel 'qualified' to 'comment on aspects of science itself' but felt 'strongly' that they should be involved in ethical decisions and sharing personal experiences to help inform research [P5].

One participant asked 'there will be many interested groups so which ones will be listened to?' [P4]. Another noted they felt that certain pharmaceutical companies were responsible for 'bullying', contributing to 'disinformation; ignorance and inflexibility of medical and scientific professions' [P12].

Participant views about proposed research with sibling group

Participants were supportive of future genomics research with the sibling group. One participant stated it would be ‘worth the effort’, and another stated they ‘wholeheartedly support the involvement of the next generation’ in any future research with the sibling group and noted any study design should ensure new siblings and their offspring should be involved to ensure they can ‘become part of the research’ [P9] [P5].

In reference to future research with the sibling group one participant stated that ideally 'we would be able to exert control over the use' of data [P7]. Conversely, one participant stated in the follow-up survey that they ‘could not at all care whether my genomics are public or not. I do not see that my genome is a matter for privacy concerns’ and recognised that others may feel differently [P9].

Participants spontaneously shared their views about what possible areas of research topics could be explored in studies they could participate in and how these could be conducted. These included 'mental health' [P6] and pharmacogenomics [P4], with non-health related topics including 'career choices' and hobbies [P4]. One participant felt they should be involved in 'seeking answers to old, or not yet thought of questions' and 'looking beyond the known into a murky unknown' [P6].

Stakeholder views about the process

Participants reported their motivation for participating in this research process was altruistic, to help researchers and the wider public. Some also recognised that their participation might directly benefit members of the sibling group itself. One participant also stated ‘it is commendable’ that ‘participatory research’ was being used, in reference to the research methods used by the study

team [P9]. While online discussions were perceived as having advantages and 'worked well'[P7], two participants reported usability issues with the online platform.

Four participants identified specific things about the way this study was conducted that enabled their involvement. One said the entire process was 'assiduous' and that the 'intent of this project' was 'obviously thoughtful and interesting' [P9]. One participant said the 'system seemed to work well' [P7]. Another added that being used to online platforms like Loomio, or having previous experience of similar platforms and 'used to' that way of communicating might facilitate involvement using such a communication mode [P5].

Four participants reported specific things about the way this study was conducted that were barriers to their involvement. The pace of the discussions was mentioned as moving 'too quickly' with another adding 'more time' was needed and that the study team should 'reconsider the pace of the research' [P7] [P4] [P5]. A discussion about creating boundaries in the discussion revealed that some participants felt that they should avoid 'topics which might trigger emotions which are stressful' whereas others thought this could be viewed as 'restrictive, even censorious' [P7].
The study team noted a 'critical mass' effect in online discussions, with the pace of comments seemingly affected by the number of posts. One study team member noted the difficulty in achieving 'the balance of being prescriptive' (for consistency) and giving freedom to facilitators to initiate discussions and follow emergent themes. Support for the facilitator by the study team was identified as an important enabler of the research process by the facilitator. Further barriers and enablers are summarised in the accompanying STARDIT report (Additional File 2).

Discussion

In this study we used a participatory action research process to explore the views of a group of 18 known donor-conceived siblings, with participants reporting enjoying the overall experience of the surveys and discussion forum. Online text-based discussion forums were reported as an inclusive way of involving people which was more flexible for communities which exist across time zones. Participants reported it also gave them more time to reflect on answers, learn collectively as a group and provided the freedom to ask questions and share ideas throughout the process. Participants reported that the participatory process described here appears to have resulted in members of the sibling group feeling they will have more influence over research done with them[295].

Co-designing the study ensured it was more likely to meet the needs of potential participants. Involving participants in co-designing the research process resulted in a number of improvements to the study design, including improving language used in recruitment and learning resources and co-creating acceptable online discussion boundaries. The process improved participants' understanding about genomics and research. However, data from participants indicates that increasing the time allowed for any future discussions would help ensure the process does not move 'too quickly' [P7].

Participation in the process led to eight outcomes, including participants 'widening' their views about who should be involved in research to include more people. Participants reported changed views about the value of involvement and an improved understanding about how to be involved in genomics research. Some participants reported via the co-authored STARDIT report that learning from this process informed subsequent discussions in the sibling group about participation in research, including a proposed self-managed biobank and helping them make informed decisions about participating in genomics research.

During the online discussion, the facilitator made significantly more comments than any one participant, reflecting the level of work and engagement required to facilitate a discussion. The 'critical mass' effect of a certain number of posts in a discussion encouraging others to post aligns with findings from other studies which have explored participants' hesitancy in posting in online discussion forums[453]. Only one participant used a pseudonym during online discussions, and this may reflect that the other group members felt comfortable sharing views with others in the group and trusted the security and privacy of the platform, however it is unclear why some participants logged in but did not comment.

Throughout the process, the study team was faced with ambiguous policies for the ethical involvement of people in co-designing participatory research, which hamper the degree of control that potential participants had in the research process. Members of the study team reported they felt that limitations in the ethics process affected the extent to which the sibling group could be involved in the participatory action research process. This is reported in more detail in the accompanying STARDIT report (Additional File 2). Additionally, an unplanned delay of 9 months in collecting follow-up data (related to ethics processes) may have affected the quality of data collected post-discussion, with one participant adding 'given the lapse in time, I cannot answer' in response to a question about their experience of participating in the online discussion [P6].

Further clarity from ethics committees and relevant organisations about the ethics of participatory action research will enhance power sharing at this crucial stage of research, with standardised reporting of data allowing direct comparison of ethical methods of involvement. One participant's suggestion that the sibling group should form their own ethics committee to guide future research reinforces the importance of transparent ways of sharing power and control in genomics research [P6], in particular for sub-populations more likely to be exploited. This includes other sub-populations where people share recent ancestry such as some Indigenous populations[2,3,5,434]; populations of people affected by rare diseases caused by similar genomic variations[58]; and other sub-populations where there is a perceived power-imbalance between researchers and potential participants.

While the focus of the online discussion was involvement in genomics research, the discussion entered additional areas. This reflected the interconnected nature of the subject, including the ethical, legal and social implications of all genomics research. The discussions thus served as an exploratory focus group, mapping participants' views in these diverse areas. For example, four participants spontaneously raised the 'ugly' issue of eugenics and eugenic attitudes to genomic variations, including the distinction between perceived disability and lived-experience of having certain genomics variations [P6]. Noting the history of eugenics, participants expressed fears that genomics research would continue to be used to reinforce racism and assist with genocide. One participant cited the well-documented historical precedent of a large technology company being complicit in enabling regimes to carry out negative eugenics policies[454], and also raised concerns about contemporary and future 'misuse' of genomic data 'for immigration' [P6].

The complex ethical, legal and social implications of genomics research reinforce the importance of further research to evaluate data from discussion methods such as the ones described in this article. Evidence-informed methods are urgently required in order to inform education, debate and develop international consensus on the ethical use of data from genomics research.

Strengths

Measuring outcomes such as 'transformative learning' can be challenging, but is an essential requirement for understanding processes such as involving stakeholders in research and knowledge translation[131,171,445]. The process followed in this study approached such challenges by collecting standardised baseline and follow-up data about views. The findings from these measures suggested

that involving people in online discussions about involvement in research can change their views about who should be involved in research, including 'widening' such views towards support for the inclusion of a wider category of stakeholders. Similarly, using STARDIT for standardised preference mapping and reporting of involvement meant outcomes from the process could be mapped more precisely[130,196], including outcomes beyond the date of the initial data collection. Participants reported their views and perspectives changed as a result of their participation, suggesting that online discussion facilitated collaborative learning; changes in views; and 'transformative learning'.[131,171,445]

Participants reported their understanding about genomics research increased as a result of participating in the study. Data from the STARDIT report completed at the end of evaluation stage of the research process indicated that learning from being involved in online discussions informed subsequent discussions in the sibling group about participation in research, including a proposed co-managed biobank.[295] Since participating in this study, some known siblings have received invitations to participate in genomics research and share their genomes with researchers unconnected to this study. The research project described in this article helped inform their decisions to participate[295], and how they would like to be involved in shaping the future of other research with the group. In addition, the sibling group has also begun discussions within the group about future research with the group, including a self-managed biobank, with discussion informed by learning from this process.[295]

Involving potential participants in the co-design of any future research is essential to ensure it is appropriate and acceptable. The importance of using STARDIT-PM for standardised preference mapping when co-designing involvement was also demonstrated by participants stating their preference for online discussion methods over face-to-face discussion or interviews. Such preference mapping helped ensure the research design met the needs of potential participants.

Providing an 'updateable' way to report ongoing impacts and outcomes is impossible or, at best, impractical with traditional academic publishing. Since starting the study, the number of known living relatives has grown to 46 half-siblings and 24 half-cousins, owing to previously unknown siblings and cousins taking direct-to-consumer DNA tests. The updatable 'living' report provides a way for these additional siblings to integrate the views and preferences into future STARDIT reports, and report any ongoing impacts or outcomes.

While this study was co-designed and conducted before the COVID-19 pandemic, the learning outcomes from the process (summarised in Table 6) have relevance to research disciplines beyond genomics. The methods of involving people aged 50-74 in an online discussion described here now have an unexpected relevance to many disciplines. As research projects around the world seek to involve people online in novel ways, using a system such as STARDIT to report and evaluate such methods (including co-design) in a standardised and updateable way that works across languages is more important than ever.[442] In particular, as more people become aware of the importance of storing data according to the preferences of research participants, hosting online discussions on platforms where data is stored according to these preferences is vital to ethical research conduct.[6,7] The detail and transparency of the methods described by this article and the accompanying STARDIT report provide a repeatable method for co-designing such discussions, using free and open-source software.

Limitations

While 14 of the 18 known siblings were involved in either co-design or as participants, only 12 siblings gave consent to join the study, and only 6 of these participants made comments in the online discussion. Some of those who did not make comments logged-in multiple times and read comments. While the follow-up survey attempted to capture the views of all 12, only the 6 active members responded, so it is hard to understand the behaviour patterns of the 6 members who did not comment. The relatively small starting cohort (18) for this study and a smaller number of active participants (6) means that although there is useful learning from our findings, their statistical significance can only be enhanced by standardised data sharing that can then combine with results with other studies.[196]

As the study was designed in parallel with other studies, some aspects of the study were inflexible during the co-design process (such as the specific open-source discussion platform that was used). Some aspects could more accurately be described as 'co-refined' rather than 'co-designed', thus limiting some areas which could be influenced by the co-design process. Using STARDIT to report which stakeholders did which tasks has attempted to overcome subjective distinctions between such terms.

In the time between the start of this research process and the publication date of this thesis (December 2021), there are now 46 half-siblings and 24 half-cousins known. Ethical constraints limited recruitment of newly discovered siblings. Further involvement to understand views and

preferences about research with the now much larger sibling group would help ensure any future participatory research meets the needs and expectations of the group.

Conclusions

The process described here provides a replicable and practical method of involving potential participants in co-designing genomics research using online discussions, with reported positive outcomes. Co-designing the study ensured it was more likely to meet the needs of potential participants and resulted in improvements to the study design. Reporting this study using 'Standardised Data on Initiatives (STARDIT)' to report preferences, plan and report involvement, evaluate participatory research methods and report 'updateable' outcomes allows ongoing comparison with other studies. Such reporting facilitates learning from this case study and contributes to data to inform evidence-based decision making when planning future research. Learning from this study contributes to the current evidence-base used to inform future ways of involving people in genomics research. Such evidence can be applied in the context of research such as self-managed biobanks for sub-populations more likely to be exploited and other sub-populations where there is a perceived power-imbalance between researchers and potential participants.

This is the end of the extract from the article 'Co-designing genomics research with a large group of donor-conceived siblings': https://doi.org/10.1186/s40900-021-00325-7

STARDIT MICRO Report: Co-designing genomics research with donor conceived siblings

This STARDIT MICRO report contains the minimum data required for a Standardised Data on Initiatives Alpha version report (STARDIT Alpha).[196] A full version of this STARDIT report can be found in the supplementary resources of the article available here:

https://doi.org/10.1186/s40900-021-00325-7

The full prospective STARDIT report for this article can be found in the Appendices under the heading 'Standardised Data on Initiatives (STARDIT) report – Alpha Version: Co-designing genomics research with a large group of donor-conceived siblings' with a link to the online 'living' Beta version of the STARDIT report available in the references.[295]

Name	STARDIT MICRO Report: Co-designing genomics research with donor conceived siblings
Involvement	
Who was involved	Group 1: Academic research investigators (Jack Nunn, Marilyn Crawshaw and Paul Lacaze) Group 2: Members of the sibling group who gave feedback during the co-design stage (including but not limited to David Gollancz and Michael Bywater) Group 3: Members of the sibling group who participated in the research and gave feedback as part of the co-design process, the manuscript checking stage or contributed data to the STARDIT report (including but not limited to Shirley Brailey, Barbara Nunn, Adrianne Smith and Barry Stevens)
Specific tasks of this person or group (list as many as possible) – including any information about why certain people were included or excluded in certain tasks	Group 1: Involved in co-designing every stage of the process, analysing data Group 2: Members of the sibling group were involved in refining wording of participant information, sharing views and advice about the process, proof-reading documents, providing feedback on surveys, analysing data, informing planning, and providing feedback on planned online discussions.

	Group 3: Participants were also involved in checking the content of Genetics Society UK podcast[455], with the recording shared with all participants before dissemination to ask them to check the content was accurate and acceptable. Group 2 and 3: Participants were sent the article and additional files to check the analysis and content and were invited to be authors of the STARDIT report.
What was the outcome or output of the involvement of these people? What changed as a result of involving people?	Improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the sibling group to describe biological relations), improved online discussion, improved learning resources for participants, improved co-design process.
Mapping financial or other 'interests'	
Describe any financial relationship or other interest this person has to this project	One investigator (Jack Nunn) is biologically related to participants from the sibling group, with one being his mother and all being half-aunts or uncles.
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	It will be published open access in peer reviewed journals with identifying information removed in order to anonymise it as much as possible.
How is it stored and hosted?	It will be shared on a public domain repository.
Who is analysing the data?	Group 1: The study team described above Group 2: participants were invited to review the analysis and give feedback to ensure they felt it reflected their experience of the process
How is information about this data disseminated?	It will be published in an open access journal It will be shared with participants of the research and also other members of the sibling group who have joined it since the study commenced. Learning from this process has been presented at conferences, and will be shared on social media and through other channels. Preliminary learning was shared in a UK Genetics Society podcast[455].
How is the data FAIR[85]?	Shared in the public domain licensed under a Creative Commons.
Impacts and outcomes	

What new knowledge has been generated? (if appropriate, include effect size, relevant statistics and level or evidence)	Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'.
Outcomes - Describe how the learning or knowledge generated from this initiative has or will be used	1. Co-design changed study design 2. The process improved participants understanding about genomics and research 3. Learning from this process informed subsequent discussions in the sibling group about participation in research 4. Participants reported finding the learning resources useful 5. Participants changed views and perspectives about genomics research as a result of participating. 6. Participants asked to stay involved in the research. 7. Participants enjoyed the online discussions.

Ethics approval and consent to participate

The La Trobe University Human Ethics Committee approved this study.

Project number: HEC18256

Project Title: Genomics Research and Involving People: Exploring the views and perspectives of people with shared ancestry about being involved in genomics research

Chapter 8 – Involving Australian Indigenous peoples in precision medicine

About this chapter

The UN has raised human rights concerns about Australian Aboriginal and Torres Strait Islander peoples' (hereafter 'Aboriginal' peoples) access to health.[456] In this chapter, I present a reflective case study in association with Sydney University's Poche Centre for Indigenous Health,[167,457] and their proposed 'Aboriginal Precision Medicine Project'. The project aimed to involve local community members in its design, data collection and management, in line with international best practice in community-controlled research. My participation included working with Aboriginal people in a remote community in Australia to co-design a protocol for genomics research.

This chapter summarises the background, methods, results, reflections and conclusions from my work, including learning and reflection points in the Discussion section. This chapter aligns with research aim 2: "apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the process of involving people in genomic research" Unlike the preceding three results chapters, this chapter as a whole is not published in peer-reviewed journals as a single case study. It is an overview of multiple different activities, represented in part by three publications in the area of genomics research and involving Indigenous peoples, which I authored or co-authored.

For clarity, where content from published articles is used, this is denoted with *italics.* The STARDIT Alpha version was shared as a pre-print for public consultation during this process and a number of authors from the Poche Centre for Indigenous Health joined as authors. A STARDIT MICRO report is included at the end of this chapter, with the full prospective STARDIT report in the Appendices under the heading 'Prospective STARDIT Report: A Pathway to precision medicine for Aboriginal Australians', with a link to the online 'living' STARDIT report in the references.[458]

Chapter summary

Background

Precision medicine, involving knowledge of a person's genome, offers a new way to improve the prevention, diagnosis and treatment of diseases. If implemented without consideration of equity issues, it could increase health inequalities. To improve the quality of healthcare and precision medicine for Indigenous peoples worldwide, improved ways of involving people that are culturally competent and safe are urgently needed.

Method

A reflective case study method is used here to describe my work with the Poche Centre for Indigenous Health at the University of Sydney, helping to advise on how the proposed 'Aboriginal Precision Medicine Project' can involve local community members in its design, data collection and management. The method and structure of this reflective case study is informed by the 'reflection cycle', and other relevant reflective case studies.

Results

Results from the participatory action research process included learning from the co-design process which informed the protocol creation, and the co-creation of the published protocol itself in 2021. In parallel to this participatory process, work with the National Centre for Indigenous Genomics (NCIG) on an article is also summarised, as discussions with staff from the NCIG informed Poche's participatory action research process, and learning from this process also informed the article led by the NCIG.

Discussion and reflections

Co-defining appropriate and acceptable terminology is a crucial stage of the co-design process. STARDIT has been designed to align with models of self-identification, and can be used to report how people have been involved in an initiative, with people able to chose any labels they self-identify with, including 'Aboriginal', 'Indigenous' or 'community member'. This removes the requirement (and the associated power) of researchers assigning such terms to people.

The published protocol demonstrates how such co-design methods can be reported, helping ensure future ethical oversight can be managed in partnership with Aboriginal people. STARDIT can be used to describe who was involved in co-designing every aspect of the research, and who was involved in making ethical decisions, providing more transparency to the entire process of ethical oversight.

Conclusion

The work described in this chapter provides a clear methodology for both planning and reporting co-design methods in genomic research with Aboriginal people. STARDIT can be used to create public records of every stage of the co-design process, including data about who was involved, how, and in which tasks. Information about the co-design method, the data analysis methods, reported enablers and barriers for involvement, and any other impacts and outcomes from the co-design process can also be reported. Such data improves transparency of terms such as 'co-design' and allows improved scrutiny and ethical oversight, including from potential or current research participants.

Introduction

This section is adapted from an invited article I authored, published in 'Health Voices', the Journal of the Consumers Health Forum of Australia. Titled 'Reducing health inequalities by involving indigenous people in genomics research', the article was reviewed by the editorial board prior to publication in 2019. [2]

Background

Precision medicine, involving knowledge of a person's genome, offers a new way to improve the prevention, diagnosis and treatment of diseases. If implemented without consideration of equity issues, it could increase health inequalities. To improve the quality of healthcare and precision medicine for Indigenous peoples worldwide, improved ways of involving people that are culturally competent and safe are urgently needed.[459,460]

Health inequality for Indigenous Australians

The UN states that Indigenous people have "social, cultural, economic and political characteristics that are distinct from those of the dominant societies in which they live", and that "special measures are required to protect their rights".[98] The UN has raised human rights concerns about Australian Aboriginal and Torres Strait Islander peoples' (hereafter 'Aboriginal' peoples) access to health.[456] The World Health Organization (WHO) and many other bodies have reported that Aboriginal peoples have lower than average life expectancies.[461] A 10-year gap in life expectancy for Aboriginal peoples compared with other Australians has remained largely unchanged between 2001 and 2015, despite attempts to improve health outcomes.[462] The nation's target to close the gap in life expectancy by 2031 "is not on track".[463] Depending on how it is implemented among Aboriginal peoples, precision medicine has potential to help close or widen that gap.

What is precision medicine?

Precision medicine refers to specialised and personal diagnosis and treatment of people. It requires knowledge of an individual's DNA (their genome) in order to make comparisons with what we already know about how genetic variations contribute to disease. There are significant differences in the type, distribution and frequency of variations in the genomes of human populations across the world.[464] For precision medicine to be effective, variations in the genome should be interpreted in the context of 'reference genomes' from appropriate ancestry groups.[189] These reference genomes are developed by combining the genomes of different individuals.[404] The more genetically similar people are to the reference genome, the more helpful precision medicine will be.

Why is there potential for further inequality in precision medicine?

There is a paucity of genomic data about Aboriginal ancestry groups as there are currently no reference genomes from these populations.[404] This lack of genomic data for specific ancestry groups may increase existing health inequalities in Aboriginal communities, as these populations will not be able to use precision medicine as effectively as others.[465]

How can inequality be reduced?

To improve the quality of healthcare and precision medicine for Aboriginal peoples in Australia, it is necessary to improve the research done with these communities. Building trust and working in partnership at all stages of the research process can minimise the risks of group and cultural harms associated with genomic research.[466] However, some researchers see the complexities of working with Aboriginal communities to co-design and govern data collection, and use it in inclusive, culturally appropriate ways, as a barrier to research. The 'unacceptably high level of disadvantage' and the low-quality of healthcare has prompted the UN to recommend that Australia "reset the relationship with Aboriginal people based on genuine consultation, engagement and partnership".[456] Improved ways of involving people that are culturally appropriate and genuinely share power are urgently needed.[456,459,467–470]

A strategic priority of the Australian 'National Health Genomics Policy Framework' is to "promote culturally safe and appropriate data collection and sharing" for Aboriginal peoples. The Framework also states that the value of genomic research "needs to be compared to the range of alternative options in terms of its ability to deliver health benefits, its capacity for harm and the costs of testing."[471] While existing unequal access to quality primary health care and other basic needs must be reduced,[472] to avoid amplifying inequalities further, the benefits of genomic medicine must be delivered equitably to Aboriginal communities, alongside other healthcare interventions.
The WHO has highlighted the importance of creating the conditions that enable people to take control of their lives.[473] In the USA, the 'All of Us' 'Precision Medicine Initiative' showed that "precision engagement" is required in genomics research.[68] If the Australian Government is "committed to working in genuine partnership" with Aboriginal peoples,[463] this must be demonstrated by adequate funding for working in partnership, including supporting communities to evaluate their involvement and creating an evidence base for best practice. If lessons from the Belmont Report on ethical principles for research involving human subjects are to be heeded,[474] vigilance is needed from the public, policy makers and researchers to ensure that the concept of 'involvement' is not reduced to a method of improving recruitment and participation in research which does not reflect the health priorities of those participating.[475] Everyone should be involved in

shaping the future of genomic research, and everyone should have access to the benefits of this new knowledge.

This is the end of the content adapted from the article 'Reducing health inequalities by involving indigenous people in genomics research'.[2]

Aboriginal Precision Medicine Project

The proposed 'Aboriginal Precision Medicine Project' (hereafter 'the Precision Medicine project') will be led by the Poche Centre for Indigenous Health at the University of Sydney (hereafter the Poche Centre), whose staff are 62% Aboriginal. The Centre works to influence genuine change in public policy and identify approaches to improving Aboriginal and Torres Strait Islander health. As part of my doctoral research, I was invited to volunteer with the Poche Centre, helping to advise on how the Precision Medicine project can involve local community members in its design, data collection and management, in line with international best practice in community-controlled research.
The Centre has been working with some Aboriginal communities on oral health and cardiovascular research since 2017.[476,477] The trust that members of Aboriginal communities developed for Poche Centre researchers was an enabling condition for the first stage of planning the precision medicine project.

Building on this trust and previous work, the Centre has involved local Aboriginal people in co-designing a proposed precision medicine research project, including how the community will be involved as partners throughout the project. The Precision Medicine project aims to sequence participants' DNA in order to create a clinically useful reference genome. This reference genome will contribute to improved health outcomes by improving the quality of precision medicine for Aboriginal people from communities in New South Wales. For example, it has the potential to improve prediction of cardiovascular disease and enable more targeted interventions for certain cancers and rare genetic conditions.

Methods

Case study method

A reflective case study method is used here to describe my work. The method and structure of this reflective case study is informed by the 'reflection cycle', and other relevant reflective case studies.[167,457,478] The stages of a reflective case study are (1) describing what happened; (2) reflective observation, including thoughts and feelings arising from what happened; (3) summarising relevant theory in order to explore what happened in the context of contemporary knowledge and evidence; (4) summarising any future action, and how this was informed by the previous stages. Data sources for this case study include a reflexive diary written throughout the research process, emails, meeting notes and relevant documents associated with the project. While methodology for data analysis for a reflective case study is not prescriptive, in order to improve validity, a colleague from the Poche Centre (KG) was involved in checking themes, reflections and learning points from the process.

Paradigms

The scoping review I conducted of international genomic research identified community-based participatory action research as the method with the most potential in this respect.[28] The work of the Poche Centre is informed by 'collective impact' method of co-design, which is a process for solving problems based on agreeing a common agenda, communication, "mutually reinforcing" activities, project support and involving multiple stakeholders in evaluation.[479,480]

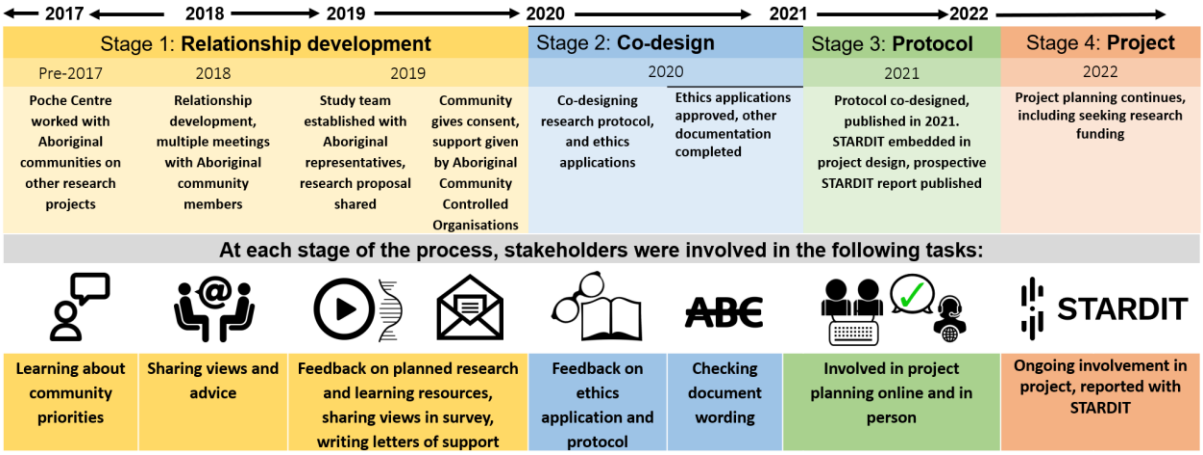
In this thesis, participatory action research is used as an umbrella term which describes a number of related approaches, including forms of action research which embrace a participatory philosophy and include 'co-design', 'collective impact' and 'co-production' of research. [88(p1)] During the participatory process, Poche researchers ensured researchers and other relevant stakeholders (including community members) "work together, sharing power and responsibility from the start to the end of the project",[89] including knowledge generation and translation. [89,330] The project will continue to use the paradigm of participatory action research, which attempts to reduce health inequalities by supporting people to be involved in research protocol design, ethical review, data collection, reflection and, ultimately, in action to improve their own health.[132]

It is important to note that the co-design process described here is a starting point for future research co-design and planning, and that only once careful and detailed co-design has taken place, are ethics applications and research protocols produced. This is in contrast to other kinds of research projects which gain ethics approval, publish protocols and then seek feedback through consultation with potential participants.

Stages of research

In 2018 I was invited to volunteer with Sydney University's Poche Centre for Indigenous Health in order to support with their planned precision medicine project. I attended online meetings and commented on shared documents in order to support the co-design of the methodology; helped create learning materials and videos; made presentations to the Poche Centre Board about my research, including STARDIT; and supported with ethics applications. The stages of the co-design process are summarised in Figure 8.1.

Figure 8.1: Stages of research co-design with Aboriginal communities



As part of the planning and co-design process, I was invited in 2019 to travel with staff members of the Poche Centre to meet members of a remote New South Wales community (see Figure 8.2, reused here with permission).[481] This meeting was one of a number of meetings with this community which took place before and after this particular one. The purpose of the meeting I attended was to meet with community representatives and members of the community to discuss any next steps regarding partnership and co-design.

The meeting itself was a formal meeting, and thus followed respectful cultural protocols. It was attended by an Aboriginal elder who 'loaned their trust' to the Poche Centre researchers. The Elder welcomed the Poche Centre Director Boe Rambaldini, who formally responded, and then introduced members of the Poche Centre team, including Kylie Gwynne, who presented to the meeting attendees about the proposed project.

My tasks at the meeting included talking with participants, making notes and sharing any recommendations with the Poche Centre staff, including assisting with writing a research protocol and integrating STARDIT into planned ways of working. After the event, in 2019, formal consent from the community was obtained via a community survey. In addition, letters of support were given from two Aboriginal Community Controlled Organisations. Learning from this event informed subsequent research planning, governance planning and associated ethics applications in 2019 and 2020, and the published protocol in 2021.

In parallel with this work, I was invited by the staff from The National Centre for Indigenous Genomics to be a co-author on a peer reviewed article, along with a number of Poche Centre staff connected with this project. The article was published in 2020 and is titled 'Equitable Expanded Carrier Screening Needs Indigenous Clinical and Population Genomic Data'.[5]

Figure 8.2: Precision Medicine project meeting participants 2019

Photo published by the Poche Centre for Indigenous Health, 2019, showing Precision Medicine project meeting participants. [481]

Results

Results from the participatory action research process included learning from the co-design process which informed the protocol creation, and the co-creation of the protocol itself. In parallel to this participatory process, work with the National Centre for Indigenous Genomics (NCIG) on an article is also summarised, as discussions with staff from the NCIG informed Poche's participatory action research process, and learning from this process also informed the article led by the NCIG.

Learning from the co-design process

Working with staff from the Poche Centre to co-design participant information provided an opportunity to explore the attitudes and values of both the Poche Centre staff, and members of the communities they hoped to involve. While my initial work was limited to online meetings and commenting on shared documents, it provided an insight into linguistic expectations, agreed terminology and the values which are associated with that. For example, discussions on the usage of the word 'race', and discussions about appropriate terminology with regard to the use of terms such as 'Indigenous', 'Aboriginal' or 'community member'.

The in-person co-design process helped elucidate concerns regarding the proposed genomic research. Some members of the community demonstrated a good understanding of how genomic

variations can contribute to risk of disease, and how these can be inherited. While community members appeared supportive, a number of concerns were raised during the co-design process (by either Poche Centre staff or community members). These concerns included genetic discrimination in employment, difficulty in obtaining insurance, and inappropriate use of genomic data in law enforcement. It was therefore necessary for the co-design process to balance these concerns with the researchers' requirements with regards to combining data.

Co-created study protocol

While subsequent planned work was delayed and disrupted by the COVID-19 pandemic, in 2021 the study protocol 'A Pathway to Precision Medicine for Aboriginal Australians: A Study Protocol' was published.[3] It is the result of the co-design process conducted by the Poche Centre with a number of Aboriginal communities and other relevant experts. The protocol outlines the background to the study, planned methods and phases and discusses the implications of the study protocol.

By working closely with the Poche Centre staff, I was able to ensure that while I was myself learning, knowledge and experience gained from my PhD were also able to inform the study protocol. This included results from the scoping review, learning from STARDIT and other relevant professional experience informing my feedback. I worked on multiple versions of the research protocol, giving feedback, writing content and editing final versions. I was able to explain STARDIT to Poche Centre colleagues (including via invited presentations to the Board), and was able to integrate plans to use STARDIT in the study protocol. The study protocol is summarised in 'Table 8.1: Study protocol summary', which is adapted from content from the peer-reviewed protocol and the article I wrote for the Journal of the Consumers Health Forum of Australia.[2,3]

Table 8.1: Study protocol summary

Background	Genomic precision medicine (PM) utilises people's genomic data to inform the delivery of preventive and therapeutic health care. PM has not been well-established for use with people of Aboriginal and Torres Strait Islander ancestry due to the paucity of genomic data from these communities. We report the development of a new protocol using co-design methods to enhance the potential use of PM for Aboriginal Australians.
Methods	This iterative qualitative study consists of five main phases:

	Phase-I will ensure appropriate governance of the project and establishment of a Project Advisory Committee, following an initial consultation with the Aboriginal community. **Phase-II** will invite community members to participate in co-design workshops. Using a combination of face-to-face events and online tools, Aboriginal community members and the Armajun Aboriginal Community Controlled Health Service will be involved in co-designing every stage of the project, including the ways in which people are involved. **Phase-III** Chief Investigators will participate in co-design workshops and document generated ideas. **Phase-IV** notes will be analysed thematically with Aboriginal community representatives, and the summary will be disseminated to the communities. **Phase-V** we will evaluate the co-design process and adapt our protocol for use in partnership with other communities. We propose the use of Standardised Data on Initiatives (STARDIT) to report, evaluate, and keep public records of every stage of the co-design process. Aboriginal community members will help evaluate the project, including how people were involved. This will help appraise effective ways of working and identify learning points and best practice.
Discussion	This study protocol represents a crucial first step to ensure that PM research is relevant and acceptable to Aboriginal Australians. Without fair access to PM, the gap in health outcome between Aboriginal and non-Aboriginal Australians will continue to widen

Influencing articles by the National Centre for Indigenous Genomics

The article 'Equitable Expanded Carrier Screening Needs Indigenous Clinical and Population Genomic Data' had over 40 co-authors, and accordingly, there were multiple perspectives which we attempted to incorporate, and not all feedback from co-authors could be integrated. While my tasks were limited to commenting on the multiple versions shared with the co-authors, my feedback that concepts of culturally safe and community led involvement should be recommended was included, and featured as follows in the abstract:

> [Aboriginal and/or Torres Strait Islander Australians] require culturally safe, community-led research and community involvement embedded within national health and medical genomics programs to ensure that new knowledge is integrated into medicine and health services in ways that address the specific and articulated cultural and health needs of Indigenous people.[5]

While my recommendation that STARDIT be proposed in the article as a way of supporting this work was not integrated, the subsequent adoption of STARDIT by the Australian Genomics project 'Involve Australia' shows the work has relevance, as the remit of that project includes involving Aboriginal peoples in genomics research.

Discussion and reflections

Exploring perceived risk and benefits

When working with Indigenous peoples in countries such as Australia, it is important to understand and acknowledge both the history and continued evidence of systemic racism and structural violence towards Aboriginal peoples.[482,483]

Some issues are common concerns of Aboriginal people in genomics research, including sovereignty over samples, and benefits and risks of genomic research to communities.[484–486] One concern shared by Aboriginal community members and study team members was the risk of DNA data being be used for law enforcement and criminal justice, rather than strictly research. Owing to Australian law, such use could not be ruled out by researchers, and was thus an important risk to consider when co-designing this protocol. Accordingly, the project was co-designed with samples which were de-identified, and data analysis methods which combined reference genomes from multiple individuals.

Getting the language right

In an early version of a participant information sheet which I was asked to proof-read, the second sentence stated, "DNA is different but a lot of our DNA is shared, especially by people of the same race". While the word 'race' was used as a plain English short-hand, I challenged the use of it as, by the very act of using the word, it implied the researchers, and thus the project itself, believed in and endorsed the concept of 'race'. Concepts described by words such as 'race' and 'ethnicity' have contested boundaries.[487] A word like 'ethnicity' can be an unhelpful grouping in public health, as it "functions like a wild-card which can readily be inserted into a string of related concepts", blurring inherited characteristics with learned behaviour, often used as a euphemism for race.[488] For

example, ethnicity can be defined as referring to 'cultural traits' exclusively[489], while other definitions also state that 'racial' origins can define it. [490]

Moreover, words like 'ethnicity' and 'race' have amorphous meanings with parameters that are "context-dependent"[435], with the usage of terms like 'race', 'ethnicity' and 'ancestry' changing over time.[491] The Nuffield Council on Bioethics points out (in a report about pharmacogenomics) that "Race and ethnicity cannot be given precise biological or genetic definitions".[492] Similarly, other authors agree that the idea of race is "a social invention with no biological validity"[493] or a "socially constructed" category.[436]

Targeting public health messages at specific populations defined by race "may reinforce tendencies to view race as a biologically defined phenomenon".[492] The Nuffield Council on Bioethics recommend being "sensitive to the potential for misunderstanding and prejudice arising from racial stereotyping". [492] Biological determinism and racial essentialism both posit that the concept of 'race' can be defined precisely.[493] Once regarded as "an ugly component of a bygone era of social Darwinism and race-based eugenics research supported by racially exclusionary and discriminatory laws, overt policies, and hostile attitudes", such modes of thinking have "invaded contemporary study of genetics and genomics".[493]

It should be carefully noted that while there is no agreed biological definition of 'race', racism (a result of a belief in 'race') clearly does exist, and has profound implications for people's human rights, social and health outcomes around the world. For example, while there is a rejection of race concepts among professional anthropologists and a consensus that there are no human biological races, there is a recognition that race exists as lived social experiences that can have important effects on health. [494(p325)] As a result, whether biologically definable, the social realities of people's attitudes about race and ethnicity also influence attitudes about engagement or involvement in genomics research.

While contemporary language would talk of concepts of 'ancestry', the Australian constitution uses the word 'race' on a number of occasions and gives "special legislative treatment" to people perceived as different races.[495] Other legislation from 2013 defines 'Indigenous' as "a member of the Aboriginal race of Australia".[496] Historically, in particular during the Stolen Generations era of Australian history, people used to use fractions to try to determine and describe an individual's ancestry, and to what extent they are "descended from the original inhabitants of Australia".[497] As

recently as 2020 the Australian Minister for Indigenous Australians cited that there are people in society still "who live with the notion that we take still the old practice of full bloods, half cast, quarter cast, quadroon",[498] labels that were used in Australian legislation up to 1936.[497] Controversial campaigns by some political parties in 2019 have sought publicity by calling for a biological definition of race in order to label people as 'Aboriginal' according to an external objective measure, rather than the current system of self-identification, despite there being no agreed ethical foundation for doing this, let alone a method.[499,500]

However, the precision medicine projects seeking to ensure data is useful for people of Aboriginal and Torres Strait Islander ancestry, and other Indigenous peoples around the world, must navigate the complex historical and contemporary contexts of racism carefully. Involving potential participants and community members in every stage of an initiative means it is more likely to be acceptable, relevant and culturally safe. This includes involving people in checking any information resources that will be shared and assessing if planned data access protocols are acceptable.

After discussion, the word 'race' was removed in the final version of the document sent out. For me, the incident reinforced the importance of not only involving potential stakeholders in co-designing resources and checking language, but also reporting transparently how people will be involved, were involved and any impacts.

In order to help navigate the complex language surrounding biological ancestry and learned culture, STARDIT has been designed to align with models of self-identification. Empowering people to chose their own labels and groupings (rather than be passively labelled or grouped) can help ensure such research aligns with the values of potential participants and does not perpetuate racist constructs and labels. For example, when STARDIT is used to report how people have been involved in an initiative, people can choose to self-identify with labels of their own choice, including 'Aboriginal', 'Indigenous' or 'community member', removing the requirement (and the associated power) of researchers to assign these terms to people. STARDIT can be used to both plan and report each stage of any such involvement, providing transparency for potential participants, and helping ensure accountability to those protocols by the future research projects.

Similarly, involving people in deciding research priorities in a transparent way can also ensure research aligns with perceived priorities. Deciding what gets researched and what does not can create inequality. For example, a paucity of data about Aboriginal peoples means that existing health

inequalities might be amplified. Reasons for a lack of research in a certain field (contributing to ascertainment bias in genomic data) might be cultural, financial or geographic. Using systems like STARDIT can help map both priorities, linguistic terms, co-define meanings and the preferences and values of multiple stakeholders, allowing an evidence-informed approach to prioritising, planning, doing and evaluating future genomic research.

Co-defining ethical research

Ensuring research is conducted ethically requires an ongoing and transparent process to involve people in defining what ethical means, and ensuring research is conducted within those defined boundaries. While such ethical processes need oversight, they also need to ensure that they are also not inhibiting research from being done which could save and improve lives. The Australian Department of Health's 2021 consultation on 'Aboriginal and Torres Strait Islander Genomics Health Guiding Principles' is a demonstration of involving people in helping shape such ethical principles for future genomic research with Aboriginal peoples.[501] One principle states that "projects should be conducted through co-design processes". [501] The published protocol demonstrates how such co-design methods can be reported, helping ensure future ethical oversight can be managed in partnership with Aboriginal people.

While undoubtedly ethical oversight is essential, it is also important to note the barriers perceived by both researchers and Aboriginal communities by current ethics processes. There is a risk that by being too complex or inaccessible (to both researchers and the public), ethics processes risk having unintended unethical consequences. For example, researchers and Aboriginal communities might be unable to navigate ethical processes for a number of reasons including limited resources (such as time, finance, people with expertise who can support the project before it is funded). An unintended consequence of such complex processes is that research with Aboriginal communities does not take place, or its not even attempted owing to perceived barriers. The risk is 'widening the gap' of life expectancy of Aboriginal communities, with ethical processes having the unintended consequence of applying health inequalities. This issue is discussed further in the section 'Implications for ethics and participatory action research', and should be urgently explored by multiple stakeholders.

It is important to note that STARDIT can be used to describe who was involved in co-designing every aspect of the research, including who was involved in making ethical decisions, providing more transparency to the entire process of ethical oversight, and helping address the questions 'who decides who decides what is ethical'.

Strengths and limitations

This case study was designed to describe the careful, complicated and often time-consuming work involved in following best-practice when working with Indigenous peoples to co-design genomics research. While a protocol for genomics research might not seem like a research output in itself in some disciplines, in the participatory action research paradigm, this protocol represents an important output in itself. It represents a free publicly accessible, peer-reviewed plan for research, co-created with a community, alongside different experts. The planned reporting of the case study using STARDIT also ensures the project will continue to provide publicly accessible information about how people will be and have been involved.

While this case study is not an appraisal of the protocol itself, it is worth noting that Aboriginal community members were not only involved in every stage of the co-design of the protocol, but according to the protocol, will remain involved throughout every stage of the planned research. The data sources for this case study were limited by ethical considerations. Other than the protocol itself, data was limited to emails, conversations and documents shared between the study team. Using any data generated by people from the communities involved, including using direct quotations, would require an entirely separate ethics process, and was never part of the plan for this case study. While this limits the depth of the data that this case study could access, it also provides an additional learning point about the complexity of ethics processes, and how planning any co-designed project needs to allow for both the workload and the time required for the ethical conduct and ethical approval of research.

Conclusions

The work described in this chapter provides a clear methodology for both planning and reporting co-design methods in genomic research with Aboriginal people. STARDIT can be used to create public records of every stage of the co-design process, including data about who was involved, how, and in which tasks. Information about the co-design method, the data analysis methods, reported enablers and barriers for involvement, and any other impacts and outcomes from the co-design process can also be reported. Such data improves transparency of terms such as ‘co-design’ and allows improved scrutiny and ethical oversight, including from potential or current research participants.

STARDIT MICRO Report: A Pathway to precision medicine for Aboriginal Australians

This STARDIT MICRO report contains the minimum data required for a Standardised Data on Initiatives Alpha version report (STARDIT Alpha).[196] This STARDIT MICRO report contains additional data relevant to the protocol 'A pathway to precision medicine for Aboriginal Australians'. The full prospective STARDIT report for this protocol can be found in the Appendices under the heading 'Prospective STARDIT Report: A Pathway to precision medicine for Aboriginal Australians', with a link to the online 'living' Beta version of the STARDIT report available in the references.[458]

Name	STARDIT MICRO Report: A Pathway to precision medicine for Aboriginal Australians
Involvement	
Who will be involved	Group 1: Academic investigators. Group 2: Aboriginal community representatives. Group 3: Aboriginal community members involved in co-design and consultation activities.
Specific tasks	Group 1 and Group 2: Project design, ethics applications, planning and delivering co-design processes and analysing data from co-design activities. Group 3: Face-to-face and online events, consultation processes, and checking data analysis.
Outcomes (of involvement)	Involving Aboriginal community representatives and members will ensure planned activities (including research methods) are appropriate, culturally safe and effective.
Mapping financial or other interests	
Describe any financial relationship or other interest this person has to this project.	Group 1: Academic investigators will be volunteering their time and may be named as authors in peer-reviewed publications. Group 2 and 3: Aboriginal community representatives and community members may be paid for their time.
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	As part of the co-design process, data sharing (including returning data and results to participants), governance (including how to plan the management and storage of sample and DNA data), and data sovereignty will be agreed with participants and affected Aboriginal communities. Data transference processes used in the project will be consistent with the principles of participatory action research, where stakeholders collectively decide upon roles, responsibilities and data access.
How is it stored and hosted?	As part of the co-design process, stakeholders will collectively decide this, along with appropriate biobanking methods.
Who is analysing the data?	Stakeholders will collectively decide this as part of the co-design process.
How is information about this data disseminated?	Social media, mass media communications, policy papers, conference presentations and open access journal publications, short video, podcasts and public events.

How is the data FAIR?[85]	Stakeholders will collectively decide this as part of the co-design process.
Impacts and outcomes	
What new knowledge has been generated?	The project intends to: 1. inform best practice in co-design with Aboriginal communities 2. answer important questions about people's preferences about data ethics, security and quality associated with genomic research 3. map people's preferences using the STARDIT-Preference Mapping tool.
Outcomes – Describe how the learning or knowledge generated from this initiative has or will be used.	Using a co-design process: 1. promotes usage of health services, and has the potential to elicit superior health outcomes and save time 2. builds strong and committed community partners 3. enhances skills and knowledge in the Aboriginal community about genomics and health.

Chapter 9 – Results: Comparison of all case studies

Comparison of all case studies

This chapter will summarise the results from all case studies and answer the questions:

- What does a comparison of the data analysed from each project reveal?
- What are the themes, generalisations, similarities and differences between these case studies?
- What are the significant observations and learnings from these observations?

It also includes a comparison of results from STARDIT reports from this doctoral research.

As this chapter refers to participants from multiple case studies, when participants are quoted they will be identified using the following format:

- Shared Ancestry - Participant 1 = [P1-SA]
- ausEE Participant 1 = [P1-ausEE]
- ASPREE participant 1 = [P1-ASPREE]

Quantitative cross-case analysis

During this thesis, I developed and used a standardised way of planning, reporting and evaluating stakeholder involvement in order to improve future genomics research using standardised data (STARDIT), including mapping the preferences of different stakeholders using standardised preference mapping (STARDIT-PM).

Baseline and follow-up preference mapping

Below is a comparison of the quantitative data gathered from all case studies where both baseline and follow-up data was collected, in order to map changes in preferences about involvement in research. Data came from the question 'Which aspects of any future research genomics research should be influenced by the following?'. Participants answered using a tick-box grid to indicate which aspects of research they felt should be influenced by which stakeholder groups. Figure 3.2.0 shows this grid.

In the ausEE study, of the 26 people who gave consent and completed the initial survey, 15 participated in the online discussion and 12 completed the follow-up survey. From the 12 responses to the baseline and follow-up survey, participants gave 41 responses. In the ausEE study 59% of participants' responses showed a change towards 'widening' their view of who should be involved in research to include more people (N=24/41), 34% stayed the same (N=14/41) and 7% narrowed (N=3/41).

In the Shared Ancestry study, from the six participants who completed both the baseline and follow-up surveys, a total of 54 responses were given. Of these, 35% showed a change towards 'widening' involvement (N=19/54), 8% 'narrowed' (N=8/54) and half stayed the same (N=27/54). Table 9.1 below combines the results from the 18 participants across the two studies who completed both the baseline and follow-up surveys using identifiable data at both stages, providing a total of 95 responses to the questions. 45% of participants' responses changed to 'wider' (N=43/95), 43% stayed the same (N=41/95) and 12% narrowed (N=11/95). More detailed data can be found in the Appendices under 'Detailed baseline and follow-up data on preferences for involvement'.

Table 9.1: Summary of baseline and follow-up preferences

Who should influence which aspects of research?	Change to wider	No change	Change to narrower
ausEE sub-totals (total of 41 responses)	24	14	3
Shared Ancestry sub-totals (total of 54 responses)	19	27	8
Combined data from both studies (total of 95 responses)	43	41	11
Percentages	**45%** of responses	**43%** of responses	**12%** of responses

Standardised Preference Mapping

This analysis combines all public domain data from case studies using the preference mapping tool STARDIT-PM to categorise the data into certain areas[196], allowing a comparison of data from 83 different stakeholders from the three case studies where preferences were mapped, including the facilitators of the online discussions (ausEE and Shared Ancestry) and the study team members from the ASPREE case study.

Preferences were recorded from all data sources, including the initial surveys, online discussions with participants, online facilitator discussions, follow-up surveys with participants, surveys with facilitators and study team members and emails from the ASPREE study team. If the same person made the same point at different stages, this was counted as one view.

Questions in online discussions and surveys were designed to provide data for these categories. This parity in data collection and analysis has allowed this data to be combined and compared from all the different stakeholder groups, thus providing more statistically useful data and providing a proof of concept for using this method of data collection and analysis. Table 9.2 provides a summary of the STARDIT-PM data, with more detailed data in the Appendices 'Quantitative data from STARDIT-PM cross-case analysis'.

Table 9.2: Summary of STARDIT-PM cross-case data

Data set	ASPREE totals	ausEE participants	Shared ancestry participants	Online facilitators (ausEE and Shared Ancestry)	Totals across case studies
Total number in dataset	42	26	12	3	83
Views on who should be involved	31	17	10	0	58
Views on who should do which tasks	26	10	8	0	44
Views on modes of communication	32	3	4	0	39
Views on what methods should be used (STARDIT-PM Q3)	25	6	5	0	36
Views on enablers of involvement	25	10	7	3	45
Views on barriers of involvement	10	5	6	3	24
Views on what the outcome or output of the research or involvement in research could be	11	3	2	0	16
Views on which stage of the research people should be involved	24	0	1	0	25
Research data	19	0	3	0	22

Data set	ASPREE totals	ausEE participants	Shared ancestry participants	Online facilitators (ausEE and Shared Ancestry)	Totals across case studies
Views on how learning from this research could be used	0	0	8	0	8

Most stakeholders expressed views on who should be involved (70%, n=58/83). Views on enablers of involvement was the second highest area people expressed views in, with all three facilitators sharing views on this. Views on who should do which tasks were shared by 44 people, with all case studies having a relatively high number of responses for this category. This standardised data provides useful information about different stakeholders' preferences that could be useful in informing the design of involvement in future research.

Analysis using STARDIT-PM allows a quantitative comparison of which stakeholder groups shared views about which areas, allowing a better understanding of which groupings those preferences are from. When this kind of data is structured using STARDIT, it allows more complex statistical analysis, including giving 'weight' certain groups' preferences. For example, if more researchers than participants have shared perspectives, the researchers' preferences can be grouped and compared to the preferences of the participants.

Learning from such statistical analysis might suggest areas where people need learning and development interventions in order to support informed decision making. For example, if most research participants have not shared preferences about data sharing and who should access their genomic data, it might mean they don't understand the principles or concepts, not that they don't have preferences about who can access this data. This might indicate an area for further learning and development in order to ensure their views and preferences are able to influence future genomic research.

Raw data about how participants from the ausEE and Shared Ancestry case studies answered both the baseline and follow-up questions can be found in the Appendix in the section 'Cross-case analysis data from ausEE and Shared Ancestry case study'.

Standardised Data on Initiatives (STARDIT) data summary

This section compares the number of different methods and tasks of involvement, and the reported impacts. All studies reported impacts from the process of involving people.

Table 9.3 summarises the quantitative data from the three case studies where it was reported (ausEE, Shared Ancestry, ASPREE), noting the Indigenous Precision Medicine case study is a prospective report. A more detailed version of Table 9.3 is included in the Appendices in the section 'Detailed quantitative analysis of STARDIT data categories'.

Table 9.3: Standardised Data on Initiatives (STARDIT) quantitative summary

Study	Number of methods [STARDIT Data Category 'Method of doing task?']	Number of reported communication modes [STARDIT Data Category 'Communication modes]	Number of reported tasks [STARDIT Data Category 'Tasks']	Total number of impacts [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
ASPREE	4	3	4	7
ausEE	3	3	4	6
Shared Ancestry	3	3	5	8

While the reductive quantitative analysis in Table 9.3 can be useful for large datasets (such as the 2019 review of global genomics research projects[28]), it has been included here to indicate the limitations of such an approach with a small dataset. Noting the standardised data can be added to any future datasets, while the use of this quantitative data is limited, it could be combined in the future with other standardised data to gain more insights. For example, the charity Science for All used the STARDIT-PM tool to map preferences of the public about who should be involved in research and reported it,[502] using a licence which allows this data to be compared and combined with the data in this thesis, and future datasets in order to provider richer analysis (see Appendix 'STARDIT-PM preference mapping data from public survey for comparison').

The next section includes a qualitative thematic analysis from the standardised data, which provides more insights into the quantitative data above.

Qualitative cross-case thematic analysis

This section summarises the results of a thematic analysis of all case studies. It is divided into sections which explore themes from:

1. the two online discussion case studies (ausEE and Shared Ancestry)
2. the ASPREE, ausEE and Shared Ancestry case studies
3. all case studies (ASPREE, ausEE, Shared Ancestry and the Indigenous Precision Medicine project).

First, (1) the ausEE and Shared Ancestry case studies are compared, as the methods were very similar. Secondly, (2) the three case studies where there was standardised data collection (including STARDIT-PM) are compared, and thirdly (3) all case studies are compared, including the Aboriginal Precision Medicine project, which was a reflective case study and where STARDIT reporting is used prospectively.

1. Themes from the two online discussion case studies

Online discussions for both the ausEE and Shared Ancestry studies demonstrated a high level of genomic literacy and understanding of ethical issues among participants. In each discussion, at least one person stated that they had been involved in genomics research as an investigator, not as a participant. Participants reported learning more about genomics as a result of taking part, and indicated that the learning resources provided were helpful, particularly, the visual summary of 'Public involvement in global genomics research: A scoping review' (see Figure 4.2 in Chapter 4).[503]

Participants shared views about the limitations of one-to-one 'one way' interviews. One person commented that they can be "quite straight jacketed with circumscribed questions" [P4-SA]. Among their advantages, online discussions permitted participants to ask questions and enabled researchers to gather data they might not otherwise have planned to collect.

Some participants reported that the pace of discussion was too fast and "moved on too quickly" [P7-SA] [P4-SA] [P5-SA], which risked leaving "some people behind" [P3-ausEE]. However, owing to the asynchronous design of the study, participants were able to contribute to discussions and catch up in a flexible way, and this was reported as an important enabler of participation in both studies.

In both case studies, participants reported a desire to help researchers. Notwithstanding, in both discussions, concerns were raised about researchers' intentions regarding genomics research (generally, not specific to these studies). Participants preferred transparency about this and were concerned about "hidden motives" [P2-SA]. The importance of determining the "purpose of research" was raised in both discussions, with participants suggesting that they should be involved in "agreeing purpose" [P7-SA]. Learning from this process influenced the design of the STARDIT Beta version (0.2), in which the purpose of research was added as a data field.

In both studies, participants discussed the concept of everyone being involved, but noted that some people should be more influential than others. In the Shared Ancestry study, one participant identified medical scientists, social scientists, psychologists and the general public as groups which

should influence research, but noted that not all "groups should have equal influence" [P10-SA]. One participant from the ausEE study stated that people with a rare disease and their families are "likely to have different priorities from scientists" [P16-ausEE]. A Shared Ancestry participant concluded that "we're all biased; whoever is affected by a condition is likely to want it prioritised" [P12-SA]. Similarly, a participant from the ausEE study stated that people affected by Eosinophilic Gastrointestinal Disorder "certainly do" have a vested interest in genomics research into that condition [P3-ausEE]. These discussions highlighted the importance of being able to use STARDIT to map and report on the different interests and perspectives of diverse stakeholder groups.

One notable difference between participants in the two case studies was that participants in the Shared Ancestry study appeared to find using the online discussion platform more difficult than those from the ausEE study. The latter reported no issues with the online discussion platform. As these participants were recruited from an existing online community, it is probable that they were more accustomed to that style of discussion and more familiar with using online discussion tools. This variation in reported difficulty highlights the importance of mapping the preferences of people who will be involved in research and this can be done using the preference mapping tool, STARDIT-PM, developed as part of the STARDIT framework.

2. Themes from ASPREE, ausEE and Shared Ancestry

Participant uncertainty about what they can offer

All three case studies indicated that participants were willing to get involved and help improve research, but were uncertain about how they could get involved and, in particular, what tasks they could do. In the Shared Ancestry study, one participant stated that compared to "highly-qualified scientists who know what's possible" some "relatively ignorant lay people" will be limited in what they can offer at some stages [P7-SA]. Similar views were expressed by participants in the ASPREE study, in which participants were willing to help but were unsure as to how they could.

Forty-five per cent of ASPREE participants understood the concept of involvement, but expressed self-doubt about how they could be involved (45 per cent, 9/20). While participants were happy to be involved, this was linked with phrases such as "I don't know I'd have much to contribute" [P10-ASPREE].

Similarly, among ASPREE participants who understood the concept of involvement and were willing to be involved, 25 per cent were unclear about what tasks they could be involved in and whether they would have sufficient expertise (25 per cent, 5/20). One participant stated that "my expertise wouldn't be sufficient" [P2-ASPREE].

Participants under-valuing or not understanding what they can offer in regard to involvement suggests that learning and development may be a crucial intervention. Interventions that help people understand what involvement is and the tasks they can be involved in may help people value the perspectives they can offer and help improve research.[28]

Desire to help and to know they have helped (146)

One participant from the Shared Ancestry study wanted to know that their involvement had been "useful to the researchers" [P11-SA], while another stated that an outcome of being involved would be the "satisfaction of knowing that I may have contributed" [P10-SA]. This view was echoed in both the ASPREE and ausEE studies, where participants shared a desire to be involved in order to help both the research process and the researchers involved.

It is interesting to note that this desire to help was not considered by study team members from the ASPREE case study. In the follow-up survey, one team member reflected that they had "worried too much" about the time burden of involving participants [MS]. This important finding shows the importance of mapping people's preferences about involvement in genomic research, so that potentially incorrect assumptions about people's willingness to be involved can be avoided.

3. Themes from all case studies

Impacts from involving stakeholders

Involving research participants and study team members in all aspects of the research process yielded positive impacts in every case study (see Table 9.4 below). As the Indigenous Precision Medicine case study is a prospective protocol, it is not been included in this table.

Table 9.4: Summary of impacts from all case studies

Study	Reported methods of involving people [STARDIT Data Category 'Method of doing task?']	Reported modes [STARDIT Data Category 'Communication modes']	Reported tasks [STARDIT Data Category 'Tasks']	Involvement outcomes, impacts, learning or outputs [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
ASPREE	Commenting on documents, meetings, interviews, group discussion. Number of methods: 4	Face-to-face meeting, telephone interviews, face-to-face group discussion. Number of communication modes: 3	Reviewing documents (including participant information), commenting on research design, sharing views and perspectives, analysing data. Number of tasks: 4	**Seven impacts** on proposed study design from involving people: 1. Recruitment and sample collection plan changed. 2. Participant communication will be improved by creating resources to assist with recruiting people 3. Participants were involved in governance. 4. Participants were involved in controlling data access. 5. Participants were included on study advisory groups (including ethical oversight) using multiple communication modes. 6. Feedback was provided to participants about the research. 7. Learning and development opportunities were created for potential participants.

Study	**Reported methods of involving people** [STARDIT Data Category 'Method of doing task?']	**Reported modes** [STARDIT Data Category 'Communication modes']	**Reported tasks** [STARDIT Data Category 'Tasks']	**Involvement outcomes, impacts, learning or outputs** [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
ausEE	Commenting on documents, survey, group discussion. Number of methods: 3	Online shared documents, online survey, online text-based asynchronous discussion. Number of communication modes: 3	Reviewing documents (including participant information), commenting on research design, sharing views and perspectives, analysing data. Number of tasks: 4	**Six impacts** reported from involving people: 1. Participants reported that learning resources were useful. 2. Participants reported changed views as a result of participating. 3. Participants reported enjoying the online discussions. 4. Online discussions to be used in future research prioritisation. 5. Participants asked to stay involved in the research. 6. Participants reported improved understanding of how to get involved in research.

Study	**Reported methods of involving people** [STARDIT Data Category 'Method of doing task?']	**Reported modes** [STARDIT Data Category 'Communication modes']	**Reported tasks** [STARDIT Data Category 'Tasks']	**Involvement outcomes, impacts, learning or outputs** [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
Shared Ancestry	Commenting on documents, survey, group discussion. Number of methods: 3	Online shared documents, online survey, online text-based asynchronous discussion. Number of communication modes: 3	Reviewing documents (including participant information), commenting on research design, sharing views and perspectives, analysing data, contributing to STARDIT report data. Number of tasks: 5	**Eight impacts** reported from involving people: 1. Improved understanding of genomics informed siblings' participation in future research. 2. Participants reported that learning resources were useful. 3. Participants reported changed views as a result of participating. 4. Participants asked to stay involved in the research. 5. Participants enjoyed the online discussions. 6. Improved understanding of how to get involved in research. 7. Co-design changed study design. 8. Method for future research co-design established.

Summary of impacts

This section provides a summary of themes from the impacts of involvement across the completed three case studies. Follow-up surveys were conducted with participants in the ausEE and Shared Ancestry case studies. Due to ethical restrictions, only ASPREE study team members were surveyed, so there may be multiple unreported impacts from this research.

Potential participants in all case studies were involved in giving feedback on the planned process for involving people. Study team members from all case studies reported feeling that this improved the design of the respective studies.

Participants from all studies asked to stay involved in the case studies, and participants from each were involved in producing peer-reviewed articles about the study – either as an author or by giving feedback on the manuscript and being named in the acknowledgements. In all cases, some participants from each study agreed to be named in the respective STARDIT reports and were coupled with the specific tasks they undertook.

Participants from both the ausEE and Shared Ancestry case studies reported that the learning resources provided were useful, and helped improve their understanding of both genomics research and how people can be involved.

Stakeholders from all case studies reported changing their views about involving people, either as a result of participating or being a study team member. All changed views represented a move towards valuing the importance of involving people in genomics research.

When combined, most responses from participants in the ausEE and Shared Ancestry case studies showed a 'widening' towards a preference for more people being involved in genomics research. This preference and follow-up data were not collected for the ASPREE study.

Participants from all case studies reported enjoying being involved and being part of the research process. With regard to preferences for involvement, some participants from the Shared Ancestry and ausEE case studies highlighted their preference for online discussions, reporting that these are more flexible and accessible and, thus, more inclusive. The Chief Executive of the charity ausEE reported she would like to use online discussions as part of future research prioritisation processes.

Participants from both the ASPREE study and the Shared Ancestry study reported they would like to be involved in future data access decisions. In the ASPREE case study this was incorporated into the design of the prospective multi-generational research study (MGRS). In the Shared Ancestry case study, participants reported that learning and changed views from participating in the online discussions had influenced their decisions both about invitations to participate in genomics research, and about how their siblings would like to be involved in future research.

Different interests

Participants from all case studies recognised different "interested parties" with regard to who is using data from genomics research and how [P7-SA]. According to participants from the Shared Ancestry study [P9-SA], private companies charging or benefitting from data was "distasteful". This sentiment was echoed by a vote among ASPREE case study participants, all of whom stated that, while they were comfortable with their data being held by academics, they were not comfortable with data being held by a commercial company. However, one participant suggested not "ruling private companies out completely" from research [P9-ASPREE]. The ASPREE participants also questioned different interests in research, with one participant stating "I don't trust people or organisations easily" and adding that "there's vested interests out there – why are they doing it – what is in it for them?"

In the ausEE case study, the themes of the 'intention' and 'purpose' of research emerged, with a related discussion on vested interests. Clarifying the purpose or 'intention' "sets all direction and intention" (*sic*) of research [P3-ausEE]. The purpose or 'intention' of research should be transparent, with one participant asking if it is "'needs' based or 'reward' based" [P3-ausEE]. AusEE participants also noted that they themselves have a vested interest in research [P3-ausEE].

One Shared Ancestry participant asked, "…there will be many interested groups, so which ones will be listened to?" [P4-SA]. One participant stated that "people who are not looking for personal gain, but who have a desire to improve quality of life and help us understand ourselves" should influence research [P4-SA]. A mistrust of for-profit research and "sponsored facts" [P5-SA] was reported by a Shared Ancestry participant. This sentiment was echoed across the other case studies. Concerns about data being used for criminal or politically motivated purposes was a significant consideration in the Indigenous Precision Medicine case study.

Participants in the ausEE case study identified that the different interests of stakeholder groups affect not only research, but also health technology assessment processes. One ausEE participant

noted that rare conditions often do not get access to specialists or funded medicines. They identified that this might be because “we will never have the 'numbers' [for sufficiently powered trials]” as such trials are not possible with smaller groups of people [P3-ausEE]. Their view that the current evidence thresholds used by some health technology assessment processes may unfairly discriminate against rare diseases aligns with other studies.[504,505]

Participants also shared a mistrust of some organisations that are physically hosting data. The ASPREE participants also noted mistrust in for-profit social media companies, with one participant stating “I don’t trust any of the social media” [P17-ASPREE]. The decision to use Loomio, which is hosted on Australian Government servers, in the two online discussions for this research was vindicated by these comments and informed co-design of the research at an early stage. Attitudes to data sharing are explored further in the next section on ‘Data sharing’.

STARDIT can be used to report different interests, including financial interests or personal interests, and was used to report on stakeholder concerns in the three completed case studies. Mapping and reporting of data in this way can improve transparency and accountability in both genomics research and health technology assessment (HTA) processes.

Findings from the case studies reinforced the findings from the initial scoping review. Aspects of genomics research that are translated into clinical use can be described as genomic medicine (including genetic testing), which is then classified as ‘health technology’. Such translation of genomics research is affected by decisions about resource allocation.[506,507] In the scoping review, reports of initiatives involving people in managing real or perceived conflicts of interest had significant implications for health technology assessment (HTA) policy around the world. For example, there is evidence that some industry-funded studies are more often favourable to the funder’s products than are non-industry funded studies.[244,508] This problem is compounded when those involved in the HTA decision-making process have access only to evidence which is itself the result of research agendas and which are not always aligned with the priorities of the people who the HTA process is intended to benefit (such as ‘payers’ or members of the public).[38,244,509] The STARDIT system provides a way to report on how different stakeholders have been involved at every stage of genomics research, including in prioritising research questions and scrutinising the HTA process. Reports from STARDIT are potentially important in maintaining public trust that HTA processes represent the public interest.[356,510]

Data sharing

Data sharing in relation to genomics research is a complex and ever-developing area. This section is divided into themes identified from the case studies which relate to data sharing. All four case studies reported discussions about data sharing practices, with the most in-depth discussion taking place between people affected by a rare disease in the ausEE case study.

Poor data sharing practices negatively impact healthcare and research (100)

Two ausEE participants expressed frustration at current data sharing practices, with one stating "I get so tired of 'fighting' for our corner" [P3-ausEE]. Frustration was caused at multiple levels including at the individual care level, healthcare system level (including health insurance), and international research level. One ausEE participant highlighted the fact that poor data sharing practices in routine healthcare negatively impact both care and research, including losing opportunities to identify potential research priorities [P3-ausEE]. "Mutual respect" was identified as an enabler of improved practices, for example, listening to the concerns of affected people and recognising their expertise [P3-ausEE].

Control of information

The issue of who controls information, and to what end, was discussed by participants in a number of contexts, including in relation to open "borderless" access to information and the problems of paywalls [P3-ausEE]; "vested" interests in controlling information and technology – with people affected by a disease included in those with a vested interest [P15-ausEE]; the lack of "safeguards" in place which makes people feel unsure about sharing data on electronic health records, and how having genetic tests might affect eligibility for insurance [P15-ausEE].

Similarly, Shared Ancestry participants gave their views about with whom data should be shared. One participant expressed explicit concerns about sharing data for political or financial uses [P5-SA], with other participants agreeing. One participant stated "Research is for humankind. Its benefits should be available to all. Information should be for the most part easily available. If it's publicly funded, it must be publicly available" [P9-SA]. Cultural conventions around ownership of knowledge such as "patenting" were also challenged as forms of knowledge control which were viewed as "unethical" in some contexts [P9-SA].

Access to information, including "borderless" access, was raised as an issue affecting who can be involved in research [P3-ausEE]. Participants reported that paywalls for articles have direct negative impacts on patients, carers and parents wishing to make informed decisions and "judicious choices"

about healthcare or wishing to get involved in research [P3-ausEE]. People affected by diseases exist "across economic and political boundaries" and access to medication is affected by "who owns, profits, controls, patents" [P3-ausEE]. Open data needs to be balanced against individual privacy concerns and safeguards are needed to protect against misuse of data. One participant added that knowledge should be shared openly, "… open access data makes real sense in terms of the most efficient way to find 'like' patients and bring us ever closer to treatments and perhaps a cure." Participants from the ausEE study also shared the view that involving people affected by a disease in data sharing can "prompt" new areas of research [P3-ausEE].

Concerns about data privacy and misuse (643)

Four ausEE participants mentioned insurance as an area of concern requiring public involvement and scrutiny with regards to data sharing. One person stated that it is a "valid community fear" that research data will be used "for" insurance companies or other for-profit organisations [P3-ausEE]. For example, one participant raised a specific concern around health research being used by insurance companies or other organisations "to be able to set new prices or exclusions" [P3-ausEE]. Involving insurance industry stakeholders was highlighted as a strategy to bring about greater transparency on the risks of participation in genomics research, so that people "don't need to fear" participating in case it affects their insurance policies [P3-ausEE]. One participant stated that "security of information needs to be well protected" so it is not "used against an individual or group of people" [P3-ausEE]. Two other participants stated that they would not use electronic health records or genetic testing for their children "until the safeguards and the benefits are clearer" [P16-ausEE]. One participant stated that "progressive watering down of privacy protections" meant they were concerned their children would be "refused insurance because of a decision I have made" [P16-ausEE]. Other participants felt the same way about this [P14-ausEE][P25-ausEE].

A similar mistrust was echoed in ASPREE participants' views about private companies and social media organisations. Seven ASPREE participants said they would be interested in being involved in decisions about data access, with all participants stating they while they were comfortable with their data being held by academics, they were not happy with them being held by a for-profit company. As previously mentioned, however, one participant said not to "rule private companies out completely".

Five Shared Ancestry participants raised a number of concerns about genomics research being misused for political purposes and "wicked ends" [P5-SA]. One participant stated that research should not be used "for political purposes" but indicated that they did not believe this was easily

prevented [P5-SA]. Another participant shared a view about "realism about the likely corrupt use of genetic information for political and financial purposes" [P2-SA]. A number of participants cited historical examples of genetic discrimination by regimes that had provided important learning for future genomics research. One participant cited the well-documented case of the large technology company IBM being complicit in enabling regimes to carry out forced negative eugenics policies.[454] For this person, IBM were "the enablers for the Nazis' ability to hunt down Jews and other 'undesirables'" [P6-SA].

The Indigenous Precision Medicine case study involved co-creating a research protocol. It required much careful discussion about ensuring that Indigenous community members were equal partners in all decisions about what kind of data was collected and how the data would be collected, shared and analysed. Concerns regarding genetic research – such as genetic discrimination in employment, difficulty in obtaining insurance, and inappropriate use of genomic data in law enforcement – all needed to be considered. It was therefore necessary for the co-design process to balance these concerns with the researchers' requirements with regards to combining data.

With the convergence of for-profit companies (such as social media companies and direct-to-consumer DNA testing services) storing and selling information about people, governments around the world are increasingly partnering with such companies to provide health services. It is therefore crucial to be able to map people's preferences for how their genomic data (and other sensitive or personal data) is shared and with whom. As the STARDIT system offers a way to map and report on different stakeholders' preferences with regard to data usage, it provides a practical and scalable solution to this complex issue. The STARDIT system uses both the 'FAIR'[85] and 'CARE Principles for Indigenous Data Governance' to structure the data,[100] making it a particularly useful tool for genomics research involving Indigenous participants. Reports from STARDIT could be used by people to help them make informed decisions about sharing their own data or participating in genomic research.

Different tasks for different people (322)

Participants from all case studies recognised that it might be more appropriate to involve certain stakeholders in some tasks, rather than others, with involvement in the design-stage tasks flagged as being more practical than in later stages of the study. A Shared Ancestry participant asked "… can we be both subjects and supervisors – at the beginning yes, later, maybe not?" [P7-SA]. The ASPREE study participants shared the view that involvement at the design stage is more practicable.

The ausEE participants liked the idea of a multi-stage approach to involving people, using a combination of face-to-face and online discussions [P25-ausEE], and this aligns with other models used in research priority setting.[15,422]

One Shared Ancestry participant commented that people (the public and research participants) will have a "variety of professional and technical and creative skills" which will be useful, with the most useful being "knowing ourselves" [P7-SA]. This mirrored comments in the ASPREE case study where participants noted that certain people have relevant skills and experience, and that "different personalities" might be more appropriately involved in certain tasks [P15-ASPREE].

The Indigenous Precision Medicine project has identified distinct tasks for different stakeholders, with members of the community involved in reviewing information and being part of consultations. According to the protocol, an independent Project Advisory Committee (PAC) will be established. It will be jointly chaired by an Aboriginal Elder nominated by the community and the Director of the Poche Centre for Indigenous Health of the University of Sydney, who is an Aboriginal Elder and senior researcher. The protocol also outlines how the training of local Aboriginal people as genetic counsellors and pathology collectors will be discussed with the local community. This variation in tasks for different stakeholder groups is a best practice example of power being shared transparently and different stakeholders having clear tasks and accountabilities.

Involve people in designing involvement (117)

Planning how people will be involved in every stage of a project is a complex process and requires input from the different stakeholders who will be involved. Findings from the review of genomics projects indicated that involving people in designing involvement would improve the effectiveness of such processes.[28] The results from all case studies supported this, with involving people in designing the involvement process resulting in a number of positive impacts. These impacts are outlined in detail in each case study.

One Shared Ancestry participant added that the public should have a voice in how "science and research can better involve" people [P5-SA]. This perspective aligns with findings from both the narrative review and the scoping review of public involvement in genomics. STARDIT was developed in response to findings and recommendations from the review of genomics projects.[28] Using STARDIT-PM for standardised preference mapping was a helpful tool to map people's preferences within a standard framework. This led to development of guidance for planning and evaluating

initiatives using STARDIT which, in turn, informed the Indigenous Precision Medicine project protocol.

Choosing what to know about your genome

Participants in the Shared Ancestry study held the view that there might be important variations in the knowledge that people choose to have about themselves and their genomes. Similarly, participants from the ASPREE study said they would like to be involved in deciding how information is shared, as they had concerns about people learning things they did not want to learn. One ASPREE study participant considered that knowledge about whether someone was carrying a variation which pre-disposed them to Huntington's could be a "poisoned chalice" [P4-ASPREE]. Such considerations were central to the protocol design of the Indigenous Precision Medicine case study.

The finding that participants wished to be involved in deciding who decides, and who knows what, aligns with a 2015 United Nations report which poses questions of autonomy in genetics. Decisions must take into account both the person being tested and implications for relatives and communities of people who may share the same or similar genetic variations.[56] While there are no simple answers to these questions, involving people affected in the decision-making process is essential. In February 2021, for example, after careful consultation, results were returned to some ASPREE-XT participants.[511] The lead investigator (PL) for the ASPREE project said that data from the ASPREE case study about people's preferences about access to their own genomic data and analysis was used when making the case to ethics boards for returning data.

The STARDIT system allows standardised reporting of who has been involved (including people with specific genetic variations) using both FAIR and CARE criteria. It provides transparent data about the different preferences of people with variations. It helps inform decision making and allows others to critically appraise how different stakeholders have been involved.

Ivory towers, silos and bubbles: labelling the academic constructs and mapping ethical grey areas

Research for this thesis used the paradigm of participatory action research within a PhD structure and it is important to record the ethics structures of the organisations involved. In every case study explored in this thesis, the ethics process either had inherent 'grey areas' or some study team members felt ethics processes were incompatible with the principles of the participatory action research paradigm. While there are difficulties in mapping this within the structure of a PhD, this

section attempts to report on these incompatibilities. This is an important part of the learning process and, thus, of the results. The implications described in this section are explored in more detail in the Discussion chapter.

Codifying and co-labelling relevant power structures from the start of the research process ensures that the very concept of a PhD must also be scrutinised according to the values of those working in partnership. This is especially relevant when working with people at greater risk of exploitation. For example, Australian Indigenous communities who have traditionally been over-researched and under serviced,[2,5] people affected by rare diseases, and those populations of special interest to genomics research. In this doctoral research, these communities are represented in the Indigenous Precision Medicine case study, the ausEE case study and the Shared Ancestry case study, respectively.

The study teams worked with each of the communities using the participatory action research paradigm. Under this paradigm, it was essential to map any ethical issues identified by community or study team members and to work in partnership to co-define the issues and co-create solutions to bridge any 'disconnects".[512,513] This included addressing complex questions such as power and representation in communities,[512] which is particularly complex for genomics research, as findings might have implications beyond individuals. There are also tensions in balancing participants' autonomy while ensuring that they are protected from risks they may not be aware of – such as their data being used for purposes for which they have not given consent.[513]

Shared Ancestry

The STARDIT Alpha report for the Shared Ancestry case study was co-created by study team members and participants and provides important data.[4,295] It noted that "the co-design process took longer than expected owing to ethical 'grey areas', with no clear instruction on whether ethics approval was required to involve people in co-design".[4] As a result, an after the initial ethics approval, subsequent feedback from the co-design process was integrated by applying to modify the ethics approval to take into account the preferences of potential participants.

During the online discussions, participants identified ethics committees as having "failings", and one participant asked "who will decide who will be on the ethics committee?" [P4-SA]. Another suggested that members of the sibling group could "form and seek out participants for the Ethics Committee" [P6-SA]. Another added "I am not sure of the ethics process, but it does seem a shame that more of us cannot participate" [P5-SA]. One participant stated "I trust the scientists and the

ethical committees" [P9-SA], while another noted that "the ethics of DNA research generally will continue to be of huge importance and will continue as a political issue, triggering new laws and regulations" [P7-SA]. This person raised a concern that "the law will not be able to keep up with the research – and we as members of the general public won't either" [P7-SA]. One participant noted that while people might be experts about a process or data, that does not make them "moral guardians" [P5-SA]. This person concluded that "no single body (and that includes the church and the government) has a right to dictate moral guidelines" [P5-SA].

During the Shared Ancestry project, it became apparent that some current ethics processes are incompatible with a participatory action research paradigm. The following is a short extract from the STARDIT report written after completion of the project:

> The entire study team agreed that limitations in the ethics process affected the extent of how the sibling group could be involved in the study. Internationally, confusion still surrounds what ethical approval is required before involving potential participants in co-designing research.[514] On the advice of the La Trobe University Human Research Ethics Committee, the study team did not approach potential participants about co-designing the study until after ethics approval had been granted, with feedback from participants being incorporated by a number of subsequent modifications to the original ethics application. As a result of the complex process of modifications, the timeline for feedback was shorter than the study team had anticipated, although the process did provide useful feedback. Ambiguous policies for the ethical involvement of people in co-designing research can hamper the degree of control potential participants have in research and further clarity from ethics committees will enhance power sharing at this crucial stage of research. [295]

An unplanned change of Chief Investigator (CI) and administrative processes related to the ethics process (which were outside of the control of the study team) led to the facilitator (MC) reporting that support from outside the study team was "non-existent" and was "wholly inadequate" for the participatory research process being used. [295] For example, investigators were instructed by the outgoing CI that they could not contact participants at all, including to explain that the study had been suspended. This removed participants from the 'participatory' loop and confirmed the La Trobe University Human Research Ethics Committees as a locus of power, which is incompatible with the principles of the participatory research paradigm in which everyone is involved at every level.[512] As all of the participants of the Shared Ancestry project are biological relatives of myself (JN) (including

my mother), as one of the investigators of the study team this created unique challenges, which were recorded in the reflexive research diary I kept during the research process.

While there were complexities to working with my own biological relatives in the capacity of a researcher (including my biological mother), the main challenges were navigating ethical processes, rather than any ethical considerations about my involvement from relatives. Rather than present inherent challenges, my perception from informal (and unrecorded conversations with my relatives) was that my involvement as both a biological relative and a researcher increased the trust between the research participants and the research process, and may have meant that more people participated than would have if it was research where a relative was not involved.

ausEE

Potential participants were involved in co-designing the study and concluded that involving children affected by EGID would be too ethically complex. Consequently, only those over 18 years old were involved in co-design and participation. Like the Shared Ancestry project, the co-design process took longer than expected owing to ethical 'grey areas', with no clear advice from the ethics committee on whether ethics approval was required to involve people in co-design. As a result, after the initial ethics approval, subsequent feedback from the co-design process was integrated by applying to modify the ethics approval to take into account the preferences of potential participants. Again, the study team agreed that the limitations of the ethics process affected the extent to which potential participants group could be involved in study co-design.

ASPREE

In the ASPREE case study, participants' preferences to have their data returned to them presented ethical difficulties for the proposed multi-generational research study (MGRS). In a response to an email survey, the lead investigator for ASPREE-XT (PL) stated that it was "fascinating" that all participants wanted "a self-managed future of health information", which he described as "very at odds with the current system" and requiring "justification to the ethics committee".

In addition, some participants had concerns about themselves or their biological relatives (especially offspring) finding out information they "might not want to know" (P6-ASPREE), or having it imposed on them. This issue is common to almost all ongoing and proposed genomics research studies and should be urgently addressed by contemporary ethical frameworks, which favour individualist perspectives over collective ones.[404]

Indigenous Precision Medicine project

This initiative encountered difficulties in publishing a protocol about a planned co-design process, as ethics approval was required even before starting the co-design process.

During the co-design stage, participants raised concerns that information from research data gathered for the project could be used for law enforcement purposes, and that researchers could not guarantee that it would not be. The research protocol responded by combining genomes to create a reference genome, rather than using a biobank model in which individuals' data were recorded.

Sharing data and publishing

There are related ethical issues with regards to sharing research data and publishing participatory action research in peer-reviewed journals. The United Nations' Report of the International Bioethics Committee on updating its reflection on the human genome and human rights' explores scientific knowledge about the human genome. It characterises it as a "heritage of humanity", states that it is "a common good", and that "open access should be therefore guaranteed".[56]

Data from genomics research require continuous evaluation of the scope of informed consent, including the balance between confidentiality, data protection, and equitable sharing of research results with all participants.[56] This point was echoed most articulately by ausEE case study participants. Four ausEE participants mentioned insurance as an area of concern that requires public involvement and scrutiny with regards to data sharing, in particular in relation to data being misused by for-profit companies. One participant stated that "progressive watering down of privacy protections" meant they were concerned their children would be "refused insurance because of a decision I have made" [P16-ausEE].

The United Nations report also calls on the international community, governments and researchers to address the very complex issue of the conflict between the right to access scientific knowledge and other relevant principles, including "patents for protection of intellectual property".[56] This issue was also raised by a participant in the Shared Ancestry case study who, unprompted, stated that "Research is for humankind. Its benefits should be available to all. Information should be for the most part easily available. If it's publicly funded, it must be publicly available. Patents should be limited in time, and patenting new life forms is unethical" [P9-SA].

Emerging forms of 'citizen' or 'participatory research' mean that people are increasingly empowered to "collect their medical information and even to analyse it".[56] Some ausEE case study participants commented that people affected by diseases exist "across economic and political boundaries" and that access to medication is affected by "who owns, profits, controls, patents" [P3-ausEE]. These concerns were echoed in the United Nations report, which states that the trend for limiting access to research data "is fostered by the secrecy of the pharmaceutical companies and academic medicine about the results of their research, so that individuals with rare diseases are using the existing resources like the internet and Citizen Science to find out more about their conditions and diseases and share this knowledge".[56]

Access to information, including 'borderless' access, was raised as an issue that affects who can be involved in genomics research [P3-ausEE]. The participants from the ausEE case study also stated that paywalls for articles have direct negative impacts on patients, carers and parents who wish to make informed decisions and "judicious choices" about healthcare, or to get involved in research [P3-ausEE]. Participants from both the Shared Ancestry and ausEE case studies shared the view that if public money is paying for research, participants should have control of these data, and research about participants should not be published behind paywalls, which participants often cannot afford to access.

The STARDIT system was developed in partnership with the WikiJournals, a Wikimedia Foundation project that provides free access to both public and peer-reviewed research. While STARDIT was used to report aspects of this doctoral research in the public domain, financial tensions exist as most open access publishers charge researchers fees to publish open access articles. For example, universities encourage PhD candidates to publish in journals as they are perceived to be prestigious or have a high 'impact factor'. This is often motivated by universities' desires to meet academic outcome measures, which have a strong connection to their business models, which in turn may reflect policies and performance measures of governments and other funders. In addition, within universities there are often not accompanying budgets to pay open access fees.[515] This situation makes it difficult to publish results from participatory action research in a format that participants themselves can freely access.

Academic structures can thus create competing, conflicting or even perverse incentives to publish research behind paywalls in order to meet perceived prestige thresholds. While there is nothing inherently wrong with publishing in pay-walled journals because they are considered 'prestigious', in

the context of the participatory action research paradigm, these arrangements raise the question of whether the priority is sharing knowledge openly, or sharing it in a way that is perceived as beneficial to the host or funding organisation.

A further advantage of STARDIT relates to sharing of data about the ways in which participants were involved in initiatives. Creating STARDIT in partnership with WikiJournals and other organisations allows people to self-identify with certain 'labels' when describing how they were involved in research, and the preference mapping tool provides a way to map different stakeholders' preferences for data sharing and publishing.

Chapter 10 – Discussion

Introduction

In this chapter, I will:

- summarise the aims and main findings of the thesis (see Table 10.1)
- summarise the outputs from this thesis
- evaluate and appraise the research design, methods and limitations of this thesis
- synthesise some findings from the results and discuss their relevance to communities of shared interest
- summarise new knowledge generated by this thesis and dissemination of findings, demonstrating how some findings and outputs from this thesis are already being used
- discuss the implications of the findings and new knowledge for genomics research and other related areas
- summarise my recommendations based on these findings, including areas for future research.

Table 10.1: Summary of aims and findings

Aims	Activity	Output	Findings
To understand when and how people have been involved in human genomics research to date.	Reviewed the literature to assess methods of public involvement that may be applicable for human genomics research (or those that are already being used).	Published scoping review, with 20 citations (as of 1st November 2021, as per Google Scholar).	Improved understanding of contemporary involvement in genomic research. Lack of a standardised way of reporting involvement in human genomics and of transparently evaluating ways people are involved, has resulted in no evidence base to inform best-practice.
To apply a participatory action research (PAR) method to human genomic research using four case studies, in order to learn more about the practicalities of involving people in genomic research.	Applied a participatory action research paradigm to studies of four different groups of people associated with human genomics research (underway or in planning) and used participatory action research to co-produce STARDIT. Planned and conducted the involvement activities and, where appropriate, shared learning experiences, and described how involving people affected or informed future genomic research.	Three published case studies and a published research protocol for a genomics research project.	Participants reported positive impacts from involvement in all four case studies, in addition to important learning relevant to each community. Stakeholders from all case studies reported changed views regarding involving people in research as a result of either participating or being a study team member. All changed views represented a move towards valuing the importance of involving people in genomics research.
To develop a standardised way of planning, reporting on and evaluating involvement (STARDIT) in order to improve future genomics research.	Initiated and then led the facilitation of the process with multiple international stakeholders. Co-created a standardised data sharing system to compare the views and perspectives of different stakeholders about genomics research across the four case studies,	(1) An Alpha version pre-print (cited 5 times as of November 2021); (2) published a public consultation report about co-design of STARDIT Beta;[284] (3)	STARDIT provides a useful way to report data about genomics research and other initiatives, including data about how different stakeholders are involved.

including views about involvement, methods of involvement, and any impacts and outcomes of the involvement.

published a working Beta version Standardised Data on Initiatives (STARDIT);[9] (4) and a final version submitted for review (under review as of November 2021).[9]

STARDIT has been used beyond genomics research, including by Cochrane,[296,297] citizen science projects[217,516] and the Wiki Journals[17]

Statement of main findings

In this thesis, I have demonstrated ways of:

1. mapping and reporting how current genomics research initiatives are involving people, including presenting this as a scoping review
2. mapping different stakeholders' preferences for involvement (researchers, participants and the wider public)
3. involving potential participants in designing how they will be involved in the research process and reporting these data
4. reporting how stakeholders have been involved in the research process using standardised, structured data which is both human and machine readable
5. comparing standardised data about involving people in order to evaluate the processes and to help inform future decision making for those planning to involve people in research.

Scoping review

The scoping review improved understanding of contemporary involvement in genomic research. The review has been well read at 7929 views, and has been cited by 20 other peer-reviewed papers and five Wikipedia pages (as of 1st November 2021, as per Google Scholar). Learning from the scoping review and the methodological techniques developed has informed subsequent scoping of reviews in the field.[33]

The scoping review found that that "without a standardised framework to report and transparently evaluate ways people are involved, it will be difficult to create an evidence base to inform best practice". This finding informed the initiation and subsequent co-creation of the 'Standardised Data on Initiatives' (STARDIT) framework.

Standardised Data on Initiatives (STARDIT)

Co-created in parallel with the case studies, STARDIT provided a consistent way to report the planning, methods and evaluation of the case studies. This work facilitated a cross-case analysis that provided standardised quantitative and qualitative data and allowed comparisons between the case studies to be made. The STARDIT system also facilitated the collection and comparison of impact data, including impacts from transformative learning, which would otherwise have been 'challenging to quantify', record, report and compare.[171]

Case studies

Participants enjoyed the experience of being involved in the case studies and participating in the online discussions, telephone interviews and face-to-face events. The reported impacts and the fact that participants enjoyed taking part in the online discussions have added relevance in a post-COVID-19 world, in which use of online tools to involve people in accessible and evidence-informed ways will be a priority for genomics initiatives and other kinds of initiatives around the world.

According to data gathered from research teams and participants, participatory action research (PAR) proved to be a successful paradigm to guide the research processes. However, participatory action research methodology was sometimes incompatible, or practically difficult to achieve, within existing ethical frameworks. The implications of these results are discussed in detail in the section 'Implications for ethics and participatory action research'.

Positive impacts and important learning relevant to each community were reported from all case studies. Stakeholders from all the studies reported changed views about involving people in research as a result of either participating or being a study team member. Where quantitative data were gathered, all changed views represented a move towards valuing the importance of involving people in genomics research.

Study team members also reported changed views as result of involving people. For example, some ASPREE study team members who reported being hesitant about involving people at the start of the process, reported changed views about the value of involving people as a result of being part of the process. While such 'transformational learning' is a valuable outcome, it is extremely difficult to capture and express in quantitative data. This demonstrates the importance of both learning by doing and using platforms such as STARDIT to capture that learning.

The STARDIT platform provides a way to record data about impacts and outcomes both quantitatively and qualitatively. However, the skills and knowledge required to plan, do and report involvement will require further research into the learning and development needs of stakeholders for each project. Mapping the knowledge and skills of stakeholders, and any areas where these might need development, can be done by using STARDIT-PM as part of the cycle outlined in the STARDIT Beta version (Figure 3.5).[9] This provides ways to embed learning and development needs into the design of a project, to report on needs assessment, and to evaluate the impact of any learning interventions. For example, application of STARDIT-PM might identify that certain communities are currently not included in any planned involvement activities. The mapping process might identify that study team members need to develop their communication and community

outreach skills or knowledge of the community in order to involve a community more effectively. Any learning and development intervention to improve these skills and knowledge, any impacts or outcomes of it, could be reported using STARDIT. Being informed about impact data from previous projects may help people planning future research to make evidence-informed decisions.

Outputs and dissemination

The published outputs from this thesis have collectively received more than 43 citations, demonstrating the interest in the issues explored in this thesis.

Review of Public Involvement in Global Genomics Research

This review has been cited 31 times (as of 11th August 2022, Google Scholar),[28] and the methods of categorising tasks, methods, stages of involvement have been adopted by other reviews, including an in progress review by Australian Genomics (as of 2022). The data extraction methods and findings from this review also directly influenced the inception of Standardised Data on Initiatives (STARDIT).

The review was cited by an article written with support from the Global Alliance for Genomics and Health (GA4GH), noting specifically:[517]

> 'As part of the process of considering engagement approaches, we also recommend building in time and resources to evaluate those approaches, though we acknowledging more work is needed to guide how best to effectively evaluate engagement and involvement (Nunn et al., 2019)'

Such important acknowledgements demonstrate that the urgent need for standardised data and evidence-informed approaches to involvement are increasingly being understood globally by genomics researchers.

Alternative processes of reporting involvement

This short section was added post-examination in August 2022, in order to further elucidate the context for why STARDIT was created and used, rather than using existing data reporting tools such as GRIPP.

A number of weeks after the publication of the 'Review of Public Involvement in Global Genomics Research', the article 'Frameworks for supporting patient and public involvement in research: Systematic review and co-design pilot' was published.[209] The findings from this supported those from the scoping review conducted as part of this thesis, including that while there was a 'plethora of frameworks', none were specific to reporting involvement in genomic research and there was no 'one' framework which could be used across disciplines. In addition the review found that "most

published frameworks have been little used beyond the groups that developed them", indicating usability issues and suggesting that creating more accessible or automated ways of generating reports might be advantageous. [209]

Standardised Data on Initiatives: Working Beta version

The STARDIT platform has helped plan and evaluate different projects, showing impacts from the processes. STARDIT is already being tested by the Wikipedia-integrated open access peer reviewed WikiJournals, which has articles which are integrated into Wikidpedia.[17] This include STARDIT being used by the Wiki Journal of Medicine to report data about an article on systematic reviews and scoping reviews. STARDIT has been used by other research initiatives beyond this PhD thesis, and has been suggested as a tool for reporting involvement in biobanks.[33]

Prospective STARDIT reports have also been created by:

- people involved with the prospective project 'DNA Voices - a platform for knowledge translation and precision medicine' in Brazil.[518]
- staff from the Manbhum Ananda Ashram Nityananda Trust, India, for a prospective tobacco control project working with the Indian public, including tribal communities will be involved in co-designing methods of measuring impact, and collecting and reporting data.[519]
- Investigators from the 'COM-IC – collaborative development of core outcome measures for dementia care' research project, which is involving people affected by dementia in developing a 'comprehensive and appropriate suite of core outcome measures accompanied by guidelines to monitor and evaluate impacts of care delivery models at individual, organisation and system levels'.[520]

People from multiple organisations continue to be involved in the co-creation, including people from Cochrane, Johns Hopkins University, Health Research Authority UK, European Organisation for Rare Diseases, Australian Citizen Science Association, The Poche Centre for Indigenous Health, and multiple Universities.

STARDIT adopted by Australian Genomics

Organisations beyond the partner organisations from this doctoral research have also used the STARDIT Beta version. The organisation Australian Genomics has established a working group to establish 'Involve Australia', an organisation which aims to "produce a set of health public involvement guidelines and resources for genomic research, prioritising the public perspective".[298] The project also plans to "conduct learning and development interventions to ensure genomic researchers learn how to effectively involve the public in their research".[298] I was invited to be a

member of the working group, which voted in favour of using STARDIT and created a prospective report to describe their planned work. The report will be updated throughout the lifetime of the project, and represents the first example of STARDIT being used by a national genomics organisation to report planned involvement of the public and patients in their work.

STARDIT cited by Global Alliance for Genomics and Health (GA4GH)

STARDIT (Alpha version) was cited as 'useful' as a way of 'evaluating engagement' in an article supported by the Global Alliance for Genomics and Health (GA4GH).[517] The STARDIT Alpha version is also cited in the GA4GH 'Framework For Involving And Engaging Participants, Patients and Publics In Genomics Research And Health Implementation' as a useful way of 'conducting evaluations of engagement'.[521]

A lack of guidelines for reporting and evaluating involvement and engagement, but an acknowledgement of this gap,[521] demonstrates the potential usefulness of STARDIT has been recognised by the researchers working with the GA4GH.

Work in Australian Government health technology assessment

During my PhD candidature, I was appointed to the Medical Services Advisory Committee Evaluation Sub-committee (MSAC-ESC) by the Australian Government Minister for Health.[522] I have given multiple presentations at five meetings since 2019, giving presentations about the public interest and patient perspective on medical technologies, including genetic testing. My own personal experience of bereavement when my partner died from sudden adult death syndrome in her early twenties (from what was likely an inherited cardiac rhythm disorders), informed my response, along with my professional experience from this doctoral research. The feedback I have provided has influenced Australian Government health technology decisions in relation to the use of genomic testing and related diagnostic tools. This included contributing to the public summary document for an item about 'Genetic testing for diagnosis of inheritable cardiac rhythm disorders'.[70] During meetings and in associated documentation, I made the point that as well as allowing better clinical diagnosis and access to potentially life-saving treatments, free genetic testing may also allow people who currently do not have treatment options to 'join communities of shared interest'[70], a concept informed by my own personal experiences, and my work on this doctoral research.

Voluntary work for Cochrane

I was invited to be part of both the Cochrane Advocacy Advisory Group, co-opted onto the Cochrane Consumer Network Executive and elected as a member of the Cochrane Council. In these roles, I have been able to inform Cochrane's work in relation to both involving people in genomics research

and reporting involvement in systematic reviews using STARDIT. In addition, I have been invited to co-lead work on developing a Cochrane Values Statement. The co-design process for this work is being reported by Cochrane using STARDIT.[296,523]

Voluntary work for the Wikimedia Foundation

The Wikimedia Foundation hosts open-access peer reviewed journals, which are Wikipedia integrated. As well as being a member of WikiJournal of Science I was elected as the Strategy Liaison for the WikiJournals. Working in these two voluntary positions, I was able to work with the Editorial team to co-design STARDIT and integrate it into the WikiJournal system, using WikiData. As a result, STARDIT is already being tested by the WikiJournals. This includes STARDIT being used by the WikiJournal of Medicine to report data about an article on systematic reviews and scoping reviews.[17]

Presentations and contributions to public debate

I have given a number of in person and online presentations, including webinars, PhD presentations[290], and winning prizes in two Three Minute Thesis competitions. My entry for the 2018 'Visualise Your Thesis' competitions was used as part of the learning resources for the two online discussion case studies and has received over 500 views. [524] I have also been invited to give two presentations to the Board of the Poche Centre for Indigenous Health to present about both my PhD findings and about STARDIT, in order to inform future reporting of genomics research by the Poche Centre. I was also invited to give a presentation to Ludwig Boltzmann Gesellschaft (Austria) in 2020 on participatory action research, informed by my PhD.[525,526]

I have informed public debate, by participating in discussion on the UK's Genetic Society's podcast as a guest, discussing my PhD and STARDIT. The podcast was the most listened to episode of the Genetic Society's podcast in 2020. In summary of my contribution on the podcast, a review from BioNews (part of the Progress Educational Trust) stated "the public must be given more power, both in how this research is conducted and how this data is used".[527] It continued "it's time for us as the public to get involved. If you would like to expand your perspective on one of the most relevant scientific breakthroughs of the past century, this podcast is well worth a listen".[527]

Table 10.2: Summary of presentations during doctoral research

Title	Invited by	Given by	Date
Standardised Data on Initiatives (STARDIT)[287]	WikiCite 2020 Virtual conference	Jack Nunn and Thomas Shafee (recording, resource and transcript)	27th October 2020
Standardised Data on Initiatives (STARDIT)[288]	Poche Centre for Indigenous Health, 9th Annual Research Showcase Program	Jack Nunn	18th November 2020
Involving people in DNA research	Poche Centre for Indigenous Health, Research Advisory Board	Jack Nunn	8th September 2020
Involving People In DNA Research	Ludwig Boltzmann Gesellschaft	Jack Nunn (recording, resource, presentation)	24th September 2020
Genomics Research and Involving People[290]	La Trobe University	Jack Nunn	13th October 2020
Involving everyone in research: Creating the evidence[291]	Australian Citizen Science Association	Jack Nunn	1st April 2021
Involving People in Rare Disease Research	Rare Voices Australia	Jack Nunn	5th August 2021
Science Communication in a Pandemic	World Evidence-based healthcare day	Jack Nunn (recording)	20th October 2021
STARDIT and InGeNA	Industry Genomics Network Alliance	Jack Nunn	25th October 2021
Ideas Forum - Stem Cells Australia	Stem Cells Australia	Jack Nunn	9th December 2021

I have also co-produced two podcasts in my capacity as Director of Science for All which discuss using STARDIT to report participatory action research, and how it integrates into WikiData.

Synthesis of ideas

Through synthesising knowledge from the literature review and learning from the participatory action research process used for the four case studies, this section will answer the following questions:

1. What did we learn from involving people in genomics research, that we would not otherwise have known, and that could inform future genomics research?
2. Is enough being done to involve different stakeholders in shaping genomics research? If not, what should be done?

Ways of protecting people from exploitation in genomics research

The findings from this thesis have particular relevance for people at greater risk of exploitation. In this thesis I describe working with three populations at greater risk of exploitation, to map their preferences about involvement and to involve them in research. Members of the first population share a common sperm donor. This large group has potential to provide new research insights into medical, genetic, sociological and psychological studies. As they are of inherently high interest to research, they are at greater risk of exploitation by researchers and those looking to profit from discoveries made about the population.

During my candidature I discovered that I was part of this population, and initiated the co-design process described in the case study. After that was complete, this population of half siblings was approached by a research organisation (which will not be named) inviting them to participate in some genomics research. Participants were asked to either give consent to participate in one particular study or have their tissue and DNA stored “for future use in research projects that are an extension of this research project”. As the consent document put it, “Alternatively, we may use your sample for future research that is closely related to the original research project or as a control tissue sample”.

This invitation came after some of the siblings had participated in the online discussions for this doctoral research project. In the subsequent STARDIT report, the Shared Ancestry Case study participants recorded that the online discussions had helped them make an informed decision about participation in the study. In some cases, participants decided not to share their DNA and tissue samples beyond the scope of the specific project they were invited to participate in. This is an important example of a significant real-world impact from a co-created learning intervention that facilitated and moderated discussion. Such interventions, and the processes they can enable, can

help people at greater risk of exploitation explore their own preferences about research and make more informed decisions about participation.

The second population I worked with and describe in this thesis comprised people affected by Eosinophilic Gastrointestinal Disorders (EGID), including Eosinophilic Oesophagitis (EoE), which is a rare disease.[58] These participants were concerned that research priorities were driven by a desire for a return on investment. Four people mentioned insurance as an area of concern requiring public involvement and scrutiny in regard to data sharing. As already stated, one participant commented that it is a "valid community fear" that research data will be used "for" insurance companies or other for-profit organisations [P3-ausEE]. Participants' concerns about misuse of data by insurance companies, governments and industry indicate a power imbalance. They clearly demonstrate the importance of involving potential research participants in making decisions about how their data will be shared and who will make decisions about data access. Both the STARDIT-PM tool and STARDIT reports provide standardised, human and machine-readable data about people's preferences, empower people to create a public record of such preferences, and hold researchers and other stakeholders to account should those preferences not be respected.

The final population I worked with and report on as part of this thesis are potential participants of the Indigenous Precision Medicine initiative, which is ongoing. In common with Indigenous peoples worldwide, they can be considered at greater risk of exploitation than other populations.[2,5,434] The reasons for this are explored in detail in Chapter 7. The study team's decision to adopt STARDIT as the reporting system for the protocol indicates that it is valued by researchers working on co-designed genomics research with Aboriginal peoples. By reporting the planned research, and how the project plans to involve people, the study team have created a public record to which people from the Aboriginal communities can refer and, if necessary, hold researchers and other people from dominant groupings or cultures to account.[98]

Learning from the three completed case studies has implications for involving people in research around the world. This learning is particularly valuable in developing countries and countries where people may not be literate (or 'health literate') or able to give informed consent to participate in research. Creation of public records on how people have been involved in research, how data will be used, and who is involved in decision making may allow people at greater risk of exploitation to make more informed decisions about participation in research. Just as there are social determinants of health,[528] there are social determinants of involvement which can weaken or strengthen "people's control over the factors that affect their health".[529]

The scoping review of involvement in genomics research conducted for this doctoral research identified a number of government-led initiatives that have reported on recruiting research participants from populations at greater risk of exploitation.[28] For example, initiatives in the USA and UK sought to improve recruitment to genomics research initiatives by involving participants who identified as members of specific 'ethnic' communities. Genomics England published the following advertisement:[530]

> Become a Genomic Patient Champion: In order to help tackle health inequalities in some communities, we are keen to hear from people who are involved in community-based work or activities. Could you draw upon your experience of participation to offer support and advice about the 100,000 Genomes Project within community groups such as Black and Minority Ethnic (BME)?

This advertisement is similar to others used by government-led genomics research initiatives in UK and USA. Of concern, these often combine the concepts of 'ethnicity' and cultural communities. While involving people from diverse communities in recruitment can be effective at improving recruitment,[35] leveraging existing partnerships or community engagement solely to increase participation raises concerns. Where people are only involved in recruiting others to research (rather than in other aspects such as governance) this might raise ethical concerns. Similarly, the questions of who defines 'communities' and whether people self-identity as members of them (such as the 'Black and Minority Ethnic' 'community') have important implications, in particular for countries with a history of systemic racism, including Australia.[482,483]

If lessons from the Belmont Report on ethical principles for research involving human subjects are to be heeded,[474] to ensure that people are not exploited, involving them in recruitment must be done alongside involving them in other aspects of the research. Involving people can help ensure power imbalances do not exist and that individuals' and communities' health priorities are met.[34] Vigilance from the public, policy makers and researchers is needed to ensure that the concept of 'involvement' is not reduced to a method of improving recruitment in populations at greater risk of exploitation, rather than of sharing power with them throughout the research process. Systems like STARDIT can empower more people to be involved in creating, sharing, analysing and using data to help inform decision making and, ultimately, to improve both their own health and healthcare systems.

People's decisions to participate in research (either publicly or privately funded) may be influenced by public domain data generated from STARDIT reports and similar platforms. Governments and

health service providers who are paying third party providers to store and access data can now create public domain reports about how data will be shared, sold or analysed. By extension, this has important implications for people around the world who are paying for-profit companies to provide direct-to-consumer DNA testing. Often these services use a 'tick-box' consent process or the requirement for users to consent to sale of their data to third-party organisations are embedded in user agreements. Systems like STARDIT can empower anyone in the world to create transparent reports about how such data are being used or planned to be used. Using machine learning and other analysis tools, potential consumers of DNA testing from for-profit companies may be able to make informed decisions about which service they choose. These decisions could be based on publicly available data that show how each company will use DNA and other data, making it easy to compare the practices of competing services. The availability of such data may prompt companies to improve their consent processes so that people continue to share their data. The STARDIT and similar platforms may thus empower those who are currently disempowered by relatively unaccountable companies operating in international free markets.

Strengths and limitations

The aims of this thesis were:

1. To understand when and how people have been involved in human genomics research to date
2. To apply a participatory action research method to human genomic research, using four case studies, in order to learn more about the practicalities of involving people in genomic research
3. To develop a standardised way of planning, reporting and evaluating involvement in order to improve future genomics research

While all of the aims of this thesis have been achieved, the purpose of this section is to evaluate the strengths and limitations of different paradigms and methods used within the thesis to achieve these aims. As this thesis was guided by a number of paradigms and used a number of different methods, this section is divided into sub-sections which evaluate each paradigm and method.

Strengths and limitations of paradigms used

This doctoral research was guided by a number of paradigms, which in themselves guided the theoretical frameworks and methods. This section is a short evaluation of those paradigms.

Evaluation of the rights-based paradigm

The application of the rights-based paradigm was helpful during this thesis. The detailed documentation provided by the United Nations, World Health Organisations and other organisations provides a clearly articulated framework on which to both justify and build a participatory action research process. By relying on internationally ratified frameworks, it releases the individual researcher from the burden of both providing and justifying their own values, and asking others to agree. Concepts such as universal human rights are so universally accepted, that they are thus a helpful starting point to frame all other discussions.

While the tension between ‘consumer’ rights, ‘individual human rights’ and concepts of ‘collective solidarity’ were naturally impossible to solve with doctoral research, the methods described here provide a foundation to provide exact and multi-lingual descriptions of actions, conflicts of interest and labels to which people might self-identify. STARDIT can be used to report these kinds of data in relation to the participatory action research process. This data, when combined with data about

impacts and outcomes, can have an empowering effect and help promote individual, consumer and collective rights in the context of genomics research and beyond.

When working with Indigenous peoples in countries such as Australia where there is both a history and continued evidence of systemic racism and structural violence towards Aboriginal peoples[482,483], international paradigms such as the UN 'Declaration on the Rights of Indigenous Peoples' provide helpful guides when co-designing a participatory action research process, and a helpful reference when both Federal and State level legislation, policy and funding is lacking.[2] The use of the STARDIT system to also publicly state how the participatory action research process would work, including shared control and Aboriginal people involved in data access decisions, means there is a public record which any research participants can use to hold researchers to account, should the protocol waiver from the original co-defined principles. STARDIT reports may provide a useful way of building trust between researchers and genomics research participants at risk of exploitation (such as Indigenous peoples), by providing a clear record of how research has been designed, and with who. When people are asked to provide informed consent to participate, such information can provide clear and unambiguous information about every aspect of such a study, and how it aligns with both FAIR[85] and CARE[100] data sharing principles.

Applying the open access paradigm meant that all of the papers in this thesis were shared using open access publishing. This was reported in detail in the section 'Sharing data and publishing' of the Results chapter. It is important to note there can be difficulty in achieving this when University budget policies (including that of La Trobe University) state that PhD budgets cannot be used for open access fees. This can mean that PhD researchers are forced to either publish behind paywalls, or seek alternative means of funding, which is not always possible. Publishing the peer-reviewed articles from this thesis in open access journals relied on me volunteering as a member of the Editorial board for a journal and funding from another University, in order to access free publishing. The debate about open access publishing and who should pay for it is wider than the scope of this thesis[531,532], but has serious implications for participatory action research led by communities with limited financial resources to pay open access fees. The Wiki Journals offer free peer-review and publishing of articles, and such volunteer-led models, backed by large charities to provide IT infrastructure, might be one such viable model.

While the rights-based paradigm is a helpful starting point, it is worth noting that such frameworks are still limited by language, and are often attempts to express concepts, values and moral stances which are not easily expressible in language, or where important meanings can be lost when translated. While 'real-world data' about applications of such rights frameworks in healthcare and

beyond can help articulate concepts[533], working to co-define such concepts across multiple human languages remains an ongoing challenge in this paradigm.

Evaluation of the participatory action research paradigm

This thesis was guided by the paradigm of participatory action research where possible. While it was not always pragmatic and was ethically complex to apply the paradigm in the context of a PhD, the participatory paradigm guided every aspect of the thesis and is evaluated in this section.

Informed by the participatory action research paradigm, we created a private online space for the facilitators from both case studies which used online discussion, in order to provide a place for self-critical reflection, share experiences, and offer support to one another. Feedback from facilitators from both case studies was that this online space was extremely helpful and provided both practical and emotional support. For examples, technical issues with using the online platform could be quickly solved with group discussion. Similarly, issues with participant engagement could be discussed and compared across the two studies, providing facilitators with reassurance that a relatively slow reply rate was occurring in the other online discussion too, and that it wasn't their own facilitation that was causing this.

While the co-design process worked well, it was inherently difficult working within academic structures and attempting to do true participatory research. For example, the ethics processes within universities are not designed for participatory action research and constrained the process by requiring the design to be submitted before people were involved, which resulted in needing to do formal modifications multiple times for each different study, in order to incorporate feedback from the different stages of the participatory action research process. Allowing sufficient time for co-design was also challenging and limited the extent to which participants could be involved in some tasks, such as co-authoring papers. Planning in more time for this process would have improved the duration given to stakeholders to give feedback as part of the participatory action research process.

Two of the case studies were originally part of the same ethics application, but on the advice of University staff, they were divided into two different studies, with the data collection, questions, and discussion structure remaining almost identical in order to allow a cross-case analysis. Designing case studies in parallel meant that in order to have some consistency (for purpose of comparison) some aspects were inflexible regarding co-design.

According to feedback from co-authors, the participatory action research process for STARDIT worked very well.[534] Facilitating this process through the legal structure of the charity Science for All

(auspiced by the Royal Society of Victoria) gave more methodological freedom to the participatory action research than would have been possible within the structure of a university. For example, organising face to face events was done according to the organisational policy, and covered by public liability insurance of the Royal Society of Victoria. However, while the participatory action research process used to develop STARDIT was enormously helpful[16], the scope of STARDIT grew beyond just genomics research. While it remains useful and relevant for planning, reporting and evaluating involvement in genomics research (as demonstrated by the uptake by Australian Genomics)[298], it can now also be used across multiple other disciplines, increasing the likelihood of any future uptake.

Involving people affected by research in how it is planned and conducted is best practice. Accordingly, from the outset of my doctoral research I began to establish a formal advisory group to guide the overall doctoral research (in addition to my supervisory team, a formal academic team which guides doctoral research on behalf of the University) in order to incorporate the perspectives of people directly affected by genomic research, such as people with diseases caused by genomic variation. However, this endeavour was actively blocked by some members of the supervisory team, and thus I could not proceed with this way of working. While it is impossible to estimate how this decision may have affected the overall doctoral research at the review stages, I was able to conduct more informal (and unpaid) involvement of people affected by genomic research, including conversations with people affected by rare diseases which influenced the design of elements of the doctoral research (see the section 'Unselected case studies' for more information). The subsequent use of the participatory action research paradigm to guide each individual case study also ensured that all research with the various communities from the four case studies actively involved people directly affected by genomics research (or who could be affected), including people affected by variations of known significance and communities of people with shared ancestry.

Strengths and limitations of methods used - summary

Using more than one theoretical approach is preferable in health research as no one single methodological framework will provide all the data and insights required in order to answer the complex questions proposed by this thesis.[158] It is noted that each chapter contains an analysis of the strengths and limitations of the methods within. Accordingly, Table 10.3 summarises an evaluation of the multiple methods used in this doctoral research.

This section relating to STARDIT in the table below is an evaluation how effective STARDIT was for enabling consistent data reporting and a cross-case analysis. It is not an evaluation of STARDIT itself.

A discussion about the strengths and limitations of STARDIT can be found in the discussion section of the peer-reviewed section 'Standardised Data on Initiatives - STARDIT: Beta Version.'[9]

A more detailed evaluation can be found in the appendix, in the section 'Strengths and limitations of methods used – detailed analysis'.

Table 10.3: Summary of strengths and limitations of methods

Method evaluated	Section	Strengths	Limitations and future research
Reviews	Narrative review of systematic reviews: How are the public involved in health research and what are the impacts?	1: Demonstrated there is not currently enough data to complete a meta-analysis of quantitative or qualitative data about involvement in genomics research. 2: Informed decision that a systematic scoping review was the most appropriate method to search for relevant data. 3: Findings suggested that methods of involving people guided by the paradigm of participatory action research were most likely to have impacts.	1: No systematic reviews specific to genomics research exist. 2: Number of systematic reviews in this area was limited at the time of the review.
	Public Involvement in Global Genomics Research: A Scoping Review	1: Database provided by the Global Alliance for Genomics and Health (GA4GH) provided a useful 'snapshot' of current international genomics research projects.	1: Authors were aware of initiatives not represented in database. 2: Updated database after the completion of the review added 125 new initiatives[48], creating an impetus for an updated review.

Method evaluated	Section	Strengths	Limitations and future research
		2: A decision on whether an initiative was reporting involvement required individual judgement, which was checked by multiple authors. 3: 'Involvement indicators' were developed and integrated into STARDIT so methods were more repeatable. 4: A subsequent review which explored reporting the impact of involvement in biobanks cited this review. It used a similar method and terminology, and suggested STARDIT could be used for reporting the impact of any involvement.[33]	3: The review extracted data at the level projects articulated in the database and did not analyse the structure of organisations which sat above multiple projects. 4: The search was related to the wider issue that concepts such as 'involvement' cannot always be expressed fully in linguistic constructs. 5: It was not always possible to determine whether involvement was 'consequential', so an assumption was made that all methods reported resulted were 'consequential'.
	Guidance for planning, reporting and evaluating initiatives: A multidisciplinary scoping review	1: This review explored guidance for planning, reporting and evaluating initiatives beyond the scope of genomics research in order to find relevant models and frameworks.[195] 2: This review showed the current variation in guidance on planning, reporting and evaluating initiatives in order to inform both future systematic	1: This review was carried out by a small team, so the capacity of the team limited the search design to certain methods which were pragmatic, rather than systematic. For example, we did not search the citations of results from the database search owing to the limited capacity of the team.

Method evaluated	Section	Strengths	Limitations and future research
		reviews and proposed standardised ways of reporting data on initiatives.	
Online discussions		1: While the research projects described in the case studies in this doctoral research were planned and completed before the COVID-19 pandemic, the methods of involving people online described in this thesis now have an unexpected relevance to many disciplines, as research projects around the world seek to involve people online in novel ways, and evaluate such methods in a standard way. 2: The ASPREE study showed that people preferred not to use social media companies to get involved in research, and this method aligned with the data storage expectations of participants (safe, secure and not owned by third party company). 3: The online text-based discussion method itself provided a flexible way for people in different time zones to get involved and interact.	1: Some participants logged-in multiple times and read comments but did not comment. While the follow-up survey attempted to capture the views of participants, not everyone responded, so it is hard to understand the behaviour patterns of those who did not comment. 2: One study team member noted the difficulty in achieving 'the balance of being prescriptive' (for consistency) and giving freedom to facilitators to initiate discussions and follow emergent themes. 3: Two participants from both online discussion case studies reported finding the platform 'complicated' and problematic [P5].

Method evaluated	Section	Strengths	Limitations and future research
		4: The facilitators reported that the support they received improved their ability to facilitate discussions, as did the shared online discussion space for the facilitators from both the Shared Ancestry study and the ausEE study. 5: Owing to how data was reported and shared (including using STARDIT), such learnings from these online discussions can be used to inform others who are planning similar methods. [535]	
Case study and cross-case analysis		1: The case studies selected for this doctoral research represent four unique and real-world communities of people, where genomics research affects their lives directly. 2: By ensuring that communities of people affected were involved in shaping this research, it has helped ensure learning is anchored in reality, rather than theoretical models of involvement in genomics research.	1: The sample sizes for the case studies in this thesis were variable, as was the percentage of people recruited from the known populations. 2: While datasets are still too small to draw any statistically significant conclusions, the mixed-methods approach meant that the interpretative analysis was able to combine themes and provide useful data, including comparing differences and generalisations.

Method evaluated	Section	Strengths	Limitations and future research
		3: The case study methods used in this thesis, guided by the paradigms described, allowed the collection and analysis of both qualitative and quantitative data from multiple sources, which provided a richer dataset. 4: The collection of data from study team members and participants provided a more holistic perspective, and meant data and impacts were collected that would not have been if data was just collected from participants. 5: Using STARDIT to combine data demonstrated a way to overcome this variation in both sample size and percentage of people recruited in the case studies, by combining standardised data. 6: While the case studies were variable in size and each had both 'representative features' and 'deviant features' (see the section 'Case selection'), the most significant learning for others planning genomics	3: The selection of case studies was influenced by pragmatic considerations. It is important to note that the unselected case studies also occupied a 'work-load' grey area, with conversations with potential partner organisations identifying areas for support in the organisations I was approaching **Future research** Future combinations of data could articulate more data to help understand any causal relationships for what 'widened' people's preferences for who should be involved. For example, data about whether it was being involved in the process, or a specific learning and development resource which changed people's perspective. Future applications of data collection using STARDIT could provide this level of articulation, which was not possible within the scope of this doctoral research. Such data could aid machine learning to help establish

Method evaluated	Section	Strengths	Limitations and future research
		research was likely to be from the 'generalisable' features of the case studies. 7: The cross-case analysis provided a successful way to apply the post-positivist paradigm and explore common themes across the case studies, and combine the quantitative data to allow cross-case quantitative analysis.	evidence informed ways of involving people with the most impact.
Standardised data reporting	Standardised Data on Initiatives - STARDIT: Beta Version	1: The preference mapping tool (STARDIT-PM) allowed consistent mapping of different stakeholder preferences across all case studies where it was used. 2: Applying a quantitative analysis to the identical questions asked at the start and the end of the online discussion case studies allowed an investigation of baseline preferences about involvement in genomics research and preferences after the online discussions. This enabled an analysis which showed how people's preferences changed, including showing a 'widening'	1: The co-creation process for STARDIT occurred during this doctoral research, with Alpha versions initially used to report. While Alpha reports were converted to Beta reports, Alpha reports were only published as PDFs, not structured data, requiring manual data extraction.

Method evaluated	Section	Strengths	Limitations and future research
		towards people preferring more kinds of stakeholders involved in genomic research. 3: Data was published online in the public domain in a consistent way, allowing it to be combined in the cross-case analysis, and open to future datasets which might use the same question structure. 4: STARDIT allowed consistent data on case studies to be reported (including self-reported outcomes), facilitating any future analysis and allowing future statistical analysis to begin to draw any correlation between certain learning interventions and learning outcomes, which may suggest causality.	
Outcomes and impact assessment		1: While time for longer term impact assessment is not possible within time limits of a PhD, it was still possible to measure outcomes and impacts immediately after the participatory action research process.	1: A follow-up survey was conducted with the study team for the ASPREE study, but not the participants, owing to ethical restrictions. As a result, there may be multiple unreported impacts.

Method evaluated	Section	Strengths	Limitations and future research
		2: Some impacts were able to be measured over two years after the online discussions had finished, as participants from the Shared Ancestry study were involved in co-creating the Alpha version STARDIT report in October 2020, and able to edit the Beta version from August 2021 onwards.	2: Preference and follow-up data was also not collected for the ASPREE study for the same reasons.

New knowledge

This section contains a short summary of the new knowledge generated from this thesis, which is explored in more detail in each respective chapter.

The scoping review showed that there was inconsistent reporting of involvement in genomics research. This included the finding that there is no standardised way to report on how people with certain lived experiences (for example, mental health phenotypes) have been involved in genomic research.

The scoping review and case studies also revealed the importance of involving multiple stakeholders, especially study team members and staff working with people affected by diseases as part of health services and research (not just the 'public' or patients). The perspectives of people with a professional relationship to those affected by diseases can provide valuable insights for research design. This is especially the case when potential research participants are affected by neuro-degenerative diseases; are too young; or unable for other reasons to be involved in co-design processes.

The case studies and the subsequent cross-case analysis showed that involving people in the participatory action research process has multiple impacts, including a 'widening' of people's preferences about who should be involved in research to include more people: potential research participants, members of communities of shared interest defined by genomic variations, health professionals and research study team members.

The STARDIT system was informed by both the scoping review and the participatory action research methods used in the case studies. This thesis, and associated peer-reviewed publications, have demonstrated STARDIT as a way to report on the preferences of all stakeholders and on planned or completed participatory methods. By providing both a standardised way to report on involvement, and a way to report how people with certain lived experiences of phenotypes have been involved in an initiative, STARDIT addresses gaps identified in the scoping review. Such data, combined with data on methods of involving people, on modes of communication, and on tasks people were involved in, can be combined with impact data to help those planning involvement in research make evidence-informed decisions about research design.

Implications and recommendations

Learning from this doctoral research thesis has implications within and beyond human genomic research, with applications in both wider genomics research, participatory research methods and citizen science across all disciplines. This section discusses the implications of this doctoral research on genomics research and beyond, and embeds the associated recommendations. As this thesis concerns human genomic research, the recommendations concern only the multiple domains of human genomics research (including research translation and health technology assessment).

The recommended use of systems such as STARDIT for reporting in these domains are numbered as follows:

1. report how people with lived experiences have been involved in annotating phenotype data
2. report how people have been involved in designing, managing and evaluating genomic research
3. report genomics research with populations at greater risk of exploitation
4. report health technology assessment processes around the world, including evidence assessment processes and how different stakeholders have been involved.

It is further recommended that STARDIT is used to:

5. transparently plan and report international debate on yōushēng and eugenics.

Implications for future human genomics health research

The unique features of genomics require new evidence-informed methods for informing involvement and power sharing in research. Tools such as "CTRL"[75] offer genomic research participants more control over how they participate in genomics research. CTRL is an "internet-based platforms to create a communication interface to support ongoing participant-led management of their involvement in research studies".[75]

Systems like STARDIT can be used to report how different stakeholders will be or have been involved in genomics research. Combining platforms like CTRL with reporting systems such as STARDIT will allow research participants, potential research participants, research funders and the wider public to appraise how their data will be used. Systems such as STARDIT can therefore be used by individuals wishing to make informed decisions about participating in genomic research or sharing genomic data, including participating in biobanks. As it provides a system to report genomics research and involvement in a standardised way, STARDIT reports can help people make informed decisions about

participation in research, for example, by allowing potential participants to view data about who is involved in data access decisions and any financial ownership of data. Such standardised information about genomics research can also be used by research funders or potential research funders in order to make decisions which align with any organisational values or policies.

STARDIT data can also be used to help assess the methods of genomic analysis, including who was involved in articulating how people with lived experiences have been involved in annotating phenotype data.

Recommendation 1: Report how people with lived experiences have been involved in annotating phenotype data in a standard way

The 2019 scoping review highlighted the importance of being able to show who was involved in labelling phenotypes for genomic variations where a subjective lived experience is important.[28] For example, subjective experiences of depression or dementia can only be articulated by the person experiencing them. The better our understanding of such subjective lived experiences, the more useful such data are when combined with genomic data.

In 2018, in the 'The Power Threat Meaning Framework', the British Psychological Society stated that "we need to take meaning, narrative and subjective experience seriously" in relation to defining, diagnosing and treating mental health.[536] The WHO estimates that 264 million people suffer from depression, with 800,000 people annually committing suicide. [537] Suicide is the second leading cause of death in 15 to 29-year-olds.[537] Similarly conditions such as dementia are recognised by the WHO as global research priorities.[538] Accordingly, there is a new imperative to combine data about the subjective lived experiences of people with more objective measures such as genomic analysis, medical records and data from standardised assessments (such as those used in the ASPREE study).

The methods used in the case studies for this thesis show how participatory action research processes informed by post-positivist and constructivist theoretical frameworks can integrate subjective perspectives into research. Systems such as STARDIT can be used to report who will be or who was involved in such data articulation, and allows people to self-identify with particular groupings or categories. Compared with relying solely on a diagnosis by a healthcare professional or on objective measures, this provides more power in the process of constructing data to describe lived experiences.[196] Involving people in the co-creation and structuring of such data will enable better quality data on phenotypes to be produced.

The findings of this doctoral research support other studies which suggest the need for more opportunities to actively involve people affected by genomic variations in the process of articulating phenotypes.[539] Involving people in reclassifying clinically ascertained variants of uncertain significance (VUS) can "impact risk assessment, medical management, and psychological outcomes for patients and their families."[539] The STARDIT system uses structured data (including standardised taxonomies and ontologies) to facilitate the articulation of common human experiences in multiple languages. It thus enables more people around the world to be involved in annotating phenotype data. In addition, STARDIT can work across cultures and human languages. This means that concepts such as 'depression' can be described in different cultural contexts and different languages, where both factors can significantly affect people's descriptions of these concepts.[540]

Using systems such as STARDIT to report on how people with specific genomic variations self-identify with standardised categorisations (such as a person with dementia or depression) can help ensure there is more machine-readable data about genomic research. In turn, this will help inform future reviews and appraisal of data quality in this field.

Recommendation 2: Report how people have been involved in designing, managing and evaluating research in a standard way

More research is required to appraise methods of co-designing and co-managing biobanks and other genomic research.[28] In coming decades, methods for managing biobanks will need to be constantly reviewed and appraised by all stakeholders to ensure cost effectiveness and accountability. The STARDIT system has been suggested as a way of reporting involvement in biobanks, and such transparent reporting should be urgently explored by all current and proposed biobanks and genomic research.[28,33]

In the Global Alliance for Genomics and Health (GA4GH) 'Framework For Involving And Engaging Participants, Patients and Publics In Genomics Research And Health Implementation' STARDIT was cited as a useful way of 'conducting evaluations of engagement'.[541] In addition, an article supported by the GA4GH, STARDIT was cited as a 'useful' as a way of 'evaluating engagement'.[517]

Recommendation 3: Report research with populations at greater risk of exploitation in a standard way

As described in the section 'Ways of protecting people from exploitation in genomics research , STARDIT can be used to plan, report and evaluate genomics research in order to ensure that research is workable, culturally safe, acceptable and effective. STARDIT should also be used to report on how people from populations at greater risk of exploitation have and will be involved in research,

allowing more people from such populations to be involved in the process, including reporting and evaluating how people were involved.

Implications for health technology assessment

What is health technology assessment?

Health technology can broadly be defined as the pharmaceuticals, devices, procedures and organisational systems used in health care. Health Technology Assessment (HTA) is a process used by governments and health insurers to assess which 'health technology' to pay for in certain populations and circumstances. The process usually considers the medical, organisational, economic and societal consequences of implementing health technologies or interventions within the health system.[542] Assessment processes and the quality of assessments vary by country, however, all require decision making based on the available evidence. As physical resources are always finite, governments worldwide use HTA as part of a 'rationing' system – that is, to decide on and justify allocation of resources to health technologies.

In the context of HTA, the definition of 'evidence' is elusive as assessment of any health technology requires an individual subjective judgement based on the available data. Health Technology Assessment can therefore be understood as the product of systematic observation or experiment.[542] In order to provide practical and useful knowledge, it often relies on data collected and analysed in accordance with a pre-established protocol.[542] As part of HTA processes, it must be agreed what kinds of data and evidence will be integrated, how they will be critically appraised, and if and how different kinds of data will be 'weighted' in the analysis. It is important to note that while HTA processes frequently uses the word 'evidence', the word 'wisdom' is often lacking.[543] As summarised by Menon, health technology policy making "cannot be reduced to a technical exercise, free from values or ethical judgement".[544]

Problems with health technology assessment and genomics research

A number of difficulties inherent in the HTA process have specific relevance for the translation of genomics research into public health genomics and other health interventions. As previously discussed in this thesis, it is difficult to collect and integrate data about subjective lived experience (including data on patient reported experience and outcome measures). This means that evidence is often drawn from more objective data sources, such as data on deaths. Recognition that people with variations of known or unknown significance need to be involved in the HTA process is essential for ensuring that people with lived experience of variations are involved as a distinct stakeholder group. When drafting a public involvement strategy for the UK Health Research Authority in 2013, I

successfully argued that “those with known genetic dispositions” should be categorised distinctly from the general public and patients.[510,545]

There can also be inherent inequity in the application of health technologies to genomics research. In small groupings of people affected by rarer variations, lack of statistical ‘power’ means that ‘gold standard’ evidence will never be available for HTA decision-making processes.[374,543] Notwithstanding, some authors have claimed that most published research findings are false and “flexibility in designs, definitions, outcomes and analytical modes” increases the chance that findings are false.[546] In Australia, nearly 50 per cent of deaths from cancer are from rare cancers, yet a US review of oncological drugs in 2015 suggested that current pricing models reflect “what the market will bear”.[58,547] Ensuring equity in the translation of genomics research into health technologies to treat ‘non-profitable’ rare diseases is a significant challenge.[548]

Financial or other interests can also increase the likelihood that research findings are false. For example, a 2017 Cochrane systematic review found that “sponsorship of drug and device studies by the manufacturing company leads to more favourable efficacy results and conclusions than sponsorship by other sources”.[508] In addition, ‘asset exchanges’ of patient groups between pharmaceutical companies may also occur when lobbying during the HTA process is led by patient groups but funded (often indirectly and not always transparently) by pharmaceutical companies. This creates potential for the most profitable diseases to be the ones approved and funded as a result of HTA processes.[549] Such inherent bias in the research done, the results published, and financial support for lobbying (which includes financial support to give feedback from the patient perspective during complex HTA processes) affects the quality of the evidence available to those making HTA decisions. In the scoping review for this thesis, the novel finding of a method of articulating variation in the perspectives of people affected by rare diseases provides a foundation for developing what may become an increasingly important model for involving these populations in every stage of genomic research, including HTA.[28]

Population screening of people’s DNA using knowledge from genomics research has already been modelled as cost effective in Australia. [550] However, there is also an inequity issue when translating genomics research for population screening for Indigenous peoples. For example, in Australia, Aboriginal peoples are under-represented in current genomics research, so the analysis of genomic data may not be as useful for these populations.[5]

DIY-Bio and health technology

As access to medicine (including genomic medicine) is not universal or affordable, the Do-it-yourself Biology (DIY-Bio) movement needs to be considered when looking at the future of HTA processes.[1] As access to technology to modify human genomes or apply genomic medicine grows, the United Nations is investigating governance challenges.[551] As for any innovation, there are risks and opportunities. Inclusion of the DIY-Bio movement into public health genomics has potential to provide access to genomic medicine for millions, as both the discovery process and creation of tools for genomic medicine are shared as part of this open-source movement. Health technology assessment processes will need to carefully consider questions of quality control and licensed "good manufacturing practice" (GMP)[552] to ensure they remains inclusive and do not only include those who can traditionally afford it, such as large pharmaceutical companies.

The risk of counterfeit, 'off-licence' or poor-quality genomic medicine being used by people who cannot afford access to better regulated interventions is very real. Systems like STARDIT can be used to report on the entire process (including manufacturing), any licenses, any competing interests, and other important data. This can help ensure that HTA processes include genomic medicine and related technology, regardless of whether it is a small community-controlled enterprise or an international company.[451]

Recommendation 4: Use STARDIT to report health technology assessment processes around the world, including evidence assessment processes and how different stakeholders have been involved

Health Technology Assessment processes around the world need to make sure that people are involved using evidence-informed methods that require standardised reporting of data about how people were involved and any impacts. Additionally, data beyond simple health outcomes data (death date) and economic evaluations are required to integrate subjective lived experiences more effectively. Improving data about quality of life and other important outcome measures identified by stakeholders is important. Standardised reporting of data about financial or other interests is also needed alongside other research data – this includes interests such as being personally affected by a disease (as articulated by the ausEE case study participants) and any financial support that patient organisations are receiving.

The STARDIT system allows sharing of standardised data about research methods, analysis, outcomes and multiple stakeholders' interests and allows both humans and machines to appraise the research studies using such meta-data. The STARDIT system also allows reporting of data about

participants involved in research or in the HTA process, including any genomic variations or diseases with an underlying genomic cause. Using STARDIT to plan, report on and evaluate HTA processes would improve evidence-informed decision making worldwide.

Implications for public discussion debate on the concepts of yōushēng (eugenics)

Defining yōushēng and eugenics

The One Health model of public health proposed by veterinarians raises no objections to humans selecting for perceived favourable characteristics in other animals based on DNA analysis. However, when applied to humans most agree that such principles are ethically complex. The comparison of selective breeding for non-human animals and humans is not made lightly. The 'First International Eugenics Congress' took place in London in 1912 and was organised by the British Eugenics Education Society.[553] It was attended by Sir Winston Churchill and presided over by Major Leonard Darwin (son of Charles Darwin). In his opening remarks, Darwin said that the principles of better breeding are known by farmers, and it would require "courage" to apply such principles to humans.[553] He said "might not we hope that the twentieth century will in like manner be known in the future as the century when the eugenic ideal is accepted as part of the creed of civilisation".[553]

In Anglophone countries, the word 'eugenics' comes from the Greek word 'Eu' for 'good' and applies to concepts of perceived genetic quality, reliant entirely on subjective judgement. Because of its association with racist applications, the word in English is highly loaded with negative associations.

Academics divide the concept of 'eugenics' into two categories:[554] 'negative eugenics' and 'positive eugenics' (which it must be noted are not value judgements, but descriptions of the mechanism of 'away from' or 'towards' certain perceived genetic qualities). In summary, positive eugenics describes a method of trying to encourage reproduction between people with perceived positive phenotypes. Negative eugenics describes trying to prevent reproduction between people with perceived negative phenotypes. It is important to note that both positive and negative eugenics can be forced on people, or entered into by choice ('opt-in').

In Mandarin Chinese, the word for the concept of eugenics is 'yōushēng xué' (hereafter shortened to yōushēng). In Mandarin Chinese, the word 'lacks cultural baggage', compared with the English word 'eugenics', which is most often associated with forced eugenics.[555] Recognising the associations of the word 'eugenics' in English, the term 'yōushēng' is used in this discussion. The Mandarin Chinese

term has positive connotation related to ideas about giving birth to children of 'better quality' or having a 'healthy birth'.[555,556(p1)] For example, smoking during pregnancy would not be considered yōushēng.[555] Some contemporary Chinese ethicists have controversially argued that, as members of society, individuals "have a duty to provide society with healthy and normal children".[556(p1)] However, as the Chinese Government has ratified the United Nations Convention on the Rights of Persons with Disabilities, it is obligated to reform laws that conflict with this convention.[557(p1)] Accordingly, clinics licensed to do pre-implantation genetic diagnosis are only permitted to do so to avoid 'serious disease' or as an infertility treatment. Selection for other traits (including gender) is not permitted.[555]

Examples of forced eugenics

An example of negative eugenics occurred in the USA in the 1920s when comparisons were drawn between the social imperative of vaccination and compulsory sterilisation.[558(p293)] This application of negative eugenics by the 'state' purported to protect its 'interests' by sterilisation of those considered to be "afflicted with an hereditary form of insanity or imbecility". [559] This was challenged in the 1927 United States Supreme Court case 'Buck v. Bell', and provides an important perspective from which to view the origins of removal of autonomy in public health genomics.[559] As recently as 1993, the Japanese Government forcibly sterilised citizens for similar reasons, with the 'Eugenics Protection Law' only repealed in 1996.[560] In the United States, California only banned coerced sterilizations of female prisoners in prisons in 2014 (distinct from forced birth control).[561] A 2022 report by the Office of the United Nations High Commissioner for Human Rights in relation to 'Uyghur and other predominantly Muslim ethnic minority communities' published in 2022 cited a 2014 report by the UN Committee on Economic, Social and Cultural Rights,[562] noting reports of 'forced abortion and forced sterilization' of women in China.[563]

Examples of positive eugenics were promoted by organisations such as the British Eugenics Society, with Mary Barton and Bertold Wiesner (biological father of the participants of the Shared Ancestry case study in Chapter 7) among many other researchers publicly supporting the concept in peer-reviewed literature and stating in a 1945 British Medical Journal article that they only took donors from "intelligent stock".[433]

One of the offspring of Wiesner, a participant in the Shared Ancestry study, shared examples of negative eugenics enabled by large technology companies such as IBM, which provided the Nazi regime with technology to carry out negative eugenics policies.454 This person was concerned that this will continue to happen into the future. The Chinese Government's alleged collection of blood samples from 'ethnic' Uighurs (or Uyghurs) – and use of their DNA to map human faces in order to

sort them into 'ethnic' categories – was initially facilitated by the technology company 'Thermo Fisher Scientific'.[564] This alarming use of forced negative eugenics in genomics research has been described by the Canadian and USA governments as 'genocide'[564], with the Office of the United Nations High Commissioner for Human Rights stating 'serious human rights violations have been committed'.[563]

Examples of contemporary 'opt-in' positive eugenics

Prenatal chromosomal diagnostic testing has been available in some countries since the 1950s,[565] and health services around the world now routinely offer parents the chance to test foetuses for conditions such as Down syndrome. Testing methods have become increasingly sophisticated and less invasive (some now only requiring a small blood sample from the pregnant woman), and countries such as Australia are exploring routinely covering the costs of non-invasive pre-natal screening (NIPS) for Down syndrome, Edward syndrome, Patau syndrome and Turner syndrome.[566] In parallel with NIPS, people undergoing in-vitro fertilisation (IVF) can increasingly access pre-implantation genetic diagnosis. Such diagnosis can help prospective parents make informed choices about which embryo they will implant, and these decisions are often based on genomic variations of known significance. However, what is classed as 'significant' is a fundamentally subjective choice, connected with both concepts of yōushēng and eugenics. Who decides who may make such decisions, and how, is central to the discussion of this thesis.

In 2015 the United Nations International Bioethics Committee stated that:

> … germline genetic interventions were the subject of science-fiction novels and scientific theoretical debate, but considered non-executable. That has changed …This new reality calls upon experts, governments and all citizens to consider all the possible consequences on human rights and fundamental freedoms as well as on the future of humanity itself. [56]

At the start of this doctoral research in 2017, editing of the human genome was still an academic discussion. The international "scandal" surrounding the alleged first editing of the human genome in China in 2018 is a clear indication that the subject of positive eugenics and gene editing is no longer theoretical.[567] While parents may 'opt-in' to use such technologies, people who have had their genomes edited cannot 'opt-in', so it can be considered forced positive eugenics. The subject needs urgent public debate, [567] and the UN has hosted recent public discussions to consider the "impact on the values and cultures of each society" of such technologies.[568]

Rapid advances in genomics analysis and falling costs mean that more people will soon have access to DNA analysis. This may allow them to make informed decisions about parenthood, including providing prospective parents and pregnant woman with more data when deciding whether to try for, or terminate, a pregnancy. Many people will seek guidance on these complex decisions from health professionals and genetic counsellors (with associated ethical oversight) or community leaders. Decision-making processes in countries such as Australia that are considering covering the costs of NIPS need to be transparent, including how decisions are made about which variations to test for, who made these decisions and how, and how it is decided who makes these decisions. For example, the views of communities with shared values (for example, communities defined by shared religion or faith or health economists) are not always compatible with the UN's human rights paradigm.

It is also important to note that while a country or culture might affirm individual choice in the matter of terminating a pregnancy, and the choice may appear to be available, in practice choice may be undermined by the social, political or clinical environment.[558(p297)] The 'social model of disability' argues that people are not disabled by impairments but by disabling barriers in societies.[569] For example, the choice of terminating a child with Down syndrome may be influenced by a perceived lack of support from society (including financial and practical support), rather than by eugenics, per se.

For example, people with specific variations in the gene GJB2 often experience what is medically described as severe-profound hearing loss. However, from the perspective of some people affected by the gene, their deafness is a way of life, not a medical problem, and society disables them by not adapting to their needs.[570(p2)] As the genetic causes of deafness are found worldwide, there is a global 'deaf culture', which has a strong sense of identity connected to living with hearing loss.[570(p5)] Using the 'right' language to describe genomic variations, their known effects, and any responses or interventions to them requires in-depth awareness of both people's personal experiences and of any sense of community that emerges when people share specific variations that affect their experience.[571]

Diverse perspectives are also evident in relation to having children with variations which may cause deafness. In order to make informed decisions, some people may wish to know more about inherited variations which may lead to deafness, and may seek information regarding carrier status, pre-implantation screening (IVF) or prenatal genetic testing. This information is important as according to a 2002 survey of over 600 people with hearing loss, some people stated they may

choose to terminate a foetus that *does not* have specific variations that may lead to deafness, while others may prefer to terminate a foetus that *does* have such a variation.[555,572]

While seeking partners based on their DNA variations is an emerging area, a number of dating websites already claim to use DNA to find a 'match'. It is not inconceivable that in the near future, a service may arise which allows people to share DNA variations they consider of significance in order to find partners they consider to be more compatible. For example, people who know they have a recessive variation known to contribute to a condition may seek partners who know they do or do not have such a variation. While this is currently a speculative service, questions of ethical oversight and subjective judgement would need careful debate.

Recommendation 5: Transparently plan and report international debate on yōushēng and eugenics

While the history and future of yōushēng and eugenics and genomics is beyond the scope of this thesis, I recommend that countries around the world urgently need to debate concepts such as yōushēng in order to co-create ethical boundaries. Further, Anglophone countries need to start by considering how concepts of eugenics can be discussed.

Systems like STARDIT can be used to map and report on the preferences and values of multiple stakeholders using standardised data about how any screening or testing decisions are made. Such reporting will allow transparent scrutiny of these processes (and of the values of those designing such processes). This has potential to create the enabling conditions for more people to be involved and to help improve decision-making processes.

Similarly, initiatives to involve the public and other stakeholders in shaping policy, law, educational interventions and ethical oversight of the use of genomic technology can be transparently reported using STARDIT, allowing them to be evaluated. STARDIT could be used to map different stakeholders, their different preferences for involvement and their values. This data can be used to inform the co-design of any planned involvement activities such as: public debate; citizens' juries and forms of public consultation.

The looming shadow of future genomics research being used to reinforce racism and assist with genocide is haunting. Urgent public education, debate and international consensus must be established on these issues and, in particular, must ensure that people at greater risk of exploitation are involved.

Implications for future genomics research beyond humans

As humanity's understanding of genomics (and other '-omics') increases, the interaction between the environment, environmental systems, and other lifeforms (including the gut biome, flora and fauna) is being increasingly explored. This is summarised in the introduction to the 2008 'One Health' report by the American Veterinary Medical Association, which stated:

> "The convergence of people, animals, and our environment has created a new dynamic in which the health of each group is inextricably interconnected." [573]

A notable example of this convergence is the connection between the COVID-19 virus, human-to-animal transmission, and the social and environmental factors that created the enabling conditions for transmission. The importance of interdisciplinary research to both understand and solve such challenges is recognised by the World Health Organization.[574]

Another example of this interconnection in the context of public health genomics was articulated clearly in a 2019 report by Genome British Columbia (Canada). It stated that:

> "our understanding of biological systems at a molecular level is transforming how society approaches solutions to complex challenges. These applications include health, forestry, fisheries, aquaculture, agrifood, energy, mining and the environment".[274]

A short video produced by Genome British Columbia explaining the concept of this interrelation was shared with case study participants in the online discussions as one of the learning resources.[575] The video was shortlisted by the study team from a 'landscape analysis' of education resources conducted by the Australian Genomics Health Alliance.[416]

As humanity is part of Earth's biological systems, there is a clear imperative for human genomics health research (including public health genomics) to be located within this wider context. The STARDIT system offers the only data sharing platform on which interdisciplinary research can be shared in a standardised way, across multiple human languages. The need for such a system of data sharing was reinforced at multiple stages of the STARDIT co-design process, with experts from public health, Johns Hopkins University, Cochrane, genomics research, environmental research and citizen science all involved and publicly supporting STARDIT. Its importance has been further highlighted by the COVID-19 pandemic.

Implications for all research

Those creating and delivering research, and those affected by it, may sometimes be the same people. Methods such as 'citizen science' and 'participatory action research' are blurring the lines between the concepts of 'researcher', 'public', 'patient' and 'citizen'.[28,134,239] Multiple stakeholder involvement (including the public; patients; communities defined by a shared interest; consumers of health services or medical products; payers; industry, and policy makers) is integral to ensuring that all perspectives are valued. The findings of the ASPREE case study supported this by demonstrating that learning involving research staff – not just senior researchers – was an important source of valuable perspectives.[6]

Some participants from the Shared Ancestry and ausEE case studies highlighted their preferences for online discussions, as they were more flexible and accessible and, thus, more inclusive. This has important implications for those planning involvement in future research, in particular, in the context of the COVID-19 pandemic, and the wider global adoption of working online.

The STARDIT system provides a way to report data about research paradigms such as participatory action research processes and, regardless of the discipline, enables comparison of methods for involving people. The adoption of STARDIT by both the Indigenous Precision Medicine project[3] and the Australian Genomics 'Involving Australia' project working group for reporting the initiatives, including planned stakeholder involvement demonstrates usability.[298]

Indigenous peoples around the world connect health and environment as linked and inter-reliant concepts.[576] Beyond the discipline of public health genomics, environmental research projects using DNA have also used STARDIT to report the initiative, including participatory action research processes. For example, two community-led citizen science projects used STARDIT to report the work.[218,516,577] The environmental DNA research project used STARDIT to report the co-design and co-management process, and reported impacts.[217] Data about these initiatives indicated that using STARDIT allows their methods and impacts to be reported across disciplines, regardless of whether it is 'health' or 'environmental' research – helping bridge this conceptual gap in reporting initiatives, in a way that Indigenous peoples (and many non-Indigenous peoples) have done for millennia.

Implications for ethics and participatory action research

The findings of a 2018 scoping review about ethical challenges in community-based participatory research were supported by the findings of this doctoral research.[514] In particular, questions of 'who is an insider or an outsider' and who decides this are central to the participatory action research process. [147,149] This was particularly important in the case study undertaken for this thesis involving communities of shared interest defined by shared ancestry. A quotation from the Shared Ancestry

study summed up this issue succinctly, with one participant asking, “who will decide who will be on the ethics committee?” [P4-SA]. The implications are similar for any genomics research which aims to recruit people from communities defined by ancestry, such as Indigenous peoples. Similarly questions such ‘who is an insider?’, ‘who decides this?’ and ‘what is the process for deciding who makes ethical decisions on behalf of others?’ are all questions which this thesis has demonstrated workable ways of answering, with a repeatable method.

For example, publication of the protocol describing the intended use of co-design to co-create a genomics research protocol for working with Aboriginal Australians took longer than predicted owing to complex ethical review processes. Although some limited guidelines are now emerging[127] from the UK’s National Health Service, internationally there is still confusion surrounding what ethical approval is required before involving potential participants as ‘specialist advisors’ in co-designing research.[578] The Indigenous Precision Medicine project embedded STARDIT reporting in the co-design process, including transparent reporting on how ethical decisions (including data access decisions) are made.[3]

The thesis describes power imbalances in ethics processes which continue to affect genomics research around the world and impact negatively on the implementation of successful participatory action research processes.[579] While answering these questions will be complex and require ongoing consideration, it is clear from the case studies conducted for this doctoral research that the current situation is no longer sustainable. Arguably, current ethics processes lack accountability in some respects and are thus unethical. In addition, the complex ethical processes were perceived as inaccessible by researchers and Aboriginal community members (as described in Chapter 8). Ethical processes which have been established to protect Aboriginal peoples risk themselves perpetuating and amplifying the gap in life expectancy by creating barriers to doing research with Aboriginal peoples.[5] This potential form of structural violence needs urgent consideration, and appropriate resourcing to ensure people can be effectively involved in ethical processes (see Recommendation 6).[580]

To ensure that power is shared effectively at all stages, these problems and barriers must be co-defined, and solutions to them co-created and co-evaluated. The STARDIT-PM tool provides a way to report the preferences of all stakeholders regarding ethical oversight, and STARDIT allows such data to be reported in a standardised way. Potential research participants are able to add data to STARDIT reports, thus removing the exclusive power of researchers, who traditionally are the only stakeholders report research in peer-reviewed publishing models and publicly accessible sources.

While traditional ethics processes have been set up with the best of intentions, new methods are emerging which give people direct control over their own data in a very real way. An example is the CTRL model created by researchers working with the Australian Genomics Health Alliance.[75] This model allows people:

> to keep personal and contact details up to date; make consent choices (including indicate preferences for return of results and future research use of biological samples, genomic and health data); follow their progress through the study; complete surveys, contact the researchers and access study news and information.[75]

Such tools offer an insight into future best practice. Among these, STARDIT offers a way to both report on the process and evaluate any impacts.

Similarly, in the area of free market 'consumer' genomic data, where data are commodified for sale, blockchain technologies are giving people practical ways to be custodians and managers of their own data. Initiatives such as 'Genecoin' and 'Encrypgen' allow individuals to have control over their genomic data and gain financial rewards if they grant access to their data for research analysis.[581,582] In contrast, people sharing genomic data with direct-to-consumer companies such as 'Ancestry DNA' and '23andMe' simply 'give away' their data as an implicit trade-off for access to services.[581–583] In addition, such companies often provide limited options regarding research participation, with some making data sharing with third-parties mandatory in order to use the service.[50,583]

To ensure initiatives have ethical oversight and protect people from exploitation, new research models are needed. Ensuring that all stakeholders have an opportunity to influence how initiatives are developed and managed (including data access decisions) will require the input of multiple experts and the implementation of complex co-design processes. STARDIT offers a way to plan, report and evaluate research processes in a standardised way. Such reporting is the first step in sharing power in genomics research in evidence-informed ways. This process will require continual evolution, including machine learning, to keep pace with the ethical challenges presented by genomics research.

Implications for genomics research policy

Organisations such as the Global Alliance for Genomics and Health which sets international standards for genomics research should urgently support standardised reporting of involvement in genomics research. Data from standardised reporting will support evidence-informed policy in this

area in which public acceptability is central to ensuring the continued expansion of genomics into public health policy. Learning from the scoping review and best-practice examples – such as the UK Biobank and Genomics England – indicate the importance of ensuring long-term funding for such involvement. This includes funding to support stakeholder (including participant) involvement in tasks such as data oversight.[28] Ongoing reporting and evaluation of such funding will allow policy makers to make evidence-informed decisions about the kinds of involvement methods, modes and tasks that are most cost-effective for payers, including tax payers and industry.

Implications for policy relating to all research

The processes described in this thesis have demonstrated a consistent way of reporting data about preferences for involvement, as well as standardised ways of co-designing and reporting on involvement in research. This has allowed comparison of preferences about involvement and a way of reporting and comparing the impacts of involvement. Policy and research funding criteria must begin to require standardised reporting of involvement so that initiatives can be evaluated in order to inform future research. The STARDIT system provides a way for initiatives to share data about involvement in research and to report impacts. This will invite scrutiny, promote transparent oversight of research and support evidence-informed policy.

Recommendation 6: Provide resources for inclusive and accessible involvement

Ensuring people can be involved in inclusive and accessible ways requires appropriate resourcing to supporting people to be involved. This can include (but is not limited to) providing practical support (transport or equipment), financial support (paying people fairly), providing learning and development opportunities (including costs for translating to multiple languages), providing emotional or mental health support, or investing in infrastructure.[584]

The ‘Human rights’ section of this thesis explored the many statements in support of involving people, and how the concept of democracy is inherently interconnected with respect for all human rights and that “fundamental freedoms are interdependent and mutually reinforcing”.[56] In health care, the “right and duty to participate individually and collectively in the planning and implementation” of health care is enshrined within the World Health Organisation’s ‘Declaration of Alma-Ata”.[19] Similarly, the United Nations has provided much guidance on working with Indigenous peoples around the world,[99] and the 'Declaration on the Rights of Indigenous Peoples United Nations' states that “Indigenous peoples have the right to be actively involved in developing and determining health, housing and other economic and social programmes affecting them”. However, without resourcing from these same organisations, these statements risk remaining just as

statements, impossible to enact in inclusive and accessible ways. Not supporting people to be involved in health care and research can therefore be viewed as a form of structural violence, and in some cases, a manifestation of systemic racism.[469,580]

Inaction in appropriately resourcing involvement activities risks not only human rights, lives and wellbeing – it risks life on Earth itself. The section 'Implications for future genomics research beyond humans' noted the interrelation of other life and the 'One Health' model,[574] but beyond genomics and human health, policies must address the largest of challenges – which is understanding our planetary ecosystems, and attempting to reduce the damage we are knowingly doing. As noted in the STARDIT Beta version article,[585] "many problems facing humans are shared by non-human life forms and ecosystems, including rapid climate change, air pollution and sea-level rise". The concept of 'planetary management' needs urgent examination in policy,[586] and ensuring that people are supported to play an active role in understanding the challenges and creating solutions requires appropriate resourcing. Some of the challenges of 'speaking the same language' and working across disciplines can be met by using STARDIT, as can reporting the impacts of any initiative and describing any participatory methods in a consistent way.[586]

The UNESCO Recommendation on Open Science offers a promising tool to 'strengthen international cooperation on open science for reducing the existing inequalities in science, technology and innovation', but such recommendations need to co-exist with commitments to resourcing (specifically funding for implementation and evaluation) in order to have impact.[237] Without appropriate funding, these statements cannot be meaningfully enacted.[109,584]

Not providing adequate resourcing (in the form of funding initiatives, providing appropriate finance, paying people, providing education or investing in infrastructure) limits how people can be involved and reduces the ways that people can get involved in shaping our shared future.[109,584]

Policy makers must demonstrate the shared values of those they represent through real action, by providing resources which ensure everyone can be involved.

Chapter 11 – Conclusions

In this thesis, I have demonstrated that there is a global imperative to improve involvement in genomics research, but there remains a lack of data to support evidence-informed policy in this area. I have led the design of several research projects with communities of people affected by genomic research and conducted case studies that explore and evaluate various methodologies for involving people in genomics research.

Learning from these case studies, and the cross-case analysis, can be applied in different settings and with different population groups. These include populations at greater risk of exploitation, people affected by rare diseases, people who are of high-interest to researchers (such as large populations of donor-conceived siblings) and Indigenous sub-populations around the world. To facilitate involvement and data collection, I have also co-created methods of using a real-world online discussion platform and used it to involve people in every stage of the research cycle.

My research has emphasised the importance of genomics research working globally across populations and human languages. Findings from the scoping review have shown that to date reporting of methods, including how stakeholders were involved in genomics research, has been both under-reported and of variable quality.

In response, I have conceived and led an international co-creation process for a standardised reporting system (STARDIT) in partnership with people from multiple organisations including Cochrane, the Wikimedia Foundation and the Poche Centre for Indigenous Health and Johns Hopkins University. The STARDIT system provides a way to share data about involvement in genomics research and a system for reporting data about genomics research. It enables those planning genomics research to make evidence-informed decisions based on any data reported. In addition, STARDIT can be used to report on how people affected by diseases such as depression or dementia have been involved in research. It allows sharing of subjective lived experience of conditions and diseases which can improve the articulation of phenotype data.

The STARDIT system can be used across human languages and cultures to co-create structured, machine-readable data which uses standardised taxonomy and ontology. The framework can also be used across other disciplines. This opportunity arises from the interdisciplinary nature of genomics research and the learning made possible by sharing information among disciplines using participatory action research processes and citizen science. As a multi-disciplinary system, STARDIT has relevance to describing initiatives in the context of global responses to urgent challenges facing

humanity such as the COVID-19 pandemic and rapid climate change. The usability and relevance of STARDIT has been demonstrated by the use of the Beta version reporting tool by the Australian Genomics working group 'Involve Australia' in 2021, and by Cochrane for reporting the co-creation process of their 'Values Statement'.[296]

In this doctoral research, I have used the participatory action research paradigm to guide the research process – including to co-define problems, co-create solutions and co-evaluate them – and to demonstrate practical ways of involving people in every stage of genomics research. Some fundamental ethical questions about power sharing have been uncovered during this research process, including the difficulties of achieving a true participatory model in an academic context where university ethics committees hold power in ways that can be incompatible with the paradigm of participatory action research.

Recognising the limits of an academic research setting for the participatory action research method, I established and run a charity (Science for All) and have volunteered with organisations (including the Wikimedia Foundation and Cochrane) and worked for the Australian Department of Health in health technology assessment. This gave me the opportunity to learn and apply alternative methods of co-design and research management and to evaluate different approaches to participatory action research in research. Focusing on involving people in genomics research and standardising data sharing allowed me to combine learning from these projects with others around the world. This included learning about organisational governance, health technology assessment, collective decision making, power sharing, data sharing and co-evaluation processes. Working within these organisations provided the necessary foundations for learning about and applying the participatory action research paradigm, which is not always possible within academia.

The most difficult work lies ahead, that is, translating the learning from this doctoral research into practice and continuing to ensure that data sharing about involvement in genomics research is standardised. Once achieved, humanity can begin to develop evidence-informed ways of sharing power in research that can be applied and evaluated by anyone, anywhere.

Afterword

> "I realized that the difference that I saw between things was the same thing as their unity, because differences (borders, lines, surfaces, boundaries) don't really divide things from each other at all, they join them together, because all boundaries are held in common"[587]
>
> Alan Watts

It has been an enormous privilege to complete this doctoral research. Along the way I've discovered new family from DNA tests, had the meaning of what 'family' means challenged, and involved them in research. I've spoken about my experience of losing my girlfriend to Sudden Adult Death Syndrome (SADS) to the Australian Department of Health in order to influence decisions on providing free genetic testing. I've worked with Aboriginal people in remote communities. I've learned that people from these communities have a deep understanding of the genetic causes of SADS. Working with them, we co-designed genomics research and explored what it means to have 'shared ancestry' and 'shared culture'. I've worked as part of a world-leading team, designing a multi-generational study, and I've worked with people affected by rare diseases and heard their concerns about data sharing. Every experience has taught me something, and I've valued each one.

As I complete this doctoral research, it has reminded me why I have this passion for involving people in genomics research, which is effectively involving people in describing themselves. Because, of course, essentially we *are* all of these subjective lived experiences when it comes to mental health or any other part of our experience of being alive. Only we can describe this 'phenotype'.

I think my upbringing in western Europe and those traditions has made me come from an individualistic way of thinking about the human experience itself, which is often defined by the concept of the 'self' and the 'I'. However, this 'I' is challenged by genomics. Just as with issues like air pollution, extinction, or eco-system collapses – genomics research unites all of us because it affects all of us. Although we might think of genomics and DNA as essentially 'us' or 'me', it is of course shared with all life on Earth. The only way to make sense of it is to put it all in together and analyse it. This is where the importance in involving people in analysing this data is central. The potential for improving lives and ecosystems that genomics has is profound, but so also are the potential harms.

Without involvement and power sharing, we get reinforcements of the social constructs of the dominant cultures, we get racist AI algorithms, we get linguistic divisions of populations based essentially on skin tone and a bit of geography. We get analysis that doesn't tell the whole picture of genomics, it doesn't include lived-experience, because often, it doesn't ask to see it.

Part of my PhD included online discussions with my half-aunts and uncles, some of whom identified as culturally Jewish and had Jewish ancestry. In a free and open discussion many of them mentioned events of the Second World War and the murder of certain minorities based on their perceived ancestry. They showed a genuine concern that genomics research is already being used to do this, and will continue to be used for this again into the future.

We know from human history that the only way to protect us from this, is recognising that we're actually protecting 'us' from 'us', there is no 'this'. That's really what science education is. One of the original authors of what became the Universal Declaration of Human Rights was H.G Wells, who wrote "human history becomes more and more a race between education and catastrophe". [588] We need to learn how to save us from ourselves. We need to start to redefine 'ourselves' as meaning everyone and everything. While still in the early stages, I sincerely hope that STARDIT can be used to help everyone in the world share trusted knowledge, and provide the evidence we need on the most effective ways of saving us from ourselves, helping share the benefits of human knowledge with all life on earth.

Jack Nunn, 16th March 2021, Melbourne

Full Acknowledgements

Formal acknowledgements

This thesis was written on Wurundjeri land, in the State of Victoria, Australia.

I acknowledge and respect the traditional owners of this land. I also acknowledge that sovereignty was never ceded by Aboriginal peoples in Australia, and that this land was not peacefully settled.

I also acknowledge the United Nations 'Declaration on the Rights of Indigenous Peoples' and acknowledge and respect the knowledge and cultures of all Indigenous peoples, leaders and Elders, past, present and future.

This doctoral research was supported by a La Trobe University Postgraduate Research Scholarship, which commenced on the 5th September 2016. Thank you to all the staff at La Trobe University who gave their support.

My family

My wife Alicia has been by my side from the start of my dream of doing a PhD to the final drafts. Since starting a PhD in 2016, we've moved house two times, bought a house, got married and brought our baby daughter Isla into this world in May 2021. Throughout all of this Alicia has given more than her fair share of attention to these grand chapters of our life while my thoughts have been in the chapters of this thesis. Thank you Alicia for your patience, support and love.

Thank you to my parents who've supported me throughout, and listened to endless hours of ideas and worries. Thank you also to my Mum for being so open to going on the adventure of finding new family and for being a very special part of this PhD. Thank you also to my parents and Alicia's mum for their financial support. Doing a PhD is not lucrative at the best of times, never mind during a pandemic. This support transformed the experience from what would otherwise have been financial hardship.

Thank you also to my "new" family of half aunts, uncles and cousins who it has been a pleasure to meet, and will likely continue to meet for the rest of my life! Thank you for enriching my life and thus this thesis.

A final thank you to the fluffiest member the family Mollie, for her sweet company during what would otherwise have been many lonely hours. And for reminding me to take breaks for belly rubs, walkies and treats.

My Supervisors

It's not an exaggeration to say that I would not have completed my PhD if it was not for Associate Professor Paul Lacaze[4]. From our first meeting in 2016 I could tell he knew exactly what I was talking about when I spoke of involving people in genomics, and more than that, he valued it. At every twist and turn he's been there with practical support, advice and patience. He's read through endless drafts and made possible my work with the ASPREE team, which was a dream come true for me. He was there for me when other supervisors no longer were, and kept me going at my lowest of points, welcoming me into his team. I will always be grateful for his support and friendship.

[4] External co-supervisor 2017-2021 https://orcid.org/0000-0002-0902-6798

From day one of becoming my Principal Supervisor, Professor Vijaya Sundararajan[5] has been supportive, kind and interested in my ideas. More importantly, she has taken the time to think about the wider implications of my research, and encouraged me to look to the future, believing in me when I myself was not always convinced. Our interesting conversations have shaped my thinking and helped enrichen this thesis. For all of this, I am very grateful.

Professor Stephen Kent[6] has encouraged me throughout his time as my supervisor. At our first meeting he asked me why I wanted to complete this PhD, and I think I talked solidly for half an hour. He listened patiently, and gave some helpful feedback; and this has been the pattern ever since. He has made the time to engage with my ideas, challenge me in helpful ways, and even pay me the compliment that some of my thinking had helped influence his own. He has also helped me navigate what is sometimes the daunting path of university administration, helping advise during a series of very complicated administrative tasks. All of this support was given generously and with kindness, and I'm grateful.

Thank you also to Dr Rebecca Ryan[7] for her supervision at the early stages of my PhD, helping shape my initial ideas at the early stages of the research. I would also like to thank Associate Professor Sophie Hill[8] for her support with shaping my scholarship application, and her supervision. Finally, thank you to my Research Progress Panel Chair Dr Rwth Stuckey.

My Co-Authors

During this PhD I was fortunate to have two co-authors who supported me almost as informal supervisors, Dr Kylie Gwynne and Dr Marilyn Crawshaw, who both generously supported me to gain experience in new areas of research, generously giving hours and hours of their time.

I was introduced to Kylie through the ausEE work, and I quickly became aware of her professional experience of working with Indigenous peoples and co-design. When we met for the first time, our shared passion for genuine participatory methods was clear. Kylie invited me to work with the Poche Centre, and on top of working together on the ausEE project, also guided and supported my work on the Indigenous Precision Medicine project. She was there for me when times were hard, and always at the end of a phone for advice and guidance. I'm proud to call her a colleague and a friend.

Marilyn was the go-to person for research with donor-conceived people, having been working in the area for over 30 years, and I was thrilled when she accepted our invitation to work together. We got on well from the start, and her eye for detail was invaluable at multiple points of the research. I would also like to thank her for reminding me of the importance of incorporating subjectivity in research, not apologising for it, or trying to remove it. Writing an acknowledgement only has value if it also acknowledges difficulty. Marylin was an enormous support when, as co-investigators on the Shared Ancestry project, we found ourselves in a complex, difficult situation which impacted on my doctoral research as a whole (which this is not the place to go into). It was two weeks before my wedding, and left me without access to my research data for two of my projects for the best part of a year, which impacted significantly on the time it took to conduct this research. While the ethical administration was navigated, Marylin was a rock. Throughout this extremely upsetting period, she reminded me of the importance of the work we were doing and to keep on persisting, when the easiest thing to do was give up and move on. Paul and Kylie (and countless family and friends) too kept my confidence buoyed at what was one of the lowest points of my entire life. I will always be grateful to them all.

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[8] Principal Supervisor 2016-2018 https://orcid.org/0000-0002-1715-5338

For the scoping review and ASPREE project, in addition to Paul, thank you to Jane Tiller, Merrin Sulovski, Bruce Holloway, Peter Fransquet and the rest of the ASPREE team for your lively discussion and support. Feeling part of a warm and friendly team was invaluable, and I really missed not seeing everyone regularly during the various and long COVID lockdowns in Melbourne. Thank you also to all the ASPREE participants who gave so generously of their time in telephone interviews, events and in other countless ways.

For the ausEE project, I would like to thank Sarah Gray for giving her time so generously, and for her thoughtful and insightful feedback throughout the project. Thank you also to everyone else at ausEE, and to all of the people who participated and supported the project.

While there are too many people to thank for the STARDIT project I would like to thank Thomas Shafee for listening patiently to my initial idea for STARDIT back in 2018 while we drank tea and ate melting chocolate in a 38°C meeting room. Thomas patiently listened to my ideas and helped me build it into the Wikipedia Journals and other Wikimedia projects over the years. Thank you also to Steven Chang for his support with STARDIT at an early stage, helping me broaden my thinking from just ‘research’ and into the ‘arts’. Thank you also too to Richard Stephens, Dr Jim Elliot, Professor Sandy Oliver, Simon Denegri and Carolyn Thompson for listening to my early ideas and assuring me I wasn't insane, when I myself was not convinced. Thank also to attendees of the London event, who are thanked in full in the STARDIT consultation report.[284]

La Trobe University

First of all, thank you to all the staff who have supported me at La Trobe University. This PhD would not have been possible without the La Trobe University Postgraduate Research Scholarship, which was kindly extended owing to the inability to access my research data from two studies, owing to the aforementioned difficulties.

I would also like to thank the staff at the various student support services and disability support services. Always chronically under-funded by any university, these teams rely on the dedication of the staff. Those at La Trobe University truly were there when I needed them. Doing a PhD is challenging at the best of times. As a ‘neuro diverse’ person (often labelled as "dyslexic" by experts) there are some things which I find very hard, in particular navigating administrative processes. The support teams provided me with the practical support I needed. There are too many to thank, but in particular I would like to thank my Disability Advisor Alex Auletta who helped ensure I was given the support I needed during this doctoral research, including organising a budget for editing and proof-reading. Thank you also to Lynda Chapple, in particular for her co-ordination of a writing circle and her feedback on an early version of a plain English summary of STARDIT.

Thank you also to the Graduate Research School's Research Education and Development (RED) team – their friendly support kept me going, along with the writing retreats (and giant lamingtons) and other excellent events and services offered by this fantastic team. In particular thank you to James Burford for his empathy and advice, and Jeanette Fyffe for her passionate support of so many students.

Thank you also to my colleagues and friends at the Centre for Health Communication and Participation, where I started my PhD journey at La Trobe. Thank you to Dr Jessica Kaufman for being the best office buddy you could have, giving great advice over a cup of tea that I will always be grateful for. Thank you also to Dr Dianne Lowe for her continued support and friendship. And thank you to Anne Parkhill for her engaging conversations and for showing me of the importance of having a good librarian when conducting a review. Thank you also to Dr Megan Prictor, Dr Bronwen Merner, Anneliese Synnot, Dr Louisa Walsh and Kowlin Leong for their support and friendship over the years.

Thank you also to the La Trobe University Student Union, La Trobe University Library and everyone else who supported me along the way.

Partner organisations

Thank you for the support of everyone at the Poche Centre for Indigenous Health and the communities they work with. There are too many people to thank individually, but I would like to add a personal thank you to Boe Rambaldini (Director, Poche Centre for Indigenous Health) for making me so welcome within the team, on behalf of the whole organisation.

Thank you to everyone at Monash University and the staff at the ASPREE study for making me so welcome, and giving me an additional inspiring place to work, think and talk at the Alfred Hospital.

Thank you also to the staff and volunteers at the Wikimedia Foundation and the Wiki Journals, EPPI-Centre, University College London, National Institute for Health Research, Wellcome Sanger Institute and Health Research Authority UK.

A final thank you to all my amazing colleagues at Science for All, including Nyssa Braid who stepped in as Assistant Director, letting me take parental leave and finish my PhD knowing that Science for All was in safe hands. Thank you to everyone else on the Steering Committee for your support – including Emma Keith, Michael Johannesen, Saloni Swaminathan, Thijs van Vlijmen, Alicia Dimovski and Anusha Budehal.

Editing and proof reading

Thank you to Pauline Sanders for her professional and thorough editing services for the STARDIT Beta article, and chapters 1-3 and 9-11. Thank you to Pauline Stobbs for her professional services editing parts of the STARDIT Alpha article.

Thank you also to Robert Smeaton for his professional services as a proof-reader, and his flexibility in completing this work.

Other people

Thank you to Dr Anna Middleton for her advice in guiding my thinking during this doctoral research, Dr Kat Arney for her sensitive and professional treatment of my story and my research on the Genetics Society Podcast, Sanjay Thakrar for his helpful correspondence and meetings, Sobia Raza for her discussions on concepts of solidarity, and the many other people who made the time to talk with me over the years about ideas relating to this thesis.

Thank you to Sir Iain Chalmers and Sally Crowe – both of whom offered their support through friendship, professional encouragement and by helping me develop my ideas at many stages. Thank you to Dr Simon J Egan for his support and friendship on our walks along Kororoit Creek.

Thank you also to my good friend Dr Thomas Faust for his moral support throughout this PhD. Thank you to all my other friends and family who have also supported me through this PhD.

This doctoral research was inspired by the memory of Hannah McDonogh (1986 – 2007).

The reader

Thank you to anyone who has read this. I welcome your thoughts and comments and hope to work with many people over the years who share the dreams of peace, wellbeing, equity and balance outlined in this thesis.

Statement acknowledging any real or perceived competing and conflicting interests

I, Jack Nunn, am the sole author of this thesis, except where otherwise stated (such as in the chapters which have been peer-reviewed and published). I was paid a La Trobe University Postgraduate Research Scholarship for most of the time I spent on this thesis (scholarship payments ceased in October 2020, the thesis was submitted in December 2021).

I was paid by La Trobe University as a casual staff member for work on some projects during my PhD candidature, including work with the Australian Department of Health up until 2018.

While completing this thesis, I founded the charity 'Science for All', which is registered in Australia (ABN: 37636063351 ACN: 636063351), and under the auspices of the Royal Society of Victoria. I volunteer my time as Director of Science for All. On occasion, Science for All pays people for consultancy work, including myself, Jack Nunn. All work is approved and voted on by the Steering Committee in a transparent process, and I abstain from any decisions relating to payment of myself. These processes are all overseen by the Royal Society of Victoria, with all financial data reported to the Australian Charities and Not-for-profits Commission. The Science for All 'Ways of Working' document codifies Science for All's ways of working,[589] and the Values statement further articulates the values of the charity.[590]

Paid work for Science for All completed by myself during my thesis included:

- Work on a project for the Victoria Comprehensive Cancer Centre to scope how people affected by cancer and the wider public can be involved in personalised cancer care
- Work on the 'Wild DNA' project and other environmental and community projects funded by the Victorian Department of Environment, Land, Water and Planning and Brimbank Council
- Webinars and presentations for a number of organisations (all not-for profit or Universities)

Pro bono work during my time as a PhD candidate included:

- Volunteering as a member of the Cochrane Consumer Executive, and a Cochrane Council member
- Volunteering as a co-chair for the UNESCO Community of Practice for Citizen Science and Open Science
- Numerous presentations, webinars and meetings (all for not-for-profit organisations, or Universities)

My work on the STARDIT project was supported by the charity Science for All, with myself as an individual donating my pro-bono time to the project, and Science for All providing administrative and secretariat support (for example, hosting online text-based discussions about STARDIT). I received no direct payment for my work on the STARDIT project, although I acknowledge that work on this project, and associated work on this thesis may result in future paid work. The STARDIT report about the co-creation process for the STARDIT Beta version has a more detailed statement on declaring interests.[534] I note that my learning from implementing STARDIT in different contexts enrichened my understanding of the potential applications, challenges and ways of overcoming them in the various contexts I have used STARDIT, including enrichening the content of this thesis.

In addition to my pro bono and paid work for Science for All, I was appointed to the Medical Services Advisory Committee Evaluation Sub-committee (MSAC-ESC) by the Australian Government Minister for Health.[522]

My ORCID record provides a more extensive record of my professional activities during this time,[591] and all payments to myself from Science for All are reported transparently by the charity, including as part of the annual reporting for the Australian Charities and Not-for-profits Commission.

List of Tables

List of Figures

Appendices

These appendices are structured in the same way as the main thesis. For example, appendices relevant to 'Chapter 3 – Methodological overview' are organised under the section 'Appendices for Chapter 3 – Methodological overview' section. The table below describe which part of the thesis each appendices relates to.

Please note – where appendices are available online, tables numbers and figures are consistent with online versions.

Summary of appendices

Related Chapter	Appendices title	Location
Chapter 3 – Methodological overview	How are the public involved in health research and what are the impacts? A narrative review	Presented in full in this appendices
	Standardised Data on Initiatives (STARDIT) Public consultation report: September 2019 to May 2021	Presented in full in this appendices, available at this URL: https://doi.org/10.26181/611dfcf12c6a9
	Standardised Data on Initiatives (STARDIT) Beta Version: Additional File 1	Available at this URL: https://doi.org/10.1186/s40900-022-00363-9
Chapter 4 - Public Involvement in Global Genomics Research: A Scoping Review	Additional file 1: Public Involvement in Global Genomics Research: A Scoping Review	A direct link to Additional File 1 can be found here: https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_1_xlsx/octet-stream/Table%201.XLSX/1/446268
	Additional file 2: Systematically searching sites for 'public involvement' and related concepts	Presented in full in this appendices, available at this URL: https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_2_docx/octet-stream/Table%202.docx/4/446268
Chapter 5 - Additional files: Involving elderly research participants in the co-design of a future multi-	Additional file 1: Involving elderly research participants in the co-design of a future multi-generational cohort study	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00271-4
	Additional File 2: Telephone Questionnaire: Version Comparison - A comparison	This file is available at this URL: https://doi.org/10.1186/s40900-021-00271-4

Related Chapter	Appendices title	Location
generational cohort study	of the versions of the telephone questionnaire used	This is a direct link to the file: https://static-content.springer.com/esm/art%3A10.1186%2Fs40900-021-00271-4/MediaObjects/40900_2021_271_MOESM2_ESM.xlsx
	Additional File 3: Quantitative results - A summary of the quantitative results	This file is available at this URL: https://doi.org/10.1186/s40900-021-00271-4 This is a direct link to the file: https://static-content.springer.com/esm/art%3A10.1186%2Fs40900-021-00271-4/MediaObjects/40900_2021_271_MOESM3_ESM.xlsx
	Additional File 4 - GRIPP2 report for 'Involving elderly research participants in the co-design of a future multi-generational cohort study'	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00271-4
	Additional File 5 - Standardised Data on Initiatives (STARDIT) report: 'Involving elderly research participants in the co-design of a future multi-generational cohort study'	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00271-4 An Alpha version of this STARDIT report is also hosted online in machine readable format: https://www.wikidata.org/wiki/Q98539361
Chapter 6 - Involving people affected by a rare genetic disorder in shaping future research	Additional file 1: Involving people affected by a rare genetic disorder in shaping future research	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00256-3
	Additional File 2: Standardised Data on Initiatives (STARDIT) report – Alpha Version: Involving people affected by a rare condition in shaping future genomic research	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00256-3 A machine readable structured data version of this report can be found here: https://www.wikidata.org/wiki/Q100403236

Related Chapter	Appendices title	Location
	Additional File 3: GRIPP2 report for 'Involving people affected by a rare condition in shaping future genomic research'	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00256-3
Chapter 7 - Co-designing genomics research with a large group of donor-conceived siblings	Additional file 1: Co-designing genomics research with a large group of donor-conceived siblings	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00325-7
	Additional File 2: Standardised Data on Initiatives (STARDIT) report – Alpha Version: Co-designing genomics research with a large group of donor-conceived siblings	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00325-7 A machine readable version of this report can be found here: https://www.wikidata.org/wiki/Q108618394 The 'living' STARDIT Beta version[296] report which relates to this project can be found in the references[587]
	Additional File 3 - GRIPP2 report: Co-designing genomics research with a large group of donor-conceived siblings	Presented in full in this appendices, available at this URL: https://doi.org/10.1186/s40900-021-00325-7
Chapter 8: Involving Australian Indigenous peoples in precision medicine	Additional file 1: Prospective STARDIT Report: A Pathway to precision medicine for Aboriginal Australians	Presented in full in this appendices. The 'living' STARDIT Beta version[588] report which relates to this project can be found in the references[89]. A machine readable version of this report can be found here: https://www.wikidata.org/wiki/Q113417061
Chapter 9 – Results: Comparison of all case studies	Detailed baseline and follow-up data on preferences for involvement	Presented in full in this appendices
Chapter 10 – Discussion	Strengths and limitations of methods used – detailed analysis	Presented in full in this appendices

Appendices for Chapter 3 – Methodological overview

Reviews

How are the public involved in health research and what are the impacts? A narrative review

This section contains the narrative review 'How are the public involved in health research and what are the impacts? A narrative review', which informed aspects of this thesis, including the use of the participatory action research methodology.

Abstract

Aim

We wanted to understand how the public are involved shaping health research and summarize the established methods of involving the public that might be applicable to genomics research. By looking at any impacts that this involvement might have had, we wanted to identify possible methods and approaches that may inform our future plans for involvement and impact assessment.

Method

A systematic search of systematic reviews relating to public involvement in research was conducted using boolean operators in CINAL, Medline and Google Scholar. Data extracted from the reviews included what kind of involvement was taking place (the type), how it was done (the method), which stage of the research cycle it occurred and any impact on the research that might have occurred.

Results

This systematic narrative review summarises five systematic reviews identified in this area. Involvement was reported at each stage of research with mostly positive impacts reported at each stage. Involvement in data collection and analysis were the most reported stages of involvement. Agenda setting was the most frequently reported stage of the research cycle for involvement. Most of the methods of involvement described specified a method involving people in a group structure, rather than individually. A total of 27 different methods of involvement were extracted, with community based participatory research having the most impacts. Other than generic positive impacts, the most impacts reported were improving the relevance of the research, improving dissemination and improving data collection. The stages of research with the most reported impacts were 'agenda setting' and 'data collection', with dissemination the next highest.

Conclusion

Involving people in health research generally has advantageous outcomes for research and health service planning. Involving people as early as possible is considered best-practice. There are a number of high impact actions which can be taken which do not necessarily require considerable budgets or time. Face to face involvement appears to have an increased cost in relation to impact, compared to other forms of involvement, including involving people using online methods. Involving people in planning how the public will be involved is crucial. While there are limitations in the data available, it is clear that involving the people at any stage of research has value, with impact at all stages being possible if people are involved early in the research cycle. More work is needed to better document the impacts of involvement in research.

Summary of narrative review

Objective

To summarise methods of public involvement in health research and any impacts these methods may have had, informing their applicability to genomics research.

Background

The is a growing body of literature about the methods of how to involve the public in health research, with a number of systematic reviews which summarise the methods and or the impacts of involvement in research. However, many reviews conclude there is limited data about involvement or low evidence of impact and are sometimes unable to draw strong conclusions.
This narrative review attempts to summarise systematic reviews in this area to identify established methods for involving people and their and assessable impacts, to inform future plans for applying involvement and impact assessment strategies to genomics.

Methods

Search method

A systematic search of systematic reviews relating to public involvement in research was conducted.

Search terms

A systematic search of systematic reviews relating to public involvement in research was conducted. The boolean operators for the searches were as follows:

1. (Community+OR+Public+OR+Consumer*+OR+Patient*+OR+Carer*+OR+Volunteer*+OR+Advocate+OR+(Civil+society)+OR+user*+OR+Group*+OR+Citizen*+OR+Lay+OR+Population*+OR+Proband+OR+Residents+OR+Stakeholder*+OR+Carrier*+OR+client*+OR+survivor*+OR+participant*)

+AND+

2. (involv*+OR+engag*+OR+participat*+OR+consult*+OR+collab*+OR+contrib*+OR+(advisory+group)+OR+co-creat*+OR+co-produc*+OR+democra*)

+AND+

3. research

+AND+

4. (scien*+OR+biomedic*+OR+health+OR+clinical+OR+trial*)

The search on CINAL returned 3,669 results. An identical search was carried out using MEDLINE and Google Scholar.

Exclusion criteria

Reviews which examined specific areas of research or specific populations were excluded. Similarly, reviews which excluded countries were excluded.

Data extraction

Data extracted from the reviews included what kind of involvement was taking place (the method), which stage of the research cycle it occurred and any impact on the research that might have occurred.
Where possible, data was extracted in order to preserve any causal relationships. For example, if a review stated that 'method X was used at stage 1, with the impact being Y', this was extracted as one line of data. In some cases, there were no causal relationships to extract, so most analysis relies on the ratio that certain methods and impacts were reported. Text was initially copied verbatim, and was then coded into categories, using existing frameworks where possible. Impacts on the people involved and the researchers were also extracted but have not been included in this review. Similarly, data on barriers and facilitators of involvement has been collected but not included in this analysis.

Results

In total, 5 systematic reviews matched the search criteria. These are summarised in Table 1.

Table 1: Included Systematic reviews

Title	Date	DOI
Patient and service user engagement in research: a systematic review and synthesized framework	2013	https://doi.org/10.1111/hex.12090
Methods of consumer involvement in developing healthcare policy and research, clinical practice guidelines and patient information material.	2006	https://doi.org/10.1002/14651858.CD004563.pub28
Patient engagement in research: a systematic review	2014	https://doi.org/10.1186/1472-6963-14-89
A Systematic Review of the Impact of Patient and Public Involvement on Service Users, Researchers and Communities	2014	https://doi.org/10.1007/s40271-014-0065-5
Mapping the impact of patient and public involvement on health and social care research: a systematic review	2012	https://doi.org/10.1111/j.1369-7625.2012.00795.x

A number of systematic reviews had limited data regarding actual methods of involvement and reported limited evidence of impact. Where impact of involvement was reported it was almost exclusively positive.

Variation in language

Variation in language makes meaningful comparisons difficult[592].

Language to describe people involved in research

In total over 46 different words and phrases were identified to describe people involved. The top four most common were variations on the word 'user' (including 'patients and service users'), with 'community' the 5th most used.

Types of involvement (what)

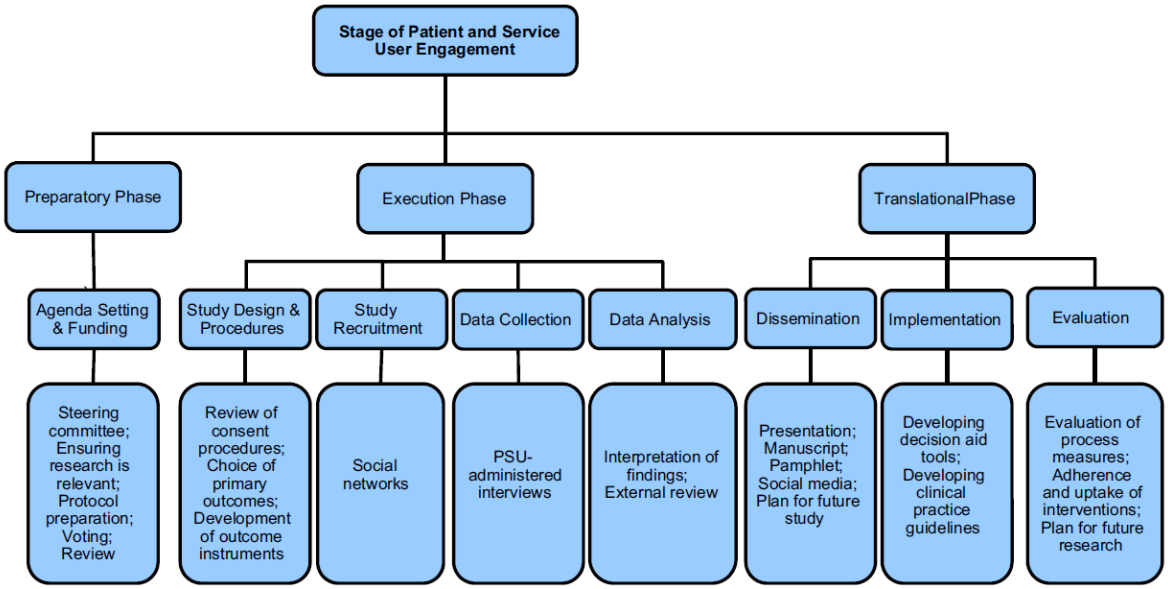
Involvement was reported at each stage of research with positive impacts were reported at each stage. Involvement in data collection and analysis were the most reported stages of involvement. While most methods of involvement did not explicitly state if it was group or individual, of those that did, more than twice as many specified that the method involved people in a group structure (31), rather than individuals (15).

Tasks of involvement

Influencing the design of the research was the most frequently reported task or 'role' (12), with interviewer (5), participant recruitment (5) and providing researchers with a perspective on the relevance of the research jointly second in occurrence.

Stages of research

In order to describe the stages of research, this review has adapted an existing framework. The most helpful framework was found in a review which describes patient and service user engagement (PSUE) at several research stages, within three larger phases: preparatory, execution and translational [592].



- Phase I: Preparatory Phase
    - Stage 1: Agenda Setting
    - Stage 2: Funding
- Phase II: Execution phase
    - Stage 1: Study Design and Procedures
    - Stage 2: Recruitment and participation
    - Stage 3: Data Collection
    - Stage 4: Data Analysis
- Phase III: Translational phase
    - Stage 1: Dissemination
    - Stage 2: Implementation
    - Stage 3: Evaluation

Number of examples of involvement at each stage

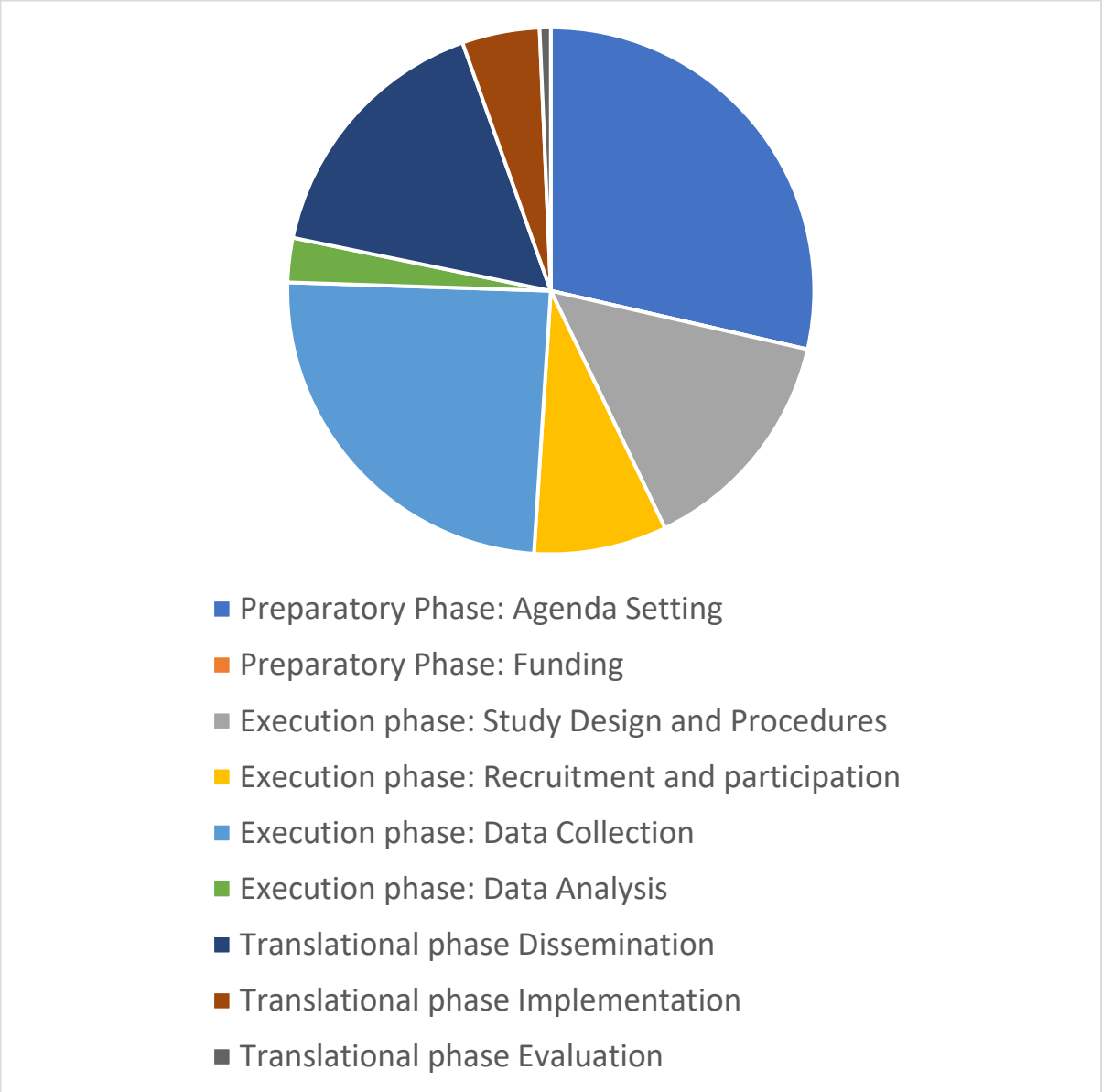
Using the framework above, the data was categorised to show the number of examples of involvement at each stage. This data is summarised in the Table 2 below and in Figure 1.

Table 2: Involvement examples reported at different stages

Stage	Number of examples at stage	Rank
Preparatory Phase: Agenda Setting	42	1
Preparatory Phase: Funding	0	8
Execution phase: Study Design and Procedures	21	4

Stage	Number of examples at stage	Rank
Execution phase: Recruitment and participation	12	5
Execution phase: Data Collection	36	2
Execution phase: Data Analysis	4	
Translational phase Dissemination	24	3
Translational phase Implementation	7	6
Translational phase Evaluation	1	7

Figure 1: Number of examples at stage



Stages of involvement

The stages of research mentioned in association with involvement most frequently were the 'Preparatory Phase: Agenda Setting' (42), 'Execution phase: Data Collection' (36), 'Translational phase Dissemination' (24) and 'Execution phase: Study Design and Procedures' (21).
Agenda setting and data collection were the stages with the most impacts (13), with dissemination the next highest (10).

Methods of involvement (how)

A total of 27 different methods of involvement were extracted, although it is likely that it does not reflect the diversity of methods used in all the studies included the systematic reviews as reporting of the methods of involvement is often imprecise or absent. For example, only 34 out of 385 lines of extracted data explicitly mentioned a method. Of all the methods, community based participatory research had the most impacts (2), with the method being reported as being used at all stages of the research.

Impacts of involvement on the research

Other than generic positive impacts, the most impacts reported were improving the relevance of the research, improving dissemination and improving data collection. It is unclear if more impacts were reported from dissemination and data collection because that is where people were most involved, or if that is because these kinds of outcomes measures are easier to measure.

Methods and impacts – causal relationships

In some reviews, explicit causal relationships between certain methods of involvement and positive impacts were made. They are summarised in Table 3 below, and ordered according to the research cycle outlined in one of the systematic reviews included in the narrative synthesis [593].

Table 3: Causal relationship between involvement and positive impacts

Type of involvement	**Stage of research cycle**	**Impact on research**
Identifying topics	Agenda Setting	Generic positive impact
Participant recruitment	Recruitment and participation	Improved recruitment
Developing questionnaires	Data Collection	Improved relevance
Interviewers		Improved data collection
Data analysis	Data Analysis	Improves interpretation
Influencing dissemination plan	Dissemination	Generic positive impact

Impacts of involvement by stage of research

The following impacts were recorded at the following stages. Where available, information about the type of involvement method used has also been included if it was present in the data line. They are summarised in the tables below and ordered according to the research cycle outlined in one of the systematic reviews included in the narrative synthesis [593]. Agenda setting and data collection were the stages with the most impacts (13), with dissemination the next highest (10). These impacts are summarised in Figure 2 and Table 3.

Figure 2: Number of impacts by stage of research

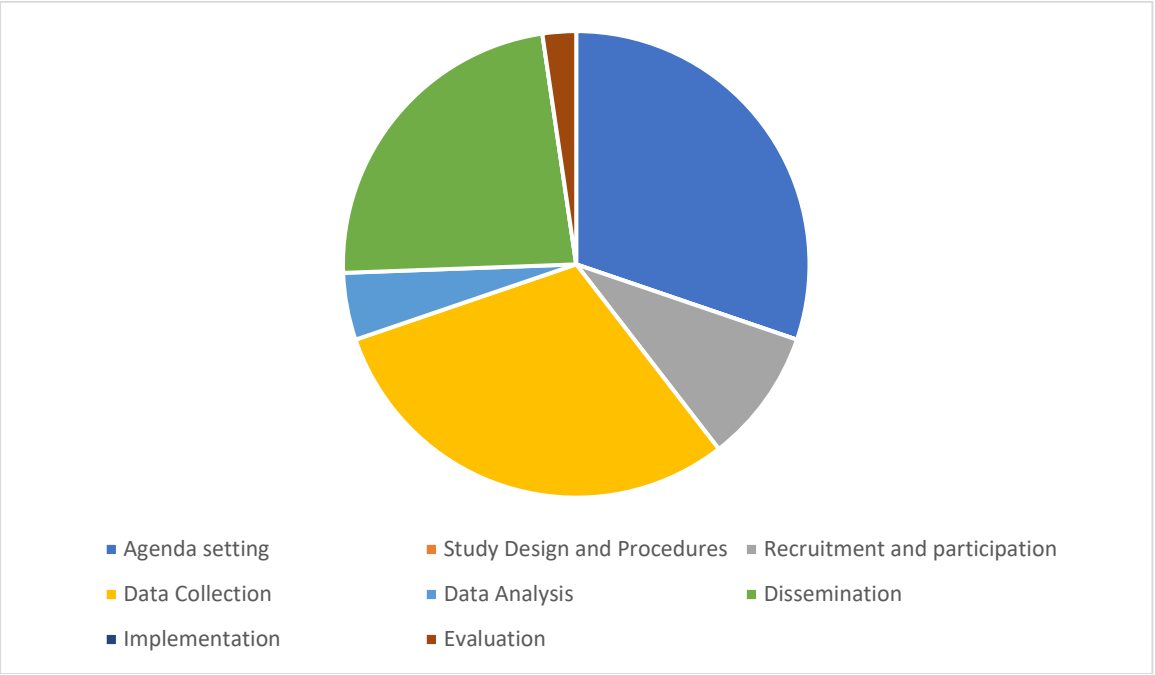


Table 3: Number of impacts by stage of research

Stage of research	**Number of impacts**
Agenda setting	13
Study Design and Procedures	0
Recruitment and participation	4
Data Collection	13
Data Analysis	2
Dissemination	10
Implementation	
Evaluation	1
All stages	2

Discussion

Variation in language

As this narrative review examines systematic reviews which themselves look at papers from the past decade, there is a natural lag in reflecting more contemporary language.
While 'user' still has a very specific and helpful meaning in some contexts (as a shortening of 'service user'), words such as community appear to be more commonly used in more recent literature to describe the wider public or community. It is likely that it appears so frequently in this analysis of systematic reviews as it is naturally more focussed on past usage of language, rather than reflecting contemporary trends, such as a shift to phrases such as 'community', 'public' or 'people'.

Types of involvement

It is unclear if involvement in data collection and analysis was most reported because this was seen as the easiest or most effective way of involving people, or because it was simply the most reported method of involvement (with other forms of involvement not being mentioned in papers as it was not strictly considered part of the research method).

Conclusions

Involving people in any way is generally advantageous, however small it may seem, there may be significant and unpredictable positive impacts. Involving people as early as possible is best-practice and will likely have significant positive impacts on every stage of the research cycle. While there may be some power shifts during this process (researchers sharing control of the agenda, outcome measures etc), if planned, resourced and facilitated well, the risk of any potential negative conflicts can be effectively reduced.

Time and budget are oft-cited perceived barriers to involvement. While the longer-term solutions to this will require a cultural change by both researchers and the public (to effectively resource the important activities required to effectively involve the public) in the short term, there are a number of high impact actions which can be taken which do not require considerable budgets or time. These include creating steering groups or committees, carrying out surveys, training people to work in partnership with communities and using events to raise the profile of the research and the ways that people can be involved.

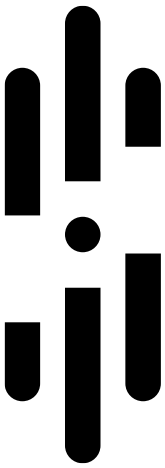
It is not clear if face to face or online involvement is more effective with regard to impact – and the definition of effective will likely change with each individual. Involving people face to face and online both have risks of excluding people. However, while the value of face to face events is clear from a number of reviews – the ratio of cost to impact is much higher for online involvement. The increasing number of people with access to online tools will mean that it is likely that in the future, the default action for involving people will initially be engagement with online communities in order to consult them about the most appropriate next steps to plan involvement.

The final principle is that involving people in planning how the public will be involved is crucial. Just as with the research itself, if people are not involved in planning how people will be involved, doing it and measuring the impact, there is a danger that the method will be ineffective and the value and impact of the research will be reduced.

Applying the principles of involvement to genomics

The narrative review, combined with the international mapping of genomic projects suggests genomic research should not be treated with exceptionalism in relation to strategies for involving people. In other words, it is unlikely that involving people needs to be approached in a radically different way when carrying out genomic research.

Some of the most important issues in genomic research are not unique to genomic research. For example, how are communities of interest defined; how do the principles of fairness and equality interact with potential conflicts of interest when prioritizing research; and privacy and data sharing concerns. However, with the number of people accessing genomic testing increasing, with direct-to-consumer testing growing every year, it is inevitable that even more people will begin to use online tools to both interpret their genomes, share their data and create communities of interest based on shared variations (including familial connections or variations of known significance). As a result, both the narrative review and mapping suggest that involving people in genomic research online will likely have a greater impact in improving research. Face to face events will be less effective as the diversity, complexity of communities of interest and geographical distribution inevitably means that the cost of such events would preclude their effectiveness. In conclusion, standard models for involving people online in every stage of genomic research are required, including standardised frameworks for both evaluation and impact assessment.



STARDIT

Standardised Data on Initiatives (STARDIT)

Sharing the 'who', 'how' and 'what'

Public consultation report

September 2019 to May 2021

About this document

This document describes how the public were invited to be involved in giving feedback on the 'Standardised Data on Initiatives (STARDIT): Alpha Version'[196] between September 24th 2019 to the end of 2019. The feedback from this process was summarised into learning points and actions which were used to inform the co-creation of the Beta version of STARDIT[442]. This report then describes the public consultation process for 'Standardised Data on Initiatives (STARDIT): Beta Version' from February 2021 to May 2021.

This report has been written by Jack Nunn, Director of Science for All and PhD researcher at La Trobe University. This project is being run in partnership with the Wikipedia Journals (Wikimedia Foundation). More information can be found at ScienceForAll.World/STARDIT

https://doi.org/10.26181/611dfcf12c6a9

This report has been included in the appendices of this thesis to indicate the detailed co-design process of STARDIT, which occurred in parallel with this doctoral research.

Public consultation

Consultation period: September 2019 to December 2019

The 'Standardised Data on Initiatives (STARDIT): Alpha Version'(1) was published in September 2019. Opportunities to be involved in co-creating this version were advertised online using social media and shared via email to potential authors. Comments from co-authors were then incorporated into a series of versions, with all co-authors reviewing the final pre-print version.
The pre-print was shared online and promoted using emails, newsletter and social media. Feedback from the public gathered by:

- Emails
- Phone and video calls
- Online discussion forums
- Online forms
- Public events in London and Melbourne
- Face-to-face conversations

More information about the public event in London is shared in the next section. Relevant learning points from a **Wikimedia Youth Salon** is also incorporated into this report.

In addition, Jack Nunn (Director of Science for All) worked with a number of people to complete STARDIT reports, in order to test how appropriate and useful the data entry was. This involved a series of phone and video calls, followed by exchanging versions of STARDIT reports in order to create finalised versions.

Data from all these sources has been collated and organised into themes using qualitative thematic analysis. Event attendees were invited to ensure this report captured comments from the event. Further information about this data (including how it was collated and analysed) will be shared in the planned peer-reviewed paper 'Standardised Data on Initiatives (STARDIT): Beta Version'.

London Event Summary

On 1st October 2019, Science for All facilitated the first public meeting about 'Standardised Data on Initiatives (STARDIT)'. The event was facilitated by Jack Nunn (Director, Science for All) and hosted by the University College London Institute of Education, London.

Registration was free and open to anyone. People could join both in person and online. The facilitated discussion lasted three hours, with breaks. A detailed facilitation plan can be found in the supplementary materials.

Learning from the discussion has been incorporated into feedback from other sources and has not been attributed to individuals.

List of attendees

In person:

- **Jack Nunn** – Director, Science for All, Strategy Liaison and Editor for the WikiJournals, member of the Cochrane Advocacy Advisory Group, PhD candidate at La Trobe University, Melbourne (Australia)
- **Sandy Oliver -** Director of the Social Science Research Unit and Deputy Director of the EPPI-Centre, Professor of Public Policy at University College London, Editor of the journal 'Research for All'
- **Carolyn Thompson** - PhD Researcher, Institute of Zoology and University College London, Postgraduate Teaching Assistant and Lecturer, University College London.
- **Mick Mullane -** Innovation Lead, National Institute for Health Research Digital Office
- **Jim Elliot -** Public Involvement Lead, Health Research Authority (England)
- **Richard Stephens** - Patient Advocate, Co-Editor-in-Chief, 'Research Involvement and Engagement', National Cancer Research Institute 'consumer' representative

Online:

- **Chloe Mayeur –** Sciensano (Belgium)
- **Wannes Van Hoof –** Sciensano (Belgium)
- **James Ansell –** Consumers Health Forum (Australia)

The first 'STARDIT' selfie at the London event
Left to right: Jack Nunn, Sandy Oliver, Carolyn Thompson, Mick Mullane, Jim Elliot, Richard Stephens

Consultation period February 2021 to May 2021

After the feedback from the Alpha version was collated, work began on the Beta version. Between January 2020 and February 2021 multiple meetings took place (with some face to face involvement cancelled owing to the COVID-19 pandemic).

Online video presentations and discussions

Online activities where feedback on STARDIT was invited and given include (but are not limited to) the following invited presentations and discussions:

Title	Invited by	Given by	Date
Standardised Data on Initiatives (STARDIT)[287]	WikiCite 2020 Virtual conference	Jack Nunn and Thomas Shafee (recording, resource and transcript)	27th October 2020
Standardised Data on Initiatives (STARDIT)[288]	Poche Centre for Indigenous Health, 9th Annual Research Showcase Program	Jack Nunn	18th November 2020
Involving people in DNA research	Poche Centre for Indigenous Health, Research Advisory Board	Jack Nunn	8th September 2020
Involving People In DNA Research	Ludwig Boltzmann Gesellschaft	Jack Nunn (recording, resource, presentation)	24th September 2020
Genomics Research and Involving People[290]	La Trobe University	Jack Nunn	13th October 2020
Involving everyone in research: Creating the evidence[291]	Australian Citizen Science Association	Jack Nunn	1st April 2021
Involving People in Rare Disease Research	Rare Voices Australia	Jack Nunn	5th August 2021

Text-based feedback and discussion

Method

A number of methods for gathering feedback and hosting online text-based discussions were used for this period of the consultation. This included using an online text-based discussion platform (hosted pro-bono by Science for All) to discuss the STARDIT Beta version, online forms for collecting feedback, online shared documents for simultaneous editing and commenting and using online pre-print servers to share stable versions[451]. A version of the Science for All STARDIT Beta webpage was archived to preserve how feedback was invited during this period[594]. Information about the consultation process was also shared by STARDIT authors via email, social media (including Twitter, Facebook and LinkedIn), and the Science for All website. Specific areas where feedback was requested included:

- Helping improve areas which are unclear or might not make sense
- Checking the STARDIT data fields are appropriate (anything missing or unclear)
- Improving the 'Example applications of STARDIT' table for your own discipline(s)
- Suggesting any relevant references that may be missing

Results

A total of 27 people provided feedback on the Beta version via the online form and collaborative document. Over 7000 words of feedback and comments were provided via the online form with 144 separate points, comments or corrections. While there were multiple small changes and comments on the collaborative document, there were 51 comments with logged changes which were 577 words in total.
All of the feedback, comments, corrections and responses by the lead authors can be found in the supplementary resources, in the section 'Anonymised Feedback on Beta Version' and 'Change log from Alpha Version'.
The final version was sent to all co-authors for checking before submitting for publication.

Learning points from the public consultations

STARDIT as a project

- The principle of standardised reporting described by STARDIT is **useful across disciplines, this is 'unique'**
- STARDIT reports will be useful for a number of disciplines, including health research, environmental research, public policy, educational interventions and community arts projects.
- Many people don't know who to trust and one participant noted that 'most of our decisions are based on trust'. **STARDIT was identified as a way of sharing data that will facilitate people to critically appraise many kinds of data**.
- STARDIT is especially **helpful for people to self-assess research** and appraise it, including supporting **informed decision making about whether to participate.**
- STARDIT was identified as a way of reporting how people were involved in defining 'shared purpose', including defining outcome measures (for example, answering the question 'what does success look like and how will we measure it')
- STARDIT could provide **an independent way for researchers and policy makers to show how people have been involved in co-producing it**
- STARDIT was identified as **a helpful system for international development**, including planning, reporting and evaluating initiatives[595]
- STARDIT was identified as **a helpful system for people planning, reporting and evaluating initiatives**, including mapping preferences for involvement, reporting involvement and impacts from involving people.

Proposed collaborative way of working

- While the project is 'ambitious', the proposed collaborative way of working balances openness with efficiency
- In order to make STARDIT happen, it was suggested to 'start small' and 'think like a start-up'
- A number of partner organisations were suggested throughout the public consultation including Academic Health Science Networks (UK), The National Cancer Research Institute (UK), Independent Cancer Patients' Voice (UK), Clinical Trials Units (UK), Patient Focused Medicines Development (global), National Institute for Health Research (England), Good Things Foundation (UK and Australia), Google ('Scholar' team), National Institute of Health (USA), Patient-Centered Outcomes Research Institute (USA) and the James Lind Alliance

Authenticity and trust

"This is so global and so big – it comes back to trust, how do I trust the people who report data using STARDIT"

STARDIT will be assessed by an editorial board and eventually, open peer review. It will use indicators from public domain sources. However, the root problem of authenticity and truthful reporting remains for all peer-reviewed data. While STARDIT provides data to facilitate critical appraisal, ongoing work will be required to ensure the authenticity of data. Partnering with the Wikidata project will ensure data is linked and machine-readable. Assigning Digital Object Identifiers to STARDIT reports will ensure that versions are immutable, but that the reports themselves can be updated should errors or inaccuracies be discovered.

Personal safety risks

Risks were identified with STARDIT for people who may share information or data which might have legal or safety implications. For example, data provided by members of the public about illegal activities (such as poaching or illegal logging) might incriminate individuals or put those sharing the data at personal risk. Ways of ensuring data is shared in ways which balance transparency with personal safety need to be carefully considered. China was identified by one researcher as an example of a country where special attention and cultural sensitivity would be required.

Life or death information

STARDIT was identified as a system which could help people critically appraise information which might be life-saving or potentially lethal if incorrect. As well as medical information, this also includes information on Wikipedia pages about things such as edible fungi and plants[596].
STARDIT should have a transparent process for redacting information which might contribute to the destruction, poaching or killing of rare or threatened species, for example, not sharing detailed location information of rare species.

Sharing power

There is 'knowledge as power and powerful knowledge', STARDIT is a way of sharing both kinds of knowledge. Some 'power brokers' might not welcome knowledge sharing, transparency and scrutiny in certain areas and may actively resist attempts to share data and power. 'Power brokers' who might be resistant were identified as people working in government and for-profit organisations.

Knowledge translation

Understanding and measuring comprehension and knowledge translation are ongoing challenges in many disciplines. While STARDIT can report data on this, ongoing work will be required to ensure reporting is aligned with international best-practice. Partnering with organisations such as Cochrane and Campbell will help ensure the reporting tool is useful. STARDIT can report transformative learning as an impact, but this will require careful tailoring to each language and culture.

Diversity and inclusion

Ensuring the process for both involving people in the development of and for using STARDIT are inclusive will need continuous reassessment, potentially requiring a group of experts and advisors. In addition, as STARDIT is developed for languages other than English, groups of people specialising in linguistic and cultural diversity will need to be involved in ensuring STARDIT is appropriate, culturally safe and inclusive. In addition, learning and development opportunities will need to be co-created with multiple stakeholders in order to ensure people are given inclusive opportunities to learn how to get involved with the STARDIT project. This was raised as a particular consideration of Indigenous peoples during one presentation to the Poche Centre for Indigenous Health.

Technical considerations

Machine learning and 'artificial intelligence' could be employed to create reports. Wikidata is built for machine learning and provides an open, public domain and free way of sharing data that anyone can access, anywhere. After providing a way to host reports, multiple ways to submit them should be co-created.

Readability and plain English

The 'Standardised Data on Initiatives (STARDIT): Alpha Version' needs to be improved for readability and plain English. In addition, the purpose and scope needs to be explained more clearly. Tailoring communication to specific disciplines should also be considered. Future versions that might be translated into other languages will require co-creation with language communities to ensure they are comprehensible to as wide an audience as possible.

Systematic Searching

Future versions of STARDIT after the Beta version will require a systematic review in order to ensure that all appropriate data sources have been consulted. As this will require a significant investment of time from those involved in the STARDIT project, it was agreed that at this stage of the co-creation process, a 'mini-review' (published in the peer-reviewed WikiJournal of Science) was an appropriate intermediate step to ensure the current search strategy is appropriate.

Indigenous knowledge

A report by Science for All written for the Wikimedia Foundation identified that there might be systematic, technical and cultural barriers to incorporating the knowledge of indigenous peoples into Wikipedia and other peer-reviewed repositories[596]. After additional meetings with staff from the Wurundjeri Woi Wurrung Cultural Heritage Aboriginal Corporation, it was agreed that it could be helpful to explore using STARDIT to co-create a way for indigenous peoples to share traditional and oral knowledge. STARDIT could be used to transparently report who created any content containing the knowledge, what tasks they had, how this knowledge was shared and any relevant concepts of 'owning' or 'property'. Members of Indigenous communities could work in partnership with the Wikimedia Foundation to create 'verified' users who formally represent relevant communities are have permission to share and verify knowledge (including stories, beliefs, medicine).
The report concluded that a detailed piece of research needs to be commissioned (potentially by the Wikimedia Foundation) to explore concepts of 'intellectual property' and 'owning knowledge', and how this respectfully interacts with the free knowledge and open access movements. Certain cultures have restricted, taboo or 'secret' knowledge[596]. This can include culturally significant sites which may be at greater risk of vandalism if they are shared in the public domain. STARDIT needs to be co-developed with the Wikimedia Foundation and with indigenous peoples to ensure that a balance is struck between sharing, storing and preserving unique intangible culture, while also remaining sensitive to respective cultural practices and attitudes regarding 'ownership'.

Actions and results

1. Create a peer-reviewed scoping review to supplement the STARDIT beta paper, in preparation for a future systematic review.

   Result: Completed and ready for submission

2. Science for All to pay developers for creation of STARDIT report hosting. Create a project brief and invite developers to apply to create a beta version of STARDIT hosting.

   Result: Completed, Beta version built by paid developers (paid by Science for All, approved by the Steering Committee)

3. Host more face to face and online events in other capital cities, including Canberra and Berlin.

   Result: Abandoned. Planned face to face events in London, Berlin and other cities in 2020 were converted to online meetings and presentations

4. Rewrite parts of the STARDIT paper to be clearer and in plain English.

   Result: Completed , Beta version ready for submission in open access journal

Supplementary resources

Facilitation plan for public event – 1st October: London

Purpose of event

Create an opportunity for people from different disciplines to talk about standardised ways of reporting initiatives, including research, education and international development.

Aims

- Summarise what different disciplines are reporting about initiatives and how
- Suggest a common framework for reporting (STARDIT)
- Host a discussion about common challenges and generate ideas

Session	Summary	Instructions	Outcomes	Timing
Introductions	A chance to learn who is in the room, and what they hope to get out of today - and what the process for the afternoon is	Ask everyone in room and online to say what their area of expertise or knowledge is and why they've come today - **Online: Facilitator will summarise comments from people**	Everyone knows who is in the room and online.	15
Jack Nunn, Director of Science for All	A short presentation from Jack Nunn, Director of Science for All - about the learning from his recent projects, including his PhD about involving people in genomics research. A short summary of 'Science for All' and what led to STARDIT			10
Carolyn Thompson, Primatologist	A primatologist investigating small ape decline in China, Vietnam and Myanmar. She's working with local people, using participatory action research to investigate the patterns and drivers of critically endangered gibbon decline. She will discuss how			10

	STARDIT could be useful for recording impacts from this kind of work.			
Open discussion (including input via Zoom)		Ask people online to type thoughts or comments to be read out (also check Twitter #stardit). Ask people in the room to share initial thoughts, **summarise comments from people online.**	People online and in the room have contributed to discussion	5
Break - mingle - tell people online to log back on in either ten minutes to hear speakers or log back on at 2:10 to join the 'Idea Vortex' - **note Australians might be going to bed and say goodnigh**t				10
Reporting the what, who and how?	Short introduction to STARDIT with 3 different speakers leading discussions on how it could be used and improved.			
	Sandy Oliver - a personal and professional perspective (including thoughts on journals) - **lessons from successful reporting tools?**			7
	Richard Stevens - a personal perspective as a cancer patient and a professional perspective in relation to genomics research and on journals) **What would make people want to use this?**			7
	Jim Elliot - a personal perspective and a professional perspective in relation to the work of the Health Research Authority. **What support would people need to use STARDIT?**			7
Idea vortex	Using the 'Idea Vortex' model - a series of questions designed to find issues and create solutions	**Welcome back people online!**		50
Open discussion and break				10

Learning so far	A chance for anyone to speak about what has been learned so far, any reflections. Also a chance to map who's not involved who should be moving forward - and who will invite them!	**Invite comments from people online**		15
Agreeing tasks, actions and discussion areas	What actions have been agreed, what tasks and areas for further discussion	Explain how Loomio will be used going forward and how actions and decisions will be made	A list of actions, tasks and decisions to be posted on Loomio.	15
Open discussion - tea - cake - 'networking'				20
Formally close event 4pm. Adjourn to nearby pub.				

Additional Discussion Points

These additional discussion points were used to supplement the discussion:

- Can anyone write STARDIT reports? People unaffiliated with projects? Can this be one data-line that contributes to a 'living report' - in other words, could people report on behalf of organisations (like people can write Wikipedia pages about organisations without their approval)
    - Solution could be that reports make it clear when people from the organisation have been involved - verified (tick like on twitter?)
- How should peer review work?
    - In the short term, it will have to be an editorial board (volunteers associated with the WikiJournals) - we will use the existing processes of the WikiJournals for the Alpha version and Beta version
    - In the longer term - post beta version - it should be an open peer review process. For discussion
    - Peer review needs to ask the question is there evidence/data to back claims in STARDIT report - does it require some kind of standard critical appraisal tool?
- In the longer term STARDIT could 'score' projects
    - STARDIT scored- a peer reviewed score for an initiative which scores it on criteria including 'power sharing/involvement', data sharing, dissemination and translation
    - Scoring could be based on 'is there a data source for this item' so that it is not subjective (Binary yes or no on indicators of involvement)
    - Scoring continually reviewed but must be future-proofed so historical scores still have validity and use
    - This function would likely require funding/grant etc to support infrastructure - while peer reviewing would be voluntary the process of editorial control and back end would need not-for-profit investment.

STAR	**Dissemination**	**Involvement**	**Translation**
4.9	5	2	3

- Can things like 'documentaries' be included - who made it, who did what, who funded it? Would this be a category of 'educational intervention' - allowing documentaries to actually measure impact
- Risk of confusion with reporting guideline: STARD http://www.equator-network.org/reporting-guidelines/stard/
    - Not considered an issue by attendees
- Create API for other journals etc to use with their site? Create badge
- Partners get accredited to improve participation and recruitment
- Citing Aboriginal stories - create STARDIT report for story?

Anonymised Feedback on Beta Version

Respondent ID	Feedback with an action for the paper	General feedback about STARDIT	Change made	Lead author's response if no change made	Second Author Response	Status
2		mention of patients/public being able to complete a STARDIT report themselves, but would research teams or those responsible for patient/public activities need to deliver training to equip them with the skills to complete the form or do you envisage the reports being a joint effort?	added to discussion: 'Similarly, based on feedback from Indigenous community leaders, patient representatives and others, it is essential to ensure access to learning and development opportunities is available to support people to both access and create STARDIT reports.'			Comment incorporated into Beta
3		132 & 133	See log from tracked changes			Comment incorporated into Beta
4	Need examples earlier on exactly WHAT you are collecting - The challenge for any non-research person reading the abstract and the lengthy preambles in the paper is always, "Yes, but WHAT data?" There are no examples given until page 10 of the main paper, where there are some very useful and comprehensive lists. Even then as a lay person I think of it as "information" not "data". All the more reason why you need examples much earlier on of exactly WHAT you are collecting - you mention responding to an epidemic, which is a great outcome, but STARDIT is about process, and that's what isn't clear.	mention responding to an epidemic, which is a great outcome, but STARDIT is about process, and that's what isn't clear.	Very helpful point, thank you. I have added this to background section 'For example, when designing a response to an epidemic, standardised data can improve retrieval of relevant information which can be used to inform which affected individuals or organisations could be involved in the design of the response and which outcomes are most important. This can include deciding which stakeholders should be involved in which tasks, such as prioritising outcomes.' I also added this para to differentiate between raw data and metadata, as both of those are included in what we refer to as 'data' - 'Hereafter, data generated by an initiative (including raw data), information about the data (meta-data) and information about the initiative will all be referred to as 'data' unless otherwise specified.'			Comment incorporated into Beta

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9	line 405, Table 4: first, regarding the Section “Involvement in initiative”, Data category “Involvement appraisal”, data field “How did the initiative change as a result of involving people (did the design or evaluation change?)”. Suggest not only ask for effects of involvement on the initiative where involvement took place per se, but also on other areas.	If these categories are supposed to be described under the data category “Involvement outcomes, impacts, learnings or outputs” I would be a bit more specific, because right now, the data field description “Where there any outcomes, impacts, learnings or outputs from people begin involved?” reads to me as if this category focuses solely on effects on the people – which I think does deserve its own category – but not on what the people (and their organizations) may do based on their involvement. I do think this aspects differs from the section “Impacts and outcomes” because it may be directly correlated to the involvement of the project. I am thinking, for example, of a research project that involved local practicioners. One of them told us that he started applying the participatory methods he experienced in the research project as a participant in his work environment at a social service organization. I would not classify this as an outcome from the project per se, but from the involvement directly. It is, however, “more” than just an outcome on the people involved, but a “spill-over” from their involvement to affect others outside the involvement process.	Thank you for this, I think all the things you identified in your comment can currently be covered by this point, however I agree in future versions perhaps this could be articulated further. I have added to the help text 'These can include impacts on people, organisations, processes or other kinds of impacts' which I hope addresses your point? I will also flag with Thomas whether this section should just be added to the other impacts categories, although I note this is more of a design interface issue rather than a structured data issue.		I think it should be possible to create structured data to indicate these sorts of outcomers something like this (e.g. using "applies to part (P518)" to indicate the person and "caused by (P828)"+"involvement (Q1671829)" to indicate that it waws the act of involvement that lead to said outcome). Indeed indicating if any outcomes apply to a specific person/group may be useful in other circumstances (e.g. applies to study participants, some governmental department, NGO, entire industry sector, etc) Having said that, I don't know whether it's going to be common enough that we'd want a whole separate section for it in terms of a data entry form. I'd be keen to have it simply as an option in the normal outputs section.	Comment incorporated into Beta

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9	line 405, Table 4: section “Impacts and outcomes”. I am wondering whether a section about who decides on the outcomes used to evaluate the impact of an initiative may be helpful.	I think it would be quite interesting to know if the outcome measures were decided on collaboratively as well or who was in charge/what was the process of agreeing on them. I found that often those initiating the project (in our case, researchers) have quite different goals and outcomes in mind than participants – sometimes these goals can even contradict themselves. If only initiators of the project decide on outcome measures, these measures may not reflect the full reality of what the project really “should” lead to, but only a limited (maybe distorted) view that reflects the interests of only one party rather than all. Therefore	Thank you - after careful thought I agree and decided this did need extra categories. While strictly this whole section could be put into methods, I think it's important to make it distinct and clear. As a result I added 'Who was or is involved in deciding on the outcomes used to evaluate any impacts or outcomes? How were they involved?' - I note this could be two data categories but for now kept them as one row in the table just more to keep table length down than anything.			Comment incorporated into Beta
10	Note- I answered ‘no’ on the first two questions even though the technical answer to the question(s) is (were) ‘yes’ but both the Abstract and Plain-English need a but of tweaking/clarification.			Please share any further changes you think should be made		comment noted but not incorporated into this version
10	Line 38: suggest ‘aims’ instead of ‘exists’ potentially a better fit		Thank you - I went with 'was created' as it's more active and concrete			Comment incorporated into Beta
10	Line 44-45: noting open-access is repeating info given in line 39+40 so is redundant. Unsure why it is being noted here that ‘authors can be verified’, suggest delete or elaborate on why this is important.		changed to 'STARDIT is free to use and data can be accessed or submitted by anyone. The authors of the data can be verified (to improve trust)'			Comment incorporated into Beta
10	Line 44-45: Data being “assessed for quality” is not plain English, needs refinement and clarification.		changed to 'checked'			Comment incorporated into Beta
10	Line 46: suggest ‘counter complex global problems’ rather than ‘improve’		changed to 'solve' as counter is not plain english			Comment incorporated into Beta
10	Line 47: delete ‘‘being”		changed			Comment incorporated into Beta
10	Line 97+98: adding a third example in the ‘pandemics and air pollution’ in line 97 (i.e. pandemics, air pollution and X’)		added 'biodiversity destruction' although climate change could be another example to use, sadly too many to choose from			Comment incorporated into Beta

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10	Line 102: putting 'research' into quote marks like the terms in line Line 103. Just for consistency and to emphasises all the terms are equally valid.		changed thank you, good spot			Comment incorporated into Beta
10	Line 49: "interventions in which affected population groups are integrally involved" is a bit convoluted. Needs tweaking/shortening.		Changed to 'STARDIT was developed on the understanding that such problems require evidence-informed collaborative methods, multidisciplinary research and interventions in which people who are affected are involved in every stage.'			Comment incorporated into Beta
10	Line 69-73: not clear as written how exactly Stardit as a mechanism leads to the benefits explained. Elaborating that it does these things through making retrieval and comparison of data easier/more efficient/faster to lead to better and quicker decisions.		Changed to 'For example, when designing a response to an epidemic, standardised data can improve retrieval of relevant information which can be used to inform which affected individuals or organisations could be involved in the design of the response and which outcomes are most important.'			Comment incorporated into Beta
10	Line 34-73: clarifying the distinction in these two sections (and in the Beta as a whole) between Stardit as a tool/concept and the Stardit repository of information.		I have worked to address this throughout but it's a valid point (and a nuanced one that's challenging to explain in plain english)			Comment incorporated into Beta
10	Line 118-120: the situation being highlighted here is bad but should tease out why exactly to lay groundwork for how Stardit will solve it.		added 'In addition to providing new standardised data categories for describing who was involved in which tasks, STARDIT can also incorporate existing data standards (see the supplementary resources 'Using Standardised Data on Initiatives (STARDIT): Beta Version Manual'), creating a unifying system for data hosting, linking and analysis. '			Comment incorporated into Beta
10	Line 115-120: This paragraph is a bit confusing and the three statements/information bits don't seem strongly linked with each other. Starts by talking about the importance of involving broad groups in initiatives generally but then ends talking about a problem of too many reporting tools in a specific type of data.					Comment incorporated into Beta
10	Line 127: unsure if 'compete' is the right word. Do we mean 'conflict'?		added 'compete or conflict' as these are distinct and important			Comment incorporated into Beta
10	Line 202: should 'Participatory Action Research' be capitalised?		changed to 'STARDIT development is guided by participatory action research (PAR) paradigms' as it's not a proper noun and the singula use implied there's only one paradigm			Comment incorporated into Beta
10	Line 208: Critical reflexifivity should maybe be in quote marks and/or be capitalised given it's the name of a, uhhh, theory(?) (methodology?)		whatever it is it's certainly a jargon term (but one that can't be avoided, as that's what it's called!) so I've changed to 'At the core of participatory research is 'critical reflexivity''			Comment incorporated into Beta
10	Line 215-216: unsure what 'media freedom' means in this context and how it is relevant to Stardit. Presume that it is referring to a free media not controlled by the State but that doesn't really seem to clearly link to Stardit processes or aims.		changed to 'To uphold human rights and 'environmental rights'64, and for 'the maintenance of peace', people require 'media freedom' in order to 'seek, receive and impart information'63, free of unaccountable censorship' - I would say media freedom and censorship are likely to be at the very core of some of the reasons STARDIT might be challenged by some people with power to challenge such things			Comment incorporated into Beta

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10	Line 216-217: How will/does Stardit held to 'uphold these universal rights'?		Thank you yes that was missing - added 'STARDIT has been created in order to help anyone uphold these universal rights, by providing a way to share open access information in a structured way with a transparent process for quality checking'			Comment incorporated into Beta
10	Line 281: there is an errant * against 'Genomic research' that does seem to correspond to any post-table notes		removed			Comment incorporated into Beta
10	Line 292: unclear where people are meant to start in this Figure i.e. which is step 1. adding numbers like in line 303. the line 'Report planned initiative' have the addendum 'into Stardit' into it. Similailrly 'preference mapping' should maybe be standardised/expanded to be 'Stakeholder preference mapping' as currently	it isn't clear what the term means within the figure.	Added 'Figure 2 describes how STARDIT can be used to map how people might be involved in designing, doing, reporting and evaluating initiatives, starting with 'idea sharing', to clarify the cycle, but I guess the point is you can start at any stage of the cycle RE Pref mapping, reworded to this for clarity 'The STARDIT Preference Mapping (STARDIT-PM) tool provides a standardised way to report the preference of multiple stakeholders.' I think adding the word 'stakeholder' is redundant, as who else would be having preferences mapped? Will check with Thomas on this one		It may be worth including the redundant 'stakeholder' just to emphasise that it is for stakeholders generally (or indeed a subset of stakeholders) as readers may have implicit assumptions that preference mapping might just be for a specific group (e.g. investors) or something like that.	Comment incorporated into Beta
10	Line 303+305: Ditto prior comment about saying 'Stakeholder preference mapping' consistently for clarity					Comment incorporated into Beta
10	Line 317: citation needed for the study being referred to		added			Comment incorporated into Beta
10	Line 326: Table X needs to be updated with relevant number		Thank you good spot			Comment incorporated into Beta
10	Line 346: In the fourth row of the table, the second column should maybe read "To establish the purpose, motivations and values of the research from different viewpoints" or similar		agreed - changed to 'To establish the purpose of the research, and the motivations and values of the initiative from multiple perspectives'			Comment incorporated into Beta
10	Line 406: Maybe title should read 'Discussion and Next Steps'?		changed to 'Discussion and future versions' to reflect the content			Comment incorporated into Beta
10	Page 75- errant highlighted text Page 83- errant track change spell check to be approved		eagle-eyed! changed			Comment incorporated into Beta
10	Page 160- in the bottom row of the table, righthand column, on the second last line it should read 'there may be formal' not 'the may be formal'		Incredible spot, you really did read it all! Thank you, corrected			Comment incorporated into Beta
10	Page 105, line 164- another 'Table X' needs number inserted		Changed!			Comment incorporated into Beta

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12	Line 281: Table 1: Example applications of STARDIT. involve different parties is the evaluation of marketing authorisations, and also appraisal. Health Technology assessment is mentionned, but not medicines or medical devices regulation (authorisation) and/or appraisal/pricing.		added 'regulation and authorisation processes (for example medicines and medical devices)' to 'Production, consumerism and business ' sub category 'Other products (medical devices, electronics)'. I also added 'code and algorithm checking (for example, autonomous vehicles)' as I think this fits here			Comment incorporated into Beta
12	Line 403 - Table 4: Summary of STARDIT Beta Version data fields In data, there are different methods used to anonymise data so that individual cannot be re-identified.	It would be important to mention 1/ the risk of re-identification, 2/ the method used to de-identify data	Super important points, thank you. Reworded to 'Who controls access to the data, how are decisions about data access made? Is data anonymised or de-identified? What methods are used for re-identification? What is the risk of unauthorised re-identification? '			Comment incorporated into Beta
12	Line 403 - Table 4: Summary of STARDIT Beta Version data fields There is a varying risk of re-identification (from 0.5 to 0.05 or less, depending on which anonymization method is used and of the context).	It would be important to mention 1/ the risk of re-identification, 2/ the method used to de-identify data				Comment incorporated into Beta
12		line 346: financial relationships and other interests are mentioned; however other interests are not too detailed. I think they're key, sometimes more important to know that financial interests. You might be involved in the early phase of an exciting project, and continune until the end. When final results are disappointing, you might not be totally objective when intepreting them (participatory type conflict of interest. Other types are carrier interests, intellectual interests, conflicts between persons etc.) . Instead of simply "other interests", create a list of all types we can think of, otherwise people will not realise they should declare some.	Changed to 'Financial or other interests (including personal or professional interests)' and also added 'Describe any conflicting or competing interests (including any relevant information about authors of this report), or any other 'interests', including personal interest or (for example, how you may be personally or professionally affected by the outcome of the initiative)'. I'm keen to keep 'interests' open ended as an exhaustive list is impossible, but certainly in future versions we can work on standardising more types of interests			Comment incorporated into Beta
18		ABSTRACT Given how important framing at the onset is for ensuring the initiative is inviting for people (feeling like they can relate to it) and setting expectations, as I find abstracts above 250-300 words and with subsections quite rare across the 3 disciplines I work in, and some of the information seems pretty detailed for an abstract. It's also good to end an abstract with a conclusion-focused line about contributions, impacts, and/or future directions, based on what results show to date. I thought it equally worthwhile for me to consolidate a list of initiatives, people, and resources I thought you would be interested in, as well as help you to make contact with some of those folks too. I hope you find the below comments helpful for the paper more broadly as well.				Comment incorporated into Beta
	LINE 63: Make this far more generic and inviting to anyone engaged in activities that have potential to inform science. Talk about the range of activities later, with broad descriptions of each, so that those unfamiliar with any or all terms can look them up and acknowledge.		reworded abstract to 'Current reporting methods lack information about the ways in which different people are involved in initiatives, making it difficult to collate and appraise data about the most effective ways to involve different people. For example, forms of participatory action research where anyone can be involved in any aspect of research (including 'citizen science') are increasingly recognised as crucial paradigms for solving global problems, as they can help ensure that initiatives are aligned with the priorities of those affected, thus redefining what it means to be a 'researcher'.' also reworded and re-order background section so it doesn't focus on health/citizen science too early but uses them as illustrated examples.			Comment incorporated into Beta

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18	**cite the following refs with the last point, as they both give an overview of terms commonly used for citsci:** **Eitzel, M. V., Cappadonna, J. L., Santos-Lang, C., Duerr, R. E., Virapongse, A., West, S. E., Kyba, C. C. M., Bowser, A., Cooper, C. B., Sforzi, A., Metcalfe, A. N., Harris, E. S., Thiel, M., Haklay, M., Ponciano, L., Roche, J., Ceccaroni, L., Shilling, F. M., Dörler, D., Heigl, F., Kiessling, T., Davis, B. Y., & Jiang, Q. (2017). Citizen Science Terminology Matters: Exploring Key Terms. Citizen Science: Theory and Practice., 2(1). https://doi.org/10.5334/cstp.113** **Kullenberg, C., & Kasperowski, D. (2016). What is citizen science? – A scientometric meta-analysis. PLoS One, 11(1), e0147152. https://doi.org/10.1371/journal.pone.0147152l.**		added			Comment incorporated into Beta
18	LINE 66: I am not sure why researcher is in brackets, it might be more fruitful to talk about different forms of knowledge that everyone has, which could make valuable contributions to scientific research people's knowledge.	In my experience, involved in activities that inform science may not feel comfortable with being labelled as a researcher or a scientist, but are happy to share their knowledge to help us I know more collectively. Professional researchers likewise may feel threatened by language that may be perceived as diminishing their work too, and framing around diverse forms of knowledge (e.g. experiential and cosmopolitan [vetter]) Vetter, J. (2011). Introduction: Lay Participation in the History of Scientific Observation. Science in Context, 24(2), 127-141. https://doi.org/10.1017/S0269889711000032	Changed abstract to read 'blurring the lines between concepts such as 'researcher', 'public', 'patient' and 'citizen''			Comment incorporated into Beta
18	LINE 71: [food for thought] The epidemic example seems very specific while also vague. Perhaps also adding a biodiversity or social science example could help people invasion the broad scope of this effort and how standardisation of all projects could allow for innovative cross pollination?	If wanting a medical example, projects looking at spread of mosquito-borne diseases can include medical, biodiversity (e.g. mozzies spread animal diseases like avian flu too), habitat assessment, and social impacts (e.g. who has access to different forms of preventative tools and meds).	Thank you - at the moment I thought to keep it one worked example throughout and changed it from air pollution to a pandemic, as most people naturally have a better understanding of that now - happy to use more examples throughout but for now have kept as it is as I worry it's too confusing to have too many worked examples too soon?			Comment incorporated into Beta

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18	LINE 78: “co-created in a collaborative way” is redundant as “co” created is cooperative, by definition, but what you actually mean by “co-design” is the key part,	LINE 78: “co-created in a collaborative way” is redundant as “co” created is cooperative, by definition, but what you actually mean by “co-design” is the key part, as people is the term in VERY broadly and often ambiguously.	reworded: STARDIT has been co-created, involving collaboration with people from around the world in multiple ways. Informed by a number of literature reviews and guidelines, methods of involving people have included public events, online discussions and a consultation process.			Comment incorporated into Beta
18	TABLE 1: I find it interesting that there is “environmental research” but there is no mention of other activities that aren’t research based necessarily (e.g. natural resource management, conservation) . It might be worth acknowledgement explicitly that are diverse ways of gaining knowledge beyond reductionist scientific methods?	This implies all projects on environmental must be research, but at a community level, this is often not the case. I also find it odd that this is under the initiative “science for all” and yet science isn’t explicitly discussed much, or a key component of Table 1. I also feel it would be useful to This table seems pretty health dominant, which surprised me a bit.	I agreed with these two comments. I have created the category in table 1 called 'Management and monitoring' where I include both environmental and natural resource management, and alsp public and private essential services (e.g. water/power blur that line between natural resource and essential critical infrastructure) - but how both of these are 'managed' and monitored (or not managed and monitored) is important data to have. I also included data management and monitoring too - but note there's already a section about data and code etc.			Comment incorporated into Beta
18	have an area explicitly talking about technology, particularly in the digital age of it transforming how we gain knowledge beyond traditional conventions of science and the scientific method (e.g. e-science involves data mining, which is very different)?					Comment incorporated into Beta
18	FIGURE 2: This figure is interesting to me, and I would really like to know more about what you mean by each of the variables included, as many of those could be interpreted in a variety of ways that may not have been intent. These aspects also apply to Figure 3.	**As a designer myself, I feel like it’s missing some key aspects around understanding the practices and cultures of people, their actions in a particular context, and their use of technologies, with interactive design as new practices develop. Without considering existing conditions, constraints, variables, etc in a local context, projects often fall down. The paper exemplifies exploring peoples practices and actions to understand feasibility and needs for future projects: Oliver, J. L., Brereton, M., Watson, D. M., & Roe, P. (2019). Listening to Save Wildlife: Lessons Learnt from Use of Acoustic Technology by a Species Recovery Team Proceedings of the 2019 on Designing Interactive Systems Conference - DIS '19 https://doi.org/10.1145/3322276.3322360**	**Thank you for this comment - reference added. I feel that the design stage is covered in more detail in figure 3, figure 2 being the standard 'plan, do, evaluate' cycle, figure 3 being the 'design' cycle which incorporates those elements you've mentioned. If you can think of ways of making this more clear please let me know**			Comment incorporated into Beta
18	ADDITIONAL ETHICAL CONSIDERATIONS Ethical considerations: I wonder, how we make sure that people running the initiatives become aware of it being added?	I have seen in project finders that projects are added by people not necessarily closely involved, and this leads to inaccurate information being in there, but if project leaders have no awareness they may not know it needs correcting. Alternatively, what if a project doesn’t want to be listed? I know this to be true for a few community-driven projects that don’t want to recruit broader interest, but would rather keep the project very locally focused without being contacted but those outside of the community.	Thank you for this comment, I've given this a great deal of thought too and hadn't included explicit information in this version about that scenario. The challenge is to stop the more powerful from censoring the voices of the less powerful, but also to prevent baseless slander etc (the challenge of all societies really!). I have now corrected this in the main text and further elaborated in the supplementary In future versions it will be necessary to further develop a transparent process if a report has been created about an initiative with no involvement from anyone associated with the project, or only one subset of stakeholders. In such cases, the Editorial team might give a standard period of time for any other stakeholders to be involved in checking and editing any information (similar to the concept of ‘right of reply’) before the report is published, or given the status ‘human reviewed’. However, the process for deciding which stakeholders to contact and how to ensure equity (alongside capacity considerations of the Editorial team) is an area for active discussion and development in future versions. For example, ensuring Indigenous peoples who are stakeholders in a mining initiative have been involved in checking a STARDIT report about a mining initiative created by a company with a declared financial interest in the mining initiative might present challenges that it is not possible for the Editorial team to overcome. Such challenges could be labelled and incorporated as future data categories in the STARDIT reports, for			Comment incorporated into Beta

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			example labels such as 'report not checked by all stakeholders' could be updated if more stakeholders have subsequently been involved in checking any report.'			
18	FOLKSONOMY VS ONOLOGY I also wonder if there has been any consideration to creating mechanisms for a folksonomy approach to tagging projects? I have suggested with the US and Euopean groups for citsci both.		from my understanding of the term 'folksonomy' (which I only learned from your comment!) both Wikidata and therefore STARDIT are a form of folksonomy. However, as I consider it a jargon term, I'll add the reference only and have incorporated the main points in into a minor re write of this section pasted below. Perhaps it is something that can be explored in more detail in future versions of the manual, as it's important to highlight this aspect of STARDIT. 'The current Beta Version of STARDIT maps terms and concepts using the Wikidata initiative (part of the Wikimedia Foundation)36, which includes definitions (taxonomy), a way of describing relationships between concepts (ontology)37, and a system to translate definitions and ontology between many languages. Examples of existing taxonomies include the National Library of Medicine's Medical Subject Headings (MeSH), which are used extensively in multiple kinds of literature reviews 38. How to involve people in combining or merging overlapping taxonomies for different subsets of data has been identified as an important question in the process of taxonomy development 5960. By using Wikidata, STARDIT can be used by anyone store both public domain data and metadata (data about data), and link to hosted structured linked data. While it is a novel element set, where possible it will also incorporate element sets from established data standards and map them where possible (see Table X in appendix for examples of data standards which could be incorporated). This includes standard elements and value sets and controlled vocabularies 61. The terms used in this paper are working terms, which will be progressively standardised over the lifetime of the project. Structured Wikidata can help define terms and concepts clearly and unambiguously, in a transparent and open way.			Comment incorporated into Beta
18		CITSCI GROUPS I WORK WITH DOING SIMILAR THINGS & ASSOCIATED RESOURCES WORK CONSIDERING CITING I have already directly shared this info with Jack and put him in contact with associated leaders, but just so you have it all in one place, citsci initiatives worth including: SciStarter [global citsci project finder]: https://scistarter.org/ Australian project finder: https://citizenscience.org.au/ala-project-finder/ [I helped facilitate SciStarter & ALA developing an API] The EU Citizen Science Cost Action CA15212 [https://cs-eu.net/] and it's working group on Interoperability Working Group [https://cs-eu.net/wgs/wg5]; The working group's outputs are reports [https://cs-eu.net/wgs/wg5/resources] and for context, I contributed to the workshop in Geneva and subsequent report "On the citizen-science ontology, standards & data [https://cs-eu.net/news/workshop-report-wg-5-geneva-declaration-citizen-science-data-and-metadata-standards]. The initiative recently rapped up and an associated book was published [https://cs-eu.net/news/book-science-citizen-science]. Several chapters may be of interest but around data and metadata efforts for citsci, see the chapter led by Rob Lemmens. I am currently working with him, Xeni, and Ina to progress some testing of the standards based on existing projects (included Jack in an email including them and Luigi as the COST Action working group). There is also a citizen science community of practice on data interoperability that is ending this month through the EU initiative, WeObserve. There is a lot of overlap in people with the COST Action group. See the resources created on their page: https://www.weobserve.eu/weobserve-cop3-interoperability-and-standards-for-citizen-observatories/ From the US there is the CSA Data and Metadata working group [be sure and see overhead tabs too: https://www.citizenscience.org/get-involved/working-groups/data-and-metadata-working-	Thank you - any new projects will be added to table 5			Comment incorporated into Beta

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		group/], and a subset of that group, including myself has been exploring development of the PPSR Core standards. I also CCed Greg Newman as chair of that group and Brandon Budnicki who is largely responsible for pulling all of our years discussions to date into creating the publicly accessible website very recently with ability to contribute via GitHub. PPSR Core: A Data Standard for Public Participation in Scientific Research (Citizen Science) [https://core.citizenscience.org/]				
19	Two other potential examples for health and social care include: process for identifying patterns of sub-optimal service, patterns for evaluating service improvement initiatives		Thank you, added			Comment incorporated into Beta
20		overall really impressed with where this is at and the progress that has been made since I last read this work.	Thank you - all your comments addressed and responded to			
21		Added comments to the Google Drive current version	Thank you - all your comments addressed and responded to			
24		Feedback on Google doc manuscript and in email.	Thank you - all your comments addressed and responded to			
23		Feedback is attached as a separate document (see 'STARDIT edits_feedback ABorda 24 03 21.docx'				
23	Building on Figures 2-3. Possibly a Flow chart visual to support readers/practitioners in the development of data for a STARDIT Report using the microcategories? For example, a chart breaking down steps and possible decision points with refs to sections in Table 4, for example.			I think this is a fantastic idea. Sadly it is out of my personal capacity at this stage to create that although I think this would be good for future versions. I have flagged this with Thomas.	Agree it could be good. Alternative (or related) implementations could include: - In the technical manual a summary of questions you should ask yourself for each section - A stardit report for this current startit beta community feedback process - An annotated video of the above stardit form being filled out	comment noted for future versions
23	(2) Due to the size of the STARDIT document – possibly divide into 3 separate standalone documents: About STARDIT, MANUAL, RESOURCES.		Thank you - yes I think the idea would be (once published) the main body of the document is the STARDIT paper and then the supplementary resource is the manual, which will be as a PDF with references and additional information			Comment incorporated into Beta
23	Table 5 resources to separate into thematic/domain sections based on field categorisations? Standardise these themes possibly aligning with Tables 2 and 4 in Manual.		Yes, this is a good idea. While there are categories at the moment, perhaps a further level of categorization would be helpful? The amount of work to do this means it might need to be something for a future version but I will investigate this.			comment noted for future versions
23	(4) Consider expanding participatory action research section with co-design and related methods of engagement? See some suggested resources below.					
23	· Line 38 - STARDIT (Standardised Data on Initiatives) 'R' needs to be bold?Also see: Lines 69, 105		Corrected thank you			Comment incorporated into Beta
23	· Line 124 - The word stakeholders includes the public.. = use single quotes? ='stakeholders'		Thank you, done.			Comment incorporated into Beta
23	· Line 142 - used in health, environment, manufacturing , publishing, government policy, education, arts and international development - link to Table 1. ?		Thank you, done.			Comment incorporated into Beta

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23	**· Line 288 - Across all disciplines, 'plan', 'do' and 'evaluate' are recognised as distinct stages of initiatives. Consider use of 'PDSA' – Plan, Do, study, Act ? a standardised iterative, four-stage problem-solving model. Check NHS publication: https://improvement.nhs.uk/documents/2142/plan-do-study-act.pdf**		**Thank you, very helpful illustration - I have added this reference to a systematic review calling for standardised reporting of PDSAs: https://qualitysafety.bmj.com/content/23/4/290.short (your link was expired)**			Comment incorporated into Beta
23	· Line 326 - Table X summarises questions - Table X?		Corrected thank you			Comment incorporated into Beta
23	ADDITIONAL RESOURCES ON FRAMEWORKS, etc		Thank you- novel references will be added into text or Table 5			Comment incorporated into Beta
23	'Proposed policy (manifestoes)' = should be: manifestos		Changed to 'Proposed policy (including draft policy and manifestoes)'			Comment incorporated into Beta
23	Suggestion to organize using a knowledge organization system? Consider Dewey Decimal 10 main classes to organize themes as outlined in table.			Good suggestion, noted for future versions as no capacity to implement at this stage		comment noted for future versions
23	'cultural heritage ' : Suggestion to add cultural heritage to Arts section or on its own? - Large category • Tangible cultural heritage: movable cultural heritage (paintings, sculptures, coins, manuscripts) immovable cultural heritage (monuments, archaeological sites, and so on) underwater cultural heritage (shipwrecks, underwater ruins and cities) • Intangible cultural heritage: oral traditions, performing arts, rituals. Consider revised header: 'Information, media and cultural heritage' – to : Information, media and local traditional knowledge. See comment above		Thank you - rejigged categories and created tangible cultural heritage, using UNESCO terminology			Comment incorporated into Beta
23	Referring to Table 3 Questions for mapping preferences for involvement p. 17: To establish which group the person identifies as being part of – for example 'researcher' or 'participant' Is there a table of definitions? There are several definitions in the introduction but these may be better highlighted in a table. Also note this Line 122 which suggests a blurring across some definitional boundaries?		Thank you - reworded 'To establish which grouping(s) the person identifies as being part of – for example 'researcher' or 'participant' (noting any groupings should be co-defined)'			Comment incorporated into Beta
23	Referring to Table 4: Summary of STARDIT Beta Version data fields p. 21 Microcategory Section on 'Methods' - perhaps breakdown further e.g. quantitative, qualitative? Examples under each ? Link to methods of approach, such as PAR, co-design, referring to relevant sections…		Change category to 'Methods and paradigms' - recognising that participatory action research and associated terms might be considered guiding paradigms not strictly methods themselves. Also added quant and qual as an e.g.			Comment incorporated into Beta
14	Why only 'involvement' (line 247)? In our systematic review/qualitative evidence synthesis (https://aagts.brasilia.fiocruz.br/wp-content/uploads/2020/10/Relatorio_POPART_final.pdf) on models and methods of social participation in R&D, policy, HTA, monitoring decision-making, we outlined engagement as the more comprehensive terminology, following, especially: Woolley, J.P., McGowan, M.L., Teare, H.J.A. et al. Citizen science or scientific citizenship? Disentangling the uses of public engagement rhetoric in national research initiatives. BMC Med Ethics 17, 33 (2016). https://doi.org/10.1186/s12910-016-0117-1		Thank you for this comment, the confusion around terms such as 'involvement' and 'engagement' is central to this paper and while it acknowledges there are different terms for the same thing, this paper articulates what this paper means by involvement in the section 'While meanings of these terms are often imprecise and can be used interchangeably, 'involvement' here is distinct from 'engagement', which is where which information and knowledge about initiatives is shared, for example, with study participants who remain passive recipients of interventions' - however I have added the ref 'Disentangling the uses of public engagement rhetoric in national research initiatives'			Comment incorporated into Beta
14	I understand the use of 'co-creation' here (line 253) and it is obviously adequate for the communal development approach deployed for STARDIT, but why not outline Sheila Jasanoff (2004) 'co-production' conceptualisation? It brings the systemic idea that there is the co-production of a culture that sustains and promotes the scientific knowledge productions and technological innovation while feedbacking the culture for STARDIT.		Ref added to supplementary information as we can't say co creation was informed by this but I've said 'Future co-creation processes should be decided collaboratively'.			Comment incorporated into Beta
14	line 263: I am happy to support this development and future development phases as well as operationalisation stages of STARDIT - especially regarding the proposed conceptualisation/terminology development abovementioned, if there is interest.			Thank you - support gladly recieved!		Comment incorporated into Beta
14	Table 1 line 281 - Research reporting: Why is there not 'data analysis' as a descriptor? This is a key 'research' activity to which I have been most often been asked about when I talk for experts and general public audiences to demystify misconceptions about citizen science around Brazil (especially high-ranking officers at the MoH and decision-makers at research foundations in Sao Paulo) [entire		Thank you, added			Comment incorporated into Beta

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	research process]: 'data validation' is also a concern for professionaly-trained researchers and decision-makers around Brazil. - as per my previous comment.					
14	Coding and algorythms reporting: Information about Data Privacy, Laws, Regulations, Directives and data security (not only pertaining to data ownership) should be mentioned here as several countries either follow GDPR, HIPPAA, GINA and Brazil, for instance adapted GDPR to its own Lei Geral de Proteção de Dados that came into effect Sept/2020.		Thank you - added to 'Management and monitoring' section			Comment incorporated into Beta
14	Other services: Jasanoff (2004) co-production conceptualisation here would be very beneficial as it outlines in which ways STARDIT can contribute to both identification and geolocalisation of impact indicators that should inform in real-time (ongoing manner) about indicators that are context-specific and may be used to develop glocally-relevent, local, regional, national indicators on various issues that remain as an unmet need somewhere, which requires data-intensive publications - it's the concept of change the culture behind the research-to-practice gap.		added ', process for identifying impact indicators (including geolocation data)'			Comment incorporated into Beta
14	involvement' (line 307): Again, do reffer back to our qualitative evidence synthesis as you may find how we have updated Rowe & Frewer's (2004) typology from Arnstein's ladder of social participation to outline that engagement is a more comprehensive terminology when it comes to citizen science and active research methods: https://aagts.brasilia.fiocruz.br/wp-content/uploads/2020/10/Relatorio_POPART_final.pdf line		Thank you -as per previous response this is covered in detail and we're using consistent terminology in this article. Have added ref - please check it's correct			Comment incorporated into Beta
14	337: OTHER OPTION: people who are not affected directly or indirectly but who hold citizenship to a certain region, country, municipality, neighbourhood and has interest in participating in decision/policy-making and/or conttributing with R&D that might be of relevance to improoving the livelihood of fellow citizens from the same region, country, municipality, neighbourhood.			thank you - I think this is covered by 'everyone' but I note this distinction. These categories are fairly fixed in this now as they were used accross the preference mapping for the alpha version but noted for future versions		comment noted for future versions
14	line 169 - REFERENCES: I have looked through your references and I see that you are familiar with Muki Haklay's amazing work on 'extreme citizen science', but I found that you may not be aware of Barbara Prainsack's work on citizen science (she had proposed in this chapter from 2013 that I have revised something very similar to what you propose here with STARDIT, but not as developed as you have done - Prainsack, B. Understanding Participation: The 'citizen science' of genetics. In: Prainsack, B., Werner-Felmayer, G., Schicktanz, S. (eds). Genetics as Social Practice. Farnham: Ashgate. Available at: https://www.researchgate.net/publication/236850804_Prainsack_B_Understanding_Participation_The_'citizen_science'_of_genetics_In_Prainsack_B_Werner-Felmayer_G_Schicktanz_S_eds_Genetics_as_Social_Practice_Farnham_Ashgate) and other publications - you may want to ask her to review STARDIT (I can mediate connection, if need be).		Thank you for offer of connection with Barbara - only just seen this - yes please for future versions! Reference added to table 5			
26	1) The definition of "initiative" is a bit novel – are there ways of highlighting what is meant by it in the context of STARDIT? One suggestion would be to have it explained in a text box, where additional level of detail and examples could be given for those that need it without disrupting the flow of the text.			I feel this is hopefully this is covered in the section 'Defining 'initiative' and 'involvement' - but a list of terms used in the paper could be a useful additional table. For consideration		comment noted but not incorporated into this version
26	1) I think STARDIT has a lot of potentially very significant contributions that could really make a difference, but I find it a bit hard to pick them out from the text. Would it be possible to make a box just highlighting the (potential) benefits/value added of STARDIT? This would be in addition to the table on the applications of STARDIT in different disciplines, and basically just be X number of bullet points explaining what STARDIT can do and why it is important.			Noted - for consideration		comment noted but not incorporated into this version
26	3) It might be helpful to go into more detail, if possible, of the benefits of getting standardized data about initiatives? I.e. what does it help us to know the values of people involved in a policy initiative.		added 'Transparent acknowledgement of differing values and perspectives is critically important, in particular when mapping if different stakeholders' values are complementary or opposing'			Comment incorporated into Beta
26	4) The abstract seemed to me to be very strongly focused on methodology (citizen science and participation action research), but as I understood it STARDIT goes well beyond this. The phrasing in para two in the abstract (line 64 onwards) seems to me to suggest a delimitation of STARDIT to standardised data about who and how people are involved in initiatives, and I feel that underscores the potential contributions of STARDIT.		thank you - reworded for clarity			
26	5) A lot of initiatives will focus on evaluating another initiative, and it would be great to cross-reference all initiatives related to a specific topic or theme, especially to see how outcomes are assessed by different initiatives. I might have missed this in the paper, or the added material, but I think a way to examine the results of all evaluations of outcomes for project X would be a great benefit, including for policy		added sentence 'In addition it allows comparison of both evaluation methods and any impacts or outcomes in relation to standardised terminology.'			Comment incorporated into Beta

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	and future option parts of regional and global assessments, such as those produced by IPBES and the IPCC.					
26	Accurate and reliable data is of critical importance for all planned, collective human initiatives (see box 1). Whether we are implementing government policies, undertaking research, or developing new industry, better decisions improve outcomes – and the quality of our decisions is inextricably linked to the quality of the information we have at our disposal. The amount and quality of data we need to make good decisions is increasing exponentially as the problems we are trying to solve become increasingly complex, global and and cross-sectorial. This is reflected in the range of on-going efforts to mobilise, standardise and share data in a number of fields. However, what is currently lacking is a standardized way to share information about initiatives themselves. What was the objective, who were involved, who did which tasks, what methods were used, what was the governance structure, which results were achieved? Answers to all of these questions contain important data that could, and should, provide valuable insight and inform design and implementation of future initiatives. STARDIT' (Standardised Data on Initiatives) aims to address this gap by standardising and sharing data about human collective actions. STARDIT is a free open access data sharing system that works across fields, disciplines and languages. Data about initiatives can be submitted by anyone, and updated throughout the lifetime of an initiative, from planning to evaluation and reporting any impacts. The authors of the data can be verified, and the data assessed for quality. STARDIT is being developed on the understanding that the complex global problems we are facing today require evidence-informed collaborative methods, multidisciplinary research and interventions in which affected population groups are integrally involved in every stage. Among its main benefits (see box 2 for further information), STARDIT offers those carrying out research and interventions access to standardised information which enables well-founded comparisons of the effectiveness of different methods. Uniquely, STARDIT also enables sharing of information about stakeholder involvement in initiatives that works in many languages, using the Wikidata system. This article outlines how STARDIT works and how contributors from multiple disciplines and organisations globally might continue to be involved in the development of the current Beta Version.		Thank you for this fantastic plain english summary. I have incorporated some words. Noting the word limit I will save this plain english summary to try and incorporate more if possible.			comment noted for future versions
26	44-47: Lines 38-39 gives a very wide definition of what is understood by 'initiatives', but lines 44-47 can be read as only linking to one set of problems (i.e. complex global problems). Would it be possible to open this up slightly to stress from the very beginning the wide possible application and relevance of STARDIT? For instance: The authors of the data can be 45 verified, and the data assessed for quality, offering a potentially important source of high-quality 46 standardised information on initiatives trying to improve complex global problems (responses to which 47 transcend the capacity of any single discipline).		Thank you, changed plain english summary: 'There is currently no standardised way to share information across disciplines about initiatives, including fields such as health, environment, basic science, manufacturing, media and international development. All problems, including complex global problems such as air pollution and pandemics require reliable data sharing between disciplines in order to respond effectively.			Comment incorporated into Beta
26	59-63: Is distinctions the right word here? Maybe rewrite to something like "sectorial and disciplinary barriers can limit"?		changed to 'As we face increasingly complex problems, such as global air and water pollution, disciplinary and sectorial distinctions can limit our ability to respond effectively'			Comment incorporated into Beta
26	64-65: I would perhaps not lead with this focus on citizen science and participatory action research, but if we do it would probably be best to rewrite this part: "crucial methods to use to solve"		rewritten as 'Current reporting methods lack information about the ways in which different people are involved in initiatives, making it difficult to collate and appraise data about the most effective ways to involve different people. For example, forms of participatory action research where anyone can be involved in any aspect of research (including 'citizen science') are increasingly recognised as crucial paradigms for solving global problems, as they can help ensure that initiatives are aligned with the priorities of those affected, thus blurring the lines between concepts such as 'researcher', 'public', 'patient' and 'citizen'. '			Comment incorporated into Beta

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26	66-68: Does it also miss other things that STARDIT provides? Highlighting that here might help make the value added of STARDIT even clearer.		rewritten as 'Standardised data can inform effective ways to share power during the design, implementation and evaluation stage of initiatives. For example, when designing a response to an epidemic, standardised data can improve retrieval of relevant information which can be used to inform which affected individuals or organisations could be involved in the design of the response and which outcomes are most important.'			Comment incorporated into Beta
26	69-71: "Standardised data can inform effective ways to share power during the design…" sounds a bit strange to me – I understand what is meant, but it probably isn't quite correct (e.g. might need to specify which standardised data) and could probably be rewritten to make it more precise.		reworded to 'Standardised data can inform effective ways to plan, implement and evaluate all stages of initiatives. '			Comment incorporated into Beta
26	75-76: Is it possible to be more precise? Or, alternatively, sound more precise than "many kinds of data"?		reworded to 'STARDIT will enable multiple categories of data to be reported in a standardised way across disciplines, facilitating appraisal of initiatives and synthesising evidence for the most effective for people to be involved in initiatives. '			Comment incorporated into Beta
26	86-88: Would it be better of simply having an annex with a list of everyone that participated, and their affiliation, rather than highlighting a few organizations? It comes across as a bit strange to me (possibly because I do not understand these organizations are highlighted – but that will probably be the case for most readers).		good point. I added them for credibility but highlighting only a few is potentially sending the wrong message. removed.			Comment incorporated into Beta
26	95-98: This is so well established that it might be better to not mention any specific organization, but just to have a several of the most relevant references for it. Then you could also remove the "For example".		agreed. also reworded to 'Many problems facing life on earth transcend the capacity of any single discipline to address' so it's not so human specific in relation to the problems			Comment incorporated into Beta
26	100-102: The sentence starting with "A scientific…" seems to be a put on its head somehow, especially with the part saying "in many contexts". Maybe write: A scientific evidence-informed approach is often the most appropriate model for analysing the effectiveness of interventions.		Excellent pick up, done.			Comment incorporated into Beta
26	100-104: This para is a bit unclear to me – both the way it is phrased and its message. As I read it the message seems to be: 1. People need access to valid and reliable information. 2. Analysing the effectiveness of interventions typically requires an evidence-based approach. 3. This evidence-based approach goes under many names, including evaluation, international development, education or initiative. 4. We use the term initiative to refer to all of the above.		thank you - I have reworded as suggested.			Comment incorporated into Beta
26	109: The united Nations secretary-general stated that '…..		thank you, corrected			Comment incorporated into Beta
26	115: Is this wording a bit strong? Because the 'initiative'-category is so large it probably contains a lot of examples of initiatives where inclusion it is not strictly "essential"?		changed to 'It is often essential' noting probably better wording is 'always best practice' - but that's wordy			Comment incorporated into Beta
26	117: What does "international consensus statement" mean? Could it be spelled out or explained in the sentence?		now just called it a 'statement'			Comment incorporated into Beta
26	123-124: Would it be possible to clarify what is meant by "STARDIT can report any different 'interests' and ways of sharing power among different stakeholders"?		**I thought about this a lot and added this sentence with a ref: An interest can include a kind of commitment, goal, obligation, duty or sense of connection which relates to a particular social role, practice, profession, experience or medical diagnosis 31. I also tried to create a wikidata entry as I don't feel there is one currently: https://www.wikidata.org/wiki/Lexeme:L483913**			Comment incorporated into Beta
26	129-131: Substitute meanwhile with another word?		changed to 'Other examples include'			Comment incorporated into Beta
26	132: Convoluted sentence? Change to: Sharing data in a consistent manner may help ensure that benefits of initiatives are shared more equitably?		changed as suggested			Comment incorporated into Beta
26	141: Move "is", so that the sentence says: … a culture of partnership across disciplines and is, whenever possible, aligned and…."		changed as suggested			Comment incorporated into Beta
26	142: Specify that these are examples?		changed to 'such as those used in health, environment, manufacturing , publishing, government policy, education, arts and international development '			Comment incorporated into Beta

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26	143: Be specific about how it works across human languages or is this evident to all/most?		sentence reordered to 'The working Beta Version of STARDIT uses Wikidata to enable definitions to be co-created by contributors anywhere in the world, and therefore works across human languages, with interoperability with other platforms planned for future versions'			Comment incorporated into Beta
26	147: Sounds slightly underwhelming – could the benefits be highlighted better?		whole section rewritten			Comment incorporated into Beta
26	222-223: What exactly would this entail? Sounds very ambitious.		added example ' For example, STARDIT has already been used to map the varying perspectives of multiple stakeholders when planning a multi-generational cohort study 73'			Comment incorporated into Beta
26	234-235: How are they mapped and reported?		reworded to 'The participatory process used for developing STARDIT has attempted to be transparent about how different stakeholders have been involved in shaping it in order to improve how the system can be used to map values and provide more culturally neutral guidance for planning and evaluating involvement in initiatives. '			Comment incorporated into Beta
26	415: Change to: is expected to take at least 5 years, and will likely involve?		changed to 'amassing sufficient reports to create a useful database is estimated to take at least 5 years, and will likely require machine learning'			Comment incorporated into Beta
26	438-439: are is paramount		changed			Comment incorporated into Beta
26	440: in the future		changed			Comment incorporated into Beta
27	1. The current text uses terms such as “intellectual property”, “ownership”, “authorship”, and “license” in a way that is ambiguous, misleading, and internally contradictory. Unfortunately, if this critical issue is not clarified early on (i.e. now), it will create a ticking time-bomb that will go off at a later point during this project. I have been involved in open science initiatives for many years, co-founded a citizen science project, edited a guide on best practices for researchers, and received official certification from the Creative Commons organisation on copyright and licensing. In addition to my specific comments further below, please let me know if and how I can assist in fixing this problem to enable the wider sharing and implementation of STARDIT that it clearly deserves.		Any further support in correcting this would be much appreciated			
27	2. I respectfully take issue with the claim that STARDIT is useful for “any type of initiative, across any discipline” (line 278). Reading the text and tables (such as Tables 1 and 5), it seems that STARDIT is heavily informed by public health, citizen science, education, environmental, and/or international development initiatives. While commendable, they do not encompass “any discipline”. For example, would STARDIT be useful for an astrophysicist, chemical engineer, archaeologist, or historian? If so, how? If not, then the wording of “any initiative” or “any discipline” should be changed and constrained.			I would say that yes, STARDIT could be very useful for all those other diciplines and initiatives, although naturally the development so far has been by people from health and enviroment, we want to 'leave the door open' to all diciplines. While we have provided mulitple examlpes in the table of how it could be applied, I acknowledge this table is not exhaustive. I have however attempted to include data management (part of astrophysics, chemical engineering) and also cultural management in there too. I've yet to think of a collective human action that couldn't be explained with a STARDIT report, so I respectfully acknowlege this point and will keep the text as is.		comment noted but not incorporated into this version
27	3. Broadly speaking, I would like to see more details on how STARDIT will continue to evolve, adapt, and improve after version 1.0.		more information has been added to the supplementary section, noting that providing any specifics past version 1 would be inconsistent with the co-creation process - but the values which guide it would point towards many more versions we would hope.			Comment incorporated into Beta

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27	4. Exactly who is the intended audience of this paper? As described in the "Beta Version interface" section, substantial technical skills and knowledge (such as familiarity with the usage of APIs, data science, RDF structure, etc.) are needed to make full use of STARDIT. Therefore, I don't think literally anyone running an initiative can just jump in. Are there plans to make STARDIT more accessible and useable? If so, to whom and how?			"In short, yes I agree some of the language is very technical two parts acknowledge we need to be more inclusive (all, pending funding!) - ' and may require creating additional tools to create more inclusive ways of involving people in developing taxonomies.40' and ', ongoing co-design will be required to ensure STARDIT is as accessible and inclusive as possible. ' - which I hope are sufficient It's hard to answer who is the audience. In summary probably anyone with a professional role in planning or managing an initiative, in particular those with a focus on involving people/citizen science "		comment noted for future versions
27	5. There is terminology not accessible to a broad, non-technical audience. For example, Wikidata is not defined on its first use on line 144.		definition of wikidata included in objectives			comment noted for future versions
27	It is not clear what the "Preferred Reporting Items for Systematic Reviews and Meta-Analyses" is which should at least come with a citation.		citation added and comma added to clarify it's a definition of what is 'Future versions should be informed by a regular, systematic search, review and appraisal processes, using the Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) data set, used for reporting in systematic reviews and meta-analyses.'			comment noted for future versions
27	Importantly, the Internet Archive is mentioned several times without explaining what it really is and why it is used.		definition added and ref			comment noted for future versions
27	Also, ORCID is not clearly defined and will be confusing for a non-technical reader. These things should be clarified.			ORCID explained in full in table 4 and not used in text anywhere else in main body of paper		comment noted but not incorporated into this version
27	The document emphasised a desire to make STARDIT "always be open access". I wholeheartedly support the general sentiment, but the terminology and concepts employed throughout the text are unintentionally but highly misleading and sometimes contradictory: The term "public domain" is used many times throughout the text. "Public domain" has a specific legal definition meaning something that is without copyright. According to national laws and international agreements such as the Berne Convention with 179 signatories, copyright is automatically applied to any intellectual work at the moment of creation with no way to opt-out. Copyright gives the copyright holder monopoly privileges and powers to dictate how the copyrighted work is to be used. This automatically applies to STARDIT and any other information/material mentioned in the current document. Colloquially, "public domain" might not be used with such specific meaning, but in the context of this STARDIT Beta version manuscript – which explicitly mentions "open access" and licensing – it is imperative to use unambiguous language. There are too many uses of "public domain" throughout the current text, but I strongly suggest replacing those instances with "publicly accessible", "in public view", or "in the public" as appropriate (unless, of course, when the intention is to refer to the legally-defined term "public domain"). If additions have been made after the version at https://osf.io/w5xj6/, I suggest running a search-and-replace operation to find and remove ambiguous uses of "public domain".		This is incredibly helpful feedback. I've replaced all uses of 'public domain' with 'publicly accessible' and note that these terms have been used incorrectly in a colloquial way (as I'm not qualified in this area) and this terminology absolutely needs to be unambiguous.			Comment incorporated into Beta
27	Line 91 claims that the STARDIT beta version is released under "a Creative Commons license." There are six separate Creative Commons licenses with vastly different implications on how the licensed work can be shared and used. Please be clear about which one STARDIT uses. Is it the Creative Commons Attribution license, the Creative Commons Attribution-ShareAlike license, or another one?		Thank you for this detailed and valuable feedback. I think the entire STARDIT project should take further advice on this, but for now the most sensible decision I feel is going with a Creative Commons license and reviewing any next steps for version one - as			Comment incorporated into Beta

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	This point applies to other mentions of these licenses throughout the rest of the text. On that note, a license is granted by the copyright holder to others who wish to make use of a work. If something is in the public domain, then by definition no license can be applied to it. In addition, please avoid the ambiguous term “intellectual property” which has no specific definition. Generally speaking, “intellectual property” includes legal concepts such as copyright, patents, trademarks, trade secrets, among others. Text such as this manuscript or software code are mostly covered by copyright while physical designs like that for a machine are commonly dealt with through patents. Logos, such as the one proposed for STARDIT, are often handled through trademark registrations. Please be specific. To be clear, open access usually does not mean a lack of copyright and being in the public domain. Open access is very much enabled by using open licenses (such as among the six Creative Commons licenses) to expressly grant freedoms to share and reuse information while requiring attribution. On a more pedantic note, it is possible to use the CC0 Public Domain Dedication to explicitly relinquish all copyright associated with a work and it would still technically be considered open access. However, this also means that no attribution is necessary when the work is being shared and reused, so that might not be desirable in many cases. I recognise that lines 230-233 states the desire to avoid imposing a set of values on what constitutes “ownership”. However, STARDIT explicitly adopts the Creative Commons licenses which operate within a copyright regime that has (unfortunately) been imposed on almost the whole world. Therefore, I stress again that terminology around copyright and licensing be made clear and unambiguous.		this step gives us some control over usage of logo and other work associated with this project, without it being in danger of becoming a commercial commodity that could be 'bought out' as it were. My instinct is to go with this one (Attribution-NonCommercial-NoDerivs CC BY-NC-ND - https://creativecommons.org/licenses/by-nc-nd/3.0/au/) currently as I think not allowing anyone to change it would hopefully prevent the project 'forking' - centralising the control of what STARDIT is inside the steering committee (and any other subsequent governance processes decided upon) rather than having a free-for-all, which could work against the very thing we're trying to achieve (standardisation!). Multiple derivations of STARDIT would be a nightmare!			
27	Line 91 – Replace “a Creative Commons license” with “the Creative Commons x license” where x is the specific license that has been applied to STARDIT.		I have added information including this sentence 'STARDIT and all associated work and logos are currently licensed under Creative Commons license (Attribution-NonCommercial-NoDerivs CC BY-NC-ND), with the quality of any future iterations being the responsibility of not-for-profit host organisations and future licensing decisions to be made transparent, with anyone invited to be involved.' - note I did not add specific information to abstract for word count reasons			Comment incorporated into Beta
27	Table 1 sub-area “Coding and algorithms” – Replace “concepts of intellectual property and copyright” with “license information”.		changed to this, noting that copyright etc is not a universally accepted construct or way of working '(including concepts of intellectual property, copyright and license information, relevant blockchains and non-fungible tokens), evaluating knowledge translation, reporting impacts and outcomes			Comment incorporated into Beta

Respondent ID	Feedback with an action for the paper	General feedback about STARDIT	Change made	Lead author's response if no change made	Second Author Response	Status
27	Table 1 area “Information, media and cultural heritage” – Many initiatives also produce physical artefacts such as medical devices to treat malaria or a do-it-yourself solar cooker. These designs are also valuable information to be published. I suggest a sub-area titled “Hardware designs” with this “Relevant data categories” text: Reporting: Who created the designs, who reviewed them, what formats are the designs shared as and in what medium, information on license(s), outcomes and impact of the hardware.		agree this is a distinct category, added			Comment incorporated into Beta
27	Line 360 – Replace “public domain reports” with “publicly-viewable reports		done			Comment incorporated into Beta
27	• Line 366 – Replace “public domain URLs” with “public URLs”.		changed to 'publicly accessible URLs '			Comment incorporated into Beta
27	• Line 387 – Replace “will also be archived in the public domain” with “will also be archived in a publicly-accessible location online”.		done			Comment incorporated into Beta
27	• Line 393 – Replace “public domain sources” with “public sources”.		done			Comment incorporated into Beta
27	• Table 4 section “Initiative context” data category “Identifying information” – Replace “public domain URL with” with “URL”.		done			Comment incorporated into Beta
27	• Table 4 section “Initiative context” data category “Methods” – Replace “include a link to a public domain document” with “include a link to a published document”.		done			Comment incorporated into Beta
27	• Table 4 section “Report authorship” data category “Identifying information for each author” – Replace “public domain profiles” with “public profiles”.		done			Comment incorporated into Beta
27	• Table 4 section “Data” - Rename this section to “Data, software code, and hardware designs”.		changed to 'Data (including code, hardware designs or other relevant information)'			Comment incorporated into Beta
27	• Table 4 section “Data” data category “Ownership and control” – Replace “Who ‘owns’ the data or claims any kind of ‘intellectual property’ or rights (include relevant licensing information)” with “detailed licensing information”.		I've kept it plain english but added brackets 'Who ‘owns’ the data or claims any kind of ‘intellectual property’ or rights (include relevant licensing information)'			Comment incorporated into Beta
27	• Table 4 section “Data” data category “Ownership and control” – Replace “public domain URL” with “public URL”.		done			Comment incorporated into Beta
27	• Table 4 section “Information completed by Editoris” (misspelling of “Editors”?) - Replace “public domain URL” with “public URL”.		done and corrected, thank you			Comment incorporated into Beta
27	• Line 485 – Replace “shared in the public domain” with “publicly shared”.		done			Comment incorporated into Beta
27	• Page 28 section “Who is involved in STARDIT?” - Replace “all decisions made transparently and in the public domain” with “all decisions made transparently and in the public”.		changed to 'In plain English, anyone can get involved and have a say in how it should be designed and run, with all decisions made transparently and stored in a publicly accessible way.'			Comment incorporated into Beta

Respondee ID	Feedback with an action for the paper	General feedback about STARDIT	Change made	Lead author's response if no change made	Second Author Response	Status
27	• Page 31 “Additional values and paradigms” – This section mentions “STARDIT design and code should always be open access and relevant licenses should always be Creative Commons”. One of the six Creative Commons licenses must be specified here. The Creative Commons Attribution 4.0 or Creative Commons Attribution-ShareAlike 4.0 licenses are the most often used in open science. In addition, there is a separate set of licenses used for software code such as the GNU General Public License (GPL) 3.0 and that should be specified as well. It is legally highly problematic to apply any of the Creative Commons licenses to code and it should be avoided.		Changed to '• STARDIT designs and code should always be open access and relevant licenses should always be those which allow others to build on and improve the project, while maintain central control over quality (such as the Creative Commons Attribution-NonCommercial-NoDerivs CC BY-NC-ND license and the GNU General Public License (GPL) 3.0 for code)'			Comment incorporated into Beta
27	• Page 31 “Immutable values” – Replace “They will always be shared in the public domain” with “They will always be shared publicly”.		changed to 'While these values will evolve, we will keep an immutable record of our values. They will always be shared via a publicly accessible URL and regularly archived on the ‘Internet Archive’ for future reference 120.'			Comment incorporated into Beta
27	• Page 44 line 17-20 (under “Data ownership and hosting” section) – Here it states a STARDIT report cannot contain any information that is not already in the public domain. If the current authors are referring to the legal term “public domain”, then this will be almost impossible. For example, the vast majority of open access scientific peer-reviewed publications are under copyright and released under one of the Creative Commons licenses (usually the Creative Commons Attribution license). By definition they are not in the public domain and the information they contain cannot be included in a STARDIT report. In addition, “to avoid ‘intellectual property’ issues” is highly ambiguous and misleading. If the current authors are not referring to “public domain” in the legal sense of the term, the I suggest replacing the first sentence of this section with “To reduce data sharing barriers and encourage reuse, a STARDIT report cannot contain any proprietary information that is not open access”.		Thank you - does this work? 'To reduce data sharing barriers and encourage reuse, a STARDIT report cannot currently contain any proprietary information that is not open access or publicly accessible, except for information volunteered by the report authors (such as institutional email addresses), much like a ‘corresponding author’ on a peer reviewed paper. '			Comment incorporated into Beta

Respondee ID	Feedback with an action for the paper	General feedback about STARDIT	Change made	Lead author's response if no change made	Second Author Response	Status
27	• Page 47 lines 110-115 – The current paragraph implies that Science for All will hold the trademarks for the STARDIT logos and the copyright over the STARDIT specification, this manuscript, and other outputs from the participatory action research process. Is this the case (e.g. did Science for All register the trademark for the logo)? If so, please be specific. Also, if the intent is for Science for All to hold the copyrights stated above, then that implies the authors do not. Is this also intentional? The copyright holder(s) and author(s) are not always the same people. I am happy to suggest edits to this important paragraph but cannot do so without more specific knowledge of what the intent is.		Thank you - working backwards, what I'm trying to achieve is a stepping stone stage - the end goal being STARDIT set up formally as its own thing - but for now it needs a host organisation, which is Science for All (as I'm Director, so it makes things simple). In the future, if it's another charitable/not for profit organisation which takes this over, great (e.g. 'Standardised Data International - STARDIT'). What I'm trying to prevent is a situation where someone uses the STARDIT name or logo and we have no legal way of preventing it (e.g used for commercial purposes or an extreme political group). So I think for now, for all practical purposes, Science for All will be the copyright holder until a better solution is found. I'm really no expert on this so legal advice would be very welcome Changed to 'For the purposes of concepts of intellectual property (including trademarks), and to protect STARDIT from being used by people in ways which are outside of the values defined in this document, any intellectual property (including logos or code associated with STARDIT) are currently owned by the charity Science for All, which is currently hosting the participatory action research process to create and manage STARDIT. Any decisions relating to the above (including transfer of ownership of any intellectual property) are to be made by the STARDIT Steering Committee, which hosted by Science for All, but independent of Science for All. In the future ownership may be transferred to an appropriate organisation established specifically for the purpose of owning such intellectual property, for example, establishing a charity called 'Standardised Data International – STARDIT).'			Comment incorporated into Beta
27	• Table 5 – The "Access" column in this table makes a distinction between "public domain" and "open access". This is confusing because material in the public domain counts as open access.		corrected as either Publicly accessible website or Open access			Comment incorporated into Beta
27	• Citizen science is briefly mentioned in the abstract but not defined or elaborated on in the text. I suggest just removing the mention of it in the abstract. Otherwise it needs to be defined in the text with relevant citations such as, but not limited to: Auerbach, J., Barthelmess, E. L., Cavalier, D., Cooper, C. B., Fenyk, H., Haklay, M., Hulbert, J. M., Kyba, C. C. M., Larson, L. R., Lewandowski, E., & Shanley, L. (2019). The problem with delineating narrow criteria for citizen science. Proceedings of the National Academy of Sciences, 116(31), 15336–15337. https://doi.org/10.1073/pnas.1909278116 Silvertown, J. (2009). A new dawn for citizen science. Trends in Ecology & Evolution, 24(9), 467–471. https://doi.org/10.1016/j.tree.2009.03.017		the term 'citizen science' is used in inverted commas in the abstract, and there's no space for explanation - I think the term is familiar enough to be used in this context in the abstract, although a more full explanation is now included in the background section with added refs			Comment incorporated into Beta
27	• Line 415-416 (page 25 in Discussion section) – Why and how might machine learning be applied to STARDIT data? Without elaborating more this feels like a throwaway sentence.		added extra sentence and ref 'STARDIT seeks to be an easy-to-use way for people from multiple disciplines to share data about initiatives. However, amassing sufficient reports to create a useful database is estimated to take at least 5 years, and will likely require machine learning. For example, adversarial machine learning may be used in parallel with humans (for verifying data) to generate STARDIT reports from existing publicly accessible data at a scale and speed impossible for humans alone to achieve. '			Comment incorporated into Beta

Respondee ID	Feedback with an action for the paper	General feedback about STARDIT	Change made	Lead author's response if no change made	Second Author Response	Status
27	• Lines 97-100 on page 46 section "How does STARDIT work within 'law' and 'lore?" (missing ' after lore?) - In addition to indigenous peoples, there are entire sovereign states that are not recognised by the United Nations. So, if "STARDIT conduct will be guided by the United Nations on all matters of law", how will citizens of non-UN-recognised states be heard, represented, and included?		Thank you - added this 'STARDIT will always defer to United Nations declarations in favour of any sovereign laws, and does not recognise itself as a legal entity which is acting in any one sovereign state. Citizens of non-UN-recognised states will be recognised as individual legal persons and included equally alongside any other person, regardless of status (including citizens, residents, asylum seekers and refugees). STARDIT is an initiative to support individuals to self-organise ways of sharing information, and it is the responsibility of each individual (including individuals working on behalf of organisations) to act both within the values of STARDIT, and any laws to which they may be subject. '			Comment incorporated into Beta

Change log from Alpha Version

Section	Change	Lead Author comment (Jack)	Lead author's response if no change made (Jack)	Second Author's response (Thomas)
Plain English summary	Improved first para in line with feedback			
	Made it clearer in the phases section that initial audiences might need targeting	Although I like this concept, in practice it won't be "anyone" that is submitting or bothering to standardise data. The people you are ultimately trying to target are researchers, NGOs, governments etc., so maybe worth mentioning who the specific target audiences are (including the encouragement of citizen scientists). With that in mind, it is worth considering that those people (like me!) will not be experts in standardising jargon so it may be good to be as specific as possible (using examples) throughout. Also, make it clear how STARDIT will run alongside government and academic outputs.	I think this is a wider point about STARDIT that is touched on later in the development phases and also a point about messaging and audience targeting, which I think is something to address for version One	
	Added use case for NGOs in table 1 examples of use and added this ref: https://www.brookings.edu/wp-content/uploads/2016/06/11_development_aid_kharas.pdf	Agreed, it says all those carrying out research and interventions, which covers what I say next. It may be worth highlighting somewhere in the paper that another really important factor is that it provides information to various development aid agencies (governments, NGOs etc.) about what initiatives has and are being done, which can save precious time and money for regions that can ill afford to have limited aid budgets blown on projects already done elsewhere.		
		The three main issues with aid architecture include: poor information sharing, coordination and planning; no info on results effectiveness and aid allocation rules that lead to a limited ability to scale up, learning and innovation. STARDITs design can help with all these issues! See Page 17 of this highlights this issue:https://www.brookings.edu/wp-content/uploads/2016/06/11_development_aid_kharas.pdf I am not saying that this should be mentioned here, but it might be worth adding as a benefit somewhere else? I know from my experience in the Pacific, there was so much money being wasted everywhere - with aid money being splashed everywhere on projects that often duplicated each other. No one had any real idea who was doing what in the Pacific? Often we would hear about a project that was closely matched to our own, but could never access documents, info etc. Different countries involved. With STARDIT there could be greater information sharing, leading to greater coordination and planning and improvements from access to info on outcomes e.g., use lessons learnt and increase the likelihood of greater success in aid projects.		
	have added 'biodiversity loss' at later point, as habitat loss is a jargon term (some people might think it means places for humans to live) but this point is about how health and enviroment are interlinked	...global air pollution and habitat loss,		
	Attempted to work in more examples	Also, examples throughout would be really helpful.		
		If a project has access to STARDIT would that have any benefits? E.g. lowered costs, improved results? Can we put some numbers/values in to add strength to the initiative?	I think this is a great idea, but beyond my capacity and expertise for the Beta version and certainly a good	

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			thing to do for version one	
	Done	Could this be framed positively as "By reporting data in a standardised way, essential information generated can provide opportunities for collaboration and comparison"		
	revised sentence strcuture but kept 'citizen science' in inverted commas as it requires defining, and using it outside commas suggests it has a fixed meaning the reader should understand, which often it doesn't.	Why is citizen science in inverted commas? Is it to suggest it is still a developing science, not yet established? Not saying it is, the inverted commas infer that.		
	clarified wording	Maybe clarify what it means to "involve people": involve people in research? Involve people in action-planning? Also, ultimately standardising data saves time/money and results in more accessible evidence-based action planning (whether that is by governments or NGOs etc.).		
		See my previous comments above. Really like the opening paragraphs, but now it seems STARDIT is aimed at 'governments, industry, research organisations and people around the world'. Needs to be clarified earlier who STARDIT is for, and how it will work alongside/in place of existing publication platforms.	the audience is 'everyone', have attempted to make use cases for different audiences clearer in the table of examlpe use cases	
		Who will do this? Will it be peer-reviewed? How is quality in the database assured? Will it have a Wikipedia model of self-review?	This is covered in detail in additional resources	
Background	added reference	This is also similar to realist evaluation, good to make this link here or later		
	excellent. Included	I know the UN and data is mentioned above but seeing that STARDIT is about multiple sectors & disciplines and that Climate Change is the biggest threat humanity faces, it might also be worth mentioning somewhere an environmental body (i.e., not just health and economics) that also outlines the importance that information and knowledge sharing has to overcoming this challenge. This could be included further down where you mention the benefits of knowledge sharing. For example, the Paris Agreement highlighted the critical role of Information and Knowledge Management to Climate Change Adaptation in Article 7 that Parties "strengthen cooperation and enhance action" to share information, experience and lessons; strengthen institutional arrangements; strengthen scientific knowledge; assist developing countries to identify effective adaptation actions and improve effectiveness of adaptation actions (UNFCCC 2015). From: UNFCCC (2015) Paris Agreement. FCCC/CP/2015/10/Add.1.1_32. See: http://unfccc.int/files/essential_background/convention/application/pdf/english_paris_agreement.pdf		
	added references to GBIF and references about the limitations of such platforms	how about adding something about previous successful initiatives for standardizing data. you could also include current trends like GBIF which have a similar rationale and also open data/ open source initiatives.		
	Thank you, noted. Vulnerable has a very specific meaning in certain contexts (for example, vulneralbe adults in the mental health context) but whether this term should be applied to whole populations is a good question. I would prefer soemthing like 'at greater risk of exploitation'. Minority is problematic, as in what frame of reference is someone in a minority, and who is doing that framing? Also, very rich people are in a minority, but are not 'vulnerable'. Lots of lethal euphemisms here!	There has been criticism of this term recently eg. https://www.tandfonline.com/doi/abs/10.1080/09581596.2019.1656800?journalCode=ccph20 Perhaps "minority"?		
	added comments about data access and citations.	1. Could it be appropriate to also add in a sentence about data ownership? For example, which stakeholders/projects are more likely to share data? Comment that we need to encourage stakeholders (that own the data) and educate on the importance of sharing data to enable others to take control over their lives/decisions etc.		

Section	Change	Lead Author comment (Jack)	Lead author's response if no change made (Jack)	Second Author's response (Thomas)
		2. Agree this would be important to define/describe before the STARDIT part and to touch on opportunities for community data ownership, sovereignty		
Objective	covered later in paper but added 'with interoperability with other platforms planned for future versions.'	How could STARDIT continue if Wikidata suddenly disappeared? Suggest describing how STARDIT is held on a platform that enables accessible data sharing and that Wikidata is currently used		
Current usage		1. There is a growing call for scientific journals to make reviewers and editors anonymous comments and reviews part of the official scientific record. A few journals are taking it up, but resistance is there and uptake is slow, STARDIT could help to build a stronger case for it. See: Polka et al. (2018). Publish peer reviews. Nature: https://www.nature.com/articles/d41586-018-06032-w. I am all for it to help improve transparency and help with better public perceptions of science. Although it is worth factoring in that asking reviewers to consent to being identified to the author has no important effect on the quality of the review, the recommendation regarding publication, or the time taken to review, but it significantly increased the likelihood of reviewers declining to review. See: Van Rooyen, S., Godlee, F., Evans, S., Black, N. and Smith, R., 1999. Effect of open peer review on quality of reviews and on reviewers' recommendations: a randomised trial. Bmj, 318(7175), pp.23-27. link: https://www.bmj.com/content/bmj/318/7175/23.full.pdf?casa_token=y5JgdYfuEIcAAAAA:RsmsQAgdYlUglSnUQ-JIQHfIEVQnJA2yEwHmLIKkK5F-KSyBKrHCavVYKOd9YIgUx6c-WeZltsmo1A 2. I agree with Roan's comments. I'm all for publishing the reviewer comments, as this will give much better transparency. However, if you don't make them anonymous you are going to find it very difficult to find people to review articles, and people will start to be less critical, especially when the community is small. It's already incredibly difficult to find experts with free time to review.	I think this is an important issue (open peer review) but one that is outside the scope of this paper. In other words, open peer review is already happending (and in fact is used by the Wiki Journals and the journal where this will be submitted) and in most cases where I've been part of open peer review process you can chose to be anonysmous or not. Regardless of the benefits or cons - the point is that STARDIT can be used to report information about peer review process - it's not in itself an open peer review process. How the editorial process works for future STARDIT reports is also an open discussion. Comments flagged with Thomas for his input as he has superior expertise in this area	Partial solutions also involve embargoing comments and/or reviewer identities
	added more examples with references	This is great! Could we mention an example to explain the benefit of STARDIT? E.g. What new information was shared though STARDIT that wasn't published by the journal (assuming this is where it was published)? How could this information be useful to future initiatives?		

Section	Change	Lead Author comment (Jack)	Lead author's response if no change made (Jack)	Second Author's response (Thomas)
Potential applications		This application could be mentioned earlier as a way of describing the benefit of STARDIT, e.g. 'one of the advantages of standardizing data reporting is that comparisons between methods and impacts across multiple initiatives can easily be made.'	mentioned in abstract - repitition if mentioned in introduction?	
	Added this para with references 'In addition, STARDIT could be used to share information which makes research more reproducible43,44, improving accessibility to the information required to critically appraise research and evidence and thus improving trust in processes such as the scientific method45,46, and facilitate an appraisal of different knowledge systems, including Indigenous knowledge systems47.	STARDIT might also help with the scourge of irreproducible scientific studies. Tens of billions of dollars are wasted each year on irreproducible research, where insufficient information on methodology and data prevents many studies from being reproduced. With 14% of scientists reporting that they have witnessed scientific fraud (Fanelli 2009), there is a push for more stringent demands of proof from investigators/ scientists (Clark 2017) e.g., video taping of experiments and full raw data. STARDIT could facilitate this. Examples of the issue, see: Clark, T.D., 2017. Science, lies and video-taped experiments. Nature News, 542(7640), p.139. Link: https://www.nature.com/news/science-lies-and-video-taped-experiments-1.21432 AND Fanelli, D., 2009. How many scientists fabricate and falsify research? A systematic review and meta-analysis of survey data. PloS one, 4(5), p.e5738. link: https://journals.plos.org/plosone/article?id=10.1371/journal.pone.0005738		
		there is also an opportunity to improve our overall understanding of social and environmental problems by embracing the strengths and weaknesses of the different knowledge systems (UNESCO 2017) UNESCO, 2017: Local knowledge, global goals. Local and Indigenous Knowledge Systems Programme Rep., UNESCO, 48 pp., http://unesdoc.unesco.org/images/0025/002595/259599E.pdf		
		...also help to improve public trust in scientific method and outcomes. e.g., https://www.nature.com/articles/nbt1199supp2_14 and https://journals.sagepub.com/doi/pdf/10.1177/0963662519869097		
	Added in this sentence: Such data sharing could also improve the translation of trusted, quality research and data, by empowering people to both access and appraise relevant data. For example, improved access to more standardised information (in multiple languages) about data and outcomes, could help to facilitate more informed collaborations between researchers and those monitoring and protecting critically endangered species48–50.	For a multidisciplinary environmental benefit that STARDIT could also help with: As with health and other disciplines, there is a big issue with a lack of knowledge translation between scientists and conservation managers (see Linklater 2003for an example). With the ongoing biodiversity crisis (Ripple et al. 2017), having improved access to more standardised shared project information, data and outcomes, could help to facilitate a better understanding of the quality of what work has been done and lead to more informed collaborations between managers and researchers, perhaps to more applied projects on conservation management focussed research for improved decision making when managing critically endangered species. An example of how a lack of standardised info and data can lead to poor outcomes, rhino conservation managers had to rely on several non-standardised home range study sources for black rhino, incorrectly interpreting the impact of the non-standardised studies, and initiated actions that had significant detrimental impacts for a key black rhino population (See Plotz et al. 2016). References: Linklater, W.L., 2003. Science and management in a conservation crisis: a case study with rhinoceros. Conservation Biology, 17(4), pp.968-975.Doi: https://doi.org/10.1046/j.1523-1739.2003.01449.x Plotz, R.D., Grecian, W.J., Kerley, G.I. and Linklater, W.L., 2016. Standardising home range studies for improved management of the critically endangered black rhinoceros. PLoS One, 11(3), p.e0150571. Doi: https://doi.org/10.1371/journal.pone.0150571 William J. Ripple, Christopher Wolf, Thomas M. Newsome, Mauro Galetti, Mohammed Alamgir, Eileen Crist, Mahmoud I. Mahmoud, William F. Laurance, 15,364 scientist signatories from 184 countries, World Scientists' Warning to Humanity: A Second Notice, BioScience, Volume 67, Issue 12, December 2017, Pages 1026–1028, https://doi.org/10.1093/biosci/bix125		
	made it clearer that sometimes meanings are vague and that we are therefore defining what they mean in this article: While meanings of these terms are often imprecise and can be used interchangeably, 'involvement' here is distinct from 'engagement', which is where which information and knowledge about initiatives is shared, for example, with study participants who remain passive recipients of interventions.54	Not sure this is the best example, engagement is surely more than just being a study participant with no input or contribution. Agree involvement is a more active term than engagement		

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	Added 'see Table X in appendix for examples of data standards which could be incorporated' as there's a big list in what is currently table 5	Such as? As someone who comes from a very niche background, it would be useful to know what other established data standards can/are used.		
	added 'Development has also been influenced by existing work in health research, including the multidisciplinary area of public health, which incorporates social, environmental and economic research. '	and say that this can be applied across disciplines? Public health is already multidisciplinary, incorporating economics, environment, social etc.		
	added ref	Another example of community ownership and TEK principles in WA: https://researchonline.jcu.edu.au/24751/1/ES-2012-5165.pdf		
	added ref	also relevant is O'Donnell, E.L. and Talbot-Jones, J., 2018. Creating legal rights for rivers. Ecology and Society, 23(1). http://www.ecologyandsociety.org/vol23/iss1/art7/; https://www.routledge.com/Legal-Rights-for-Rivers-Competition-Collaboration-and-Water-Governance/ODonnell/p/book/9780367584160		
	added 'In addition, ongoing co-design will be required to ensure STARDIT is as accessible and inclusive as possible. '	I think this is such an important paragraph! Also, the article touches on this, but are we not excluding different types of people/cultures already by asking them to fill in these reports? I am just thinking of people I work with in Indonesia and China, many of whom, would not respond well to a wordy document/guidelines and would need strong benefits to encourage them to use STARDIT (which goes back to one of my original points).	This is a very important point, and one raised by an aboriginal community member too, it comes down to accebility and if there's training in place. I've mentioned this in limitations but also, frankly, it's a limiation of ALL kinds of publishing and any online tool. If anything, being able to complete it in other langauges makes it more accecible but ongoing co-creation is needed to keep improving it (including training)	
	added '	Is this the first mention of Science for All? Maybe some more information on who they are, their values and objectives?		
Version One implementation	changed to 'Once STARDIT Beta (version 0.2) has been **submitted** for publication, work will begin on the next version, (version 1.0). '	Perhaps confidence in the current version should enable version 1.0 work to commence regardless of publication status. The journal may not want to publish an interim version		
	I have added 'more detail in appendicies'	would be useful to describe the make-up of the working group, in categories e.g. researcher, citizen scientists or as a figure?	I'm keen to keep it brief in the main body and also open ended (not too prescriptive) but I have added 'more detail in appendicies'	
		How did Wikipedia become so universally adopted? Any lessons or examples there we can use to improve adoption and reporting into STARDIT?		

Section	Change	Lead Author comment (Jack)	Lead author's response if no change made (Jack)	Second Author's response (Thomas)
		How did Wikipedia become so universally adopted? Any lessons or examples there we can use to improve adoption and reporting into STARDIT?	I think this is outside of the scope of the paper but the short answer from me would be transparent governance and editirial processes, which we've emulated with STARDIT	
Scope and applications		Agreed. As an fyi, there is a regional information and knowledge management system called iCLIM. It is not across any discipline but many of the principles align with STARDIT's objectives. The Pacific iCLIM Project aims to enable better climate change resilience and adaptation planning in the Pacific by improving the discoverability, storage, access, and utilisation of climate change data and information. For more info see here: https://www.griffith.edu.au/research/research-excellence/griffith-climate-change-response-program/pacific-iclim and a situation analysis here: https://www.redicomar.com/wp-content/uploads/2019/12/Pacific-Situation-Analysis_Pacific-iCLIM-Project-2019.pdf		
Table 1: Example applications of STARDIT		The Table is great. In the first two pages of it, there is a lot of repetition of categories that overlap across all the different Areas & sub-areas (e.g., people affected/ involved; process for deciding and measuring outcomes; experts involved; in most cases 'funding'). Just a suggestion: could all the similar categories that occur within and across all the areas/ sub-areas be collated in a separate table to say - 'these categories will be recorded as standard across all areas, and then have this table show any additional categories that are uniquely/ specifically recorded for each of these different application areas. It might make it easier for the reader to understand what is being recorded for their area of interest and make more meaningful comparisons. As it is now, I found it a bit hard to absorb all the various categories relevant to specific areas with all the repetition. Also, an added benefit once all the similar/standard categories across areas are evaluated and collated, allows easier comparisons to evaluate whether any of the categories currently listed should actually be recorded for that area or if a category currently not considered in area should actually be recorded and vice versa. The added complication is that this is an evolving tool and categories that are currently listed within an area might shift, and others likely to be added and removed over time - we could add an appropriate caveat to indicate that.	I think this is a great idea but I just didn't have the time to do this right now. Perhaps this could be something we work on for future version when trying to communicate it to dofferent audiences?	
		important to engage with Indigenous people around this content, hope this has occurred through Poche Centre engagement but if not yet, suggest engaging directly with Boe	Thank you - yes, STARDIT presented at multiple Poche Centre meetings, and feedback invited and incorporated	
	Thank you - incorporated and added refs	Data custodianship' might cover this but might be worth mentioning whether the initiative followed any local, national and international legislation and policies that might be in place to govern the way in which TK is collected, documented, and stored. When projects involve TK, there is a need to be aware of any legal frameworks that may apply, including those designed to protect cultural and intellectual property (IP). This can be in the form of national policies and Acts and /or local cultural restrictions. This will vary across the globe. In short, have initiatives documenting Traditional and Indigenous Knowledge considered the following key issues: (1) legal and national contexts; (2) prior informed consent; (3) cultural restrictions; and (4) IP rights. For example, the use of prior informed consent is part of a best practice approach. In the Pacific, we provided information to participants about the project, including project purpose, who		

Section	Change	Lead Author comment (Jack)	Lead author's response if no change made (Jack)	Second Author's response (Thomas)
		was involved, methods of collection and dissemination and consent was asked, including the level of sensitivity of the information e.g., low =publicly available, medium = known only to community or knowledge holder and can only be shared with requested permission from the knowledge holders; High sensitivity (spiritual info/ customary laws) = to remain with knowledge holder and within database. For specific example see: Malsale, P., Sanau, N., Tofaeono, T.I., Kavisi, Z., Willy, A., Mitiepo, R., Lui, S., Chambers, L.E. and Plotz, R.D., 2018. Protocols and partnerships for engaging Pacific Island communities in the collection and use of traditional climate knowledge. Bulletin of the American Meteorological Society, 99(12), pp.2471-2489. DOI: DOI: https://doi.org/10.1175/BAMS-D-17-0163.1		
		I think a measure of gender representation/ inclusiveness in initiatives involving IK needs specific consideration here? Indigenous Knowledge, and use, of environmental information can vary and are often segregated according to gender. In many IK projects and outputs the female voice is absent. It is therefore important to consider gender inclusiveness when collecting IK information, particularly as the impacts of environmental variability can impact genders differently in many remote and vulnerable regions. E.g., Balakrishnan, R., 1998: The Pacific. Rural Women and Food Security: Current Situation and Perspectives, Food and Agriculture Organization of the United Nations, 50–60, https://www.fao.org/3/W8376E/w8376e05.htm. AND Lane, R., and R. J. G. McNaught, 2009: Building gendered approaches to adaptation in the Pacific. Gend. Dev., 17, 67–80, https://doi.org/10.1080/13552070802696920.		
		There is an example of a database for Traditional Knowledge forecast indicators in the Pacific Islands that was designed with such added levels of security to culturally sensitive information, through limiting access according to restrictions imposed by the TK expert/ community who initially provided the information. For info see: Chambers, L.E., Plotz, R.D., Dossis, T., Hiriasia, D.H., Malsale, P., Martin, D.J., Mitiepo, R., Tahera, K. and Tofaeono, T.I., 2017. A database for traditional knowledge of weather and climate in the Pacific. Meteorological Applications, 24(3), pp.491-502. doi: https://doi.org/10.1002/met.1648 The TK Database needed to enable restrictions based on things such as: user's membership of a clan or tribe; user's status/role within the tribe; user's gender, and the context in which the resource will be reused or reproduced etc.		
	added	ethics important to clarify here, with additional ethics requirements for research involving Indigenous people		
	Thank you - incorporated and added refs	I have said some of this before in another comment here. e.g., free, prior and informed consent. Also worth considering sui generis systems based upon customary law (ref: https://mckinneylaw.iu.edu/iiclr/pdf/vol17p67.pdf). For example, this is straight from from UNESCO 2017 Indigenous Knowledge document: "Many communities are calling for the protection of their knowledge from inappropriate use, emphasising the need for free, prior and informed consent and benefit sharing. Existing intellectual property regimes are ill-adapted to indigenous knowledge. More appropriate methods are being developed, such as sui generis systems based upon customary law. There is also knowledge that the community may want to keep for themselves (e.g. locations of sacred groves and preferred harvesting areas). It is important to understand the different types of knowledge, and both the individual's and community's right to control access. " UNESCO, 2017: Local knowledge, global goals. Local and Indigenous Knowledge Systems Programme Rep., UNESCO, 48 pp., http://unesdoc.unesco.org/images/0025/002595/259599E.pdf✎ EditSign		
	done - good suggestion	Add a referenced definition? e.g. UK MRC guidance?		

Section	Change	Lead Author comment (Jack)	Lead author's response if no change made (Jack)	Second Author's response (Thomas)
	added	Indigenous knowledge might need a caveat here, due to cultural sensitivity and ownership issues limiting full immediate transparency.		
Mapping preferences for involvement	added to objectives	I think it would be good to touch on/briefly introduce this tool earlier on e.g in the aims		
	added	Reference missing.		
	Done - and changed strucuture of discussion	Highlight more STARDIT strengths in these early paragraphs e.g. the strength of co-design in its development, interdisciplinary nature etc.		
	changed to 'estimated' - there's no preceedant really, but GRIPP2 is quite old now and one review says it's not very well used	How or on what basis is 5-years deemed the likely time-frame? Any precedence or examples to compare -e.g., wikijournal?		

Standardised Data on Initiatives (STARDIT) Beta Version: Additional File 1

About this document

This document contains additional information relevant to the article 'Standardised Data on Initiatives (STARDIT) Beta Version', which published and is available at this link: https://doi.org/10.1186/s40900-022-00363-9

A direct link to Additional File 1 can be found here: https://static-content.springer.com/esm/art%3A10.1186%2Fs40900-022-00363-9/MediaObjects/40900_2022_363_MOESM1_ESM.pdf

Results

Appendices for Chapter 4 – Additional file: Public Involvement in Global Genomics Research: A Scoping Review

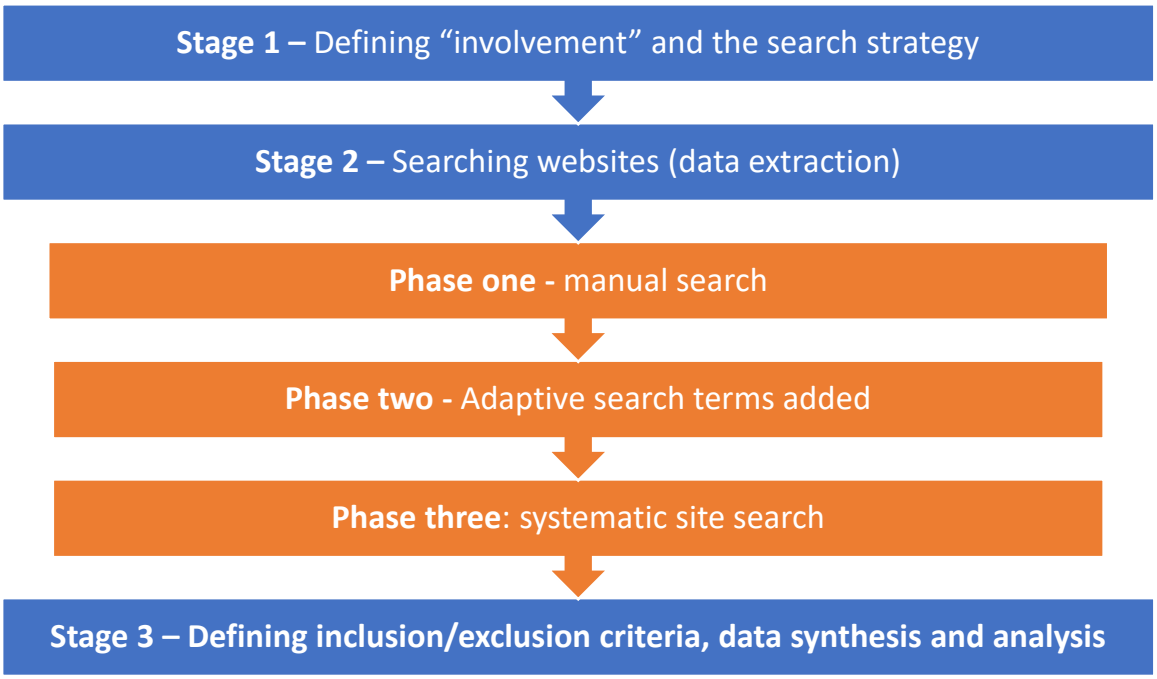
Additional file 1: GA4GH data and results

A direct link to Additional File 1 can be found here:
https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_1_xlsx/octet-stream/Table%201.XLSX/1/446268

Additional file 2: Systematically searching sites for 'public involvement' and related concepts

A direct link to Additional File 2 can be found here:
https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_2_docx/octet-stream/Table%202.docx/4/446268

Using the search strategy described below, the public domain websites of all the included initiatives in the GA4GH database were searched for reports of involvement and any impacts. This document describes Stage 1 and Stage 2 of the scoping review in more detail. The graphic below visually represents the different phases at each stage of the review.


Stage 1 – Defining "involvement" and the search strategy
Stage 2 – Searching websites (data extraction)
Phase one - manual search
Phase two - Adaptive search terms added
Phase three: systematic site search
Stage 3 – Defining inclusion/exclusion criteria, data synthesis and analysis

30
31

Stage 1 – Defining "involvement" and the search strategy

We conducted a narrative review of systematic reviews to inform our search terms. In addition we used similar studies and existing frameworks to inform the word list [35,36,597–600,37,78,79,91,351,358,363,371]. We developed a criteria to define 'involvement' based on the International Association for Public Participation's participation spectrum and other studies[36,184,349,351]. The final list was checked by the research team after being tested. Below are the search terms included in the main search.

Main search terms to describe 'involvement'

An asterisk (*) denotes that all possible grammatical variations of the nouns used to describe people involved.

- Involvement (involv*)
- Engagement (engag*)
- Partnering (partner*)

Main search terms to describe people involved

An asterisk (*) denotes that all possible grammatical variations of the nouns used to describe people involved.

- public*
- communit*
- consumer*
- patient*
- stakeholder
- user*
- citizen*
- Lay ('people' included to exclude phrases such as 'involved laying down')
- Patient
- PPI (an acronym commonly used in the UK which stands for 'patient and public involvement')

Stage 2 – Searching websites (data extraction)

Public domain websites of all the initiatives in the GA4GH database were searched for reports of involvement and associated impacts.

Phase one: manual search

Public domain websites of all initiatives in the GA4GH database were manually searched for reports of involvement and associated impacts.

Phase two: Adaptive search terms added

During the manual search, adaptive (context dependent) search terms are added to phrase generation table. The adaptive search terms which were added to the phrase generation table were:

- lay*
- carer*
- volunteer*
- population* [358]
- group*
- residents (geographical grouping) [91]
- participa* (context dependant)

- representative
- payer*
- taxpayer*
- customer* [363]
- client* [363]
- advocate*
- civil societ*

When any of these terms returned a result that was within the inclusion criteria, the exact search string was recorded.

Phase three: systematic site search

Phrase-generation table

Commercial search engines like Google do not allow an asterisk (*) to denote all variations in grammatical endings, so searching for all possible variations must be done manually. Search engines like Google do allow the 'OR' operator (up to 35 words per search), meaning multiple variations can be searched at once. Once the list of words was finalised, the variations were generated in order to systematically create search strings. This checklist was used to ensure all possible verb and noun forms variations were used.

Verb forms

- Base (root) form or infinitive (involve, engage, partner)
- Active or plural form (involves, engages, partners), used as the present indicative in the third-person singular
- Past tense (involved, engaged, partnered)
- present participle, gerund, verbal and deverbal nouns (involving, engaging, partnering)

Nouns

- Singular (involvement, engagement, partnership)
- Plural (involvements*, engagements*, partnerships)

*Not found in prototype searches

The table below demonstrates this process for systematically combining the various words to describe people involved with the terms used to describe involvement.

106

Term for 'the public'	Involvement	Engagement	Partnering
public*	"public involvement" OR "public involved" OR "involving public" OR "involves public" OR "involving the public"	"public engagement" OR "public engaged" OR "engaged public" OR "engaging the public"	"public partner" OR "public partners" OR "public partnership" OR "public partnerships" OR "public partnering" OR "partnering with the public"
communit*	"community involvement" OR "communities involved" OR "involving communities" OR "involved communities" OR "involve communities" OR "involves the community"	"community engagement" OR "community engaged" OR "engaged community" OR "community engages" OR "engaged community" OR "engaging the community" OR "community engaging"	"community partner" OR "community partners" OR "community partnership" OR "community partnerships" OR "community partnering" OR "community partnered" OR "partnering with the community"
consumer*	"consumer involvement" OR "consumers involved" OR "involving consumers" OR "involved consumers" OR "involve consumers" OR "involves consumers"	"consumer engagement" OR "consumers engaged" OR "engaged consumer" OR "engaged consumers" OR "consumer engages" OR "engaged consumer" OR "engaging the consumer" OR "engaging the consumers" OR "consumer engaging"	"consumer partner" OR "consumer partners" OR "consumer partnership" OR "consumer partnerships" OR "consumer partnering" OR "consumers partnered" OR "partnering with the consumers"
patient	"patient involvement" OR "patients involved" OR "involving patients" OR "involved patients" OR "involve patients" OR "involves patients"	"patient engagement" OR "patients engaged" OR "engaged patient" OR "engaged patients" OR "patient engages" OR "engaged patient" OR "engaging the patient" OR "engaging the patients" OR "patients engaging"	"patient partner" OR "patient partners" OR "patient partnership" OR "patient partnerships" OR "patient partnering" OR "patients partnered" OR "partnering with the patients"

Term for 'the public'	Involvement	Engagement	Partnering
stakeholder	"stakeholder involvement" OR "stakeholder involved" OR "involving stakeholders" OR "involved stakeholders" OR "involve stakeholders" OR "involves stakeholders"	"stakeholder engagement" OR "stakeholders engaged" OR "engaged stakeholder" OR "engaged stakeholders" OR "stakeholders engage" OR "stakeholder engages" OR "engaged stakeholder" OR "engaging the stakeholder" OR "engaging the stakeholder" OR "stakeholders engaging"	"stakeholder partner" OR "stakeholder partners" OR "stakeholder partnership" OR "stakeholder partnerships" OR "stakeholder partnering" OR "stakeholder partnered" OR "partnering with the stakeholders"
user*	"user involvement" OR "user involved" OR "involving users" OR "involved users" OR "involve users" OR "involves users"	"user engagement" OR "user engaged" OR "engaged user" OR "engaged users" OR "users engage" OR "user engages" OR "engaged users" OR "engaging the users" OR "engaging the user" OR "user engaging"	"user partner" OR "user partners" OR "user partnership" OR "user partnerships" OR "user partnering" OR "user partnered" OR "partnering with the users"
citizen*	"citizen involvement" OR "citizen involved" OR "involving citizens" OR "involved citizens" OR "involve citizens" OR "involves citizens"	"citizen engagement" OR "citizen engaged" OR "engaged citizen" OR "engaged citizens" OR "citizens engage" OR "citizen engages" OR "engaged citizens" OR "engaging the citizens" OR "engaging the citizen" OR "citizen engaging	"citizen partner" OR "citizen partners" OR "citizen partnership" OR "citizen partnerships" OR "citizen partnering" OR "citizen partnered" OR "partnering with the citizens"
Lay ('people' included to exlude phrases such as 'involved laying down')	"lay involvement" OR "lay involved" OR "involving lay people" OR "involved lay people" OR "involve lay people" OR "involves lay people"	"lay engagement" OR "lay engaged" OR "engaged lay" OR "engaged lay" OR "lay engage" OR "lay engages" OR "engaged lay" OR "engaging the lay" OR	"lay partner" OR "lay partners" OR "lay partnership" OR "lay partnerships" OR "lay partnering" OR "lay partnered" OR "partnering with lay"

Term for 'the public'	Involvement	Engagement	Partnering
		"engaging the lay" OR "lay engaging"	
Public and/& patient Patient and& Public "PPI involvement" included as it is often misused	"public and patient involvement" OR "patient and public involvement" or "involving patients and the public" OR "involving the public and patients" OR "ppi involvement"	"public and patient engagement" OR "patient and public engagement" or "engaging patients and the public" OR "engaging the public and patients" OR "ppi engagement"	"public and patient partnerships" OR "patient and public partnerships" or "partnering with patients and the public" OR "partnering with the public and patients" OR "ppi partnerships" OR "ppi partnering"

107

108 The words 'dialogue', 'consultation' and variations of 'co-production' are sometimes used to describe
109 involving people, so these terms were also searched for.

Dialogue	Consult	co-
"public dialogue" OR "public dialogues" OR "dialogue with the public"	"public consultation"	"co-production" OR "co-produced" OR "co-created" OR "co-designed" OR "co-design"

110 *Systematic site search*

111 Once the manual search was completed, websites were searched using the 'site search' function on the
112 Google search engine. This method relies on Google servers having carried out a 'website crawl', where
113 data from the website is indexed [601]. While this method cannot be called 'exhaustive', it is an
114 appropriate sampling technique for this scoping review. In addition, the limitations of the Google 'site
115 search' function (with regards to Boolean operators and a maximum character limit of 35 characters per
116 search) were partially overcome by the 'phrase generation table' which was used by authors to manually
117 create an exhaustive list of search operators.

118 Once the search string was generated in the phrase generation table it was entered into the Google
119 search engine. For example, this search string returned 4 results:

120 *site:www.ukbiobank.ac.uk/* "public involvement" OR "public involved" OR "involving public" OR
121 "involves public" OR "involving the public"

122 If a term such as 'patient' was commonly used on a site (and thus had more than 10 pages of search
123 results) 'intext' operators were used to refine the search to only return page results where another
124 specific term appears. For example:

125 *site:www.irdirc.org patient intext:participation*

126 When search strings returned a result, these were logged in the extraction document.

Appendices for Chapter 5 - Additional files: Involving elderly research participants in the co-design of a future multi-generational cohort study

Additional File 1: Data and Analysis

About this document

This document contains additional data relevant to the case study 'Involving elderly research participants in the co-design of a future multi-generational cohort study'. In addition it contains a more detailed description of the data sources in this case study. This document includes the preferences mapping data (STARDIT-PM), and other data about this initiative[196]. The corresponding Standardised Data on Initiatives Alpha Version (STARDIT) of the report can be found in Additional File 5.

A version of this file can be found online here: https://doi.org/10.1186/s40900-021-00271-4

A direct link to the file is here: https://static-content.springer.com/esm/art%3A10.1186%2Fs40900-021-00271-4/MediaObjects/40900_2021_271_MOESM1_ESM.pdf

Data mapping

This table summarises all the data sources used for the case study.

Data Category	Data point description
Diary	ASPREE research diary – including reflections
ASPREE Newsletter	Advert in the newsletter for all participants asking for those 'interested in providing input on the design of possible future genetic, family and multi-generational studies' to get in touch
Meeting records	Meetings with the study team, including notes, audio recordings and relevant emails
Email discussions	Emails about updated versions of the questionnaire May 2018 and June 2018 Email discussion with study team members
Reports on progress	Interview reports from MS – June 2018
Interview participant initial feedback	Interviews with participants asking for feedback on questionnaire design changes
Meeting about interviews	Discussion with MS and JN based on interviews conducted by mid-June
Interview data	Interview recordings (audio and PDF notes) Interview response highlights identified by MS
Interview summary	Interview contents are summarised in a spreadsheet by MS
Meeting about event	Study team meeting about event, informed by interviews – August 2018
Interview	Email interview with MS about interviews, including early identification of themes and learning points
Event planning feedback	Feedback from participant advisor on event facilitation plan – August 2018
Pre and post event information and questions	Information and questions sent to participants before and after the event

Data Category	Data point description
Facilitation plan and relevant reflections	The final facilitation plan and relevant reflections in JN's research diary
Event recording	Audio Video of event Short video interviews with 4 event participants
Participant feedback about event	Participant feedback about the event, including feedback forms
Email summaries of event	Email summaries about event
Notes from event	Notes from the event by MS, PL, BH and JN
Meeting notes	Meeting notes from discussion with PL and BH
Email discussions	Emails to study team after event about survey – mid September
Discussion about newsletter	Feedback on newsletter by study team members – late September
Final newsletter	Final newsletter sent out to event participants and other people interviewed
Budget of involvement	Budget documents
Study team interviews	Interviews carried out by email with study team members 6 months after the event - March 2019

Advert in Winter/Spring 2017 newsletter

CALL OUT FOR INPUT ON GENETIC FAMILY AND MULTIGENERATIONAL STUDIES

ASPREE has provided an opportunity to investigate factors affecting health and well-being in older people. What is not understood is how factors affecting health, such as lifestyle and genes, can run in families and across generations.

Your thoughts on how to best undertake future studies related to the genetics, health and well-being of families are most welcome.

If you are an ASPREE participant (or have a family member) who is interested in providing input on the design of possible future genetic, family and multi-generational studies, please register your interest by email: aspree@monash.edu or ring 1800 728 745 (toll free from a landline).

Upon receipt of your expression of interest, you may be contacted by a member of the Public Health Genomics Program at Monash University, who works closely with ASPREE on current genomic sub-studies such as the Resilience Project.

Your contribution to the design of genetic family studies can be as little or as much as you wish and you can cease involvement at any time.

Budget

The budget of the process is itemised below.

Item	Cost ($AUD)
Room hire and food for event	1000
Staff time (estimated)	9000
Total	10000

149
150

Study team survey

The following questions were emailed to the study team members six months after the event:

1. Please describe your tasks in the process of involving people in planning of the new ASPREE multi-generational study

2. What did you learn from the process of involving ASPREE participants in the research planning phase?

3. Please describe specifically what worked well or was useful about the way people were involved

4. Please describe specifically what did not work well or was not useful about the way people were involved

5. Were there any barriers or facilitators to conducting the involvement activities? (institutional or otherwise)

6. Do you think the involvement activity achieved its intended aim(s)?

7. Do you have any advice to other researchers planning participant involvement for their research?

8. Describe the impact you think involving people had (positive/negative - on the research, staff or participants)

9. Who do you think should influence the kind of human genomic research done in the future, and why? (e.g. the public, participants of research studies, doctors, school children, politicians etc)

10. Which stages of future genomic research should be influenced by people other than researchers (if any)? (e.g. concept planning of new studies, study design, conducting the research, presenting the results etc)

11. Other comments

STARDIT Preference Mapping (STARDIT-PM) – Alpha Version

This table uses the Alpha version of the Standardised Data on Initiatives Preference Mapping (STARDIT-PM) to categorise the data into certain areas[196].

Area (quantitative data about responses in this area)	Qualitative summary of participants' views and perspectives	Qualitative summary of study team's views and perspectives
Views on who should be involved: 50% (10/20) of interview participants, 100% (18/18) of event participants and three (75%, 3/4) study team members shared a view or perspective about this area.	Two participants noted that the purpose of involving people needed to be clear in order to avoid 'wasting time'. One participant felt only researchers should be involved as they are 'the qualified people', two others stated participants should be involved as researchers only 'see it from their point of view and nobody else's', and participants bring 'new perspectives'. One participant mentioned 'vested interests' and suggested involving participants was a way of overcoming this. Transparency from the project about who is involved (specific professions) might help participants identify 'different directions'. Event participants were unanimous that there was no aspect of the research that they should not be involved in. One participant stated that 'funding' decisions may be better being made by experts, although participants agreed they should be involved in the oversight of research funding.	One study team member (an ASPREE participant assessor) reflected that the 'increased autonomy' of involving other staff equally (rather than just senior research staff) made them feel valued and gave the opportunity to 'think creatively' and 'engage in controversial or difficult discussions'.
Views on who should do which tasks:	Participants stated they should be involved in research design. A participant commented that feedback is needed from participants. Another participant suggested that	The lead investigator stated that participant involvement "significantly improves the researchers' ability to make sound decisions

Area (quantitative data about responses in this area)	Qualitative summary of participants' views and perspectives	Qualitative summary of study team's views and perspectives
25% (5/20) of interview participants, 100% (18/18) of event participants and two (50%, 2/4) study team members shared a view or perspective about this area.	participant information can be confusing and that a layperson can have the task of simplifying it. A third participant expressed a willingness to be involved as long as the task had purpose and was not 'just for the sake of chatting'. 10 event participants said they would be interested in being involved in recruitment and communication, 7 were willing to be involved in data access decisions and 2 in ethical decisions.	regarding the fundamental research questions, study design, ethics and funding applications". The participant advisor stated ,'I believe that researchers should have the dominant responsibility to plan and complete genomic research'.
Views on modes of communication: 65% (13/20) of interview participants, 100% (18/18) of event participants and 25% (1/4) of study team members shared a view or perspective about this area.	There was much variation in views and perspectives about communication mode. For example, some participants stated a preference for face-to-face discussion, while others preferred online questionnaires, commenting on documents online or joining online text-based discussion groups. Participants reported perceived advantages and disadvantages for each communication mode. Two thirds of event participants said that they would be happy to be involved both face-to-face and online (using computers and smartphones). Event participants felt certain tasks (such as reviewing information) could be done 'more online', and that face-to-face meetings were helpful when there was an 'occasional need'. Online text-based discussions were stated to have advantages by 'opening up more discussion' as it gave a chance for people to reflect on other participants' views and perspectives, meaning discussion could be more in-depth.	Referring to the face-to-face event, one study team member stated, 'participants really enjoyed the opportunity to be heard and put their views forward'. After the event, the lead investigator noted that the planned research 'must use mobile/internet technology'.

Area (quantitative data about responses in this area)	Qualitative summary of participants' views and perspectives	Qualitative summary of study team's views and perspectives
Views on what methods should be used: 15% (3/20) of interview participants, 100% (18/18) of event participants and all (100%, 4/4) study team members shared a view or perspective about this area.	One participant stated that they did not feel comfortable being part of a face-to-face group, while another described a method of being involved which was a small group conversation with a researcher leading a discussion to gather views and ideas. Another participant suggested having information sent out which could be read, with participants providing feedback. Event participants spontaneously suggested using an online discussion platform and shared views on what is good moderation and the advantages of online discussion, although some shared concerns about for-profit social media platforms being used.	After the face-to-face event, most of the study team members felt that dividing the event discussion into small groups facilitated discussion and gave more people a chance to share views and perspectives. One study team member felt that asking focussed questions and requesting a show of hands was a time-efficient way to gauge perspectives.
Views on facilitators of involvement: 15% (3/20) of interview participants, 100% (18/18) of event participants and 100% (4/4) of study team members shared a view or perspective about this area.	Giving people early notice of events and clear advice about the purpose and expectations were identified as important by two participants. One participant identified 'personality' as distinct from skills and knowledge – which could be considered a facilitator if managed appropriately. 100% of event participants felt that if they were involved in recruiting participants from their immediate family, a short explanatory video would be helpful and improve their confidence in explaining the study. When asked about support, two event participants identified it as helpful having a person act as an independent facilitator when involved in working in groups	One study team member felt education was essential, and that assuming a limited knowledge of a subject and explaining the basic concepts at the start of the event was important, as this appeared to support people to make informed decisions when contributing to group discussions. Another study team member stated that adequate funding for involvement was required so that it can become 'a requirement, rather than a luxury'. Having a lead investigator 'who valued the unique experiences of each team member and participants' was identified as a facilitator by one study team member.

Area (quantitative data about responses in this area)	Qualitative summary of participants' views and perspectives	Qualitative summary of study team's views and perspectives
	on tasks such as ethical oversight (either face-to-face or online).	
Views on barriers of involvement: 40% (8/20) of interview participants and 50% (2/4) of the study team shared a view or perspective about this area.	Living in rural areas and other travel logistics were considered a barrier to participation in face-to-face events by a number of participants. A lack of clarity about expected time-commitments or timing of events was identified by four participants. Not having the skills or knowledge was identified as a barrier. One participant felt they lacked literacy in using computers and online tools.	One study team member observed after the event that they perceived participants from 'professional backgrounds' dominating group discussions, which may have inhibited others. Another study team member stated the 'cost' of involvement in terms of time and financial commitment might be a barrier for some research projects.
Views on what the outcome or output of the research or involvement in research could be: 15% (3/20) of interview participants, 22% (4/18) event participants and 100% of study team members (4/4) shared a view or perspective about this area.	Participants raised the issue of wanting to know outcomes and outputs of involvement, with one seeking clarity on what the purpose of involvement was. Participants shared many views about the outcomes of research and felt being involved in clarifying the aims of the future study was important. Involving participants in helping answer what the research 'hoped to achieve' was an outcome identified by one participant. It was stated that 'responses from participants could cause the experts to ask new questions' or lead the research in 'different directions'. One event participant said that her preference was not to participate in research which was 'about the aggrandisement of the professor', while another stated	All members of the study team thought the involvement process achieved the intended aims, and that process had a positive impact. None reported negative impacts. One study team member stated that involving participants could help researchers make decisions about 'fundamental research questions, study design, ethics and funding applications'.

Area (quantitative data about responses in this area)	Qualitative summary of participants' views and perspectives	Qualitative summary of study team's views and perspectives
	that the most important thing to 'get right' was having clear study aims with regard to what it is trying to achieve. Interview participants suggested that their motivation for participating was altruistic, in the hope that the research would contribute to positive outcomes for future generations.	
Views on which stage of the research people should be involved: 10% (2/20) of interview participants, 100% (18/18) of event participants and 100% (4/4) of study team members shared a view or perspective about this area.	Most participants agreed that people other than researchers should be involved in research design, including designing the research question. A participant commented that feedback is needed from participants.	One study team member stated that 'participant involvement is vital, especially in the early stages' of research, with 'less involvement' needed in executing the study, collecting data and analysing results. Participants could then be more involved in the 'publishing and communication' of results.
Research data: 0% (0/20) of interview participants, 100% of event participants (18/18) and 25% of study team members (1/4) shared a view or perspective about this area.	Seven event participants said they would be interested in being involved in decisions about data access. 100% of event participants were comfortable with their data being held by academics. 100% were not comfortable with it being held by a for-profit company, although one participant said not to 'rule private companies out completely'. All event participants were interested in having pharmacogenomic results returned. Two thirds of event participants wanted access to their own genomic data, and had mixed views about who else should have access. GPs were generally trusted to access and interpret genomic data, but participants felt GPs shouldn't have access to data that they did not. All but 2 participants agreed they should have access to their own data, with those disagreeing mentioning cognitive decline as a reason for a co-managed model. Some participants had concerns about them or their relatives (especially offspring) finding	During the event, the lead investigator noted that while participants overwhelmingly wanted 'a self-managed future of health information', this was 'at odds' with the current healthcare professional managed information paradigm.

Area (quantitative data about responses in this area)	Qualitative summary of participants' views and perspectives	Qualitative summary of study team's views and perspectives
	out information they 'might not want to know'. Questions about duty of disclosure and how this might affect 'employability' were also asked, with participants seeking clarity about how these issues would affect the research design regarding return of data.	

Additional File 2: Telephone Questionnaire: Version Comparison - A comparison of the versions of the telephone questionnaire used

This file is available at this DOI: https://doi.org/10.1186/s40900-021-00271-4

This is a direct link to the file: https://static-content.springer.com/esm/art%3A10.1186%2Fs40900-021-00271-4/MediaObjects/40900_2021_271_MOESM2_ESM.xlsx

Additional File 3: Quantitative results - A summary of the quantitative results

This file is available at this DOI: https://doi.org/10.1186/s40900-021-00271-4

This is a direct link to the file: https://static-content.springer.com/esm/art%3A10.1186%2Fs40900-021-00271-4/MediaObjects/40900_2021_271_MOESM3_ESM.xlsx

Additional File 4 - GRIPP2 report for 'Involving elderly research participants in the co-design of a future multi-generational cohort study'

This report has been completed using the 'GRIPP2 reporting checklists: tools to improve reporting of patient and public involvement in research' available at: https://doi.org/10.1136/bmj.j3453.

GRIPP2 short form

Category	Category description	Data
1a: Aim	Report the aim of the study	Participatory action research to involve elderly research participants in the co-design of a proposed multi-generational cohort study, in order to improve research design, relevance, acceptability and recruitment.
1b: Methods	Describe the methods used by which patients and the public were involved	Participatory action research to involve elderly research participants in the co-design of a proposed multi-generational cohort study.
1c: Results	Report the impacts and outcomes of PPI in the study	Improved participant information resources, improved wording that is culturally appropriate, improved question design for interviews, improved learning resources for participants, improved co-design process.
1d: Conclusions	Summarise the main conclusions of the study	Involving participants in co-designing a proposed study resulted in changes to the design of the proposed study The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participant and study team members' views about the value of involvement, which can be viewed as an impact of 'transformative learning'.
1e: Keywords	Include PPI, "patient and public involvement," or alternative terms as keywords	Public Health; Epidemiology; Preventive Medicine; Medical Ethics; Medical Education & Training; public involvement; participatory research; genomics; patient involvement;

Category	Category description	Data
2a: Definition	Report the definition of PPI used in the study and how it links to comparable studies	The words 'involvement' or 'being involved' describe the concept of people being 'involved' in research. This is when research is carried out 'with' people rather than 'on' them. 'Involvement' can also be defined as when other people aside from the research team, such as the public, patients, research participants and other stakeholders, actively contribute to the research process. It is the 'active involvement' in shaping and guiding research, rather than only providing data.
2b: Theoretical underpinnings	Report the theoretical rationale and any theoretical influences relating to PPI in the study	The process was guided by a number of international participatory action research methodology frameworks, including the International Collaboration for Participatory Health Research and INVOLVE guidance on co-design. An Alpha version of 'Standardised Data on Initiatives' (STARDIT) was used to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. STARDIT was then used to guide co-design of the process, and to subsequently report how people were involved, using standardised data.
2c: Concepts and theory development	Report any conceptual or theoretical models, or influences, used in the study	We used a case study research methodology to record and describe the process of involving participants in the co-design. The process was guided by a number of international participatory action research methodology frameworks, including the International Collaboration for Participatory Health Research and INVOLVE guidance on co-design. An Alpha version of 'Standardised Data on Initiatives' (STARDIT) was used to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. STARDIT was then used to guide co-design of the process, and to subsequently report how people were involved, using standardised data.
3: Aim	Report the aim of the study	Participatory action research to involve elderly research participants in the co-design of a proposed multi-generational cohort study, in order to improve research design, relevance, acceptability and recruitment.
4a: Design	Provide a clear description of methods by which patients and the public were involved	The study team held four meetings to co-design the involvement activities. One participant advisor was involved in a number of tasks including reviewing and improving the written information, telephone interview questions, and the facilitation plan for the event. After the recruitment and consent process, participants were interviewed by telephone. Participants were asked about their willingness to provide feedback throughout the study, and to be involved in study design, as well as preferences for modes of communication.

Category	Category description	Data
		Eighteen participants attended a four-hour workshop event in central Melbourne. The event was co-designed by the study team, and was informed by interview data and international best-practices for involvement events.
4b: People involved	Provide a description of patients, carers, and the public involved with the PPI activity in the study	3 academic research investigators An ASPREE participant assessor An ASPREE-XT participant
4c: Stages of involvement	Report on how PPI is used at different stages of the study	Stage 1: Planning The study team held four meetings to co-design the involvement activities. One participant advisor was involved in a number of tasks including reviewing and improving the written information, telephone interview questions, and the facilitation plan for the event. Stage 2: Recruitment and telephone interviews An advert was placed in a newsletter to 14,268 ASPREE participants. After the recruitment and consent process, participants were interviewed by telephone. Participants were asked about their willingness to provide feedback throughout the study, and to be involved in study design, as well as preferences for modes of communication. The definition of involvement below used in the script was co-designed with participants for subsequent interviewees. Stage 3: Event Eighteen participants attended a four-hour workshop event in central Melbourne. The event was co-designed by the study team, and was informed by interview data and international best-practices for involvement events. The event included an introduction to the proposed MGRS by the lead ASPREE-XT genomics researcher (PL); a plain-English introduction to genomics by an expert in genomics who is also an ASPREE-XT participant (BH); a summary of the telephone interview results by the interviewer (MS); and an

Category	Category description	Data
		interactive session which included open questions about the types of information participants would like returned and recruitment of family members. The final session included a presentation and interactive discussion about involvement in research, led by the event facilitator (Jack Nunn). This session explored preferences about how people would like to be involved, with open and closed questions. Questions included preferences about tasks and modes of communication. Throughout the event, participants shared their views on a range of issues through interactive discussions, voting (by show of hands) and anonymous written feedback. Stage 4: Evaluation and analysis Members of the study team were surveyed six months after the face-to-face event in order to integrate the valuable views and perspectives of those involved in co-designing and delivering the process. Design of surveys was informed by frameworks for planning and reporting public involvement (GRIPP2 and PiiAF). The study team were asked 11 questions and the data from the four interviews was coded and categorised using Standardised Data on Initiatives (STARDIT). The stages of qualitative data analysis included data mapping and familiarisation; transcription; coding; searching for themes; reviewing themes with study team members (including a participant representative); labelling and summarising themes; and reporting the findings. In order to enhance validity of the analysis, two authors independently analysed the data thematically, which was then checked by a third author (triangulation). Standardised categories (STARDIT) were used during content analysis of the data in order to facilitate comparison with other research projects. More information about the data sources and a STARDIT report available.
4d: Level or nature of involvement	Report the level or nature of PPI used at various stages of the study	Participants were involved at every level of every stage, with more information in section 4C. Everyone listed in 4B was involved in co-designing every stage of the process. This included refining wording of participant information, sharing views and advice about the process, proof-reading documents, providing feedback on questionnaires, analysing data, informing planning, presenting information to participants, interpreting data, and participating in email surveys.

Category	Category description	Data
5a: Qualitative evidence of impact	If applicable, report the methods used to qualitatively explore the impact of PPI in the study	The stages of qualitative data analysis included data mapping and familiarisation; transcription; coding; searching for themes; reviewing themes with study team members; labelling and summarising themes; and reporting the findings. In order to enhance validity of the analysis, two authors independently analysed the data thematically, which was then checked by a third author (triangulation). Standardised categories (STARDIT) were used during content analysis of the data in order to facilitate comparison with other research projects. Involving stakeholders in the co-design process impacted the study in seven specific impacts ways. By asking for participants' views on aspects of the proposed study design, the study team gained insight into participant preferences and opinions. While there was diversity in views, the process allowed the study team to improve aspects of the study design.
5b: Quantitative evidence of impact	If applicable, report the methods used to quantitatively measure or assess the impact of PPI	Twenty relevant interviews were transcribed, coded and categorised, with relevant interviews identified by two investigators independently. To reduce any unconscious selection bias, a sample of over 10% of the interviews was selected at random.
5c: Robustness of measure	If applicable, report the rigour of the method used to capture or measure the impact of PPI	We used the 'Standardised Data on Initiatives (STARDIT)' Alpha Version to plan and report how participant involvement activities positively impacted the study design. STARDIT includes a tool to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. STARDIT was then used to guide co-design of the process, and to subsequently report how people were involved, using standardised data. An Alpha version of the STARDIT framework was also used in parallel with the thematic analysis to organise data into pre-defined 'super-categories' which allow consistent comparison with other data using this reporting framework.
6: Economic assessment	If applicable, report the method used for an economic assessment of PPI	The entire process of involving people was estimated to cost $10,000 AUD, including staff time, catering and event venue hire. The value of the process was summarised by the lead investigator who stated "I learnt a lot from the process and am very glad we made the effort".
7a: Outcomes of PPI	Report the results of PPI in the study, including both	Improved participant information resources, improved wording that is culturally appropriate, improved question design for interviews, improved learning resources for participants, improved co-design process.

Category	Category description	Data
	positive and negative outcomes	
7b: Impacts of PPI	Report the positive and negative impacts that PPI has had on the research, the individuals involved (including patients and researchers), and wider impacts	Involving stakeholders in the co-design process impacted the study in seven specific impacts ways. By asking for participants' views on aspects of the proposed study design, the study team gained insight into participant preferences and opinions. While there was diversity in views, the process allowed the study team to improve aspects of the study design. 1: Recruitment and sample collection Recruitment and consent for the MGRS will occur online wherever possible, and salvia samples (rather than blood) will sent by post to be used as biospecimens for DNA analysis. 2: Participant communication A short video and 'information pack', which will explain the MGRS study, will be created to assist with recruiting family members. 3: Participant involvement in governance Participants will be invited to be involved in overseeing governance, including funding decisions. 4: Data access Study participants should be involved in controlling data access decisions and policies. 5: Communication and ways of involving participants Participants will be included on study advisory groups, including for study recruitment and communication, data access and ethical oversight using multiple communication modes. 6: Provide feedback to participants about the research Participants will be informed about the impact of the research, and how their involvement has affected the design and management of the study. 7: Create learning and development opportunities

Category	Category description	Data
		Learning and development opportunities will be created for potential participants, researchers and other stakeholders.
7c: Context of PPI	Report the influence of any contextual factors that enabled or hindered the process or impact of PPI	The process took longer than expected. There is confusion over what ethics approval is required in order to involve people, especially people who are participants in an ongoing study. Involving field staff (as well as senior researchers and academics) provided a valuable perspective, as some staff knew some participants personally and had knowledge that senior research staff did not. Some study team members worried about over-burdening participants by asking them to do too much, however this concern did not seem to be backed up by the data collected, and may be considered a barrier to involvement. Enablers of involvement - Giving people time to read resources. Clear communication about the intention of involving people. Barriers of involvement - Face-to-face meetings were difficult to organise. Some participants were elderly or lived in remote areas, so face-to-face meetings needed to be minimised where possible.
7d: Process of PPI	Report the influence of any process factors, that enabled or hindered the impact of PPI	The process took longer than expected. There is confusion over what ethics approval is required in order to involve people, especially people who are participants in an ongoing study. Involving field staff (as well as senior researchers and academics) provided a valuable perspective, as some staff knew some participants personally and had knowledge that senior research staff did not. Some study team members worried about over-burdening participants by asking them to do too much, however this concern did not seem to be backed up by the data collected, and may be considered a barrier to involvement.

Category	Category description	Data
7ei: Theory development	Report any conceptual or theoretical development in PPI that have emerged	The effective involvement of 'stakeholders' also includes involving all relevant staff and health professionals at all levels of an initiative, who may have important knowledge or perspectives that senior research staff do not. In other words, the 'PPI' label for this question does not incorporate all relevant stakeholders who are not described by this acronym. The participatory action research method gave insights into participants' preferences that measurably impacted on the proposed study design. The improvement of the interview design using the co-design process illustrates the value of a flexible and iterative approach to involvement in a study. By asking participants their preferences, the study team gained useful insights to inform the design of the proposed study. Participants preference for being involved in decision making about funding sources, data management and ownership, and what information to share with participants will help ensure any future study design aligns with participants' values, ensuring the design is culturally safe and culturally competent
7eii: Theory development	Report evaluation of theoretical models, if any	The 'transformative learning' during the process reported from both study participants and the study team was an important impact captured by the participatory action research (PAR) method. The process showed that it was valuable to create regular involvement opportunities for each stakeholder. Reporting this process in a standardised way using 'Standardised Data on Initiatives' (STARDIT) meant that impacts such as transformative learning could be reported and that this case study can be compared to similar studies in the future.
7f: Measurement	If applicable, report all aspects of instrument development and testing (eg, validity, reliability, feasibility, acceptability, responsiveness, interpretability, appropriateness, precision)	'Standardised Data on Initiatives' (STARDIT)was used to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. STARDIT was then used to guide co-design of the process, and to subsequently report how people were involved, using standardised data. An Alpha version of the STARDIT framework was also used in parallel with the thematic analysis to organise data into pre-defined 'super-categories' which allow consistent comparison with other data using this reporting framework.
7 g: Economic assessment	Report any information on the costs or benefit of PPI	The entire process of involving people was estimated to cost $10,000 AUD, including staff time, catering and event venue hire. The value of the process was summarised by the lead investigator who stated "I learnt a lot from the process and am very glad we made the effort".

Category	Category description	Data
8a: Outcomes	Comment on how PPI influenced the study overall. Describe positive and negative effects	No negative impacts were reported from any participants or study team members at any stage of the process. Involving stakeholders in the co-design process impacted the study in seven specific impacts ways. By asking for participants' views on aspects of the proposed study design, the study team gained insight into participant preferences and opinions. While there was diversity in views, the process allowed the study team to improve aspects of the study design. Outcomes included Improved participant information resources, improved wording that is culturally appropriate, improved question design for interviews, improved learning resources for participants, improved co-design process.
8b: Impacts	Comment on the different impacts of PPI identified in this study and how they contribute to new knowledge	Involving stakeholders in the co-design process impacted the study in seven specific impacts ways. By asking for participants' views on aspects of the proposed study design, the study team gained insight into participant preferences and opinions. While there was diversity in views, the process allowed the study team to improve aspects of the study design. 1: Recruitment and sample collection Recruitment and consent for the MGRS will occur online wherever possible, and salvia samples (rather than blood) will sent by post to be used as biospecimens for DNA analysis. 2: Participant communication A short video and 'information pack', which will explain the MGRS study, will be created to assist with recruiting family members. 3: Participant involvement in governance Participants will be invited to be involved in overseeing governance, including funding decisions. 4: Data access Study participants should be involved in controlling data access decisions and policies. 5: Communication and ways of involving participants Participants will be included on study advisory groups, including for study recruitment and communication, data access and ethical oversight using multiple communication modes.

Category	Category description	Data
		6: Provide feedback to participants about the research Participants will be informed about the impact of the research, and how their involvement has affected the design and management of the study. 7: Create learning and development opportunities Learning and development opportunities will be created for potential participants, researchers and other stakeholders.
8c: Definition	Comment on the definition of PPI used (reported in the Background section) and whether or not you would suggest any changes	The acronym 'patient public involvement' here is limiting, as it does not incorporate research participants and other stakeholders such as study staff, who might have unique insights into study design. The words 'involvement' or 'being involved' describe the concept of people being 'involved' in research. This is when research is carried out 'with' people rather than 'on' them. 'Involvement' can also be defined as when other people aside from the research team, such as the public, patients, research participants and other stakeholders, actively contribute to the research process. It is the 'active involvement' in shaping and guiding research, rather than only providing data.
8d: Theoretical underpinnings	Comment on any way your study adds to the theoretical development of PPI	An Alpha version of 'Standardised Data on Initiatives' (STARDIT) was used to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. STARDIT was then used to guide co-design of the process, and to subsequently report how people were involved, using standardised data. STARDIT includes a tool to map people's preferences for involvement in a standardised way, including mapping views on who should be involved and how. An Alpha version of the STARDIT framework was also used in parallel with the thematic analysis to organise data into pre-defined 'super-categories' which allow consistent comparison with other data using this reporting framework. Creating consistency in terminology to describe the 'who', 'what' and 'how' of involvement allows better comparison.
8e: Context	Comment on how context factors influenced PPI in the study	The process took longer than expected. There is confusion over what ethics approval is required in order to involve people, especially people who are participants in an ongoing study. Involving field staff (as well as senior researchers and academics) provided a valuable perspective, as some staff knew some participants personally and had knowledge that senior research staff did not.

Category	Category description	Data
		Some study team members worried about over-burdening participants by asking them to do too much, however this concern did not seem to be backed up by the data collected, and may be considered a barrier to involvement.
8f: Process	Comment on how process factors influenced PPI in the study	During the process, both participants and study team members reported changed views about the value of involvement in research, demonstrating 'transformative learning' and co-construction of knowledge.
8 g: Measurement and capture of PPI impact	If applicable, comment on how well PPI impact was evaluated or measured in the study	Detailed data was collected and shared. Participants were supportive about being involved, with all participants supportive of being involved by providing feedback throughout the research process (100%, 32/32), with a typical participant response being 'I'd be happy to be involved'. Views about enablers were shared in three of the 20 interviews coded, by all 18 of the event participants and all study team members surveyed. Views about barriers were shared in eight of the interviews coded and by half of the study team surveys. Mapping of preferences for involvement was completed using the STARDIT-PM tool, with the involvement reported using STARDIT Alpha.
8 h: Economic assessment	If applicable, discuss any aspects of the economic cost or benefit of PPI, particularly any suggestions for future economic modelling.	Economic assessment needs to be widened to 'assessing value', with one way of this being measured being financial. The entire process of involving people was estimated to cost $10,000 AUD, including staff time, catering and event venue hire. The value of the process was summarised by the lead investigator who stated "I learnt a lot from the process and am very glad we made the effort".
8i: Reflections/critical perspective	Comment critically on the study, reflecting on the things that went well and those that did not, so that others can learn from this study	A number of significant learning points were identified by the study team when responding to the question "do you have any advice to other researchers planning involvement for their research". Significant learning points are were: 1. Fund and prioritise involvement, make it a requirement 2. Ethics processes take time, but can improve plans 3. Know your audience – don't make assumptions 4. Value diversity in experience and knowledge 5. A supportive team improves the experience for all

Additional File 5 - Standardised Data on Initiatives (STARDIT) report: 'Involving elderly research participants in the co-design of a future multi-generational cohort study'

About this report

This report uses the Standardised Data on Initiatives Alpha version (STARDIT)[196]. An Alpha version of this STARDIT report is also hosted online in machine readable format.[410]

A 'living' version of the report can be found here:
https://wikispore.wmflabs.org/wiki/STARDIT/Involving_ASPREE-XT_participants_in_co-design_of_a_future_multi-generational_cohort_study

STARDIT Report Alpha Version

Identifying information	
Initiative name	Involving ASPREE-XT participants in co-design of a future multi-generational cohort study
Geographic location or scope	Australia
Date range (planned start and end dates of initiative)	2017-2019
Purpose of the initiative	Participatory action research to involve elderly research participants in the co-design of a proposed multi-generational cohort study, in order to improve research design, relevance, acceptability and recruitment.
Organisations or other initiatives involved (list all if multi-centre)	1. Department of Epidemiology and Preventive Medicine, School of Public Health and Preventive Medicine, Monash University 2. School of Psychology and Public Health, La Trobe University
Funding sources	Department of Epidemiology and Preventive Medicine, School of Public Health and Preventive Medicine, Monash University
Clinical trial registration details (if applicable)	https://clinicaltrials.gov/show/NCT01038583
Ethics approval (if applicable)	Monash University
Other relevant information (free text)	This report describes involving potential participants in co-designing a proposed multi-generational research study. It would recruit participants from the existing ASPREE-XT study.
At which stage of the research project has this report been written? (Select from: 1. **Before** the intervention or initiative– this report is prospective or describes planned activity 2. **Ongoing** – the intervention or initiative is still taking place 3. **After** the research project or initiative has occurred	**After** the co-design process occurred, but before the proposed multi-generational research study has been approved or funded.

Methods of the initiative (what is planned to be done, or is being reported as done)	Participatory action research to involve elderly research participants in the co-design of a proposed multi-generational cohort study.
Report authorship	
Name	Jack Nunn
Public domain profiles, institutional pages	https://scholars.latrobe.edu.au/display/j2nunn
Open Researcher and Contributor ID (orcid.org)	https://orcid.org/0000-0003-0316-3254
Tasks in report completion	Main author
Date of report authorship	22nd July 2020
Key contact at initiative for confirming report content	Paul Lacaze, PhD, Head, Public Health Genomics Program, Paul.Lacaze@monash.edu
Involvement	
Who was involved	4. 3 academic research investigators 5. An ASPREE participant assessor 6. An ASPREE-XT participant
Specific tasks of this person or group (list as many as possible) – *including any information about why certain people were included or excluded in certain tasks*	Everyone listed above was involved in co-designing every stage of the process. This included refining wording of participant information, sharing views and advice about the process, proof-reading documents, providing feedback on questionnaires, analysing data, informing planning, presenting information to participants, interpreting data, and participating in email surveys.
How were these people involved (what methods were used)	Face to face meetings, email communication, shared online documents, teleconferences.
Enablers of involvement (what do you expect will help these people get involved – or what helped them get involved)	Giving people time to read resources. Clear communication about the intention of involving people.
Barriers of involvement (what do you expect will inhibit these people from getting involved – or what inhibited them from getting involved). Are there any known equity issues which may contribute?	Face-to-face meetings were difficult to organise. Some participants and participant representatives were elderly or lived in remote areas, so face-to-face meetings needed to be minimised where possible.
What was the outcome or output of the involvement of these people? What changed as a result of involving people?	Improved participant information resources, improved wording that is culturally appropriate, improved question design for interviews, improved learning resources for participants, improved co-design process.
Which stage of the initiative were these people involved? (select from list of pre-defined stages or allow 'other')	All stages
What was the estimated financial cost for involving people. How much time did it take. Were there any costs that cannot be measured financially?	$10,000 AUD was the estimated cost for the process. The total number of hours, including staff time was estimated to be around 200,

	including telephone interviews and excluding data analysis. Some people who attended events were unable to work or care for people on that day, and these costs were not calculated.
What worked well, what could have been improved? Was anything learned from the process of involving these people?	The process took longer than expected. There was confusion over what ethics approval was required in order to involve people, especially people who are participants in an ongoing study. Involving field staff (as well as senior researchers and academics) provided a valuable perspective, as some staff knew some participants personally and had knowledge that senior research staff did not. Some study team members worried about over-burdening participants by asking them to do too much, however this concern did not seem to be backed up by the data collected, and may be considered a barrier to involvement.
Mapping financial or other 'interests'	
Describe any financial relationship or other interest this person has to this project	Three members of the study team were employed by Monash University during this process.
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	It will be published open access
How is it stored and hosted?	It will be shared on a public domain repository.
Who is analysing the data?	The study team described above
What methods will be used to analyse the data (including a link to any relevant code and information about validity)	We used case study methodology to describe our experience involving participants in the co-design of the proposed study. We collected and analysed both qualitative and quantitative data during the involvement activities. We analysed data from audio recordings of interviews and events, meeting notes, emails, reflexive diary entries and survey responses of study investigators. Coding and thematic analysis of qualitative data was carried out by two authors independently and checked by other authors.
How is information about this data disseminated?	4. It will be published in an open access journal 5. It will be shared as an item in a newsletter to participants of the ASPREE-XT study

	6. Learning from this process will be presented at conferences, shared on social media and through other channels (such as podcasts).
Who 'owns' the data or claims any kind of 'intellectual property' (include relevant licensing information)	Monash University
Who controls access to the data	Monash University
How is/will the data be 'Findable, Accessible, Interoperable, Reusable' according to the FAIR criteria?	Data will be shared in the public domain and licensed under a Creative Commons license.
Impacts and outcomes	
What new knowledge has been generated? (if appropriate, include effect size, relevant statistics and level or evidence)	6. Involving participants in co-designing a proposed study resulted in changes to the design of the proposed study 7. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participant and study team members' views about the value of involvement, which can be viewed as an impact of 'transformative learning'.
Describe how the learning or knowledge generated from this initiative has or will be used	3. Knowledge from this process will inform the design of a future multi-generational study 4. Learning from this process can inform future involvement activities
How has or how will this be measured?	Future STARDIT reports
Who is involved in measuring this?	The study team

Appendices for Chapter 6 - Additional files: Involving people affected by a rare condition in shaping future genomic research

Additional File 1: Data and Analysis

About this document

This document contains additional data relevant to the case study 'Involving people affected by a rare condition in shaping future genomic research'. In addition it contains a more detailed description of the data sources in this case study. This document includes the preferences mapping data (STARDIT-PM), and other data about this initiative[196]. The corresponding Standardised Data on Initiatives Alpha Version (STARDIT) of the report can be found in 'Additional File 2 - STARDIT report'.

This file is available at this URL: https://doi.org/10.1186/s40900-021-00256-3

Survey questions

Pre-discussion survey questions

After participants had read the Participant Information and given consent to participate, they were asked to complete the following information. The questions below are worded exactly as participants read them.

1. Full Name (if you would prefer to not use your real name you may use a pseudonym)
2. Email address (Please note this needs to be a working email address. If you do not have one leave this blank and we will contact you by your preferred method of communication.)
3. Phone number (optional)
4. Any other preferred method of communication? (Please share any other preferred method of communication if email or phone are not preferred)
5. Age
6. Gender (Choose from 'Male', 'Female', 'Transgender', 'Intersex', 'Other', 'Prefer not to say')
7. Educational background (tick all that apply)
    a. Middle school qualifications (up to age 16) ('lower')
    b. High school qualifications (ages 16-19) ('middle')
    c. Degree (bachelors), diploma or post-graduate ('higher')
    d. I have qualifications or professional experience in genomics (professional)
    e. Prefer not to say
8. How would you describe yourself? (Please tick all that apply)
    a. A person with EGID
    b. A parent of a person with EGID
    c. A carer of a person with EGID
    d. A partner, family member or loved one of a person with EGID
    e. Other (please describe)

f. Prefer not to say

9. In which country do you live (or spend most time)?
10. Please tick which statement applies
    a. I am a parent, a carer, a partner, family member or loved one of someone with EGID who is under 18
    b. I am a parent, a carer, a partner, family member or loved one of someone with EGID who is 18+
    c. I am over 18 and am representing myself
11. What made you decide to respond to our invitation to participate in this project?
12. What do you hope to get out of participating in this discussion? Do you have any specific expectations?
13. There are many benefits of involving people other than researchers in the co-design of research studies at every stage of the research cycle. Research suggests that involving people improves the quality and the relevance of the research. Involving people can also improve participant experience and increase participation. **Who do you think should influence what kind of genomic research should be done in the future?**
14. What makes you say that? (why did you give that answer?)
15. Do you have any ideas about how the people from your previous answer could influence future research? (For example, what tasks could people affected by EGID be involved in?)
16. **Which aspects of any future research genomic research should be influenced by the following (**participants were presented with a grid of tick boxes, the horizontal axis being who should be involved, the vertical a list of tasks. The horizontal was as follows)

Everyone (any member of the public who is interested)	Anyone who might be indirectly affected by the research	Only people who are directly affected by the research	Only people who are participating in the research	Only people with a professional role in research

    a. All aspects mentioned below (leave others blank if ticking this)
    b. Finding questions to ask (identifying research topics)
    c. Deciding which questions to prioritize and fund
    d. Deciding how to try and answer the question (the research method)
    e. Attempting to answer the question (carrying out the research, including collecting information)
    f. Trying to understand if it is possible to answer the question (analyzing the information)

g. Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)
h. Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)
i. Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)
j. Designing how people are involved in the research

17. Have you ever participated in research in the past? (by participation, we mean as a research subject – for example part of a trial)
    a. Yes
    b. No
    c. Prefer not to say
    d. Unsure
18. Have you ever participated in research in the past? (by participation, we mean as a research subject – for example part of a trial)
    a. Yes
    b. No
    c. Prefer not to say
    d. Unsure

Post discussion survey questions

1. How would you rate the following? (chose from 'Excellent', 'Somewhat good', 'Neither good nor bad', 'Somewhat poor', 'Extremely poor')
    a. Your overall experience of participating in the online discussion
    b. Your assessment of how we conducted the survey and discussion format
    c. The support you received to be involved (for example, practical support such as instructions for using the online tools)?
    d. Information and learning materials you were given before the event
2. Did you feel you meaningfully contributed to the discussion?
    a. Yes
    b. No
    c. Unsure
3. Is there anything in particular you liked or thought was helpful about how the discussion was conducted?
4. Is there anything you didn't like, thought was unhelpful or could have been improved about how the discussion was conducted?
5. Do you have any other thoughts, ideas or comments?
6. Would you like to be updated about the progress of the research and offered chances to be involved where possible? (Chose 'yes' or 'no')
7. Did you have any expectations from participating in this research that were met or not met?
8. Have any of your views and perspectives about involving people in genomic research changed since participating in this research? If so, please describe.
9. There are many benefits of involving people other than researchers in the co-design of research studies at every stage of the research cycle.
10. Research suggests that involving people improves the quality and the relevance of the research. Involving people can also improve participant experience and increase participation. **Who do you think should influence what kind of genomic research should be done in the future?**
11. What makes you say that? (why did you give that answer?)
12. Do you have any ideas about how the people from your previous answer could influence future research? For example, what tasks could people affected by EGID be involved in?
13. **Which aspects of any future research genomic research should be influenced by the following (**participants were presented with a grid of tick boxes, the horizontal axis being who should be involved, the vertical a list of tasks. The horizontal was as follows)

Everyone (any member of the public who is interested)	Anyone who might be indirectly affected by the research	Only people who are directly affected by the research	Only people who are participating in the research	Only people with a professional role in research

a. All aspects mentioned below (leave others blank if ticking this)
b. Finding questions to ask (identifying research topics)
c. Deciding which questions to prioritize and fund
d. Deciding how to try and answer the question (the research method)
e. Attempting to answer the question (carrying out the research, including collecting information)
f. Trying to understand if it is possible to answer the question (analyzing the information)
g. Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)
h. Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)
i. Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)
j. Designing how people are involved in the research

14. Full Name (Optional- if you would prefer to not use your real name you may use a pseudonym)
15. Email address (optional)
16. Phone number (optional)
17. Age
18. Gender (Choose from ‘Male’, ‘Female’, ‘Transgender’, ‘Intersex’, ‘Other’, ‘Prefer not to say’)
19. Educational background (tick all that apply)
    a. Middle school qualifications (up to age 16) ('lower')
    b. High school qualifications (ages 16-19) (‘middle’)
    c. Degree (bachelors), diploma or post-graduate ('higher')
    d. I have qualifications or professional experience in genomics (professional)
    e. Prefer not to say
20. How would you describe yourself? (Please tick all that apply)
    a. A person with EGID
    b. A parent of a person with EGID

155 c. A carer of a person with EGID
156 d. A partner, family member or loved one of a person with EGID
157 e. Other (please describe)
158 f. Prefer not to say
159 21. In which country do you live (or spend most time)?
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Facilitator survey questions

1. Please describe your tasks in the process of involving people in planning of the study
2. What did you learn from the process of involving participants in the research planning phase?
3. Please describe specifically what worked well or was useful about the way the study was conducted (including how people were involved)
4. Please describe specifically what did not work well or was not useful about the way the study was conducted (including how people were involved)
5. Were there any barriers or enablers to conducting the study or involvement activities? (institutional or otherwise)
6. Do you think the involvement activity achieved its intended aim(s)?
7. Do you think the study achieved its intended aim(s)?
8. Do you have any advice to other researchers planning involvement for their research?
9. Do you have any advice to other researchers planning to involve people using online discussions?
10. Describe the impact you think involving people had (positive/negative - on the research, staff or participants)
11. Who do you think should influence the kind of human genomic research done in the future, and why? (e.g. the public, participants of research studies, doctors, school children, politicians etc)
12. Which stages of future genomic research should be influenced by people other than researchers (if any)? (e.g. concept planning of new studies, study design, conducting the research, presenting the results etc)
13. Other comments

STARDIT Preference Mapping (STARDIT-PM)

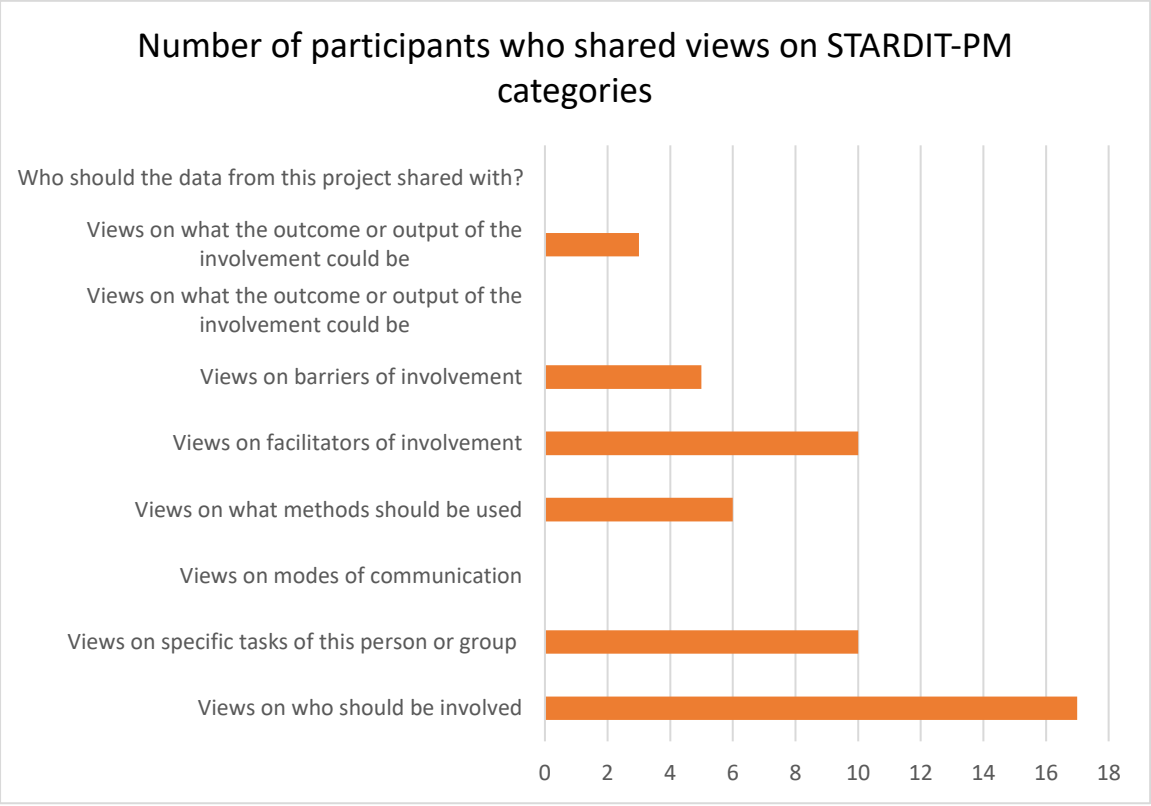
This table uses the Alpha version of the Standardised Data on Initiatives Preference Mapping (STARDIT-PM) to categorise the data into certain areas [196]. Preferences were recorded from all data sources, including the initial survey, online discussion with participants, online Facilitator discussions, follow-up surveys with participants and with facilitators. Facilitator comments were only included from one Facilitator with personal experience (KG), and comments from the other Facilitator were excluded from analysis (JN). If the same participant made the same point at different stages, this was counted as one view. The standardised categorisation is intended to facilitate comparison with other studies. Accordingly, the content may be similar to other sections of the qualitative thematic analysis.

STARDIT-PM area and quantitative data	Qualitative summary
Views on who should be involved: **17 participants** shared views about who should be involved	One participant wrote that asking 'Who should be 'excluded' is a helpful starting point when answering this question' [P3]. Participants contributed 17 different statements saying patients and their families should be involved. The word 'collaboration' was used to describe how people should work together. Besides patients and families and those 'directly affected' [P3], other 'specialists' and groups were mentioned by participants [P23]. These included including doctors, medical professionals, researchers, patient advocacy groups, immunologists, gastroenterologists and IT experts. One participant stated 'drug companies will have a part to play' and went on to say 'this would need supervision and strict guidelines' [P9].
Views on specific tasks people involved could do: **10 participants** shared views about specific tasks people involved could do	Participants shared multiple tasks which they felt patients should be involved in. Identifying topics and 'what's a priority' was mentioned 5 times [P3]. One participant wrote 'medical researchers should take the opportunity to be guided more from the patient themselves on an idea/direction for a research project rather than the other way round' [P21]. Involving people affected in setting outcomes was also mentioned by participants, including being involved in saying what 'would be useful' to patients [P22]. Involving people affected 'at the design stage' of research was mentioned, with two participants mentioning surveys as an example. For example, a 'quality of life survey' was mentioned as a way people could make sure the right questions were being asked [P21] [P25]. Fundraising, campaigning and 'advocating' were mentioned as ways of 'raising awareness' by two participants [P5] [P9]. Being involved in 'doing' research was mentioned by one participant [P3]. Being involved in working with insurance companies and government to explore the legal, financial and privacy impacts of how a diagnosis might affect people was mentioned by one participant [P3]. One participant mentioned involving patients in discussions about health technology assessment, including 'health economics and the consequences'[P3].
Views on modes of communication: **2 participants** shared views about	Two participants mentioned websites, with 'public government research websites' and 'patient advocacy groups' as ways of advertising opportunities for involvement or participation. Face to face research and online surveys were also mentioned as other modes. Online discussion, face to face communication were also mentioned.

preferred communication modes	
Views on what methods should be used to involve people: **6 participants** shared views about what methods should be used to involve people	One participant mentioned **surveys**, stating that 'short surveys' completed on computers or phones are 'easy' and cost effective, especially while people 'wait for their medical appointment' [P25]. Groups such as '**volunteer committees'** were suggested as a method. One participant suggested that public awareness 'facilitates involvement' [P25] and that **people sharing stories** can be a source of 'support or inspiration', in particular 'public figures' [P25]. **Online and face to face support groups** were identified as an area for 'uncovering trends', the first stage in identifying research topics [P3] 'Community discussions' [P16] and 'working collaboratively' with professors and 'those directly affected' [P3] was mentioned by two participants. Similarly one participant stated 'patient advocacy groups partnering with researchers is valuable for all involved' [P28]. One participant stated she liked 'focus groups as face to face' as when working in an online group the 'intent of the writer' can be interpreted differently to as it was intended (for example, presuming an incorrect tone of voice)' [P21]. Another participant preferred a combination of face to face events once or twice a year with most work being done by teleconferencing, owing to geographical separation [P3]. Online video teleconferencing was also suggested as a good method as participants 'get a much better feel for people and their thoughts because you can see them (get all the cues), then break away for specific follow-up' [P3]. Another participant agreed a 'two stage' approach would be good, starting with involving people online, then meeting face to face [P25]. Ensuring online discussions have a mixture of both open and closed questions allows the open questions to 'create another idea' and novel discussion [P3].
Views on facilitators of involvement: **10 participants** and **3 facilitators** shared views about facilitators' of involvement	One participant noted that 'Participants need to be able to contribute without putting their personal situation at risk'. One participated noted that 'more respect for the patient and their family, their knowledge & experience' would facilitate involvement. Two participants noted 'Power and knowledge' as facilitators [P20], highlighting not having 'payment for access' to information as a facilitator [P3]. Emotional connection to an issue can be both a facilitator and a barrier to involvement, with a 'balance' highlighted as a facilitator of involvement [P21]. For those who are unable to travel or live in remote areas, online discussions are 'good to help' people get involved, with one participant stating 'It is hard for us to help with research' if it is not in their area and they are 'unable to afford to travel' [P20]. Good facilitation of face to face events was highlighted by one participant as face to face discussions 'can be endless so need to be governed well' [P3]. Public awareness facilitates involvement, especially stories from people affected. This also can be a source of 'support or inspiration' for some people [P25].

	Six participants reported specific things about the way this study was conducted that facilitated their involvement. One participant 'enjoyed the interaction' and four found it 'helpful' to have '**links with information about genomics'** relevant to the topic threads' to 'explain a concept' [P21, P3, P29, P28]. One participant also stated '**being able to read others thought processes** on each topic' helped [P3]. Another responded that responses in the **discussion 'provoked further questions that made me think in new directions'** [P25]. Another participant added that other **participants were 'supportive, positive and open'** [P3]. One participant noted that the **flexibility of being able to join at any time** 'was great' as she was often 'busy caring for her son with EoE and travelling'. They stated that the way the discussion was hosted meant that they 'could still catch up and learn things' and also provide input [P30]. Two participants mentioned having **different topics threads with a lead question was helpful** and a 'good format' [P29] [P28]. One participant stated 'This process has been really interesting because we can only write so much, get **a variety of input which makes us think'** [P3]. Facilitators reported that they 'needed more support and advice than expected', stating that the training and ongoing support given before and during facilitation was essential. One Facilitator reported that 'touching base' and learning the experiences of other facilitators and feeling 'part of the team' all facilitated their facilitation of online discussion.
Views on barriers of involvement: **5 participants** and **3 facilitators** shared views about barriers of involvement	One participant stated barriers included '**payment for access' to information,** such as paywalls for peer-reviewed information [P3]. Researchers promising '**feedback that never came'** was considered a barrier to involvement by one participant [P25]. One participant stated that barriers included **representatives being 'undervalued and under-supported**.' [P3]. One participant noted that issues which have been identified should be addressed by 'the Ministry' and as a result of inaction, volunteers were 'working on extra time'. [P3]. **Online discussions 'can be viewed differently to the intent of the writer'** [P21]. One participant highlighted that **privacy was an issue when involving people** in research using focus groups online or face-to-face. They stated '**some people might share more if they didn't have a broader audience - especially people they don't know'**. [P21]. Two participants reported specific things about the way this study was conducted that were barriers to their involvement. One participant reported that although they like the format of the discussion, they '**didn't like the platform** it was conducted on' as it was not 'user friendly' [P15]. Another reported that the pace of new questions being added (around one every two days) was 'too quick' and done 'before a number of people had a chance to answer'. One participant felt **facilitators sometimes did not accurately summarise participants' comments,** and that some **participants might feel reluctant to correct a Facilitator** [P3]. Facilitators reported finding it a challenge to separate 'personal' experiences when facilitating. One Facilitator noted 'Separating out my

	parent role and Facilitator role has been hard. Harder than I thought it would be'. One Facilitator also noted 'The discussion was too rapid. The group needed more time to work through the process of the online conversation. It felt too rushed.'
Views on which stage of the research people should be involved: **3 participants** views on what the outcome or output of the involvement could be	One participant noted that **involving people in the design stage is more practical** as once research begins 'you can't change direction or question' [P3]. Two participants stated that **identifying topics and research development were appropriate stages for people to be involved**. One participant noted that **people 'must influence the research agenda including the questions, how they are funded, research design, data analysis, interpretation and dissemination of results'** [P16]
Who should the data from this project be shared with?: **0 participants** shared views about who data from this project shared with	
Views on what the outcome or output of the involvement could be: **0 participants** shared views about what the outcome or output of the involvement could be	



Data

This table summarises all the data sources used for the case study.

Data Category	Data point description
Diary	ausEE research diary of lead investigator (JN) – including reflections
Emails and meeting notes	Email, meeting notes and Notes from planning and discussion. This included involvement of participant representatives in co-refining study design.
Online pre-discussion survey	Informed consent and pre-discussion survey data
Learning resources for participants and facilitators	Learning resources giving information about genomics and using Loomio
Online discussion with participants	Text data from online Loomio discussion with participants
Online discussion with facilitators	Text data from online Loomio discussion between facilitators of two parallel studies
Online post-discussion survey	Post-discussion survey data from participants
Follow up survey for facilitators	Post-discussion survey data from facilitators

201

202

Demographic information

Category	Pre-discussion survey	Post discussion Survey
Gender		
Female	28	8
Male	1	0
Age		
20-24 years	1	0
25-29 years	1	0
30-34 years	3	1
35-39 years	12	0
40-44 years	7	2
45-49 years	3	1
50-54 years	3	1
Educational background ('highest' only counted)		
Prefer not to say	1	0
I have qualifications or professional experience in genomics	1	0
Degree (bachelors), diploma or post-graduate ('higher')	26	6
High school qualifications (ages 16-19)	2	1
Middle school qualifications (up to age 16) ('lower')	1	0
How would you describe yourself?		
A carer of a person with EGID	1	1
A parent of a person with EGID	22	5
A partner, family member or loved one of a person with EGID	1	0
A person with EGID	5	1
A person with EGID and a parent of a person with EGID	1	0
In which country do you live (or spend most time)?		
Australia	28	6
New Zealand	1	0

206 **Widening and narrowing for each question asked in pre and post survey**

Who should influence which aspects of research?	Change to wider	No change	Change to narrower
Finding questions to ask	3	1	1
Deciding which questions to prioritize and fund	3	1	1
Deciding how to try and answer the question (the research method)	3	2	0
Attempting to answer the question (carrying out the research, including collecting information)	2	2	1
Trying to understand if it is possible to the answer the question (analysing the information)	2	3	0
Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)	3	2	0
Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)	4	1	0
Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)	2	1	0
Designing how people are involved in the research	2	1	0
Change totals	**24**	**14**	**3**

207

208

Participant experience

How would you rate your overall experience of participating in the online discussion	
Excellent	9
Somewhat good	6

How would you rate how we conducted the survey and discussion format	
Excellent	11
Somewhat good	3
Neither good nor bad	1

How would you rate the support you received to be involved (for example, practical support such as instructions for using the online tools)?	
Excellent	11
Somewhat good	4

How would you rate the information and learning materials you were given before the event	
Excellent	11
Somewhat good	5
Neither good nor bad	1

Did you feel you meaningfully contributed to the discussion?	
Yes	8
Unsure	7

Learning resources

A number of different learning resources were shared with participants at different stages of the process. This included a short 60 second online video about the study, giving information about the context and purpose[417], a one page infographic summary of a scoping review about genomics research[418], and a short two-page summary of genomics and contemporary research relating to EoE was co-created with ausEE, the study team and experts in genomics[419].

Learning resources were both co-created and selected by the investigator team, working in partnership with the Australian Genomics Health Alliance and co-refining the selection with potential participants. In addition, in order to support the Enablers in providing good quality information, a number of were curated into a list to be available for Enablers to share during the online discussion, if they became relevant to aspects of the discussion in order to help inform people. The table below provides a summary of which learning resources were shared at which stage of the process.

223 **Summary of learning resources**

Stage	Title	Media	Summary	Authorship
Stage 2: Before consent	What is genomic testing?	Portable Document Format (PDF)	Simple infographic explaining the basics of genomics research in plain English	Australian Genomics Health Alliance[602]
	What is genomics?	Online video animation with audio narration and subtitles	A 6 minute video outlining the principles of genomics research	Genome BC[575]
Stage 3: Before online discussion	Definitions and explanations	PDF	These definitions and explanations were used as a glossary to explain the main concepts of this research project.	Investigator team
	Genomics and involvement	Online video with hard-coded text	A 2 minute video exploring why people should be involved in genomics	Jack Nunn[417]
	Infographic summary of scoping review	PDF (infographic – images and text)	A one page summary of the main findings from a recent scoping review about involving people in genomics[1]	Jack Nunn et al[418]
	A summary of EGID and relevant genomics research	PDF (text with hyperlinks)	A co-created learning resource updating people on what is known so far, what research is currently being done and what might be the future	Jack Nunn et al[419]
	Guide to using Loomio	PDF (text with hyperlinks)	A co-created learning resource giving practical advice for using the online discussion platform Loomio	Investigator team
Additional resources available to facilitator	Inheriting genomic conditions (chapter)	Webpage	An additional resource if participants wanted more information about inherited conditions	U.S National Library of Medicine[603]
	Data in the 100,000 Genomes Project	Online video animation with	An example of the bioinformatic pathway (specific to Genomics	Genomics England[604]

		voice over and subtitles	England) but generalisable (talks about access review committee)	
	Ethical issues in human genomics and genomics	PDF (text with hyperlinks)	Additional resource for a relevant ethics discussion from a medical perspective	Centre for Genomics Education[605]
	Genes, DNA and cancer	Webpage (text with hyperlinks)	Good plain English information about genes in relation to cancer.	Cancer Research UK[606]
	How to Share Genomic Test Results With Family	Webpage (text with hyperlinks)	Good information about sharing genomic test results with family.	American Society of Clinical Oncology[607]
	How do you sequence a human genome?	Image file (infographic – images and text)	Infographic about the stages of genome sequencing	Genomics England[608]

224
225

Additional File 2: Standardised Data on Initiatives (STARDIT) report – Alpha Version: Involving people affected by a rare condition in shaping future genomic research

About this report

This report uses the Standardised Data on Initiatives Alpha version (STARDIT)[196]. An Alpha version of this STARDIT report is also hosted online in machine readable format.[609]

A machine readable structured data version of this report can be found here:
https://www.wikidata.org/wiki/Q100403236

A human readable version can be found here:
https://wikispore.wmflabs.org/wiki/STARDIT/Involving_People_Affected_by_a_Rare_Condition_in_Shaping_Future_Genomic_Research

STARDIT Report Alpha Version

Identifying information	
Initiative name	Involving people affected by a rare condition in shaping future genomic research
Geographic location or scope	Australia
Date range (planned start and end dates of initiative)	2017-2020
Purpose of the initiative	Participatory action research to involve people affected by a rare disease in shaping future research, by using online discussions.
Organisations or other initiatives involved (list all if multi-centre)	3. School of Psychology and Public Health, La Trobe University
Funding sources	School of Psychology and Public Health, La Trobe University
Clinical trial registration details (if applicable)	N/A
Ethics approval (if applicable)	The La Trobe University Human Ethics Committee approved this study. Project number: HEC18242 Project Title: Genomics Research and Involving People: ausEE
Other relevant information (free text)	This report describes involving people affected by a rare disease in shaping future research, by using online discussions exploring how they would like to be involved in future research.
At which stage of the research project has this report been written?	**After** the participatory action research occurred.
Methods of the initiative (what is planned to be done, or is being reported as done)	The research process was co-designed using a participatory action research method to involve people affected by a rare disease in the co-design of online discussions to explore future genomic research with members of the group.
Report authorship	

Name	Jack Nunn
Public domain profiles, institutional pages	https://scholars.latrobe.edu.au/display/j2nunn
Open Researcher and Contributor ID (orcid.org)	https://orcid.org/0000-0003-0316-3254
Tasks in report completion	Main author
Date of report authorship	24th July 2020
Key contact at initiative for confirming report content (include institutional email address)	Jack Nunn, PhD researcher, School of Psychology and Public Health, La Trobe University, jack.nunn@latrobe.edu
Involvement	
Who was involved or how would you label groupings of those involved	Group 1: Academic research investigators (Jack Nunn and Paul Lacaze) Group 2: People affected by the rare disease representing the charity ausEE with experience of academic research (Kylie Gwynne) Group 3: People affected by the rare disease representing the charity ausEE (Sarah Gray) Group 4: People affected by the rare disease who are members of the online community and participated in the study
How many people were in each grouping label?	Group 1: 2 Group 2: 1 Group 3: 1 Group 4: 25
Specific tasks of this person or group (list as many as possible) – *including any information about why certain people were included or excluded in certain tasks*	Group 1 and 2: Involved in co-designing every stage of the process, analysing data and member checking during the thematic analysis Group 3: Involved in co-designing the recruitment and giving feedback on the proposed study design and as an author of the paper. Group 4: Invited to give feedback on the paper
How were these people involved (what methods were used)	Group 1 - 3: Face to face meetings, video calls, email communication, shared online documents, teleconferences. Group 4: Invited to give feedback on paper and be acknowledged for contribution
Enablers of involvement (what do you expect will help these people get involved – or what helped them get involved)	Giving people time to read resources. Clear communication about the intention of involving people. Have multiple modes of communication for involving people.
Barriers of involvement (what do you expect will inhibit these people from getting involved – or what inhibited them from getting involved). Are there any known equity issues which may contribute?	Face-to-face meetings were difficult to organise. The study team were located in different states of Australia. Unclear communication about intentions and purpose of the involvement contributed to confusion (explaining how involvement is distinct

	from participation was challenging). Ensuring those involved had enough time to give feedback was also a challenge.
What was the outcome or output of the involvement of these people? What changed as a result of involving people?	Improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the group to describe themselves), improved online discussion, improved learning resources for participants, improved co-design process.
Which stage of the initiative were these people involved? (select from list of pre-defined stages or allow 'other')	Group 1 and 2: All stages Group 3: Co-design, evaluation, dissemination Group 4: evaluation, dissemination
What was the estimated financial cost for involving people. How much time did it take. Were there any costs that cannot be measured financially?	$0 AUD – people volunteered their time. The total number of hours volunteered (excluding participation) is estimated to be 75.
What worked well, what could have been improved? Was anything learned from the process of involving these people?	The co-design process took longer than expected owing to ethical 'grey areas' with no clear instruction on whether ethics approval was required to involve people in co-design. As a result an ethics application was made and subsequent feedback from the co-design process was integrated using modifications to the ethics application. Involving potential participants in co-defining language used to describe the group of people affected helped ensure that language was acceptable and appropriate.
Mapping financial or other 'interests'	
Describe any financial relationship or other interest this person has to this project	Two investigators are affected by the rare disease, which is why they were invited to be part of the study
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	It will be published open access in peer reviewed journals with identifying information removed in order to anonymise it as much as possible.
How is it stored and hosted?	It will be shared on a public domain repository.
Who is analysing the data?	Group 1-3: The study team described above Group 4: participants were invited to review the analysis and give feedback to ensure they felt it reflected their experience of the process
What methods will be used to analyse the data (including a link to any relevant code and information about validity)	We used case study methodology to describe our experience involving participants in the co-design of the proposed study. We collected and analysed both qualitative and quantitative data during the involvement activities. We analysed data from online surveys and online discussions with participants. In addition, data from

	the study team communications was included, such as meeting notes, emails, reflexive diary entries and survey responses of study investigators. Coding and thematic analysis of qualitative data was carried out by two authors independently and checked by other authors.
How is information about this data disseminated?	7. It will be published in an open access journal 8. It will be shared with participants of the research and also other members of the sibling group who have joined it since the study commenced 9. Learning from this process will be presented at conferences, shared on social media and through other channels (such as podcasts).
Who 'owns' the data or claims any kind of 'intellectual property' (include relevant licensing information)	Confidential data collected as part of the study is stored according to laws and the data access plan approved by La Trobe University. The authors maintain 'ownership' of the data in the paper and is shared under the Creative Commons license used by the publishing journal.
Who controls access to the data	The study team, La Trobe University and participants will be involved in any future data access decisions.
How is/will the data be 'Findable, Accessible, Interoperable, Reusable' according to the FAIR criteria?	Data will be shared in the public domain and licensed under a Creative Commons license.
Impacts and outcomes	
What new knowledge has been generated? (if appropriate, include effect size, relevant statistics and level or evidence)	1. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources 2. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'.
What was learned	Involving people in online discussions about involvement in research changes people's views about who should be involved in research, including participants 'widening' their views about who should be involved in research to include more people.
Knowledge translation	1. Knowledge from this process will inform the design of a future genomic research 2. Learning from this process can inform future involvement activities

Outcomes	Learning from this process informed subsequent discussions in the charity ausEE about involvement in research, including proposed improved ways of involving people
How has or how will this be measured?	Future STARDIT reports
Who is involved in measuring this?	The study team and participants

Additional File 3: GRIPP2 report for 'Involving people affected by a rare condition in shaping future genomic research'

This report has been completed using the 'GRIPP2 reporting checklists: tools to improve reporting of patient and public involvement in research' available at: https://doi.org/10.1136/bmj.j3453

GRIPP2 short form

Section and topic	Category description	Data
1: Aim	Report the aim of PPI in the study	Participatory action research to involve people affected by a rare disease in shaping future research, by using online discussions.
2: Methods	Provide a clear description of the methods used for PPI in the study	The research process was co-designed using a participatory action research method to involve people affected by a rare disease in the co-design of online discussions to explore future genomic research with members of the group. Participants were also involved in analysing the data and checking the final version of the paper.
3: Study results	Outcomes—Report the results of PPI in the study, including both positive and negative outcomes	The input of the representatives during the planning and co-design stage had clear positive impacts, particularly in improving educational resources and ensuring the online discussion was advertised using wording appropriate to the existing online community. For example, representatives from ausEE helped change the study design to include explicit opportunities for participants to learn more about genomics and EoE, avoiding participation being perceived as having a one-way benefit. During the co-design process it was also decided to exclude people who were under 18 and people who stated they were representing someone who was over 18, as people who were 18 and over had the choice to represent themselves. Enablers of involvement: Giving people time to read resources. Clear communication about the intention of involving people. Have multiple modes of communication for involving people. Barriers of involvement: Face-to-face meetings were difficult to organise. The study team were located in different states of Australia. Unclear communication about intentions and purpose of the involvement contributed to confusion (explaining how involvement is distinct from participation was challenging). Ensuring those involved had enough time to give feedback was also a challenge.

Section and topic	Category description	Data
4: Discussion and conclusions	Outcomes—Comment on the extent to which PPI influenced the study overall. Describe positive and negative effects	Involvement improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the group to describe themselves), improved online discussion, improved learning resources for participants, improved co-design process. Involving potential participants in co-defining language used to describe the group of people affected helped ensure that language was acceptable and appropriate. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'.
5: Reflections/critical perspective	Comment critically on the study, reflecting on the things that went well and those that did not, so others can learn from this experience	The co-design process took longer than expected owing to ethical 'grey areas' with no clear instruction on whether ethics approval was required to involve people in co-design. As a result an ethics application was made and subsequent feedback from the co-design process was integrated using modifications to the ethics application. Involving potential participants in co-defining language used to describe the group of people affected helped ensure that language was acceptable and appropriate. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'. Involving people in online discussions about involvement in research changes people's views about who should be involved in research, including participants 'widening' their views about who should be involved in research to include more people.

Appendices for Chapter 7 - Additional files: Co-designing genomics research with a large group of donor-conceived siblings

Additional File 1: Data and Analysis

About this document

This document contains additional data relevant to the case study 'Co-designing genomics research with donor-conceived siblings'[4]. In addition it contains a more detailed description of the data sources in this case study. This document includes the preferences mapping data (STARDIT-PM), and other data about this initiative[196]. The corresponding Standardised Data on Initiatives Alpha Version (STARDIT) of the report can be found in 'Additional File 2 - STARDIT report'. The 'living' STARDIT Beta version[442] report which relates to this project can be found in the references[295].

This file is available in full at this URL: https://doi.org/10.1186/s40900-021-00325-7

Definitions of terms

We have used consistent language to describe concepts throughout this case study. The list below defines important terms used throughout.

Involvement – The words **'involvement'** or 'being **involved'** describe the concept of people being 'involved' in research. This is when research is carried out 'with' people rather than 'on' them.[260] 'Involvement' can also be defined as when other people aside from the research team, such as the public, patients, research participants and other stakeholders, actively contribute to the research process.[349] It is the 'active involvement' in shaping and guiding research, rather than only providing data.[81,352,353]
Engaged – participants in the online discussions are described as 'engaged' if they are reading and commenting in discussions, and 'disengaged' if they are not.
Enablers – enablers are things which 'facilitate' certain things happening. For example, something which enables someone to participate in research.
Facilitator –in this article it refers to a person facilitating online discussions. For example, "**facilitators** shared views about **enablers** of involvement"
Online community – as the half-siblings discovered one another through various direct-to-consumer ancestry services, they self-created an online community which used a mailing list to an email group as the mode of communication. Participants were recruited from this online community.
Online discussion – the study team recruited participants to an online discussion, specifically created for this study and hosted on the secure platform Loomio.
Participant – a person who participated in the process of sharing views and perspectives about the genomics research, including sharing views about preferences for any future involvement.
Participatory action research (PAR) - is an umbrella term which describes a number of related approaches, including forms of action research which embrace a participatory philosophy. Concepts such as 'co-design', 'co-creation' and 'co-production' describe involving people in the respective tasks of designing a project or creating a learning resource, and can be considered as part of participatory action research[88]. It is a process where researchers, relevant stakeholders and sometimes the public "work together, sharing power and responsibility from the start to the end of the project"[89], including knowledge generation and translation[89].
Potential participant – before inviting people to become participants, it was necessary to involve a number of potential participants to help advise and plan the process.

Shared Ancestry groups –refers to people who have shared ancestors and have been grouped into these sub-populations by genomic researchers.
Sibling group - refers specifically to the community of shared interest defined by people who shared the same sperm-donor father, Bertold Wiesner.
Stakeholder – this term includes anyone who has a 'stake' in the research, in particular those who have important knowledge, views or perspectives that should be taken into account.[21,31] In this paper it refers to participants, representatives, patients, parents and carers of patients, potential patients and the study team (including researchers and representatives) and the wider public.
The study – the study refers to the formal research described in this case study, which was overseen by the 'Ethics, Integrity and Biosafety team' team at La Trobe University and the La Trobe University Human Research Ethics Committee.
Study team – this process was guided by the study team, who consisted of academic researchers and members of the sibling group. The work of the study team was also advised and overseen by the 'Ethics, Integrity and Biosafety team' team at La Trobe University and the La Trobe University Human Research Ethics Committee.
Sub-populations - This term refers to any grouping of people below the population level. Groupings can include communities of shared interest defined by shared genetic variation. These can include groupings of people who are half-siblings.
The process – this term will be used to describe both the study and the co-design process which involved members of the sibling group by inviting them to share views and perspectives about genomics research, including sharing views about preferences for any future involvement.

Case study background and context

The practice of artificial insemination existed in a legal and ethical grey area for many years[430]. For example, in the UK in the 1950s, the legitimacy of children conceived from a donor father was unclear as husbands were registered as the fathers, which was legally an offence[433]. Despite the recognised 'immense social, moral and medical questions' raised by this process[430], there was no legislative oversight and the practice was self-regulated by the individuals managing clinics carrying out the procedure. Members of the UK's House of Lords suggested that artificial insemination of married women with the husband's consent be classified as adultery as late as 1954[430]. Such contemporary attitudes created potential ethical, legal and social issues for families conceiving in this way[430]. Subsequently, there was a requirement for discretion for all involved, including the identities of donors which were surrounded by 'complete secrecy'[430]. Additionally, it was the view of some doctors that parents should not know the identity of the donors as it was 'incompatible with secrecy'[433]. Accordingly, many parents were encouraged to never disclose the paternity to the offspring.

The total number of donor conceived people in the UK by 1958 was estimated to be 7500, and 100,000 in the United States[430]. One pioneering clinic mentioned in debates of the UK Parliament in the 1950s was the Dr Mary Barton's medical practice, which operated in London from the 1940s to the 1960s and was responsible for at least 433 children with Dr Mary Barton stating that she had seen 600 prospective parents between 1944 and 1954[428,437,610]. The Barton practice used donors from 'intelligent stock' and ruled out donors where there was 'inheritable disease on that side' or 'criminality', introducing concepts of 'the eugenic quality of the donor's stock' into the very earliest years of the practice[433]. While attempts were made to find suitable donors that were a 'match', (including parents choosing whether or not they wanted a 'Jewish' donor[433])[610], some early practitioners of artificial insemination used donors from 'a very small panel of donors'[433], often from their own immediate social circles[610,611].

One prolific donor was Dr Barton's husband, the scientist Bertold Wiesner, a consulting biologist at the Royal Northern Hospital in the 1940s[440]. According to some estimates Wiesner may have fathered up to 1000 offspring during the time the clinic was operational[439], despite a 1945 British Medical Journal paper where Barton and Wiesner stated they set an 'arbitrary limit of 100 children

for each donor'[433]. In the UK, a government register of donors was proposed as early as 1949[430], but such a register was not established until 1991[431]. Subsequently, a number of people who have discovered they are the offspring of Wiesner have discovered each other by various means in subsequent years (including using direct-to-consumer genetic testing services) and formed an online community. Some members of the group have disclosed their biological relation to Wiesner (and thus other siblings) in the public domain through media, including documentaries[455,611–613], and advocated for the rights of people who are donor conceived[614].

Methods

Case study data collection and analysis

We used case study methodology to describe our experience of involving participants in an online discussion about genomics research together with pre and post discussion surveys.

Case study selection

The selection of this case study was informed by a number of factors which were appraised by the study team using the following questions[178]:

- Was it a population of people affected by genomics research, distinct from the general public?
- Was it pragmatic – was it possible to establish a mutually trusting and effective relationship within the time and resources of the research project?
- Was the power dynamic equal and not exploitative (would the research offer participants something rather than just being passive subjects?)
- Were there conflicting or competing interests which could negatively affect the research?
- Was the proposed case study ethical (including a consideration of creating a capacity burden on populations or partner organisations)

As part of a doctorate in public health genomics exploring public involvement in genomics research, one member study team (JN) began simultaneously planning a number of groups to work with to explore this area using participatory action research methodology. Unrelated to his PhD, he bought a direct-to-consumer DNA genetic test and subsequently discovered his grandfather was Bertold Wiesner and his mother was a half-sibling over up to 1000 other people[455]. After seeking advice from relevant ethics advisors, a proto-study team was assembled and began planning how to include the sibling group in co-designing a study to explore their views about involvement in genomics research. The study team contacted a researcher who had previously worked with members of this group (MC) and invited her to join the team in order to inform study design. The study team worked closely with both potential participants and experts from the La Trobe University human ethics department to ensure the method was acceptable and no one (including the study team) would be exposed to avoidable risk.

Case study method

The case study is presented as an instrumental case study, where the purpose is to understand the particular case and can attempt to provide data that could produce useful generalisations by using inferences from the data[163]. The codesign of the case study was informed best practices for enhancing validity and rigour in the case study methodolgy[153,160,452,180–182,397,399,447–449]. The data collection and analysis was also informed by a number of frameworks for reporting involvement in research[21,80,91,183–185]. In addition to quantitative analysis, each source was analysed using the method of thematic analysis, which involved stages including data mapping and familiarisation, transcription, coding, searching for themes, reviewing themes with study team members, labelling and summarising themes and reporting the findings[182].
We collected and analysed both qualitative and quantitative data during the involvement activities. We also attempted to measure impacts, which can be outcomes from the participatory research

process which have had an impact on individuals, the research process itself or wider society and other areas[130,155,329]. For example, participation in research might be shown to have a transformative impact on participants knowledge or views at an individual level, involving participants might have an impact on the research design or learning from the research may change policy or practice[91,196,615,616].

Two members of the study team were involved in analysing data from multiple sources including participant survey responses and online discussions (JN,MC). In addition, meeting notes, emails, surveys of the study team and reflexive diary entries of one member of study team (JN) were also analysed (JN). Coding and thematic analysis of qualitative data was carried out by two authors independently (JN, MC) and checked by another author (PL). Two authors of this paper also shared comments in the online study team discussion (JN, MC). Once a draft version of this article was created, it was shared with all participants who were invited to give feedback on the case study and contribute to STARDIT reports.

An alpha version of the STARDIT framework was also used in parallel with the thematic analysis to organise data into pre-defined 'super-categories' which allow consistent comparison with other data using this reporting framework[196], including other case studies.

Study team survey

The Facilitator (MC) was surveyed 6 months after the online discussion in order to integrate the valuable views and perspectives of those involved in planning and delivering the process. The survey questions can be found in the section 'Facilitator survey questions'. Design of surveys was informed by best practice frameworks for public involvement[80,91]. This method was informed by the Public Involvement Impact Assessment Framework Guidance (PiiAF)[91] and the questions were informed by sections 7 and 8 of the GRIPP2 reporting checklist[80]. The Facilitator was asked 11 questions and the data was coded and categorised, including using the STARDIT framework [196]. The data was then compared and integrated with the other data from the interviews and checked by other study team members (JN and PL).

Investigator shared learning group

During the online facilitation of the two online discussions, a shared learning group was established for facilitators and the study teams of two similar projects being run in parallel. The study teams shared reflections and learning about the process of facilitation online, as well as offering and receiving support regarding technical and practical issues. The data was coded and categorised, including using the STARDIT framework[196].

Survey questions

Pre-discussion survey questions

After participants had read the Participant Information and given consent to participate, they were asked to complete the following information. The questions below are worded exactly as participants read them.

19. Full Name (if you would prefer to not use your real name you may use a pseudonym)
20. Email address (Please note this needs to be a working email address. If you do not have one leave this blank and we will contact you by your preferred method of communication.)
21. Phone number (optional)
22. Any other preferred method of communication? (Please share any other preferred method of communication if email or phone are not preferred)
23. Age
24. Gender (Choose from 'Male', 'Female', 'Transgender', 'Intersex', 'Other', 'Prefer not to say')
25. Educational background (tick all that apply)
    a. Middle school qualifications (up to age 16) ('lower')
    b. High school qualifications (ages 16-19) ('middle')
    c. Degree (bachelors), diploma or post-graduate ('higher')
    d. I have qualifications or professional experience in genomics (professional)
    e. Prefer not to say
26. Please tick which of the following statements that you agree with:
    a. I feel comfortable describing other descendants of my biological father as 'half-siblings'
    b. I would describe our email group as an 'online community'
    c. Members of this email group potentially have a shared interest in discussing future research which might affect them, including genomic research
    d. If you do not feel comfortable describing other descendants of your biological father as 'half-siblings' please share any term (or terms) you prefer.
27. In which country do you live (or spend most time)?
28. What made you decide to respond to our invitation to participate in this project?
29. What do you hope to get out of participating in this discussion? Do you have any specific expectations?
30. There are many benefits of involving people other than researchers in the co-design of research studies at every stage of the research cycle. Research suggests that involving people improves the quality and the relevance of the research. Involving people can also improve participant experience and increase participation. **Who do you think should influence what kind of genomic research should be done in the future?**
31. What makes you say that? (why did you give that answer?)
32. Do you have any ideas about how the people from your previous answer could influence future research? (For example, what tasks could people affected by EGID be involved in?)
33. **Which aspects of any future research genomic research should be influenced by the following (**participants were presented with a grid of tick boxes, the horizontal axis being who should be involved, the vertical a list of tasks. The horizontal was as follows)

Everyone (any member of the public	Anyone who might be indirectly	Only people who are directly	Only people who are participating	Only people with a professional

who is interested)	affected by the research	affected by the research	in the research	role in research

a. All aspects mentioned below (leave others blank if ticking this)
b. Finding questions to ask (identifying research topics)
c. Deciding which questions to prioritize and fund
d. Deciding how to try and answer the question (the research method)
e. Attempting to answer the question (carrying out the research, including collecting information)
f. Trying to understand if it is possible to answer the question (analyzing the information)
g. Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)
h. Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)
i. Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)
j. Designing how people are involved in the research

34. Have you ever participated in research in the past? (by participation, we mean as a research subject – for example part of a trial)
    a. Yes
    b. No
    c. Prefer not to say
    d. Unsure
35. Have you ever participated in research in the past? (by participation, we mean as a research subject – for example part of a trial)
    a. Yes
    b. No
    c. Prefer not to say
    d. Unsure

Post-discussion survey questions

22. How would you rate the following? (chose from 'Excellent', 'Somewhat good', 'Neither good nor bad', 'Somewhat poor', 'Extremely poor')
    a. Your overall experience of participating in the online discussion
    b. Your assessment of how we conducted the survey and discussion format
    c. The support you received to be involved (for example, practical support such as instructions for using the online tools)?
    d. Information and learning materials you were given before the event
23. Did you feel you meaningfully contributed to the discussion?
    a. Yes
    b. No
    c. Unsure
24. Is there anything in particular you liked or thought was helpful about how the discussion was conducted?
25. Is there anything you didn't like, thought was unhelpful. or could have been improved about how the discussion was conducted?

26. Do you have any other thoughts, ideas or comments?
27. Would you like to be updated about the progress of the research and offered chances to be involved where possible? (Choose 'yes' or 'no')
28. Did you have any expectations from participating in this research that were met or not met?
29. Have any of your views and perspectives about involving people in genomic research changed since participating in this research? If so, please describe.
30. There are many benefits of involving people other than researchers in the co-design of research studies at every stage of the research cycle. Research suggests that involving people improves the quality and the relevance of the research. Involving people can also improve participant experience and increase participation. **Who do you think should influence what kind of genomic research should be done in the future?**
31. What makes you say that? (why did you give that answer?)
32. Do you have any ideas about how the people from your previous answer could influence future research?
33. For example, what tasks could people be involved in?
34. **Which aspects of any future research genomic research should be influenced by the following (**participants were presented with a grid of tick boxes, the horizontal axis being who should be involved, the vertical a list of tasks. The horizontal was as follows)

Everyone (any member of the public who is interested)	Anyone who might be indirectly affected by the research	Only people who are directly affected by the research	Only people who are participating in the research	Only people with a professional role in research

    a. All aspects mentioned below (leave others blank if ticking this)
    b. Finding questions to ask (identifying research topics)
    c. Deciding which questions to prioritize and fund
    d. Deciding how to try and answer the question (the research method)
    e. Attempting to answer the question (carrying out the research, including collecting information)
    f. Trying to understand if it is possible to answer the question (analyzing the information)
    g. Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)
    h. Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)
    i. Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)
    j. Designing how people are involved in the research
35. Full Name (Optional- if you would prefer to not use your real name you may use a pseudonym)
36. Email address (optional)
37. Phone number (optional)
38. Age
39. Gender (Choose from 'Male', 'Female', 'Transgender', 'Intersex', 'Other', 'Prefer not to say')
40. Educational background (tick all that apply)
    a. Middle school qualifications (up to age 16) ('lower')
    b. High school qualifications (ages 16-19) ('middle')

    c. Degree (bachelors), diploma or post-graduate ('higher')
    d. I have qualifications or professional experience in genomics (professional)
    e. Prefer not to say
41. Please tick which of the following statements that you agree with:
    a. I feel comfortable describing other descendants of my biological father as 'half-siblings'
    b. I would describe our email group as an 'online community'
    c. Members of this email group potentially have a shared interest in discussing future research which might affect them, including genomic research
    d. If you do not feel comfortable describing other descendants of your biological father as 'half-siblings' please share any term (or terms) you prefer.
42. In which country do you live (or spend most time)?

Facilitator survey questions

The Facilitator (MC) was surveyed 6 months after the online discussion in order to integrate the valuable views and perspectives of those involved in planning and delivering the process. Design of surveys was informed by best practice frameworks for public involvement[80,91]. This method was informed by the Public Involvement Impact Assessment Framework Guidance (PiiAF)[91] and the questions were informed by sections 7 and 8 of the GRIPP2 reporting checklist[80]. The Facilitator was asked 11 questions and the data was coded and categorised, including using the STARDIT framework [196]. The data was then compared and integrated with the other data from the interviews and checked by other study team members (JN and PL).

14. Please describe your tasks in the process of involving people in planning of the study
15. What did you learn from the process of involving participants in the research planning phase?
16. Please describe specifically what worked well or was useful about the way the study was conducted (including how people were involved)
17. Please describe specifically what did not work well or was not useful about the way the study was conducted (including how people were involved)
18. Were there any barriers or enablers to conducting the study or involvement activities? (institutional or otherwise)
19. Do you think the involvement activity achieved its intended aim(s)?
20. Do you think the study achieved its intended aim(s)?
21. Do you have any advice to other researchers planning involvement for their research?
22. Do you have any advice to other researchers planning to involve people using online discussions?
23. Describe the impact you think involving people had (positive/negative - on the research, staff or participants)
24. Who do you think should influence the kind of human genomic research done in the future, and why? (e.g. the public, participants of research studies, doctors, school children, politicians etc)
25. Which stages of future genomic research should be influenced by people other than researchers (if any)? (e.g. concept planning of new studies, study design, conducting the research, presenting the results etc)
26. Other comments

Learning resources

A number of different learning resources were shared with participants at different stages of the process. This included a short 60-second online video about the study, giving information about the context and purpose[417], a one page infographic summary of a scoping review about genomics research[418].

Learning resources were both co-created and selected by the study team, working in partnership with the Australian Genomics Health Alliance and co-refining the selection with potential participants. In addition, in order to support the facilitators in providing good quality information, a number of were curated into a list to be available for facilitators to share during the online discussion, if they became relevant to aspects of the discussion in order to help inform people. The next section summarises the resources used, with references using Internet Archive links to future-proof the content of the learning resources as well as the URL.

Summary of Learning Resources

Stage	Title	Media	Summary	Authorship
Stage 2: Before consent	What is genomic testing?	Portable Document Format (PDF)	Simple infographic explaining the basics of genomics research in plain English	Australian Genomics Health Alliance[602]
	What is genomics?	Online video animation with audio narration and subtitles	A 6-minute video outlining the principles of genomics research	Genome BC[575]
Stage 3: Before online discussion	Definitions and explanations	PDF	These definitions and explanations were used as a glossary to explain the main concepts of this research project.	Study team (see 'Learning resource example 1' below)
	Genomics and involvement	Online video with hard-coded text	A 2-minute video exploring why people should be involved in genomics	Jack Nunn[417]
	Infographic summary of scoping review	PDF (infographic – images and text)	A one page summary of the main findings from a recent scoping review about involving people in genomics[1]	Jack Nunn et al[418]
	Guide to using Loomio	PDF (text with hyperlinks)	A co-created learning resource giving practical advice for using the online discussion platform Loomio	Study team
Additional resources available to facilitator	Inheriting genetic conditions (chapter)	Webpage	An additional resource if participants wanted more information about inherited conditions	U.S National Library of Medicine[603]
	Data in the 100,000 Genomes Project	Online video animation with voice over and subtitles	An example of the bioinformatic pathway (specific to Genomics England) but generalisable (talks about access review committee)	Genomics England[604]
	Ethical issues in human genetics and genomics	PDF (text with hyperlinks)	Additional resource for a relevant ethics discussion from a medical perspective	Centre for Genetics Education[605]
	Genes, DNA and cancer	Webpage (text with hyperlinks)	Good plain English information about genes in relation to cancer.	Cancer Research UK[606]
	How to Share Genetic Test Results With Family	Webpage (text with hyperlinks)	Good information about sharing genetic test results with family.	American Society of Clinical Oncology[607]
	How do you sequence a human genome?	Image file (infographic – images and text)	Infographic about the stages of genome sequencing	Genomics England[608]

Learning resource example: Definitions and explanations

The following definitions and explanations were used to explain the following concepts throughout this research project. This document was formatted as a PDF with references.

Genomics

The study of all the DNA in the genome together with the technologies that allow it to be sequenced, analysed and interpreted is collectively called genomics, or genomic medicine if applied to patients[32]. The study of genomics can include other types of "omics"*, such as 'proteomics' and 'metabolomics' – which for simplicity will be referred to under the term 'genomics'. When the term 'genomics' is used in this project, it refers exclusively to human genomics.

**Other types of "Omics" include transcriptomics (all the RNA molecules in cell or organism), proteomics (all the proteins in a cell or organism) and metabolomics (all the metabolites in a cell or organism).*

Genomic research

Genomic research refers to any kind of activity which is intended to increase our current understanding of genomics. It is distinct from genomic medicine or other routine services which use existing knowledge, rather than add to it. However, this distinction is not always clear.

Involvement in genomic research

Research should be conducted ethically and to benefit people. Involving people as equal partners in genomic research has been identified as the most crucial aspect, as the benefits include improved public trust [21]. The concept of 'public involvement' in research is defined as research that is carried out 'with' people rather than 'on' them [260]. This allows people to have 'active' rather than 'passive' roles in research, which can lead to better research outcomes. Involving people in research in this way is now promoted by many governments as a right and is predicted to increasingly be an obligation in biomedical research, with research funding initiatives already recognising evidence of involvement as a key criterion [36,349,617]. Involving people can give greater public influence over research directions and conduct [35], ensuring research is both acceptable, accessible and meets people's needs by reflecting and balancing the diversity of priorities [19,36,38]. Tasks that people can be involved in include identifying areas of benefit, helping design and plan studies, helping in raising funds and analysing results.

This project

This project is part of Jack Nunn's PhD, 'Genomics research and involving people' which is focused on exploring how we can better involve people in all stages of genomics research, including the best methods to do so.

Data

Data sources

This table summarises all the data sources used for the case study.

Data Category	Data point description
Diary	Research diary of lead investigator (JN) – including reflections during the process
Emails and meeting notes	Email, meeting notes and notes from planning and discussion. This included involvement of potential participants in co-designing and co-refining the study.
Online pre-discussion survey	Informed consent and pre-discussion survey data
Learning resources	Learning resources for participants and the Facilitator giving information about genomics and using Loomio (see section 'Learning resources')
Online discussion with participants	Text data from online Loomio discussion with participants
Online discussion with facilitators	Text data from online Loomio discussion between facilitators of two parallel studies
Online post-discussion survey	Post-discussion survey data from participants
Follow up survey for facilitators	Post-discussion survey data from facilitators and additional emails with further reflections

STARDIT Preference Mapping (STARDIT- PM)

This table uses the Alpha version of the Standardised Data on Initiatives Preference Mapping (STARDIT-PM) to categorise the data into certain areas[196]. Preferences were recorded from all data sources, including the initial survey, online discussion with participants, online facilitator discussions, follow-up surveys with participants and with facilitators. Facilitator comments in the online discussion were not included. If the same participant made the same point at different stages, this was counted as one view. The standardised categorisation is intended to facilitate comparison with other studies. Accordingly, the content may be similar to other sections of the qualitative thematic analysis.

STARDIT-PM area and quantitative data	**Qualitative summary**
Views on who should be involved: **10 participants** shared views about who should be involved	Participants stated that anyone should be involved in research, with a experts, people affected by the research directly and the public all sharing perspectives in the context of research carried out with ethical oversight. Six participants stated that anyone should be involved in research, with one participant stating 'everyone should have a voice not just scientists and researchers' [P5]. Another participant stated 'it needs to be a wide-ranging discussion so that the benefits and possible problems can be fully explored' [P4]. One participant said it can depend 'what kind of research it is' and what the purpose is [P7], with another adding 'we all need to have a voice' as 'we may not be "experts" in genomics but our opinions must be respected and have validity' [P5]. Six participants stated those affected by research should be involved, however others challenged this saying this could provide 'a rather one-eyed perspective' [P4]. One participant stated 'some research will benefit certain people - those people should probably influence it if they are an identifiable group'. Another participant noted that 'we're all biased; whoever is affected by a condition is likely to want it prioritised'[P12]. Another participant concluded 'we're all biased; whoever is affected by a condition is likely to want it prioritised'[P12]. One participant stated that people (including the public and research participants) will have a 'variety of professional and technical and creative skills' which will be useful, with the most useful one being 'knowing ourselves' [P7]. One participant stated 'I am a strong supporter of patient involvement in medical care' and that 'involving members of the public' in genomic research was important in order to 'have their views, reactions, interpretations, questions, concerns sought, interacted with, and considered' [P11]. Two participants stated that experts (including 'scientists'[P9]) who 'know what they are doing' should be involved [P2], with 'research reviewed by ethics boards'[P9]. There was a recognition that different groups in society might have different interests and influence. One participant articulated groups including 'medical scientists', 'social scientists', 'psychologists' and the

	general public as being groups which should influence research, but noted that not all 'groups should have equal influence'[P10]. One participant asked 'there will be many interested groups so which ones will be listened to?' [P4] One participant stated that 'people who are not looking for personal gain, but who have a desire to improve quality of life and help us understand ourselves' should influence research [P6].
Views on specific tasks people involved could do: **8 participants** shared views about specific tasks people involved could do	One participant said that research participants should be involved in 'agreeing purpose, parameters and methods' [P7]. Another asked 'whatever format is decided upon who would decide on the points for discussion?, implying that participants' tasks should include deciding this [P4]. One participant added that it is a 'good idea to involve research subjects in formulating the research questions' [P10]. Another participant stated they should be involved in 'having a say in what research is supported by public money' and 'making sure that the uses and purposes to which the research is put are responsible and allied with the laws and mores of our society' – which includes 'ethical oversight' [P7]. One participant also added that the public should have a voice in how 'science and research can better involve' people [P5]. In reference to future research with the sibling group one participant stated that ideally 'we would be able to exert control over the use' of data [P7]. One participant felt they should be involved in 'seeking answers to old, or not yet thought of questions' and 'looking beyond the known into a murky unknown' [P6]. The discussion also explored who should be involved and in which tasks. One participant noted they didn't feel 'qualified' to 'comment on aspects of science itself' but felt 'strongly' that they should be involved in ethical decisions and sharing personal experiences to help inform research [P5]. They also stated experts 'need to drive research' but they 'cannot do it in vacuum' as the public need them and they need the public [P5].
Views on modes of communication: **4 participants** shared views about preferred communication modes	One participant stated 'moderated face to face discussions (through video if need be) remain the best method in my opinion for focussed outcomes and decisions with groups of people' [P7}. Another participant stated that 'most of our group would be able' to use video-conferencing platforms [P5]. Face-to-face synchronous discussion was ruled out by another participant as there are 'too many voices' which are across multiple time-zones [P4]. Communicating via a verbal interview was perceived as taking less time than anything which 'requires a lot of writing' [P4]. One participant said they were 'happy to contribute in any way in which is practical online' [P5], with another adding 'email is still the best way' [P4]. One participant suggested there would be 'differing preferences for how such a project should be organised' and suggested agreeing on some ideas (not using email) and then sending these ideas to the group once decided [P4]. One participant concluded that 'a forum for considered comments' online can be useful as long as enough time is allowed [P7].
Views on what methods should be used to involve people:	The participatory research method was described as 'commendable' [P9]. Participants suggested the idea of using one to one interviews as way of involving people (including using telecommunications) [P5], however, one participant noted that one-to-one interviews can restrict discussion and 'be quite straight jacketed with circumscribed questions', compared to online discussions [P4]. A 'group of "special

5 participants shared views about what methods should be used to involve people	interest" people involved in a group discussion' was suggested as a 'simple but effective method of encouraging debate', if participants can 'can dip in and out' [P4]. They also stated group discussion would not work as 'there are too many voices and some would be drowned out' [P4]. This participant also stated 'we will have differing preferences for how such a project should be organised' and asked 'would it be possible to agree on some ideas and then post them to the group' in order to involve people in co-creating how they will be involved and participate [P4]. One participant suggested that discussion, interviews, surveys and 'documents and videos shared for feedback' would all be viable methods of involving people [P5]. Another participant suggested a professional 'market researcher' who was 'tasked with finding a cross-section of people' might be a helpful method to involve people [P4]. One participant shared highly-specific views about the method that should be used, stating 'It should not be a plebiscite' nor 'self-electing moral Praetorian guard', adding that 'the more diverse the debate, the more dilute the effect of irrational preconception and ethical incompetence should become. The model of representative democracy seems to me the best available' [P8]. Another participant stated that 'ongoing discussions using social media and specific pages' such as Facebook pages, could be a good method to involve people [P5]. Another participant concluded that 'Moderated face-to-face discussions (through video if need be) remain the best method in my opinion for focussed outcomes and decisions with groups of people', adding that 'a forum for considered comments which are neither binding nor meant to be conclusive such as this can certainly be online' as long as enough time is allowed' [P7].
Views on enablers (facilitators) of involvement: **7 participants** and **3 facilitators** shared views about facilitators of involvement	One participant noted that being 'highly educated' was an enabler for involvement and that having a 'bit of time on their hands' was also an enabler [P4]. Being 'respectful' when involving 'those affected by genomic research' will facilitate research as the 'more brains applied to research, the more likely answer to puzzles will be found' [P11]. Similarly, another participant stated 'the more diverse the debate, the more dilute the effect of irrational preconception and ethical incompetence should become' [P8]. One participant stated that whatever model was chosen, it should be 'as flexible as possible' [P5]. Four participants reported specific things about the way this study was conducted that facilitated their involvement. One participant said the entire process was 'assiduous' and that the 'intent of this project' was 'obviously thoughtful and interesting'[P9]. One participant said the 'system seemed to work well' [P7]. Another added that being used to online platforms like Loomio, or having previous experience of similar

	platforms and 'used to' that way of communicating might facilitate involvement using that communication mode. One participant suggested an alternative discussion format where the participants discussed a thread for 2 days and then had a 3 day break before coming to another thread [P7]. The Facilitator (MC) noted that regular contact with the study team and timely support was essential and they 'could not have done it without this'.
Views on barriers of involvement: **6 participants** and **3 facilitators** shared views about barriers of involvement	Barriers to involvement in research identified by participants included public fear and 'hysteria' caused by a lack of understanding, which may 'hamper' research, involvement and general public support for research [P5]. Synchronous discussion was highlighted as another barrier if participants 'are across time zones' [P4]. One participant mentioned that they felt that their emotional response to some issues made it difficult to get involved in some ways [P6]. Being required to watch lengthy videos was identified as a barrier by one participant. One-to-one interviews were mentioned as being 'quite straight-jacketed with circumscribed questions' compared to more open online discussions [P4]. They also stated 'I don't think that a group discussion would work as there are too many voices and some would be drowned out' [P4] Four participants reported specific things about the way this study was conducted that were barriers to their involvement. A discussion about boundaries revealed that some participants felt 'avoiding topics which might trigger emotions which are stressful or unpleasant' could be viewed as 'restrictive, even censorious' [P7]. The pace of the discussions was mentioned as moving 'too quickly' with another adding 'more time' was needed and study team should 'reconsider the pace of the research' [P7] [P4] [P5]. Updates from the discussion were sent to participants according to their preferences, and one stated they 'lost track of emails' and were sometimes unsure if they were 'responding to the right part' [P6]. Two participants stated the 'platform presented technical difficulties' [P4] and that it was 'complicated' [P5]. One participant stated the 'premise and the purpose of the study could be clearer' and that the various discussion threads were 'difficult to untangle sometimes' [P7]. They also mentioned it was 'hard to be able to guarantee to do this every day for a period' and that not doing so meant they 'got lost' [P7]. Another participant added that 'it's a difficult subject to discuss in a vacuum, without real life examples' [P4]. One participant expressed 'trepidation' at sharing views about research and compared the feeling to getting an answer wrong in an 'exam' [P6]. The facilitator (MC) stated that they felt more time was required in the co-design process. In addition, the administrative processes surrounding the unplanned change of Chief Investigator and related administrative processes in relation to the ethics process (outside of the control of the study team) meant they felt support was 'non-existent' and was 'wholly inadequate' for the participatory research process being used.

Views on what the outcome or output of the involvement could be **2 participants** shared views on what the outcome or output of the involvement could be	One participant stated they wanted to know that their involvement had been 'useful to the researchers' [P11], with another stating an outcome of being involved would be the 'satisfaction of knowing that I may have contributed' [P10]
Views on which stage of the research people should be involved: **1 participant** shared views about which stage of the research people should be involved	One participant noted that changing study design in the later stages of a study not always possible, posing the question 'can we be both subjects and supervisors - at the beginning yes, later, maybe not' [P7]. The participant also stated that compared to 'highly-qualified scientists who know what's possible' some 'relatively ignorant lay people' will be limited in what they can offer at some stages [P7]. The participant suggested that getting the 'purposes, the parameters and the methods agreed with all participants' at the design stage might be most appropriate [P7].
Views on who should the data from this project shared with?: **3 participants** shared views on who data from this project should be shared with	Participants shared views about who data should be shared with, with One participant stated explicit concerns about sharing data for political or financial use [P5], with other participants agreeing. One participant stated 'Research is for humankind. Its benefits should be available to all. Information should be for the most part easily available. If it's publicly funded, it must be publicly available [P9]'. Cultural conventions around ownership of knowledge such as 'patenting' were challenged as forms of knowledge control which are 'unethical' in some contexts.
Views on how think learning from this research could be used **8 participants** shared views about how learning from this research could be used	Eight participants shared multiple views on how learning from a proposed study could be used, including predicting human traits (for example, risk of diseases or mental health problems). One participant indicated the study could help individuals in the group understand 'what unwelcome genes we might have inherited'[P4]. Participants also indicated the research could be used to improve understanding of how things like personal experience, socioeconomic circumstances and culture interact with genomics. One participant added 'longitudinal studies might well be set up to study a cohort of babies with certain genomic sequences which predispose them to certain diseases' [P4]. Another participant stated research with the group 'could have implications for all' people [P3].

Demographic information

Category	Pre-discussion survey	Post discussion Survey
Gender		
Female	7	3
Male	5	3
Age		
50-54 years	1	0
55-59 years	2	0
60-64 years	0	0
65-69 years	6	4
70-74-years	3	2
Educational background ('highest' only counted)		
Degree (bachelors), diploma or post-graduate ('higher')	11	5
High school qualifications (ages 16-19)	1	1
Number who agreed with following statements		
I feel comfortable describing other descendants of my biological father as ‘half-siblings’	12	N/A
I would describe our email group as an ‘online community’	9	N/A
Members of this email group potentially have a shared interest in discussing future research which might affect them, including genomic research	11	N/A
In which country do you live (or spend most time)?		
Canada	3	2
Greece	1	0
Spain	1	1
United Kingdom	7	3

Views about who should be involved in research

Participants were asked the same questions before and after the online discussion. When asked who should be involved in various tasks in research, participants could choose from the categories outlined in Figure 2 in the main article. A change in direction is described as 'widening', the inverse as 'narrowing'. Widening was calculated as being a move towards an attitude that more people should be involved in research, whereas narrowing was calculated as a move towards an attitude that fewer people should be involved.
A total of 54 responses were given by 6 participants where participants completed answers to questions of both the baseline and follow-up survey. 35% of responses showed a change towards 'widening' involvement (N=19/54) while 8% 'narrowed' (N=8/54). 50% of responses stayed the same (N=27/54).

Widening and narrowing for each question

Who should influence which aspects of research?	**Change to wider**	**No change**	**Change to narrower**
Finding questions to ask	2	4	0
Deciding which questions to prioritize and fund	2	3	1
Deciding how to try and answer the question (the research method)	2	2	2
Attempting to answer the question (carrying out the research, including collecting information)	2	3	1
Trying to understand if it is possible to the answer the question (analysing the information)	3	3	0
Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)	2	3	1
Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)	2	3	1
Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)	2	4	0
Designing how people are involved in the research	2	2	2
Change totals	19	**27**	**8**

Participant experience

How would you rate your overall experience of participating in the online discussion	
Somewhat good	4
Neither good nor bad	1
How would you rate how we conducted the survey, and discussion format	
Excellent	1
Somewhat good	2
Neither good nor bad	1
Somewhat poor	1
How would you rate the support you received to be involved (for example, practical support such as instructions for using the online tools)?	
Excellent	2
Somewhat good	2
Neither good nor bad	1
How would you rate the information and learning materials you were given before the event	
Excellent	3
Neither good nor bad	2
Did you feel you meaningfully contributed to the discussion?	
Yes	3
Unsure	3

Investigator shared learning group

During the online facilitation of the two online discussions, a shared learning group was established for facilitators and the study teams of two similar projects being run in parallel. The study teams shared reflections and learning about the process of facilitation online, as well as offering and receiving support regarding technical and practical issues. The data was coded and categorised, including using the STARDIT framework[196].

Data analysis

Quantitative analysis of all themes

Theme	Number of participants
Research for profit and 'Bullying' by 'big pharma'	6
Anyone should be involved in research	6
Those affected by research should be involved	6
Research with sibling group is unique and complex but important	6
Who decides who decides what is ethical	5
Concerns about genomics research being used for political purposes	5
Finding out they are part of sibling group has been a positive experience	4
View on topics for research	4
Participants reported changed views and perspectives as a result of participating	4
Desire to improve situation for people affected by assisted conception	3
Interested in learning what other siblings think and discuss issues	3
Concerns about control of knowledge and data	3
Questioning which groups should have 'equal influence'?	3
Questioning eugenic attitudes to genomic variations	3
Views on participation in genomics research	3
Participants learned about genomics	3
Motivation for participation to help researchers and sibling group	2
Uncertainty about what they can offer but happy to help	2
What is the purpose of research?	2
Experts should be involved (over seen by ethics boards)	2
Developments in genomics have significant implications for society	2
People have responsibility to be involved in research	1
Questioning giving power to experts	1
What control do research participants have?	1
People with specific experience and skills should be involved	1
The public and research participants should be involved in research	1
How should people be involved in genomics research	1
Tasks of involvement	1
Stages of involvement	1
Questions genomic medicine as intervention before other methods	1
Questioning genomics determinism	1
Choosing what to know about your genome	1

Qualitative data analysis

Emergent themes from qualitative participant data

This section organises qualitative data from participants into themes. This analysis has been provided in order to improve transparency of the qualitative analysis process which is summarised in the accompanying article.

Detailed Summary of Thematic Area 1: Participant views about involvement in genomic research

Participants demonstrated an understanding of the difference between participation in research and involvement in research. One participant stated 'I am a strong supporter of patient involvement in medical care' and that 'involving members of the public' in genomic research was important in order to 'have their views, reactions, interpretations, questions, concerns sought, interacted with, and considered' [P11]. Participants explored ideas around the purpose of research and one stated 'if I was the researcher running the project, I would want to get the purposes, the parameters and the methods agreed with all participants' [P7]. One participant also added that the public should have a voice in how 'science and research can better involve' people, which aligns with best-practice identified in a review of public involvement in genomics research [P5][618].

Six participants of the online discussion stated that anyone should be involved in research, with one participant stating 'everyone should have a voice not just scientists and researchers' [P5]. Another participant stated 'it needs to be a wide ranging discussion so that the benefits and possible problems can be fully explored' [P4]. One participant said it can depend 'what kind of research it is' and what the purpose is [P7], with another adding 'we all need to have a voice' as 'we may not be "experts" in genomics but our opinions must be respected and have validity' [P5].

Methods of involving people were discussed in detail with a number of options explored. Participants suggested that discussion, interviews, surveys, 'representative democracy' and 'documents and videos shared for feedback' would all be viable methods of involving people [P8] [P5]. The participatory research method was described as 'commendable' and some participants suggested the idea of using one to one interviews as way of involving people (including using telecommunications) [P9] [P5], however, one participant noted that one to one interviews can restrict discussion and 'be quite straight jacketed with circumscribed questions', compared to online discussions [P4]. This participant also stated 'we will have differing preferences for how such a project should be organised' and asked 'would it be possible to agree on some ideas and then post them to the group' in order to involve people in co-creating how they will be involved and participate [P4]. Another participant concluded that 'a forum for considered comments which are neither binding nor meant to be conclusive such as this can certainly be online' as long as enough time is allowed [P7].

Participants used a number of literary references to frame ethical debates, including citing *The Tempest* by Shakespeare as offering helpful analogies in exploring the ethics of the 'brave new world' of genomic research [P9][619], with one participant stating 'I feel more like a mix of Ariel or Caliban with respect to research - willing to help (and hoping not to be enslaved) but also willing to cede leadership' [P5]. Public fear and 'hysteria' caused by 'Brave New World fantasies' and a lack of understanding may 'hamper' involvement and general public support for research. [P9] Another participant disagreed, adding 'if we say nothing, do nothing that has to be worse doesn't it?' [P5].

One participant noted that changing study design in the later stages of a study not always possible, posing the question 'can we be both subjects and supervisors - at the beginning yes, later, maybe not?', recognising that involvement at the design stage is the most practicable, which aligns with other similar studies [P7][6].

Six participants stated those affected by research should be involved, however others challenged this saying this could provide 'a rather one eyed perspective' [P4]. One participant stated 'some

research will benefit certain people - those people should probably influence it if they are an identifiable group'. Another participant noted that 'we're all biased; whoever is affected by a condition is likely to want it prioritised' [P12]. Being 'respectful' when involving 'those affected by genomic research' will facilitate research as the 'more brains applied to research, the more likely answer to puzzles will be found' [P11]. One participant said that research participants should be involved in 'agreeing purpose, parameters and methods' [P7].

Six participants expressed concern about research for profit and those with financial interests influencing research. One participant stated they would be happy to participate in research but that they 'would however be concerned if this data was ever shared for commercial purposes' [P5]. Another participant noted they felt that certain pharmaceutical companies were responsible for 'bullying', contributing to 'disinformation; ignorance and inflexibility of medical and scientific professions' [P12]. Another participant raised concerns about 'sponsored facts' and asserted that the opinions about genomics research 'must be respected' [P5]. Another participant stated that 'the people paying for the research will influence it', noting that if it is 'public money the gatekeepers will have the greatest influence' [P7]. One participant said they find it 'distasteful that a private company can benefit from public research and then withhold the data or charge for it' [P9].

While some participants felt 'trust' towards existing scrutiny for research oversight [P5], ethics committees were identified as having 'failings', and one participant asked the question 'who will decide who will be on the ethics committee?' [P4]. Another participant added 'I am not sure of the ethics process but it does seem a shame that more of us cannot participate' [P5]. Another participant stated 'I trust the scientists and the ethical committees' [P9], while another noted that 'the ethics of DNA research generally will continue to be of huge importance and will continue as a political issue, triggering new laws and regulations' and raised a concern that 'the law will not be able to keep up with the research - and we as members of the general public won't either' [P7]. One participant noted that while people might be experts about a process or data, but that does not make them 'moral guardians' [P5]. They concluded 'no single body (and that includes the church and the government) has a right to dictate moral guidelines' [P5].

Participants indicated that all health policy decisions have political power associated with them, and five participants raised a number of specific concerns about genomic research being mis-used for political purposes and 'wicked ends' [P5]. One participant stated research should not be used 'for political purposes' but indicated they did not believe this was easily prevented [P5], with another participant sharing the 'realism about the likely corrupt use of genetic information for political and financial purposes' [P2].

Multiple participants agreed that the public can get involved in publicly funded research by overseeing what research is funded (identifying and prioritising) and ethical oversight [P7] [P5]. Three participants challenged established forms of knowledge and data control, stating that if research is 'publicly funded, it must be publicly available' and challenging concepts around intellectual property and patent laws, and that 'patenting new life forms is unethical' [P9]. Another participant stated they should be involved in 'having a say in what research is supported by public money' and 'making sure that the uses and purposes to which the research is put are responsible and allied with the laws and mores of our society' – which includes 'ethical oversight' [P7].

Views about genomics research in general

Four participants spontaneously raised the 'ugly' issue of eugenics and eugenic attitudes to genomic variations [P6] [P12]. A number of participants cited historical examples of genetic discrimination by regimes such as the Nazis as providing important learning for future genomics research. One participant cited the well documented historical precedent of the large technology company IBM being complicit in enabling regimes to carry out negative eugenics policies[454], stating IBM were 'the enablers for the Nazis ability to hunt down Jews and other "undesirables"' [P6]. The same participant also raised concerns about contemporary and future 'misuse' of genomic data 'for immigration' [P6].

Another participant raised concerns about deterministic applications of genomics for the early 'detection' of mental health problems perceived as 'undesirable' to those with political power [P7]. One participant noted that political 'demagogues' can influence public opinion and as a result they 'generally trust qualified researchers and ethics committees' and 'trust "people" far less' [P9]. Similarly, another participant stated 'the more diverse the debate, the more dilute the effect of irrational preconception and ethical incompetence should become' [P8].

Another participant raised the issue of pre-birth genetic testing, asking 'isn't it less ethical not to present informed choice', while acknowledging that the perception of impairment is subjective, and gave the example of the 'Deaf and Hearing Impaired community' not viewing deafness as 'an impairment' [P6]. The participant concluded with the question 'can we speak for those who are different to us?' [P6]. Another participant responded that 'The ethics of pre birth testing will never be clear cut' and cited the example of deaf couples to selecting 'an embryo for IVF that would be deaf like them' [P4][571].

There was a recognition that different groups in society might have different interests and influence. The discussion also explored who should be involved and in which tasks. One participant articulated groups including 'medical scientists', 'social scientists', 'psychologists' and the general public as being groups which should influence research, but noted that not all 'groups should have equal influence'[P10]. One participant noted they didn't feel 'qualified' to 'comment on aspects of science itself' but felt 'strongly' that they should be involved in ethical decisions and sharing personal experiences to help inform research [P5]. One participant asked 'there will be many interested groups so which ones will be listened to?' [P4].

One participant noted that being 'highly educated' was an enabler for involvement and that having a 'bit of time on their hands' was also a enabler [P4].

Detailed Summary of Thematic Area 2: Participant views about proposed research with sibling group

Participants recognised they were part of a 'unusual cohort' and suggested they would be useful to study [P4]. One participant stated they believed the sibling group 'were part of a eugenics programme in some way' [P4]. Participants shared multiple views about proposed research with the sibling group. One participant stated that 'this field of study is so huge and our involvement would be a 'first' in many ways' [P5]. Participants recognised the complexity of research on their 'community of shared interest' owing to multiple variables and one participant stated research with the sibling group would be 'enormously complicated' owing to 'confounding variables' but would be 'worth the effort' [P9]. Another participant added that they would 'wholeheartedly support the involvement of the next generation' in any future research with the sibling group and noted any study design should ensure new siblings and their offspring should be ensure they can 'become part of the research' [P5] .

One participant noted 'there is a world of difference between the idea of a study' of the sibling group and other genomic research [P7]. Another participant concluded that 'we are on the cusp of new forms of information , study and knowledge about ourselves' and stated that in the context of genomic research 'we are important in the grand scheme of things' [P6]. In a follow-up survey one participant stated they 'could not at all care whether my genomics are public or not. I do not see that my genome is a matter for privacy concerns' but recognised that others may feel differently [P9].

One participant proposed that research with the sibling group would 'not be trying to push a set agenda or profit financially' [P5]. Another stated that 'people who are not looking for personal gain' should be involved, including those 'who have a desire to improve quality of life and help us understand ourselves' [P6]. One participant added that it is a 'good idea to involve research subjects in formulating the research questions' [P10]. Another asked 'whatever format is decided upon who would decide on the points for discussion?, implying participants' tasks should include deciding this [P4]. One participant suggested that members of the sibling group could 'form and seek out participants for the Ethics Committee' [P6].

One participant stated it was a ‘civic duty’ to participate in research and that research participants should be involved in formulating the research questions [P10]. Participants spontaneously suggested potential research topics for the group, some serious issues and some appeared to be more light-hearted comments made in jest. More serious topics included ‘mental health’ [P6] and pharmacogenomics [P4], with more light-hearted suggestions including ‘career choices’ and hobbies [P4]. One participant felt they should be involved in ‘seeking answers to old, or not yet thought of questions’ and ‘looking beyond the known into a murky unknown’ [P6].
Participants shared multiple views about what possible areas of research topics could be explored in studies they could participate in and how these could be conducted Participants spontaneously suggested potential research topics for the group, some serious and some more light-hearted comments made in jest. More serious topics included ‘mental health’ [P6] and pharmacogenomics [P4], with more light-hearted suggestions including ‘career choices’ and hobbies [P4]. One participant felt they should be involved in ‘seeking answers to old, or not yet thought of questions’ and ‘looking beyond the known into a murky unknown’ [P6].
One participant suggested genomics research ‘a subject which cries out for more public discussion by those who have been unexpectedly and deeply affected by genomics findings’ [P5]. Another participant suggested a study of ‘any intersection of phenotypes’ including ‘psychological and intellectual’ with other genomics data [P8]. The participant concluded with the question ‘I have wondered what unwelcome genes we might have inherited?’, citing their own experience of genetic testing for both Parkinson’s and Huntington’s disease [P4].
One participant reported they were ‘happy to contribute any information from my own genome that might be useful in scientific or forensic research’ [P9]. One participant suggested a study design where participants can ‘all participate or not as we wish’ [P5]. They also commented they were ‘OK to provide genetic material for further analysis’. [P5]. Another participant concluded the siblings should ‘contribute their DNA’ for research [P9] and warned people from thinking that ‘your DNA is “you”’ [P9].
A ‘group of “special interest” people involved in a group discussion’ was suggested by one participant as a ‘simple but effective method of encouraging debate’, if participants can ‘can dip in and out’ [P4]. They also stated group discussion would not work as ‘there are too many voices and some would be drowned out’ [P4]. This participant also stated ‘we will have differing preferences for how such a project should be organised’ and asked ‘would it be possible to agree on some ideas and then post them to the group’ in order to involve people in co-creating how they will be involved and participate [P4].
The participatory research method was described as ‘commendable’ [P9]. One participant stated ‘moderated face to face discussions (through video if need be) remain the best method in my opinion for focussed outcomes and decisions with groups of people’ [P7}. Another participant stated that ‘most of our group would be able’ to use video-conferencing platforms [P5]. Face to face synchronous discussion was ruled out by another participant as there are ‘too many voices’ which are across multiple time-zones [P4]. Participants suggested the idea of using one to one interviews as way of involving people (including using telecommunications) [P5], however, one participant noted that one to one interviews can restrict discussion and ‘be quite straight jacketed with circumscribed questions’, compared to online discussions [P4].
In reference to future research with the sibling group one participant stated that ideally ‘we would be able to exert control over the use’ of data [P7].

Detailed Summary of Thematic Area 3: Participant views about the online discussion

Participants reported their motivation for participating in the research was to help researchers and the sibling group. Three participants stated they were participating partly because they were interested in learning what their siblings think, and the opportunity to ‘think through’ and ‘discuss’ issues together [P4] [P6].
Participants were asked to comment on their experience of participating in this study. One participant mentioned they were ‘surprised about the number of participants’ in the study, stating

they thought more siblings would have participated [P6]. Others stated the experience was 'interesting' and they 'enjoyed thinking about the questions posed and reading the responses of others' and the 'perceptive comments' of the Facilitator [P7] [P4]. One participant stated 'it is commendable that there is a concern about participatory research' in reference to the research methods used by the study team [P9]. However the participant also noted that in the early stages of participatory action research there was more discussion than action [P9].

Four participants reported specific things about the way this study was conducted that facilitated their involvement. One participant said the entire process was 'assiduous' and that the 'intent of this project' was 'obviously thoughtful and interesting'[P9]. One participant said the 'system seemed to work well' [P7]. Another added that being used to online platforms like Loomio, or having previous experience of similar platforms and 'used to' that way of communicating might facilitate involvement using that communication mode. One participant suggested an alternative discussion format where the participants discussed a thread for 2 days and then had a 3 day break before coming to another thread [P7].
Four participants reported specific things about the way this study was conducted that were barriers to their involvement. A discussion about boundaries revealed that some participants felt 'avoiding topics which might trigger emotions which are stressful or unpleasant' could be viewed as 'restrictive, even censorious' [P7]. The pace of the discussions was mentioned as moving 'too quickly' with another adding 'more time' was needed and the study team should 'reconsider the pace of the research' [P7] [P4] [P5]. Two participants stated the 'platform presented technical difficulties' [P4] and that it was 'complicated' [P5]. One participant stated the 'premise and the purpose of the study could be clearer' and that the various discussion threads were 'difficult to untangle sometimes' [P7]. They also mentioned it was 'hard to be able to guarantee to do this every day for a period' and that not doing so meant they 'got lost' [P7]. Another participant added that 'it's a difficult subject to discuss in a vacuum, without real life examples' [P4].

Detailed summary of all other thematic areas

Finding out they are part of sibling group has been a positive experience
The study team also noted that before the main discussion began, participants shared many personal experiences and reflections on finding out they were part of the extended family. For example four participants spontaneously reported that finding out they had siblings and being part of the siblings' online community was a positive experience, with one considering themselves 'lucky' [P4]. Another participant added 'I am so happy to be part of this [group]' and that the experience 'has changed and enriched my life' [P6]. They added 'we are beyond fortunate, we have our group' [P6]. Participants' reactions also suggested the siblings group offered a form of support, and are in contrast to contemporary views of the 1950s where people feared that offspring finding out about their origins might cause 'psychological injury'[430].

People have responsibility to be involved in research
One participant stated it was a 'civic duty' to participate in research and that research participants should be involved in formulating the research questions [P10].

Motivation for participation to help researchers and sibling group
Participants reported their motivation for participating in the research was to help researchers and the sibling group.

Uncertainty about what they can offer but happy to help
Two participants stated that while they were happy to help with research, they were uncertain about what they could offer or did not believe they were 'knowledgeable enough' [P6].

Desire to improve situation for people affected by assisted conception
Three participants stated they hoped future research would improve the situation for people affected by donor conception, as 'the views of donor conceived people were not considered for

many years' [P4]. One participant stated 'there should be better and accessible structures in place to support people through the process of self-discovery' [P5].

Interested in learning what other siblings think and discuss issues

Three participants stated they were participating partly because they were interested in learning what their siblings think, and the opportunity to 'think through' and 'discuss' issues together [P4] [P6].

Concerns about power imbalance in research

Seven participants shared multiple views about power imbalances in research, including concerns about bias, conflicting interests, data breaches and 'hidden motives' [P2], with genomic research being 'used to perverted ends' for both political and financial purposes [P2].

Control of knowledge and data

Three participants challenged established forms of knowledge and data control, stating that if research is 'publicly funded, it must be publicly available' and challenging concepts around intellectual property and patent laws, and that 'patenting new life forms is unethical' [P9]. Another participant raised concerns about 'sponsored facts' and asserted that the opinions about genomics research 'must be respected' [P5].

Who decides who decides what is ethical?

While some participants felt 'trust' towards existing scrutiny for research oversight [P5], ethics committees were identified as having 'failings', and one participant asked the question 'who will decide who will be on the ethics committee?' [P4]. One participant suggested that members of the sibling group could 'form and seek out participants for the Ethics Committee' [P6]. Another participant added 'I am not sure of the ethics process but it does seem a shame that more of us cannot participate' [P5]. Another participant stated 'I trust the scientists and the ethical committees' [P9], while another noted that 'the ethics of DNA research generally will continue to be of huge importance and will continue as a political issue, triggering new laws and regulations' and raised a concern that 'the law will not be able to keep up with the research - and we as members of the general public won't either' [P7]. One participant noted that while people might be experts about a process or data, but that does not make them 'moral guardians' [P5]. They concluded 'no single body (and that includes the church and the government) has a right to dictate moral guidelines'[P5].

Genomics research used for political purposes

While some participants indicated that all health policy decisions have political power associated with them, five participants raised a number of specific concerns about genomic research being mis-used for political purposes and 'wicked ends' [P5]. One participant stated research should not be used 'for political purposes' but indicated they did not believe this was easily prevented [P5], with another participant sharing the 'realism about the likely corrupt use of genetic information for political and financial purposes'[P2]. A number of participants cited historical examples of genetic discrimination by regimes such as the Nazis as providing important learning for future genomics research. One participant cited the well documented historical precedent of the large technology company IBM being complicit in enabling regimes to carry out negative eugenics policies[454], stating IBM were 'the enablers for the Nazis ability to hunt down Jews and other "undesirables"' [P6]. The same participant also raised concerns about contemporary and future 'misuse' of genomic data 'for immigration' [P6]. Another participant raised concerns about deterministic applications of genomics for the early 'detection' of mental health problems perceived as 'undesirable' to those with political power [P7].

Questioning giving power to experts 'reinforces dependency on experts'

One participant noted that 'blind fear and reinforces dependency on experts, at the expense of genuinely holistic solutions' and that the people should challenge the 'unquestioning faith in a kind of scientific determinism' which genomics can encourage [P12]. Another participant felt experts 'need to drive research' but they 'cannot do it in vacuum' as the public need them and they need the public [P5].

Research for profit and 'Bullying' by 'big pharma'

Six participants expressed concern about research for profit and those with financial interests influencing research. One participant stated they would be happy to participate in research but that they 'would however be concerned if this data was ever shared for commercial purposes [P5]. Another participant noted they felt that certain pharmaceutical companies were responsible for 'bullying', contributing to 'disinformation; ignorance and inflexibility of medical and scientific professions' [P12]. One participant proposed that research with the sibling group would 'not be trying to push a set agenda or profit financially' [P5]. Another stated that 'people who are not looking for personal gain' should be involved, including those 'who have a desire to improve quality of life and help us understand ourselves' [P6]. Another participant stated that 'the people paying for the research will influence it', noting that if it is 'public money the gatekeepers will have the greatest influence' [P7]. One participant said they find it 'distasteful that a private company can benefit from public research and then withhold the data or charge for it' [P9].

What control do participants have?

One participant raised the question of 'what type of control the subjects of the study were allowed' [P7]. They posed the question 'Can we be both subjects and supervisors' and suggested that this would be easier at the start during the design phase but more challenging later on in the research cycle [P7].

Why do we do research?

One participant stated that research is 'for humankind' [P9], with another going further and saying 'research is for every and any aspect of life', including other forms of life [P5].

Who should be involved in research

Participants stated that anyone should be involved in research, with a experts, people affected by the research directly and the public all sharing perspectives in the context of research carried out with ethical oversight.

Anyone should be involved in research

Six participants stated that anyone should be involved in research, with one participant stating 'everyone should have a voice not just scientists and researchers' [P5]. Another participant stated 'it needs to be a wide ranging discussion so that the benefits and possible problems can be fully explored' [P4]. One participant said it can depend 'what kind of research it is' and what the purpose is [P7], with another adding 'we all need to have a voice' as 'we may not be "experts" in genomics but our opinions must be respected and have validity' [P5].

Those affected by research should be involved

Six participants stated those affected by research should be involved, however others challenged this saying this could provide 'a rather one eyed perspective' [P4]. One participant stated 'some research will benefit certain people - those people should probably influence it if they are an identifiable group'. Another participant noted that 'we're all biased; whoever is affected by a condition is likely to want it prioritised'[P12].

People with specific experience and skills should be involved

One participant stated that people (including the public and research participants) will have a 'variety of professional and technical and creative skills' which will be useful, with the most useful one being 'knowing ourselves' [P7].

The public and research participants should be involved in research

One participant stated 'I am a strong supporter of patient involvement in medical care' and that 'involving members of the public' in genomic research was important in order to 'have their views, reactions, interpretations, questions, concerns sought, interacted with, and considered' [P11].

Experts should be involved (over seen by ethics boards)

Two participants stated that experts (including 'scientists'[P9]) who 'know what they are doing' should be involved [P2], with 'research reviewed by ethics boards'[P9]. One participant stated they 'loathe this current political atmosphere that is anti-intellectual, anti-expert, anti-science' and that is why they believe that genomics research should be 'left to the qualified' [P9]. This view was

challenged by another participant who stated that it was often people who think they are qualified who in fact sometimes know 'nothing at all', for example regarding personal experiences 'in practical terms' [P5].

Which groups should have 'equal influence'?

There was a recognition that different groups in society might have different interests and influence. The discussion also explored who should be involved and in which tasks. One participant articulated groups including 'medical scientists', 'social scientists', 'psychologists' and the general public as being groups which should influence research, but noted that not all 'groups should have equal influence'[P10]. One participant noted they didn't feel 'qualified' to 'comment on aspects of science itself' but felt 'strongly' that they should be involved in ethical decisions and sharing personal experiences to help inform research [P5]. One participant asked 'there will be many interested groups so which ones will be listened to? adding that 'It is easy to say only those people who are directly affected should have an influence on research but theirs could be a rather one eyed perspective' [P4].' [P4] One participant stated that 'people who are not looking for personal gain, but who have a desire to improve quality of life and help us understand ourselves' should influence research [P6].

Views on genomics research

Participants shared views on genomics which demonstrated a high-level of medical and genomic literacy. One participant who is a qualified medical professional stated developments in genomics have 'been the most significant of my lifetime' [P8].

Another participant questioned the use of genomic medicine as an intervention before other solutions such as 'simple lifestyle changes' [P12]. They also questioned 'a kind of scientific determinism' which genomic research can encourage.

Eugenics

Four participants spontaneously raised the 'ugly' issue of eugenics and eugenic attitudes to genomic variations [P6] [P12]. One participant stated they believed the sibling group 'were part of a eugenics programme in some way' [P4]. Another participant raised the issue of pre-birth genetic testing, asking 'isn't it less ethical not to present informed choice', while acknowledging that the perception of impairment is subjective, and gave the example of the 'Deaf and Hearing Impaired community' not viewing deafness as 'an impairment' [P6]. The participant concluded with the question 'can we speak for those who are different to us?' [P6]. Another participant responded that 'The ethics of pre birth testing will never be clear cut' and cited the example of deaf couples to selecting 'an embryo for IVF that would be deaf like them' [P4][620]. The participant concluded with the question 'I have wondered what unwelcome genes we might have inherited?', citing their own experience of genetic testing for both Parkinson's and Huntington's disease [P4].

Views on participation in genomics research

One participant suggested a study design where participants can 'all participate or not as we wish' [P5]. They also commented they were 'OK to provide genetic material for further analysis'. [P5]. Another participant concluded the siblings should 'contribute their DNA' for research [P9] and warned people from thinking that 'your DNA is "you"' [P9].

Research with siblings is unique and complex but important

One participant stated research with the sibling group would be 'enormously complicated' owing to 'confounding variables' but would be 'worth the effort' [P9]. Another participant added that they would 'wholeheartedly support the involvement of the next generation' in any future research with the sibling group and noted any study design should ensure new siblings and their offspring should be ensure they can 'become part of the research' [P5] . On participant noted 'there is a world of difference between the idea of a study' of the sibling group and other genomic research [P7]. Another participant concluded that 'we are on the cusp of new forms of information , study and knowledge about ourselves' and stated that in the context of genomic research. 'we are important in the grand scheme of things' [P6]. In a follow-up survey one participant stated the 'could not at all

care whether my genomics are public or not. I do not see that my genome is a matter for privacy concerns' but recognised that others may feel differently [P9].

Research topics – 'into a murky unknown'

Participants spontaneously suggested potential research topics for the group, some serious and some more light-hearted comments made in jest. More serious topics included 'mental health' [P6] and pharmacogenomics [P4], with more light-hearted suggestions including 'career choices' and hobbies [P4]. One participant felt they should be involved in 'seeking answers to old, or not yet thought of questions' and 'looking beyond the known into a murky unknown' [P6].

One participant suggested genomics research 'a subject which cries out for more public discussion by those who have been unexpectedly and deeply affected by genomics findings' [P5]. Another participant suggested a study of 'any intersection of phenotypes' including 'psychological and intellectual' with other genomics data [P8]. One participant also indicated that involvement in research might help people make sense of their personal experience and added joining an online discussion 'will let me think beyond my emotion'[P6].

Choosing what to know about your genome

Participants shared views which recognised there might be important variations in the knowledge people might chose to have about themselves and their genomes. One participant stated knowledge about whether someone was carrying a variation which pre-disposed them to Huntington's could be a 'poisoned chalice' [P4].

Co-creating discussion boundaries

In addition to inviting potential participants to help co-design discussions, participants were themselves invited to co-create their own boundaries for the group discussion by reviewing a suggested statement and suggesting amendments. The group was invited to self-creates code of conduct based on previous negative experiences of people unintentionally causing offence. One participant stated there was difficulty in self-censoring and knowing what others might find 'emotionally stressful or unpleasant' [P7]. One participant stated 'Common courtesy should be sufficient' [P2]Another participant suggested 'Perhaps we might agree that if one of us unintentionally makes a comment perceived as offensive by another that we agree either to apologise or explain our position' [P5].

Experience of participating in research

Participants were asked to comment on their experience of participating in this study. One participant mentioned they were 'surprised about the number of participants' in the study, stating they thought more siblings would have participated [P6]. Others stated the experience was 'interesting' and they 'enjoyed thinking about the questions posed and reading the responses of others' and the 'perceptive comments' of the Facilitator [P7] [P4]. One participant stated 'it is commendable that there is a concern about participatory research' in reference to the research methods used by the study team [P9]. However the participant also noted that in the early stages of participatory action research there was more discussion than action [P9].

Enablers and barriers specific to this study

Participants reported a number of enablers and barriers for involvement which were specific to this study.

Enablers of participation and involvement

Four participants reported specific things about the way this study was conducted that facilitated their involvement. One participant said the entire process was 'assiduous' and that the 'intent of this project' was 'obviously thoughtful and interesting'[P9]. One participant said the 'system seemed to work well' [P7]. Another added that being used to online platforms like Loomio, or having previous experience of similar platforms and 'used to' that way of communicating might facilitate involvement using that communication mode. One participant suggested an alternative discussion format where the participants discussed a thread for 2 days and then had a 3 day break before coming to another thread [P7]. The Facilitator (MC) noted that regular contact with the study team and timely support was essential and they 'could not have done it without this'.

Barriers of participation and involvement

Four participants reported specific things about the way this study was conducted that were barriers to their involvement. A discussion about boundaries revealed that some participants felt 'avoiding topics which might trigger emotions which are stressful or unpleasant' could be viewed as 'restrictive, even censorious' [P7]. The pace of the discussions was mentioned as moving 'too quickly' with another adding 'more time' was needed and the study team should 'reconsider the pace of the research' [P7] [P4] [P5]. Updates from the discussion were sent to participants according to their preferences, and one stated they 'lost track of emails' and were sometimes unsure if they were 'responding to the right part' [P6]. Two participants stated the 'platform presented technical difficulties' [P4] and that it was 'complicated' [P5]. One participant stated the 'premise and the purpose of the study could be clearer' and that the various discussion threads were 'difficult to untangle sometimes' [P7]. They also mentioned it was 'hard to be able to guarantee to do this every day for a period' and that not doing so meant they 'got lost' [P7]. Another participant added that 'it's a difficult subject to discuss in a vacuum, without real life examples' [P4]. One participant expressed 'trepidation' at sharing views about research and compared the feeling to getting an answer wrong in an 'exam' [P6].

The Facilitator (MC) stated that they felt more time was required in the co-design process. In addition, the administrative processes surrounding the unplanned change of Chief Investigator and related administrative processes in relation to the ethics process (outside of the control of the study team) meant they felt support was 'non-existent' and was 'wholly inadequate' for the participatory research process being used.

Emergent themes from qualitative investigator discussion data

A number of themes were identified during a qualitative thematic analysis of the discussion. In order to maintain confidentiality, comments have not been attributed to individual facilitators and have been shared with the permission of those who participated in discussions.

A personal or professional perspective?

Facilitators reported finding it a challenge to separate 'personal' experiences from 'professional perspectives' when facilitating. The platform was new to all facilitators and a considerable amount of time was required in order to both train and support facilitators using the platform. While some issues were platform specific, any such platform will require training and ongoing support for those new to using it. This includes real-time video and voice calls.

Facilitators required guidance on using personal experience, with the confidential investigator group serving as a place for advice and support. Future research of this kind should ensure that facilitators are supported appropriately, including being part of an active and confidential community of practice in order to give and receive practical, emotional and technical support.

One members of the study team noted that facilitation is 'not a dispassionate neutral and purely intellectual' activity, and that some kind of personal motivation for facilitation can be helpful. This aligns with findings from the online discussion with participants, where the personal experience of being part of the sibling group was hard to separate from general views about genomics research. When analysing the data, one Facilitator noted 'subjectivity isn't something shameful to be avoided - it's something to be acknowledged'. They later added:

> 'I think this discussion has also explored the boundaries of 'researcher'/'neutral Facilitator'/'person with skin in the game' - stepping back, they are all social constructions - some defined by law/ethics - others by un-codified conventions - and labelling them all is quite a liberating process.'

Critical mass

Facilitators noted a 'critical mass' effect in online discussions, with the pace of comments seemingly affected by number of posts. The rate of posts to a discussion would increase exponentially, with more comments seeming to generate more comments in a 'snow-ball' effect. Conversely,

discussions with fewer comments and infrequent posts attracted fewer replies, with 'energy levels' appearing to 'drop'. Facilitators theorised people were more confident to post in forums if there were more people posting, so they were less 'exposed'. This aligns with findings from other studies which have explored participants' hesitancy in posting in online discussion forums[453].

Discussion pacing and participant engagement

When co-designing discussions, the study team attempted to balance discussion pacing between being too slow and losing engagement and being too fast and overwhelming participants. After the discussions one Facilitator reflected they feared that 'introducing a new thread might interrupt the flow' and that it was difficult to judge this using this with online discussions.
Facilitators also reported struggling to balance keeping people engaged and trying to get hesitant participants (people logging in, reading, but not commenting) to comment. Similarly, some active participants in discussions would not post for a number of days. Follow-up survey data from participants suggested that sometimes they could not post owing to other commitments, but the nature of online discussions meant they could catch up and join in when they had the time.
One Facilitator noted that they underestimated how long it would take for people to get 'properly signed up', including giving technical support to participants. This created a tension when they 'wanted the latecomers to have time to 'catch up' without being overwhelmed'. Another Facilitator also commented that they felt it took a week for trust to be established in the group, for example, for people to see that the co-created rules of the discussion were observed by everyone.
Another Facilitator noted that:

> 'an advantage of this kind of discussion is that you can have those simultaneous discussions - yes people have a finite read/write/processing time in a day - but it allows those with interests in specific areas to pursue that without feeling left out, as they might do with a more traditional linear time discussion (like face to face or synchronous webinars)'

They also noted it allowed people to 'refer back' to previous discussions, making them more rich. Participants also reported enjoying having time to reflect on comments and reconsider views in light of others' comments.

Prescribed discussion and open-ended discussion

One investigator noted the difficulty in achieving 'the balance of being prescriptive (for consistency) and giving freedom to Facilitators' to initiate discussions and follow emergent themes. Participants reported that they enjoyed being able to raise issues and that this was preferable over the more traditional scripted interviews, as it allowed areas to be discussed that researchers might not have considered. Facilitators agreed that using the 'forking' function on Loomio to 'fork' discussions moving in different directions into threads should be avoided as might confuse both participants and facilitators.

Ethical limitations for participatory research

The entire study team agreed that limitations in the ethics process affected the extent of how the sibling group could be involved in the study. Internationally, confusion still surrounds what ethical approval is required before involving potential participants in co-designing research as 'specialist advisors'[578], with some guidelines now emmerging[127]. On the advice of the La Trobe University Human Research Ethics Committee, we did not approach potential participants about co-designing the study until after ethics approval had been granted, with feedback from participants being incorporated by a number of subsequent modifications to the original ethics application. As a result of the complex process of modifications, the timeline for feedback was shorter than the study team had anticipated, although the process did provide useful feedback[127]. Ambiguous policies for the ethical involvement of people in co-designing research can hamper the degree of control potential participants have in research and further clarity from ethics committees will enhance power sharing at this crucial stage of research.

Impacts from the process: Detailed summary

Eight specific impacts were reported from this process. These are summarised in detail below.

Impact 1: Improved understanding of genomics

While participants showed a good-baseline level understanding of genomics, three reported their understanding about genomics and research increased as a result of participating in the study. However some participants demonstrated a lack of self-confidence in their understanding, in spite of demonstrating a good understanding of the principles of genomic research, citing relevant peer-reviewed literature in discussions and discussing the nuances of ethical oversight.

Impact 2: Learning resources useful

Participants reported finding the information resources and videos useful. One participant commented they had learned from the visual summary of the review of public involvement in genomic research stating 'I didn't realise there were so many research projects involving global genomics projects but it is so good to read that public involvement is becoming more and more important' [P5] However one participant stated 'I must say that personally I hate being required to watch videos but think I am in a minority' [P4].

Impact 3: Changed views and perspectives as a result of participating

Four out of the six participants who completed the follow up survey noted their views and perspectives changed as a result of participating. One participant stated 'I now realise how fast the field of genomics is changing and there are all kinds of implications especially in the field of precision medicine' [P5]. One participant also indicated that involvement in research might help people make sense of their personal experience and added joining an online discussion 'let me think beyond my emotion' [P6]. Another stated 'involving people in genomic research is crucial' as it has 'unknown consequences and needs as wider discussion as possible'[P4]. A number of participants had views about 'leaving research to the qualified' challenged by other participants [P9], with follow-up survey data suggesting that those challenged changed their views about who should be involved, towards widening. The changed views of the participants involved can be viewed as an impact of 'transformative learning' [131].

Impact 4:Participants asked to stay involved in the research

All participants who completed the follow-up survey requested to stay involved in the research process, including in analysing data and being co-authors on the paper.

Impact 5: Participants enjoyed the online discussions

Participants stated the experience of participating was 'interesting' and they 'enjoyed thinking about the questions posed and reading the responses of others' and the 'perceptive comments' of the Facilitator [P7] [P4]. Another participant added that it 'work well' as a way of involving people [P7].

Impact 6: Improved understanding of how to get involved in research

Participants reported improved understanding of how to get involved in research. However some participants demonstrated a lack of self-confidence in their usefulness in being involved in genomics research. Learning and development interventions to help people understand the valuable tasks they can complete in the research process might improve confidence.

Impact 7: Co-design changed study design

Feedback from participants resulted in changes to the study design including improving language used in recruitment, improving the online discussions and learning resources.

Impact 8: Method for future research co-design established

By co-creating methods of involving participants in proposed future genomics research, this process has demonstrated a practical and well-evaluated method of involving potential participants in co-designing research. Participants stated that the methods used in this process could be helpful when co-designing future stages of proposed genomic research with the sibling group.

Additional File 2: Standardised Data on Initiatives (STARDIT) report – Alpha Version: Co-designing genomics research with a large group of donor-conceived siblings

About this report

This report uses the Standardised Data on Initiatives Alpha version (STARDIT)[196]. The 'living' STARDIT Beta version[442] report which relates to this project can be found in the references[295].

STARDIT Report Alpha Version

Identifying information	
Initiative name	Co-designing genomics research with donor -conceived siblings
Geographic location or scope	Australia
Date range (planned start and end dates of initiative)	2017-2020
Purpose of the initiative	Participatory action research to involve members of a sibling group in online discussions about how they would like to be involved in future research.
Organisations or other initiatives involved (list all if multi-centre)	4. School of Psychology and Public Health, La Trobe University
Funding sources	School of Psychology and Public Health, La Trobe University
Clinical trial registration details (if applicable)	N/A
Ethics approval (if applicable)	La Trobe University
Other relevant information (free text)	This report describes involving members of a sibling group in online discussions about how they would like to be involved in future research.
At which stage of the research project has this report been written? (Select from: 4. **Before** the intervention or initiative– this report is prospective or describes planned activity 5. **Ongoing** – the intervention or initiative is still taking place 6. **After** the research project or initiative has occurred	**After** the participatory action research occurred.

Methods of the initiative (what is planned to be done, or is being reported as done)	The research process was co-designed using a participatory action research method to involve members of a sibling group in the co-design of online discussions to explore future genomic research with members of the group.
Report authorship	
Name	1: Jack Nunn 2: Marilyn Crawshaw 3: Paul Lacaze 4: Shirley Brailey 5: Barbara Nunn 6: Adrianne Smith 7: Barry Stevens
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Other information	Other people were involved in writing this report and contributing data but did not want to be named.
Key contact at initiative for confirming report content (include institutional email address)	Jack Nunn, PhD researcher, School of Psychology and Public Health, La Trobe University, jack.nunn@latrobe.edu
Involvement	
Who was involved or how would you label groupings of those involved	Group 1: Academic research investigators (Jack Nunn, Marilyn Crawshaw and Paul Lacaze) Group 2: Members of the sibling group who gave feedback during the co-design stage (including but not limited to Becky Gardiner, David Gollancz and Michael Bywater) Group 3: Members of the sibling group who participated in the research and gave feedback as part of the co-design process, the manuscript checking stage or contributed data to the STARDIT report (including but not limited to Shirley Brailey, Barbara Nunn, Adrianne Smith and Barry Stevens)

How many people were in each grouping label?	Group 1: 3 Group 2: 5 Group 3: 18
Specific tasks of this person or group (list as many as possible) – *including any information about why certain people were included or excluded in certain tasks*	Group 1: Involved in co-designing every stage of the process, analysing data Group 2: Members of the sibling group were involved in refining wording of participant information, sharing views and advice about the process, proof-reading documents, providing feedback on surveys, analysing data, informing planning, and providing feedback on planned online discussions. Group 3: Participants were also involved in checking the content of Genetics Society UK podcast[455], with the recording shared with all participants before dissemination to ask them to check the content was accurate and acceptable. Group 2 and 3: Participants were sent the article and additional files to check the analysis and content and were invited to be authors of the STARDIT report.
How were these people involved (what methods were used)	Group 1: Face-to-face meetings, video calls, email communication, shared online documents, teleconferences. Group 2: video calls, email communication, shared online documents, teleconferences.
Facilitators of involvement (what do you expect will help these people get involved – or what helped them get involved)	Giving people time to read resources. Clear communication about the intention of involving people. One participant noted that being 'highly educated' was an enabler for involvement and that having a 'bit of time on their hands' was also an enabler [P4]. Being 'respectful' when involving 'those affected by genomic research' will facilitate research as the 'more brains applied to research, the more likely answer to puzzles will be found' [P11]. Similarly, another participant stated 'the more diverse the debate, the more dilute the effect of irrational preconception and ethical incompetence should become' [P8]. One participant stated that whatever model was chosen, it should be 'as flexible as possible' [P5]. Four participants reported specific things about the way this study was conducted that enabled their involvement. One participant said the entire process was 'assiduous' and that the 'intent of this project' was 'obviously thoughtful and interesting'[P9]. One participant said the 'system seemed to work well' [P7]. Another added that being used to online platforms like Loomio, or having previous experience of similar platforms and 'used to' that way of communicating might facilitate involvement using that communication mode. One participant suggested an alternative discussion format where the participants discussed a thread for 2 days and then had a 3-day break before coming to another thread [P7]. The Facilitator (MC) noted that regular contact with the study team and timely support was essential and they 'could not have done it without this'.
Barriers of involvement (what do you expect will inhibit	Face-to-face meetings were difficult to organise. The study team was located in both Australia and the UK, so face-to-face meetings were not

these people from getting involved – or what inhibited them from getting involved). Are there any known equity issues which may contribute?	possible. Unclear communication about intentions and purpose of the involvement contributed to confusion (explaining how involvement is distinct from participation was challenging). Ensuring those involved had enough time to give feedback was also a challenge. Barriers to involvement in research identified by participants included public fear and 'hysteria' caused by a lack of understanding, which may 'hamper' research, involvement, and general public support for research [P5]. Synchronous discussion was highlighted as another barrier if participants 'are across time zones' [P4]. One participant mentioned that they felt that their emotional response to some issues made it difficult to get involved in some ways [P6]. Being required to watch lengthy videos was identified as a barrier by one participant. One-to-one interviews were mentioned as being 'quite straight-jacketed with circumscribed questions' compared to more open online discussions [P4]. They also stated 'I don't think that a group discussion would work as there are too many voices and some would be drowned out' [P4] Four participants reported specific things about the way this study was conducted that were barriers to their involvement. A discussion about boundaries revealed that some participants felt 'avoiding topics which might trigger emotions which are stressful or unpleasant' could be viewed as 'restrictive, even censorious' [P7]. The pace of the discussions was mentioned as moving 'too quickly' with another adding 'more time' was needed and study team should 'reconsider the pace of the research' [P7] [P4] [P5]. Updates from the discussion were sent to participants according to their preferences, and one stated they 'lost track of emails' and were sometimes unsure if they were 'responding to the right part' [P6]. Two participants stated the 'platform presented technical difficulties' [P4] and that it was 'complicated' [P5]. One participant stated the 'premise and the purpose of the study could be clearer' and that the various discussion threads were 'difficult to untangle sometimes' [P7]. They also mentioned it was 'hard to be able to guarantee to do this every day for a period' and that not doing so meant they 'got lost' [P7]. Another participant added that 'it's a difficult subject to discuss in a vacuum, without real life examples' [P4]. One participant expressed 'trepidation' at sharing views about research and compared the feeling to getting an answer wrong in an 'exam' [P6]. The Facilitator (MC) stated that they felt more time was required in the co-design process.
What was the outcome or output of the involvement of these people? What changed as a result of involving people?	Improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the sibling group to describe biological relations), improved online discussion, improved learning resources for participants, improved co-design process.
At which stage of the initiative were these people involved? (select from list of pre-defined stages or allow 'other')	Group 1: All stages Group 2: Co-design, evaluation and dissemination

What was the estimated financial cost for involving people. How much time did it take. Were there any costs that cannot be measured financially?	$0 AUD – people volunteered their time. The total number of hours volunteered (excluding participation and the contributions of co-investigator MC) is estimated to be 25.
What worked well, what could have been improved? Was anything learned from the process of involving these people?	Involving potential participants in co-defining language used to describe the sibling group helped ensure that language was acceptable and appropriate. The co-design process took longer than expected owing to ethical 'grey areas' with no clear instruction on whether ethics approval was required to involve people in co-design. As a result an ethics application was made and subsequent feedback from the co-design process was integrated using modifications to the ethics application. The entire study team agreed that limitations in the ethics process affected the extent of how the sibling group could be involved in the study. Internationally, confusion still surrounds what ethical approval is required before involving potential participants in co-designing research. On the advice of the La Trobe University Human Research Ethics Committee, the study team did not approach potential participants about co-designing the study until after ethics approval had been granted, with feedback from participants being incorporated by a number of subsequent modifications to the original ethics application. As a result of the complex process of modifications, the timeline for feedback was shorter than the study team had anticipated, although the process did provide useful feedback. Ambiguous policies for the ethical involvement of people in co-designing research can hamper the degree of control potential participants have in research and further clarity from ethics committees will enhance power sharing at this crucial stage of research.
Mapping financial or other 'interests'	
Describe any financial relationship or other interest this person has to this project	One investigator (Jack Nunn) is biologically related to participants from the sibling group, with one being his mother and all being half-aunts or uncles.
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	It will be published open access in peer reviewed journals with identifying information removed in order to anonymise it as much as possible.
How is it stored and hosted?	It will be shared on a public domain repository.
Who is analysing the data?	Group 1: The study team described above Group 2: participants were invited to review the analysis and give feedback to ensure they felt it reflected their experience of the process

What methods will be used to analyse the data (including a link to any relevant code and information about validity)	We used case study methodology to describe our experience involving participants in the co-design of the proposed study. We collected and analysed both qualitative and quantitative data during the involvement activities. We analysed data from online surveys and online discussions with participants. In addition, data from the study team communications was included, such as meeting notes, emails, reflexive diary entries and survey responses of study investigators. Coding and thematic analysis of qualitative data was carried out by two authors independently and checked by other authors.
How is information about this data disseminated?	10. It will be published in an open access journal 11. It will be shared with participants of the research and also other members of the sibling group who have joined it since the study commenced 12. Learning from this process has been presented at conferences, and will be shared on social media and through other channels. Preliminary learning was shared in a UK Genetics Society podcast[455].
Who 'owns' the data or claims any kind of 'intellectual property' (include relevant licensing information)	Confidential data collected as part of the study is stored according to laws and the data access plan approved by La Trobe University. The authors maintain 'ownership' of the data in the paper and is shared under the Creative Commons license used by the publishing journal.
Who controls access to the data	The study team, La Trobe University and participants will be involved in any future data access decisions.
How is/will the data be 'Findable, Accessible, Interoperable, Reusable' according to the FAIR criteria?	Data will be shared in the public domain and licensed under a Creative Commons license.
Impacts and outcomes	
What new knowledge has been generated? (if appropriate, include effect size, relevant statistics and level or evidence)	8. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources 9. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'.
What was learned	Involving people in online discussions about involvement in research changes people's views about who should be involved in research, including participants 'widening' their views about who should be involved in research to include more people.
Knowledge translation	3. Knowledge from this process will inform the design of a future genomic research 4. Learning from this process can inform future involvement activities

	5. Learning from this co-design process can inform future ways of involving people in genomic research including co-designing self-governed biobanks. 6. Learning from this process was shared in a Genetics Society UK podcast[455], with the recording shared with all participants before dissemination to ensure the content was accurate and acceptable.
Outcomes	8. **Co-design changed study design.** Feedback from participants resulted in changes to the study design including improving language used in recruitment, improving the online discussions and learning resources. 9. **The process improved participants understanding about genomics and research**. Participants had an improved understanding of genomics. While participants showed a good-baseline level understanding of genomics, three reported their understanding about genomics and research increased as a result of participating in the study. However, some participants demonstrated a lack of self-confidence in their understanding, in spite of demonstrating a good understanding of the principles of genomic research, citing relevant peer-reviewed literature in discussions and discussing the nuances of ethical oversight. 10. **Participants' improved understanding about genomics and research helped them make informed decisions about invitations to join genomics research studies**, which were shared with members of the sibling group after the process by researchers unconnected with this study. 11. **Learning from this process informed subsequent discussions in the sibling group about participation in research**, including a proposed self-managed biobank. Method for future research co-design established. By co-creating methods of involving participants in proposed future genomics research, this process has demonstrated a practical and well-evaluated method of involving potential participants in co-designing research. Participants stated that the methods used in this process could be helpful when co-designing future stages of proposed genomic research with the sibling group. 12. **Participants reported finding the learning resources useful** (including infographics and videos). One participant commented they had learned from the visual summary of the review of public involvement in genomic research stating 'I didn't realise there were so many research projects involving global genomics projects but it is so good to read that public involvement is becoming more and more important' [P5] 13. **Participants changed views and perspectives about genomics research as a result of participating.** Four out of the six participants who completed the follow up survey noted their views and perspectives changed as a result of participating. One participant stated 'I now realise how fast the field of genomics is changing and there are all kinds of implications especially in the field of precision medicine' [P5]. One participant also indicated

	that involvement in research might help people make sense of their personal experience and added joining an online discussion 'let me think beyond my emotion' [P6]. Another stated 'involving people in genomic research is crucial' as it has 'unknown consequences and needs as wider discussion as possible'[P4]. A number of participants had views about 'leaving research to the qualified' challenged by other participants [P9], with follow-up survey data suggesting that those challenged changed their views about who should be involved, towards widening. The changed views of the participants involved can be viewed as an impact of 'transformative learning'. 14. **Participants asked to stay involved in the research.** All participants who completed the follow-up survey requested to stay involved in the research process, including in analysing data and being co-authors on the paper 15. **Participants enjoyed the online discussions**. Participants stated the experience of participating was 'interesting' and they 'enjoyed thinking about the questions posed and reading the responses of others' and the 'perceptive comments' of the Facilitator [P7] [P4]. Another participant added that it 'worked well' as a way of involving people [P7].
How has or how will this be measured?	Future STARDIT reports
Who is involved in measuring this?	The study team and participants

Additional File 3 - GRIPP2 report: Co-designing genomics research with a large group of donor-conceived siblings

This report has been completed using the 'GRIPP2 reporting checklists: tools to improve reporting of patient and public involvement in research' available at: https://doi.org/10.1136/bmj.j3453.

GRIPP2 short form

Section and topic	Category description	Data
1: Aim	Report the aim of PPI in the study	Participatory action research to involve members of a sibling group in online discussions about how they would like to be involved in future research.
2: Methods	Provide a clear description of the methods used for PPI in the study	The research process was co-designed using a participatory action research method to involve people from the sibling group in the co-design of online discussions to explore future genomic research with members of the group. Participants were also involved in checking the final version of the paper.
3: Study results	Outcomes—Report the results of PPI in the study, including both positive and negative outcomes	Improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the sibling group to describe biological relations), improved online discussion, improved learning resources for participants, improved co-design process. Enablers of involvement: Four participants reported specific things about the way this study was conducted that enabled their involvement. One participant said the entire process was 'assiduous' and that the 'intent of this project' was 'obviously thoughtful and interesting'[P9]. One participant said the 'system seemed to work well' [P7]. Another added that being used to online platforms like Loomio, or having previous experience of similar platforms and 'used to' that way of communicating might facilitate involvement using that communication mode. One participant suggested an alternative discussion format where the participants discussed a thread for 2 days and then had a 3-day break before coming to another thread [P7]. Barriers of involvement: Four participants reported specific things about the way this study was conducted that were barriers to their involvement. A discussion about boundaries revealed that some participants felt 'avoiding topics which might trigger emotions which are stressful or unpleasant' could be viewed as 'restrictive, even censorious' [P7]. The pace of the discussions

Section and topic	Category description	Data
		was mentioned as moving 'too quickly' with another adding 'more time' was needed and study team should 'reconsider the pace of the research' [P7] [P4] [P5]. Updates from the discussion were sent to participants according to their preferences, and one stated they 'lost track of emails' and were sometimes unsure if they were 'responding to the right part' [P6]. Two participants stated the 'platform presented technical difficulties' [P4] and that it was 'complicated' [P5]. One participant stated the 'premise and the purpose of the study could be clearer' and that the various discussion threads were 'difficult to untangle sometimes' [P7]. They also mentioned it was 'hard to be able to guarantee to do this every day for a period' and that not doing so meant they 'got lost' [P7]. Another participant added that 'it's a difficult subject to discuss in a vacuum, without real life examples' [P4]. One participant expressed 'trepidation' at sharing views about research and compared the feeling to getting an answer wrong in an 'exam' [P6]. The Facilitator (MC) stated that they felt more time was required in the co-design process.
4: Discussion and conclusions	Outcomes—Comment on the extent to which PPI influenced the study overall. Describe positive and negative effects	Involvement improved participant information resources, improved wording that was culturally appropriate (using terminology preferred by the group to describe themselves), improved online discussion, improved learning resources for participants, improved co-design process. Involving potential participants in co-defining language used to describe the group of people affected helped ensure that language was acceptable and appropriate. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'.
5: Reflections/critical perspective	Comment critically on the study, reflecting on the things that	The co-design process took longer than expected owing to ethical 'grey areas' with no clear instruction on whether ethics approval was required to involve people in co-design. As a result,

Section and topic	Category description	Data
	went well and those that did not, so others can learn from this experience	an ethics application was made and subsequent feedback from the co-design process was integrated using modifications to the ethics application. Involving potential participants in co-defining language used to describe the group of people affected helped ensure that language was acceptable and appropriate. Involving participants in co-designing the research process resulted in a number of changes to the study design, including improving language used in recruitment and learning resources. The process of involving people can be viewed as a learning experience for both the participants involved and study team members. The process changed participants' views about who should be involved, which can be viewed as an impact of 'transformative learning'. Involving people in online discussions about involvement in research changes people's views about who should be involved in research, including participants 'widening' their views about who should be involved in research to include more people.

Appendices for Chapter 8 – Additional files: Involving Australian Indigenous peoples in precision medicine

Prospective STARDIT Report: A Pathway to precision medicine for Aboriginal Australians

This STARDIT report contains additional data relevant to the protocol 'A pathway to precision medicine for Aboriginal Australians'.

The 'living' STARDIT Beta version report which relates to this project can be found in the references and at this link:[458]

https://wikispore.wmflabs.org/wiki/A_Pathway_to_Precision_Medicine_for_Aboriginal_Australians:_A_Study_Protocol

A machine readable version of this report can be found here:
https://www.wikidata.org/wiki/Q113417061

Identifying information	
Initiative name	Using Co-design to Enable Precision Medicine for Aboriginal Australians
Geographic location or scope	New South Wales, Australia
Date range (planned start and end dates of initiative)	Start: February 2021 End: December 2022
Purpose of the initiative	The development of a new protocol using co-design methods to enhance the potential use of Precision Medicine for Aboriginal Australians.
Organisations or other initiatives involved (list all if multi-centre)	1. Poche Centre for Indigenous Health 2. The National Centre for Indigenous Genomics (NCIG)
Funding sources	Poche Centre for Indigenous Health
Clinical trial registration details (if applicable)	
Ethics approval (if applicable)	
Other relevant information (free text)	This project is currently in the planning phase and seeking ethics approval before beginning the co-design phase.
At which stage of the research project has this report been written?	**Before** the initiative– this report is prospective and describes planned activity
Methods of the initiative (what is planned to be done, or is being reported as done)	This is an iterative qualitative study consisting of five main phases. In Phase-I, we will ensure appropriate governance of the project, establish a team of investigators and a Project Advisory Committee which includes Aboriginal community representatives. Following an initial consultation with the Aboriginal community, we will begin Phase-II with the advertisement of the co-design workshops and invite community members to participate. In Phase-

	III, the Chief Investigators will participate in co-design workshops and document ideas generated during the workshops. The notes shall be subsequently analyzed thematically in Phase-IV with Aboriginal community representatives. Summaries of notes regarding participant recruitment, the consent process, DNA sample collection and storage, data governance and sovereignty, and the reporting of results and incidental findings are disseminated to the community. Lastly, in Phase-V we evaluate the co-design process and adapt our protocol for the use in partnership with other communities.
Report authorship	
Name	1: Jack Nunn 2: Kylie Gwynne
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Tasks in report completion	1: Main author of report 2: Checked data of report
Date of report authorship	25th August 2020
Key contact at initiative for confirming report content (include institutional email address)	Jack Nunn, PhD researcher, School of Psychology and Public Health, La Trobe University, jack.nunn@latrobe.edu
Involvement	
Who will be involved or how would you label groupings of those involved?	Group 1: Academic investigators Group 2: Aboriginal community representatives Group 3: Aboriginal community members involved in co-design and consultation activities
How many people will be in each grouping label?	Group 1: 10 Group 2: 5 Group 3: 20
Specific tasks of this person or group (list as many as possible) – *including any information about why certain people were included or excluded in certain tasks*	Group 1 and Group 2: Project design, ethics applications, planning and delivering co-design processes analysing data from co-design activities Group 3: Face to face and online events, consultation processes, checking data analysis
How will these people be involved (what methods will be used)	Group 1 and Group 2: formal investigator team, Project Advisory Committee, formal meetings, email

	discussions, commenting and editing documents, teleconferences, face to face meetings (when required) Group 3: Face to face and online events, co-design workshops and other activities to involve people
Enablers of involvement (what do you expect will help these people get involved – or what helped them get involved)	Co-designing planned involvement activities with Aboriginal community representatives will ensure they are appropriate, culturally safe and effective.
Barriers of involvement (what do you expect will inhibit these people from getting involved – or what inhibited them from getting involved). Are there any known equity issues which may contribute?	Geographic isolation and COVID restrictions will make face to face meetings difficult. Access to IT equipment and reliable and affordable internet is not universal in Australia, including the Aboriginal communities we plan to work with.
What was the outcome or output of the involvement of these people? What changed as a result of involving people?	Involving Aboriginal community representatives and community members will ensure planned activities (including research methods) are appropriate, culturally safe and effective.
Which stage of the initiative will these people be involved? (select from list of pre-defined stages or allow 'other')	All stages
What is the estimated financial cost for involving people. How much time will it take. Are there any costs that cannot be measured financially?	$10,000 Estimated time for planning and delivering involvement activity is 200 hours. The project is not planning to reimburse people for their time when involved (for example lost earnings if taking time off of work). The project may pay people for out of pocket expenses, or paid care which might be necessary for some people to be involved (for example, child care costs).
Mapping financial or other 'interests'	
Describe any financial relationship or other interest this person has to this project	Group 1: Academic investigators will be volunteering their time and may be named as authors in peer-reviewed publications Group 2 and 3: Aboriginal community representatives and community members may be paid for their time
Describe any conflicting or competing interests	N/A
Data	
Who is the data from this intervention shared with?	Data sharing (including returning data and results to participants), governance (including how to plan the management and storage of sample and DNA data) and data sovereignty will be agreed with the participants and the Aboriginal communities as part of the co-design process.

	The processes of data transference used in the project will be consistent with the principles of Participatory Action Research where stakeholders collectively decide upon roles, responsibilities and data access
How is it stored and hosted?	Stakeholders will collectively decide this as part of the co-design process, including stakeholder's view on appropriate biobanking methods
Who is analysing the data?	Stakeholders will collectively decide this as part of the co-design process
What methods will be used to analyse the data (including a link to any relevant code and information about validity)	Stakeholders will collectively decide this as part of the co-design process, including stakeholder's view on appropriate integration of data into biobanks and other data repositories
How is information about this data disseminated?	1: Public domain websites 2: Community events (online and face to face)
Who 'owns' the data or claims any kind of 'intellectual property' (include relevant licensing information)	Stakeholders will collectively decide this as part of the co-design process, with a guarantee that Aboriginal community members will be involved in any final decisions
Who controls access to the data	Stakeholders will collectively decide this as part of the co-design process
How is/will the data be 'Findable, Accessible, Interoperable, Reusable' according to the FAIR criteria?	Stakeholders will collectively decide this as part of the co-design process
What new knowledge do you expect to be generated? (if appropriate, include effect size, relevant statistics and level or evidence)	1: Inform best practice on co-design with Aboriginal communities 2: Answer important questions about people's preferences about data ethics, security, and quality associated with genomic research 3: Map people's preferences using the STARDIT-Preference Mapping tool
What do you hope to learn	1: Community preferences about the development of a new genomic research protocol 2: Community preferences about using co-design methods
How do you hope this knowledge translation will be translated?	1: Learning about community preferences and best practice can inform future co-design of genomic research and other initiatives with Aboriginal communities
Outcomes	1: Using a co-design process saves time, promotes the usage of health services, and elicits superior health outcomes 2: Benefits specific to this PM project include the building of strong and committed community partners and enhancing skills and knowledge in the Aboriginal community about genomics and health

How has or how will this be measured?	Group 1, 2 and 3 will be involved in developing outcome measures of importance, collecting baseline data and follow up data and analysing any changes.
Who is involved in measuring this?	Group 1 and 2

498

Appendices for Chapter 9 – Results: Comparison of all case studies

Detailed baseline and follow-up data on preferences for involvement

In the ausEE study, of the 26 people who gave consent and completed the initial survey, 15 participated in the online discussion and 12 completed the follow-up survey. From the 12 responses to the baseline and follow-up survey, participants gave 41 responses. In the ausEE study 59% of participants' responses showed a change towards 'widening' their view of who should be involved in research to include more people (N=24/41), 34% stayed the same (N=14/41) and 7% narrowed (N=3/41).

In the Shared Ancestry study, from the six participants who completed both the baseline and follow-up surveys, a total of 54 responses were given. Of these, 35% showed a change towards 'widening' involvement (N=19/54), 8% 'narrowed' (N=8/54) and half stayed the same (N=27/54).
This table combines the results from the 18 participants across the two studies who completed both the baseline and follow-up surveys using identifiable data at both stages, providing a total of 95 responses to the questions. 45% of participants' responses changed to 'wider' (N=43/95), 43% stayed the same (N=41/95) and 12% narrowed (N=11/95).

Study	Who should influence which aspects of research?	Change to wider	No change	Change to narrower
ausEE	Finding questions to ask	3	1	1
	Deciding which questions to prioritize and fund	3	1	1
	Deciding how to try and answer the question (the research method)	3	2	0
	Attempting to answer the question (carrying out the research, including collecting information)	2	2	1
	Trying to understand if it is possible to the answer the question (analysing the information)	2	3	0
	Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)	3	2	0
	Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)	4	1	0

Study	Who should influence which aspects of research?	Change to wider	No change	Change to narrower
	Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)	2	1	0
	Designing how people are involved in the research	2	1	0
	ausEE sub-totals (total of 41 responses)	24	14	3
Shared Ancestry	Finding questions to ask	2	4	0
	Deciding which questions to prioritize and fund	2	3	1
	Deciding how to try and answer the question (the research method)	2	2	2
	Attempting to answer the question (carrying out the research, including collecting information)	2	3	1
	Trying to understand if it is possible to the answer the question (analysing the information)	3	3	0
	Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)	2	3	1
	Ensuring that any information or answers are able to be used to help people in practice, policy or future research (sometimes called research translation)	2	3	1
	Deciding if the way of asking the question and all the other stages of the research were appropriate (evaluating the research method and any impacts)	2	4	0
	Designing how people are involved in the research	2	2	2

Study	Who should influence which aspects of research?	Change to wider	No change	Change to narrower
	Shared Ancestry sub-totals (total of 54 responses)	19	27	8
Totals	Combined data from both studies (total of 95 responses)	43	41	11
Percentages		**45%** of responses	**43%** of responses	**12%** of responses

Quantitative data from STARDIT-PM cross-case analysis

This table combines the STARDIT-PM data from all case studies in a cross-case analysis. Please note, data from the facilitators of both online discussions was combined, as the data source was a shared online discussion between facilitators from both the ausEE study and the Shared Ancestry study. In the ASPREE study, as data on interview participants was anonymised, we have no data on how many of the 59 interview participants were among the 18 who attended the event, so some people may have been counted twice.

Data set	ASPREE Interview participants	ASPREE event participants	ASPREE study team members	ASPREE totals	ausEE participants	Shared ancestry participants	Online facilitators (ausEE and Shared Ancestry)	Totals across case studies
Number in dataset	20	18	4	42	26	12	3	83
Views on who should be involved	10	18	3	31	17	10	0	58
Views on who should do which tasks	5	18	3	26	10	8	0	44
Views on modes of communication	13	18	1	32	3	4	0	39
Views on what methods should be used	3	18	4	25	6	5	0	36
Views on facilitators of involvement	3	18	4	25	10	7	3	45
Views on barriers of involvement	8	0	2	10	5	6	3	24

Data set	ASPREE Interview participants	ASPREE event participants	ASPREE study team members	ASPREE totals	ausEE participants	Shared ancestry participants	Online facilitators (ausEE and Shared Ancestry)	Totals across case studies
Views on what the outcome or output of the research or involvement in research could be	3	4	4	11	3	2	0	16
Views on which stage of the research people should be involved	2	18	4	24	0	1	0	25
Research data	0	18	1	19	0	3	0	22
Views on how learning from this research could be used	0	0	0	0	0	8	0	8

Detailed quantitative analysis of STARDIT data categories

Table 12.1 summarises the quantitative data from the three case studies where it was reported (ausEE, Shared Ancestry, ASPREE), noting the Indigenous Precision Medicine case study is a prospective report.

Table 12.1: Quantitative data from three case studies

Study	Reported methods of involving people [STARDIT Data Category 'Method of doing task?']	Reported modes [STARDIT Data Category 'Communication modes]	Reported tasks [STARDIT Data Category 'Tasks']	Involvement outcomes, impacts, learning or outputs [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
ASPREE	Commenting on documents, meetings, interviews, group discussion Total number: 4	Face to face meeting, telephone interviews, face to face group discussion Total number: 3	Reviewing documents (including participant information), commenting on research design, sharing views and perspectives, analysing data Total number: 4	Summary of impacts on study design from involving people: 1: Recruitment and sample collection plan changed 2: Participant communication improved 3: Participants will be involved in governance 4: Participants involved in controlling data access 5: Participants will be included on study advisory groups (including ethical oversight) using multiple communication modes 6: Feedback will be provided to participants about the research 7: Learning and development opportunities will be created for potential participants Total impacts: 7

Study	Reported methods of involving people [STARDIT Data Category 'Method of doing task?']	Reported modes [STARDIT Data Category 'Communication modes]	Reported tasks [STARDIT Data Category 'Tasks']	Involvement outcomes, impacts, learning or outputs [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
ausEE	Commenting on documents, survey, Group discussion Total number: 3	Online shared documents, online survey, online text-based asynchronous discussion Total number: 3	Reviewing documents (including participant information), commenting on research design, sharing views and perspectives, analysing data Total number: 4	Summary of the six impacts reported from involving people: 1. Participants reported learning resources were useful 2. Participants reported changed views as a result of participating 3. Participants reported enjoying the online discussions 4. Online discussions to be used in future research prioritisation 5. Participants asked to stay involved in the research 6. Participants reported improved understanding of how to get involved in research Total impacts: 6

Study	Reported methods of involving people [STARDIT Data Category 'Method of doing task?']	Reported modes [STARDIT Data Category 'Communication modes]	Reported tasks [STARDIT Data Category 'Tasks']	Involvement outcomes, impacts, learning or outputs [STARDIT Data Category 'Involvement outcomes, impacts, learning or outputs']
Shared Ancestry	Commenting on documents, survey, Group discussion Total number: 3	Online shared documents, online survey, online text-based asynchronous discussion Total number: 3	Reviewing documents (including participant information), commenting on research design, sharing views and perspectives, analysing data, contributing to STARDIT report data Total number: 5	Summary of the eight impacts reported from involving people: 1. Improved understanding of genomics informed sibling's participation in future research 2: Participants reported learning resources were useful 3: Participants reported changed views as a result of participating 4: Participants asked to stay involved in the research 5: Participants enjoyed the online discussions 6: Improved understanding of how to get involved in research 7: Co-design changed study design 8: Method for future research co-design established Total impacts: 8

Table 12.2: Cross-case analysis data from ausEE and Shared Ancestry case study

Preference data	Total response	Category	ausEE	Shared Ancestry
Baseline - All aspects mentioned	28	Everyone (any member of the public who is interested)	8	7
		Anyone who might be indirectly affected by the research	3	1
		Only people who are directly affected by the research	6	1
		Only people who are participating in the research	1	0
		Only people with a professional role in research	1	0
Follow-up - All aspects	12	Everyone (any member of the public who is interested)	6	5
		Only people who are directly affected by the research	1	0
Baseline - Finding questions to ask (identifying research topics)	26	Everyone (any member of the public who is interested)	4	3
		Anyone who might be indirectly affected by the research	2	0
		Only people who are directly affected by the research	2	0
		Only people who are participating in the research	0	1
		Only people with a professional role in research	2	1
Follow-up - Finding questions to ask (identifying research topics)	13	Anyone who might be indirectly affected by the research	2	1
		Everyone (any member of the public who is interested)	7	1
		Only people with a professional role in research	1	1
Baseline - Deciding which questions to prioritize and fund	26	Anyone who might be indirectly affected by the research	4	0
		Everyone (any member of the public who is interested)	1	2
		Only people who are directly affected by the research	7	1

Preference data	Total response	Category	ausEE	Shared Ancestry
		Only people who are participating in the research	1	1
		Only people with a professional role in research	6	3
Follow-up - Deciding which questions to prioritize and fund		Anyone who might be indirectly affected by the research	1	0
		Everyone (any member of the public who is interested)	5	
		Only people who are directly affected by the research	1	0
		Only people who are participating in the research	1	0
	12	Only people with a professional role in research	2	2
Baseline - Deciding how to try and answer the question (the research method)	26	Anyone who might be indirectly affected by the research	4	
		Everyone (any member of the public who is interested)	1	1
		Only people who are directly affected by the research	5	1
		Only people who are participating in the research	3	4
		Only people with a professional role in research	8	1
Follow-up - Deciding how to try and answer the question (the research method)		Anyone who might be indirectly affected by the research	2	1
		Everyone (any member of the public who is interested)	3	0
		Only people who are directly affected by the research	5	0
		Only people who are participating in the research	4	0
	13	Only people with a professional role in research	4	2
Baseline -Attempting to answer the question	27	Anyone who might be indirectly affected by the research	2	1
		Everyone (any member of the public who is interested)	2	2
		Only people who are directly affected by the research	5	0

Preference data	Total response	Category	ausEE	Shared Ancestry
		Only people who are participating in the research	2	2
		Only people with a professional role in research	9	2
		Anyone who might be indirectly affected by the research	0	0
		Everyone (any member of the public who is interested)	2	0
		Only people who are directly affected by the research	1	1
		Only people who are participating in the research	4	0
Follow-up -Attempting to answer the question	12	Only people with a professional role in research	3	1
Baseline - analyzing the information	27	Anyone who might be indirectly affected by the research	2	0
		Everyone (any member of the public who is interested)	2	1
		Only people who are directly affected by the research	1	0
		Only people who are participating in the research	2	2
		Only people with a professional role in research	13	4
		Anyone who might be indirectly affected by the research	1	0
		Everyone (any member of the public who is interested)	1	0
		Only people who are directly affected by the research	1	
		Only people who are participating in the research	1	1
Follow-up - analyzing the information	12	Only people with a professional role in research	6	1
Baseline - dissemination and publication	24	Anyone who might be indirectly affected by the research	0	0
		Everyone (any member of the public who is interested)	9	4
		Only people who are directly affected by the research	1	0

Preference data	Total response	Category	ausEE	Shared Ancestry
		Only people who are participating in the research	1	0
		Only people with a professional role in research	8	1
Follow-up - dissemination and publication	12	Anyone who might be indirectly affected by the research	1	0
		Everyone (any member of the public who is interested)	6	0
		Only people who are directly affected by the research	0	0
		Only people who are participating in the research	0	2
		Only people with a professional role in research	3	1
Baseline - research translation	11	Anyone who might be indirectly affected by the research	2	1
		Everyone (any member of the public who is interested)	2	3
		Only people who are directly affected by the research	2	0
		Only people who are participating in the research	1	0
		Only people with a professional role in research	3	0
Follow-up - research translation	7	Anyone who might be indirectly affected by the research	1	0
		Everyone (any member of the public who is interested)	3	0
		Only people who are directly affected by the research	1	0
		Only people who are participating in the research	1	0
		Only people with a professional role in research	1	1
Baseline - evaluating the research method and any impacts	22	Anyone who might be indirectly affected by the research	5	0
		Everyone (any member of the public who is interested)	2	1
		Only people who are directly affected by the research	3	3

Preference data	Total response	Category	ausEE	Shared Ancestry
		Only people who are participating in the research	1	2
		Only people with a professional role in research	9	1
Follow-up - evaluating the research method and any impacts	10	Anyone who might be indirectly affected by the research	2	0
		Everyone (any member of the public who is interested)	2	0
		Only people who are directly affected by the research	0	1
		Only people who are participating in the research	0	1
		Only people with a professional role in research	6	0
Baseline - Designing how people are involved in the research	8	Anyone who might be indirectly affected by the research	2	0
		Everyone (any member of the public who is interested)	0	1
		Only people who are directly affected by the research	2	0
		Only people who are participating in the research	1	0
		Only people with a professional role in research	4	0
Follow-up - Designing how people are involved in the research	10	Anyone who might be indirectly affected by the research	1	0
		Everyone (any member of the public who is interested)	3	0
		Only people who are directly affected by the research	1	1
		Only people who are participating in the research	1	0
		Only people with a professional role in research	3	1
Total responses			**268**	**84**

STARDIT-PM preference mapping data from public survey for comparison

This data below is included from a report about public preferences in science, shared by the charity Science For All.[502] Identical STARDIT-PM data categories were used, allowing this data to be compared with data from this thesis, and combined with other data in the future. The table below summarises the answers to the question 'Which aspects of any future research should be influenced by the following'.

	Deciding which questions to prioritize and fund	Deciding how to try and answer the question (the research method)	Attempting to answer the question (carrying out the research, including collecting information)	Trying to understand if it is possible to answer the question (analyzing the information)	Sharing the information that has been found, and any answers that may have emerged (dissemination and publication)	Ensuring that any information or answers are able to be used to help people in practice, policy or future research	Deciding if the way of asking the question and all the other stages of the research were appropriate	Designing how people are involved in the research
Everyone (any member of the public who is interested)	5	7	8	3	15	1	8	6
Anyone who might be indirectly affected by the research	4	2	2	5	0	10	1	2
Only people who are directly affected by the research	1	2	1	1	1	1	2	3
Only people with a professional role in research	6	5	5	6	1	4	8	4
Only people who are participating in the research	2	3	2	3	2	3	0	4

Appendices for Chapter 10 – Discussion

Strengths and limitations of methods used – detailed analysis

Evaluation of review methods

Narrative review of systematic reviews: How are the public involved in health research and what are the impacts?

As part of this thesis, I completed a narrative review of systematic reviews about involvement. The conclusion of many of the systematic reviews found, and thus the conclusion of the narrative review was, that there is not currently enough data to complete a meta-analysis of quantitative or qualitative data about involvement in genomics research. I published an article in the WikiJournal of Medicine which summarised and compared over 30 different types of systematic reviews[17], and concluded a systematic scoping review was the most appropriate method to search for relevant data. Findings from this review suggested that methods of involving people guided by the paradigm of participatory action research were most likely to have impacts. This finding had significant implications for this thesis, as this paradigm was used to guide the entire thesis as a result of this review.

Public Involvement in Global Genomics Research: A Scoping Review

While the scoping review method was not an exhaustive summary of all data, working with a database provided by the Global Alliance for Genomics and Health (GA4GH) provided a useful 'snapshot' of current international genomics research projects[28]. However, while the database compiled by GA4GH contained a majority of the predominant genomics research initiatives globally, the authors were aware of initiatives not represented, and a subsequent update after the completion of the review added 125 new initiatives[48], creating an impetus for an updated review. Furthermore, the review extracted data at the level projects articulated in the database and did not analyse the structure of organisations which sat above multiple projects. For example, the Broad Institute had a number of projects that were included in the database and a number that were not.

Another limitation of the scoping review search process was related to the wider issue that concepts such as 'involvement' cannot always be expressed fully in linguistic constructs even within the English language, let alone other languages. Such limits on English language reporting were explored in my presentation about participatory action research for the German-language organisation Ludwig Boltzmann Gesellschaft (LBG)[525,526]. As a result of such imprecise language, a decision on whether an initiative was reporting involvement sometimes required individual judgement. While the authors attempted to overcome this by using objective 'involvement indicators', it is never entirely possible to remove such individual judgement in this kind of review, so such decisions were checked by multiple authors[151]. The use of similar indicators in STARDIT was an attempt to combine individual judgements in order that multiple perspectives can be incorporated[442]. While a move towards more concrete and objective indicators of involvement will bring more consistency to definitions of involvement, truly 'standardised' descriptions will never be possible. By using Wikidata (which can have definitions updated by anyone at any time in any language), STARDIT can also be used to incorporate evolving language as it changes to reflect developments in different disciplines.

Another significant limitation of the scoping review was finding reports of involvement that gave a definite answer to the question 'did the involvement influence the research?'. For example, it is best practice that deliberative public dialogue and consultation should report the consequences of involving people [356(p13),357(p36)]. That is, there is an assumption that if people are asked to share views and perspectives, any new knowledge generated from such a process will be used to influence the genomics research initiative. While there were many initiatives that reported using methods where tasks included people sharing views and perspectives, only a few directly reported that these

influenced the research (for example, the UK Biobank Ethics Consultation Workshop)[621]. The issue of deciding if a method of involving people was 'consequential' was not possible to resolve. If involvement is 'consequential', it means that the process of involving people contributing to influencing research process, as distinct from involvement which is ignored or not incorporated[356–358]. It was not always possible to determine whether involvement was consequential based on the available information, so an assumption was made that all methods reported resulted were 'consequential'.

STARDIT provides a function for initiatives to report any impacts from involvement, as well as the ability to update a report throughout the project lifetime, thus allowing the consequences of involvement to be reported into the future. With some genomics research projects projected to span decades[622], such reporting will provide valuable data about how involvement has impacted and influenced genomics research projects. The uptake of STARDIT to report the Australian Genomics project 'Involve Australia', including impacts from involving people[298], demonstrates the perceived viability of such a method.

While this scoping review was the first of its kind in this field, a subsequent review exploring reporting the impact of involvement in biobanks used a similar method and recommended the use of STARDIT for reporting the impact of any involvement[33]. The choice by this team to use a similar method validates the choice of a scoping review methods, and STARDIT as a system used for reporting such data.

While future systematic reviews exploring involvement in genomics research and any impacts would greatly improve understanding in this area, at present, there is not enough data in order to complete such a review type. If STARDIT is used to report multiple genomics research projects, including involvement in those projects, it will provide standardised data from which future systematic reviews can be created, including living systematic reviews[623]. As the field of involvement in genomics research is growing fast, living systematic reviews provide a practical way for those planning involvement in genomics research to evaluate data and make evidence informed decisions about involvement.

Guidance for planning, reporting and evaluating initiatives: A multidisciplinary scoping review

As per the narrative review of systematic reviews of public involvement, this review explored guidance for planning, reporting and evaluating initiatives beyond the scope of genomics research in order to find relevant models and frameworks[195]. The co-design process of STARDIT meant that the system was developed into a way or reporting involvement across multiple disciplines including genomics research. This review attempted to show the current variation in guidance on planning, reporting and evaluating initiatives in order to inform both future systematic reviews and proposed standardised ways of reporting data on initiatives.

The review showed the multiple different ways of reporting data within and across disciplines, and informed the co-design of STARDIT, ensuring it was built in a way to incorporate the multiple different reporting standards which exist.

Evaluation of online discussion method

While the research projects described in the case studies in this doctoral research were planned and completed before the COVID-19 pandemic, the methods of involving people online described in this thesis now have an unexpected relevance to many disciplines, as research projects around the world seek to involve people online in novel ways, and evaluate such methods in a standard way.

The importance of safe online spaces is being recognised as more important than ever, with the concept of safety extending to 'data privacy' and 'cultural safety'. The ASPREE study showed that people preferred not to use social media companies to get involved in research. Using code from an open-source version of Loomio, the online forum was installed and hosted on La Trobe University virtual machines running on Australian Government servers, which provided reassurance to participants that the data would be stored ethically and in alignment with their values. This would not have been possible if using many third-party platforms.

The online text-based discussion method itself provided a flexible way for people in different time zones to get involved and interact. Hosting it as an asynchronous (people do not need to be online at the same time) discussion over two weeks also provided flexibility for those with caring or other responsibilities, allowing them to participate at times which suited them, without feeling excluded. The decision to use an asynchronous text-based discussion for two case studies was vindicated by the positive experience reported by participants. Literature suggested it was a more inclusive method, as it supports people with different cognitive needs or preferences[177]. This was echoed by a comment from an ASPREE participant at a face to face event, who was concerned that more dominant, confident or cognitively able voices would be heard in group discussions. While this was mitigated by facilitating smaller group discussions, the online discussions successfully created the enabling conditions for everyone to be heard equally and thanked individually for their contribution.

The facilitators reported that the support they received improved their ability to facilitated discussions, as did the shared online discussion space for the facilitators from both the Shared Ancestry study and the aausEE study both. The decision to create this was informed by the reflective practice articulated in the methods section[166].

Owing to how data was reported and shared (including using STARDIT), such learnings from these online discussions can be used to inform others who are planning similar methods[535]. Such learning can also inform how siblings, people with recent shared ancestry and other people at greater risk of exploitation could actually achieve workable methods to create power-sharing structures in line with the participatory action research paradigm, including to reporting any such methods in a standard way to inform future involvement.

Learning from this project also influenced how the charity 'Science For All' was set up and established by myself and others in parallel with this doctoral research (see 'Thesis Timeline'). Learning from these case studies informed how Science for All used Loomio to self-organise and run collaborative research projects with multiple stakeholders,[218,577] including the co-creation process for STARDIT[16].

Evaluation of the case study method and cross-case analysis

The case studies selected for this doctoral research represent four unique and real-world communities of people, where genomics research affects their lives directly. By ensuring that communities of people affected were involved in shaping this research, it has helped ensure learning is anchored in reality, rather than theoretical models of involvement in genomics research.

The case study methods used in this thesis, guided by the paradigms described, allowed the collection and analysis of both qualitative and quantitative data from multiple sources, which provided a richer dataset. In addition, the collection of data from study team members and participants provided a more holistic perspective, and meant data and impacts were collected that would not have been if data was just collected from participants. The collaborative analysis which was achieved by involving multiple stakeholders also ensured multiple people were involved in checking the analysis reflected their experience of the research process. The reflective analysis was

especially important during my analysis of data from the Shared Ancestry, as I am directly related to participants. This was able to be balanced and compared with the experiences of other study team members.

The sample sizes for the case studies in this thesis was variable, as was the percentage of people recruited from the known populations. While the ASPREE case study was relatively large[407], involving 59 participants and 20 interviews analysed, the total number of people was a relatively small sample from the 14,268 ASPREE participants (N=59/14268, 0.4%). The ausEE case study was medium sized with 29 participants, representing over 1% of people who were recruited from an online community of around 2000 people (N=29/2000, 1.45%). While the Shared Ancestry case study was a relatively small size compared to the other two (12 gave consent and 6 actively participated in the online discussion and surveys), it was recruiting from total known population of 18 at the time. Counting the two siblings [P8-SA, P10-SA] that were involved in the co-design process (but that did not participate in discussions), this gives a total of 15 out of 18 involved in some capacity (N=15/17, 88%), which is the highest proportion of all the case studies. It is interesting to note that after this study ended, there are now 46 known siblings (some deceased), as of 12th October 2021. Using STARDIT was an attempt to overcome this variation in both sample size and percentage of people recruited in the case studies, by combining standardised data. While datasets are still too small to draw any statistically significant conclusions, the mixed-methods approach meant that the interpretative analysis was able to combine themes and provide useful data, including comparing differences and generalisations.

As previously noted, the selection of case studies was influenced by pragmatic considerations. It is important to note that the unselected case studies also occupied a 'work-load' grey area, with conversations with potential partner organisations identifying areas for support in the organisations I was approaching. However, the line between 'research' and providing a pro-bono service to the organisation required careful elucidation. Working within the structure of a PhD to provide such pro-bono services is problematic for ethical, practical and financial reasons. Establishing partnerships through the neutral and transparent structure of a charity makes navigating such grey-areas more practical. For example, pro-bono and paid work can be articulated in working agreements, as can any data shared for research and publication. This model of working was used for developing STARDIT, rather than presenting it as a case-study in itself.

While the case studies were variable in size and each had both 'representative features' and 'deviant features' (see the section 'Case selection')[178], the most significant learning for others planning genomics research was likely to be from the 'generalisable' features of the case studies. Accordingly the cross-case analysis provided a successful way to apply the post-positivist paradigm and explore common themes across the case studies, and combine the quantitative data to allow cross-case quantitative analysis.

Evaluation of standardised data reporting method (STARDIT)

This section is an evaluation how effective STARDIT was for enabling consistent data reporting and a cross-case analysis. It is not an evaluation of STARDIT itself. A discussion about the strengths and limitations of STARDIT can be found in the discussion section of the peer-reviewed section 'Standardised Data on Initiatives - STARDIT: Beta Version'[9].

The preference mapping tool (STARDIT-PM) allowed consistent mapping of different stakeholder preferences across all case studies where it was used. This included the preferences of both potential participants and study team members. Applying a quantitative analysis to the identical questions asked at the start and the end of the ausEE case study and the Shared Ancestry study allowed an investigation of baseline preferences about involvement in genomics research and

preferences after the online discussions. It showed how people's preferences changed, including showing a 'widening' towards people preferring more kinds of stakeholders involved in genomic research. This data was published online in the public domain in a consistent way, allowing it to be combined in the cross-case analysis, and open to future datasets which might use the same question structure.

The design and reporting cycle in STARDIT was also helpful to allow consistent mapping of the participatory action research process for the different case studies. The design cycle proposed in STARDIT was also adopted and used in the protocol for the Indigenous Precision Medicine project, including proposing using STARDIT reports at various stages of the research[3]. STARDIT also demonstrated a working Beta system for reporting involvement and any impacts and has been recommended for use in reporting involvement in biobanks[33]. It has also been adopted by the Australian Genomics project 'Involve Australia', who created a prospective STARDIT report in 2021[298].

The impacts reported using STARDIT in this thesis were sometimes 'transformative learning'. While it is often difficult to attach causality to learning interventions (such as the information resources or videos used in the case studies), STARDIT allows consistent data on this to be reported (including self-reported outcomes), facilitating any future analysis and allowing future statistical analysis to begin to draw any correlation between certain learning interventions and learning outcomes, which may suggest causality.

STARDIT was also used by other projects outside the discipline of genomics research to report a participatory action research process involving citizen science and environmental DNA collection, demonstrating it can be used to allow comparison across case studies and beyond this doctoral research.[217]

Evaluation of outcomes and impact assessment methods

While time for longer term impact assessment is not possible within time limits of a PhD, it was still possible to measure outcomes and impacts immediately after the participatory action research process. Some impacts were able to be measured over two years after the online discussions had finished, as participants from the Shared Ancestry study were involved in co-creating the Alpha version STARDIT report in October 2020, and able to edit the Beta version from August 2021 onwards. These significant additional impacts about how participation in the participatory action research process helped participants make informed decisions about participating in genomics research would not have otherwise been recorded.

A follow-up survey was conducted with the study team for the ASPREE study, but not the participants, owing to ethical restrictions. As a result, there may be multiple unreported impacts. Preference and follow-up data was also not collected for the ASPREE study for the same reasons.

Appendices - Doctor of Philosophy examination

For transparency, this section contains the anonymised examiners' reports for the version of this thesis submitted for examination (found here). The tables also contain the response to the comments as a form of change log.

Examiner report 1

The candidate has carried out an innovative programme of research, making a well-defined and substantial contribution to public involvement in genomics research. They have demonstrated the ability to understand the literature and issues associated with involving members of the public in genomics research, and also developed a system to address some of these issues.

They display a good understanding of the methodologies they have used, and present the strengths and limitations of the approach.
The thesis is well-written and clearly presented throughout, although some of the language used, interpreted literally, is not based on evidence. I think some sentences need to be 'softened' or supported with references. I provide more detail below, but, for example, 'STARDIT is an effective way to plan, report and evaluate involvement in genomics research'. If it is effective, then references should be provided to demonstrate how it has been evaluated, and how it has been determined to be effective.

I do not believe this thesis needs to be re-submitted but there are a few areas where clarity, change of language, and typographical changes are required.
I am pleased to recommend that this thesis be awarded the degree of Doctor of Philosophy.

Minor content changes

Section (page number)	Paragraph/sentence	Change required	Response
Executive summary (7)	2nd paragraph, 2nd sentence	The statement 'Involving people in genomics research means sharing power…..' – why does it. This statement needs to be supported. What is it about genomics research that requires sharing of power?	The word genomics has been removed as this is not a statement specific to genomics. I note that Table 1.1 defines words and terminology more precisely and 'participatory action research' is defined as a form of sharing power.

Section (page number)	Paragraph/sentence	Change required	Response
Executive summary (11)	1st paragraph, last sentence	The statement that STARDIT has been recommended for use in describing involvement in biobanks appears throughout the thesis. The word 'recommend' suggests endorsement. The citation referenced uses the wording 'One way to solve this issue….' which is a 'suggestion' for use, rather than an 'endorsement'. I recommend softening the language to reflect the citation.	Reworded to 'suggested' and added the following: In addition STARDIT (Alpha version) was cited as 'useful' as a way of 'evaluating engagement' in an article supported by the Global Alliance for Genomics and Health (GA4GH). The STARDIT Alpha version is also cited in the GA4GH 'Framework For Involving And Engaging Participants, Patients and Publics In Genomics Research And Health Implementation' as a useful way of 'conducting evaluations of engagement'.
Executive summary (11)	2nd paragraph, 1st sentence	The statement 'As part of this doctoral research, STARDIT was also successfully used…' As above it is not clear how 'success' has been defined, please provide clarity, or remove the word 'successfully'.	Changed to 'As part of this doctoral research, STARDIT was also demonstrated as a way to map preferences (using the preference mapping tool STARDIT-PM),'
Executive summary (11)	2nd paragraph, 2nd sentence	The statement 'In each case study, the process of involving people in the research led to positive impacts and outcomes'. As above how was this defined? It is also important to include negative impacts and outcomes. Or make a statement that there weren't any.	Added mention that no negative impacts reported 'In each case study, the process of involving people in the research led to positive impacts and outcomes, with no negative impacts or outcomes reported.'

Section (page number)	Paragraph/sentence	Change required	Response
Theoretical approach used for case studies (62)	3rd paragraph, 3rd sentence	The statement 'This theoretical approach also enables everyone to be heard equally, in contrast to synchronous or face-to-face discussion that might be dominated by certain people.' 'Enables' is too definitive, should be softened with 'provides the opportunity or potential for people to be heard equally.	Reworded to 'This theoretical approach also creates the enabling conditions for everyone to be heard equally, in contrast to synchronous or face-to-face discussion that might be dominated by certain people.'
STARDIT MICRO Report: A Pathway to precision medicine for Aboriginal Australians (240)	Outcomes (final line of table)	The statement **'**Using a co-design process: promotes usage of health services, elicits superior health outcomes and saves time' needs to be softened, unless you can support with evidence, suggest 'has the potential'.	Reworded to 'promotes usage of health services, and has the potential to elicit superior health outcomes and save time'
Chapter 9 (248)	1st paragraph, 3rd sentence	Potential conflict of interests should be highlighted in the sentence – 'For example, the charity Science for All used the STARDIT-PM tool to map preferences…' suggest '(of which I am the Director)'	Created a 'Statement on real or perceived competing and conflicting interests' in thesis, referenced on contents page.
Table 9.4 (250)	ASPREE impacts	The statement 'Participant communication improved'. Based on what criteria? Evidence? Suggest 'changed'.	Wording from peer-reviewed article included

Section (page number)	Paragraph/sentence	Change required	Response
Summary of impacts (253)	6th paragraph, 1st sentence	The statement 'Most responses from participants in the ausEE and Shared Ancestry case studies showed a 'widening' towards a preference for more people being involved in genomics research.' Please check this statement is correct, from figures stated it was only 35% in Shared Ancestry group.	This statement is true when results from both case studies are combined (see table Table 9.1). Reworded for clarity to 'When combined, most responses from participants in the ausEE and Shared Ancestry case studies showed a 'widening' towards a preference for more people being involved in genomics research.)
Table 10.1 (268)	Last sentence under 'Findings'	The statement 'STARDIT is useful beyond genomics research'. Please reference, or soften with 'has the potential…'	Changed to 'STARDIT has been used beyond genomics research, including by Cochrane, citizen science projects and the Wiki Journals.[17,217,296,516]'
Standardised Data on Initiatives (270)	Last sentence	The statement 'The STARDIT system also facilitated the collection and comparison of impact data, including impacts from transformative learning, which would otherwise have been challenging to record, report and compare. ' Please provide evidence that *these 'would otherwise have been challenging to record, report and compare.'* Or soften.	References added and wording changed 'The STARDIT system also facilitated the collection and comparison of impact data, including impacts from transformative learning, which would otherwise have been 'challenging to quantify', record, report and compare'
Case studies (271)	2nd paragraph, 1st sentence	The statement 'Participatory action research (PAR) proved to be a successful paradigm to guide the research processes.' Please provide the criteria in which this was determined successful, or soften.	Changed to 'According to data gathered from research teams and participants, participatory action research (PAR) proved to be a

Section (page number)	Paragraph/sentence	Change required	Response
			successful paradigm to guide the research processes'
Standardised Data on Initiatives; Working Beta version (272)	4th sentence	The statement 'STARDIT has been used by other research initiatives beyond this PhD thesis, and has been recommended for reporting involvement in biobanks'. As per my previous comment 'The word 'recommend' suggests endorsement. The citation referenced uses the wording 'One way to solve this issue….' which is a 'suggestion' for use, rather than an 'endorsement'. I recommend softening the language to reflect the citation.	Wording changed and added three new additional projects reporting with STARDIT
Evaluation of the PAR paradigm	5th paragraph, 1st sentence	The statement 'The participatory action research process for STARDIT worked very well', please provide evidence and criteria for which this has been determined. Or soften with 'appeared'.	Changed wording to 'According to feedback from co-authors, the participatory action research process for STARDIT worked very well.' Added citation to STARDIT report [534]
Table 10.3 (286)	Point 4, under strengths	The statement '..and recommended the use of STARDIT for reporting the impact of any involvement.' As per my previous comment 'The word 'recommend' suggests endorsement. The citation referenced uses the wording 'One way to solve this issue….' which is a 'suggestion' for use, rather than an 'endorsement'. I recommend softening the language to reflect the citation	Changed

Section (page number)	Paragraph/sentence	Change required	Response
New knowledge (293)	5th paragraph, 2nd sentence	The statement 'It has been demonstrated as a proven way to report on the preferences of all stakeholders a….' Please provide evidence for 'proven' or soften language.	Changed to 'This thesis, and associated peer-reviewed publications, have demonstrated STARDIT as a way to report on the preferences of all stakeholders and on planned or completed participatory methods.'
Recommendation 2 (296)	Last sentence in that paragraph	The statement 'The STARDIT system has been recommended as a way of reporting involvement in biobanks..' As per my previous comment 'The word 'recommend' suggests endorsement. The citation referenced uses the wording 'One way to solve this issue….' which is a 'suggestion' for use, rather than an 'endorsement'. I recommend softening the language to reflect the citation.	changed
Implications for all research (306)	2nd paragraph, 3rd sentence	The statement 'For example, a community-led environmental DNA research project used STARDIT to report the co-design and co-management process, and reported impacts.' The references provided are from manuscripts uploaded to Wikispore. Is this peer reviewed? If not suggest 'softening the statement.	Reworded, added peer-reviewed citations and additional published reports

Typographical changes

Section (page number)	Paragraph/sentence	Change required	Response
Abstract (3)	3rd paragraph, 1st sentence	Missing word - 'with regard to involving people <u>with</u> the research cycle'	Changed to 'Involving each group had different challenges with regard to involving people in the research cycle'
Abstract (3)	Last paragraph, last sentence	Missing word - 'helping <u>to</u> make sure the benefits of genomic research are for everyone.'	Intentional wording, not typo. Added 'to' for clarity
Heading 'Standardised Data on Initiatives (STARDIT)' (10)	First sentence	Lack of clarity, perhaps missing word, '….involving people in human genomics research <u>and</u> other initiatives.'	Added 'and'
Executive summary, results (8)	3rd paragraph, 5th sentence	Typo – '..for improving human <u>heath…</u>'	Corrected to 'health'

Section (page number)	Paragraph/sentence	Change required	Response
Executive summary, results (10)	First sentence	This thesis has developed standardized ways (suggest singular)	Changed to 'This thesis has developed and used a standardised ways of planning, reporting on and evaluating stakeholder involvement in genomics research and other related initiatives'
How the rights based paradigm shaped this doctoral research(40)	1st paragraph, 5th sentence	Duplication – 'Terms such as 'consumer' and 'community member' also raise questions of 'who decides who decides who is in each of these categories' or groupings.'	Not an error but poorly worded. Reworded to ''who is involved in deciding who decides who is in each of these categories' or groupings.' It is seeking to ask who decides who decides who is in these categories.

Section (page number)	Paragraph/sentence	Change required	Response
			That is, if the Government decides categories, who decides who in the Government creates such categories – and who was involved in creating them?

4

Section (page number)	Paragraph/sentence	Change required	Response
Types of case study (57)	1st paragraph, 1st sentence	Comma required. 'In 1995, Stake defined three types of case study…'	Corrected
Co-defining ethical research (237)	1st paragraph, 3rd sentence	Typo - The Australian Department of Heath's 2021	Corrected
Strengths and limitations (237/238)	Last sentence	'The planned reporting of the case study using STARDIT also ensure the…' (ensures)	Corrected

Chapter 9 (241)	Bullet points	Presumed typo - ausEE Participant 1 = [P2- ausEE]	Corrected
Evaluation of rights-based paradigm (281)	2nd paragraph, 6th sentence	Typo - limited financial resources to pay open access feed	Corrected to fees
Table 10.3 (286)	Point 3, under strengths	Typo – 'Involvement indicators' were developed an..' (and)	Corrected
Table 10.3 (286)	Point 4, under strengths	Requires clarity 'A subsequent review exploring reporting the impact of involvement in biobanks cited this review	Reworded 'A subsequent review which explored reporting the impact of involvement in biobanks cited this review. It used a similar method and terminology,'
Implications for all research (306)	1st paragraph, 2nd sentence	Typo - The adoption of STARIT	corrected

General points for consideration

Comment	Response
I think the 'Discussion' section of the thesis could have been strengthened with consideration of alternative processes of reporting public involvement, in comparison to STARDIT. I was expecting you to list potential alternatives, and the pros and cons of each.	Added section 'Retrospective context for STARDIT co-creation' in methods expanding why STARDIT was developed Added further section in discussion 'Alternative processes of reporting involvement'

Comment	Response
In addition, I expected to see more discussion about the pros and cons of the PAR methodology. What other methods you might have used instead, given some of the limitations you experienced? Especially, in light of some of the members of the public perspectives. It would of added strength to discuss some of these.	Explored in narrative review why that method was chosen, as it had the most reported impacts. Made this clearer in rewording section on PAR: 'Informed by the reviews conducted as part of my doctoral research (including the narrative review), I chose a participatory action research paradigm to guide the process with co-design and reporting informed by guidance from a number of sources'
I was surprised too that one of your recommendations did not include re-imbursement for members of the public, as this surely limits who can be involved, and has the potential to introduce inequity. Purposive sampling has its place and can ensure that 'voices' are heard from underserved communities.	While I believed this was implicit in the thesis through words such as 'inclusive' I have sought to make this an explicit recommendation by adding recommendation 6 'Provide resources for inclusive and accessible involvement', noting that reimbursement is one way of many of supporting people to be involved for involving people.
In the survey questions, you make a statement 'There are many benefits of involving people other than researchers in the co-design of research studies at every stage of the research cycle. Research suggests that involving people improves the quality and the relevance of the research. Involving people can also improve participant experience and increase participation.'; prior to asking the question. I wondered if this might have affected the responses you received.	Added to the living version of STARDIT report 'By piloting different versions of the questionnaire, we were able to get feedback from participants that the wording of the question about involvement was difficult to understand. In partnership with participants and the study team, the wording was changed to include a short statement explaining what 'involvement' meant and the perceived benefits (see Additional file 2).' [410] I note this is explained in the different versions of the survey questions are available in additional file 2. The Aplha version of the STARDIT report also stated that involvement improved question design for interviews. The 'living' version of this has been updated to clarify how.
I realise that in a sense we are all 'members of the public' but I felt some of the participant responses you had were from people with a relevant professional background.	I note the line 'The process of involving people can be viewed as a learning experience for both the participants involved and study team members' This study illustrates that 'all stakeholders' – includes both the public and professionals involved in research– the thesis was about mapping different preferences of different stakeholder groups. I have attempted to make this point clearer in the thesis. STARDIT-PM is designed to map the preferences of people from different stakeholder groupings – noting they are not mutually exclusive – for example, 'patients' can also be 'researchers'

Comment	Response
	I further note this section in the introduction under the section 'What is public involvement': "Noting that 'potential patients' includes everyone, the word 'stakeholders' can be a more useful term, if the usage of this term is defined carefully. In this thesis, the term 'stakeholder' means anyone who has a 'stake' in health research, in particular those with important knowledge, experiences, expertise or views that should be taken into account. It can include, as sub-categories; researchers; research funders; policy makers, people affected by the research; people with specific health conditions, people with specific genomics variations; patients and the general public (who may also be categorised as 'tax-payers' for publicly funded research). The term 'people' and 'the public' will be used to refer specifically to patients, potential patients, carers, payers, consumers of health technology and the general public, excluding professional researchers, research funders, policy makers and anyone else with a professional connection to research."
By including study team members data in places, this distinction became blurred even further.	This comment indicates that a central tenet of the thesis has not been communicated effectively. The tenet is that it's important to involve researchers in this process, as well as other stakeholders, including the public etc. I note this section in the section 'Positivism and scientism' "Participatory action research differs from more traditional forms of research as the conceptual division between the researcher and the researched is blurred, removing the 'object' from research, making all stakeholders partners in the process.[137(p5)] By removing 'subjects' or 'respondents', the reductive methods of more traditional kinds of research are avoided, with the 'data' remaining in context as a record of subjective experience" Similarly, in the section 'Research with family members and close relatives' I explore being both a 'researcher' and connected with the research subject, exploring what is meant by concepts which divide research subjects from study team members in participatory research:

Comment	Response
	"As the concept of 'researcher objectivity' is challenged by participatory action research, having a researcher involved who 'has a stake' in the issue or is an 'insider' and is also affected may improve trust in the process among those participating. [18] In addition, as a researcher with an 'insider' status, I was more likely to be able to offer a novel interpretation of the data than someone 'outside' or unconnected to the research. While having the perspective of being an 'insider' or a person comparably affected can aid a researcher in understanding and empathising with the other research participants, the status of being an 'insider' or 'outsider' is often blurred.[19]" STARDIT can be used to report the involvement of multiple stakeholders including patients, the public and researchers (or people who identify as belonging to all those categories). STARDIT can be used to articulate some of the 'blurred' boundaries, or collaboratively label where the blurring is perceived to be and by who. I have sought to improve this point in the thesis. I further note the line from the published article 'Involving elderly research participants in the co-design of a future multi-generational cohort study' stating 'During the process, both participants and study team members reported changed views about the value of involvement in research, demonstrating 'transformative learning' and co-construction of knowledge' – data was gathered from both participants and the study team in order to assess the impact of involving participants from multiple perspectives. The distinction of data sources is articulated in detail in the article (all comments attributed to either participant IDs or study team members). Additionally, the data in Additional file 3 provides quantative data on participant responses

Examiner report 2

This is a comprehensive and impressive thesis that significantly advances our understanding of consumer and community involvement in the development and conduct of genomic research. The development of STARTDIT is an exciting initiative with international impact. The candidate has explored this topic using appropriate methods and demonstrates a deep understanding of the nuanced and complex environment of co-design, community engagement and participatory action research.

Recommendations for amendments

Recommendation	Response
Overall, the thesis could be improved with minor revisions to increase clarity, this includes: • Signposting chapter introductions for case study to include the same language description used in the thesis summary.	Created alignment with research aims and wording at the start of each chatper
Better linking between the research questions and how each of the studies/case studies addresses these questions.	I have adapted chapter intros – for example, chapter 8 saying that other chapters show initiated and completed co-design, but were the result of years of careful co-design, relationship building and evaluation. Chapter 8 shows a work in process and the complex process for best practice in research with indigenous peoples. Can the best practice in Australia be applied to other domains and learned from (including the potential barrier of so called ethical processes, having unintended unethical impacts – such as 'widening the gap' in life expectancy of Aboriginal peoples).[580] [5]
• Describing Wikidata and WikiJournal, justifying their use and outlining their suitability as repositories for content generated by this thesis.	In the adapted section 'Definitions as structured data' I have further explained how Wiki Journals and Wikidata are free, open access and independent and have included the following references: https://www.sciencedirect.com/science/article/pii/S1532046419302114 https://elifesciences.org/articles/52614
• Labelling of appendices is needed.	Improved labelling and explanatory text added, list of appendices also added a table of appendicies

Recommendation	Response
Ensuring additional files still have table titles or figure legends.	Addressed, additional text in introduction to appendices
Structurally, it would be easier to read with the contents table placed at the start of thesis rather than at the end.	There is a summary TOC at the start, the end table of contents is included as more of an index. As the TOC is more than 40 pages (!), this is too much for the start of a document, so a summary one was provided at the start, with a hyperlink to the more detailed one at the end to aid naviation.
Chapter 1/2 • A summary figure that maps your studies to your research aims to your activity would be valuable. It could be mapped across the existing Figure 1.1 thesis timeline.	It is hoped that the visual abstract at the start sufficiently provides this summary, as the numbered research aims in the thesis are included in this visual summary [8]
Chapter 3 • Better justification of why a scoping review was selected rather than a systematic review.	Added section 'Why were scoping reviews used in this thesis?' I note that this explanation is given in the narrative review section and I have further elucidated the reasoning in this new section. The main learnings from this review which informed this doctoral research were that language and terminology is inconsistent in the area of public involvement in research; systematic reviews called for improved reporting and consistency;
Figure 3.4, Figure 3.5, is the icon used the STARDIT logo or does it represent something else? This needs to be made clearer.	Explanation provided with additional figures.
• Page 122 – the author notes that Wikidata is blocked in some countries and describes how STARTDIT has been designed to be interoperable. Additional detail could be added on other platforms that could be	This point has been developed further – and an additional table of values has been added to the STARDIT article, with further information addressing this question added to Additional File 1 of the STARDIT Beta article.

Recommendation	Response
suitable and why? Linking back to its goal of being inclusive and ethical. Are there other elements of sustainability that need to be explored?	
Chapter 4 • Additional context about why GA4GH was the only suitable database to search for the review.	Added more detail in the new section 'Why were scoping reviews used in this thesis?'
• Suggest linking search terms used for searching websites to MeSH terms that would be used in a traditional literature review would help align the process used to a more traditional scoping review.	This is addressed in the section 'Why were scoping reviews used in this thesis?' In consultation with colleagues at the Cochrane Consumers and Communication review group (including professional librarians and search experts), the suggestion of the examiner here would not have been appropriate, as there are limited MeSH terms with which to conduct such a search (compared with the detailed terms searced using the systematic site search). In addition, 'traditional' Mesh headings are not fit for purpose here as they are not consistently used across anglophone countries, never mind non-English languages. This was one of the reasons that Wikidata was used to structure STARDIT, so that such consistent terms could be co-created and used.
• How was the data extracted and in what format?	This information is available in the additional files. A direct link to Additional File 1 can be found here: https://www.frontiersin.org/articles/file/downloadfile/446268_supplementary-materials_tables_1_xlsx/octet-stream/Table%201.XLSX/1/446268
• Figure 4.1 – Suggest data synthesis and analysis are removed from stage 3 and the final box be considered 'Stage 4' data synthesis and analysis as this is the state where only included studies are reviewed.	This comment is noted and appreciated, however as this article is published, these changes are unfortunately not possible
• English language as a limitation was described, additional detail on the likelihood of key non-English	This comment is noted and appreciated, however as this article is published, these changes are unfortunately not possible. However, this issue is further addressed in the new section 'Retrospective context for STARDIT co-creation'

Recommendation	Response
speaking (or publishing) countries that are known to have strong investment in genomics research?	
Chapter 5 • Add reference or rephrase statement "It has been proposed that the existing ASPirin in Reducing Events in the Elderly Extension study (ASPREE-XT) would be a good basis for a future multigenerational research study (MGRS)."	While this has been published (and references are not possible in plain English summaries, I have re-worded as follows in the thesis: It has been proposed by Paul Lacaze, an investigator on the ASPirin in Reducing Events in the Elderly Extension study (ASPREE-XT), that the study would be a good basis for a future multigenerational research study (MGRS).
Chapter 7 • Additional detail and discussion, in this chapter or wherever appropriate, of the ethical considerations and justification of why the candidate was involved as both researcher and participant	I note that this article is already published and can make no changes here, but note that this issue is explored in detail in the following sections: 1: Positivism and scientism 2: Research with family members and close relatives 3: Ivory towers, silos and bubbles: labelling the academic constructs and mapping ethical grey areas (sub-section Shared Ancestry) I note that reflections and feedback were also recorded by participants in the associated STARDIT report. I added this paragraph to the discussion section mentioned above to futher elicidate my personal reflections. "While there were complexities to working with my own biological relatives in the capacity of a researcher (including my biological mother), the main challenges were navigating ethical processes, rather than any ethical considerations about my involvement from relatives. Rather than present inherent challenges, my perception from informal (and unrecorded conversations with my relatives) was that my involvement as both a biological

Recommendation	Response
	relative and a researcher increased the trust between the research participants and the research process, and may have meant that more people participated than would have if it was research where a relative was not involved.
Chapter 8 • Elaborate on the connection between this project and the timeline for the development of STARDIT?	I have included further contextual information in this chapter, including how staff from the Poche centre joined the STARDIT project as co-authors.
Chapter 9 • Overall, this chapter feels like a combination of methods, results, and discussion, recommend a review and consolidation into other chapters where appropriate or improving the layout. It includes descriptions of the approach to analysis which should be captured in the methods chapter rather than presented as results.	Methods have been moved to the methods sections, and clearer links between the summary of results in this chapter and the detailed data in the appendices have been created. Discussion points have been moved to the relevant sections of the Discussion chapter.
.• Suggest consistent colour coding of case studies between tables.	This has been attempted where possible
• Figure 9.1 should be included and described in method/results chapters and referenced in Chapter 9.	Moved
• Table 9.1 should include original baseline and follow-up data, sample size as well as the combined data already stated.	This is included in the appendices and is linked to directly from this section

Recommendation	Response
• Page 243 heading Standardised Preference Mapping, what is (550)? Is it a reference?	Word count left in by mistake – removed!
• Table 9.2 does the methods section describe how views were mapped to questions/surveys if not add clarification on what mapped to each of the 'views' categories in the table or in the methods section refer to the appropriate section of one of the published papers.	Added numbers to PM-grid and adapted the section 'Quantitative cross-case analysis' to provide more clarity about the prefence grid and the thematic analysis using super-categories from STARDIT-PM.
Would also be helpful to reformat table to make the n=83 clearer	reformatted
• All additional detailed data/files related to the cross-case analysis should be labelled with Chapter 9.	Done
The above suggestions consider the restrictions on editing content from within published papers and appreciates that any changes may need to be included within unpublished linking text to improve clarity. As the thesis is dense given the breadth of content apologies if additional signposting or detail is present about the above dot points and has been missed.	
This is a comprehensive piece of work, and the addition of Chapter 8 may not have been necessary. Reporting on research still underway made comparisons between some of the case study results more difficult to interpret.	As stated above, Chapter 8 shows the important 'work in progress' aspect of building meaningful partnerships with communities. At this work is complex and takes time, Chapter 8 was included as the protocol itself is an important output and an important step on what is a complex co-design process.

Recommendation	Response
As the main output of this chapter is about the development of a protocol rather than the full conduct and analysis of a co-design study, this chapter may create a better narrative as Chapter 5.	Chapters are reflective of the chronology of the thesis. This point has been further highlighted in the introduction to provide clarity for the reader.

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Contact and identification

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STARDIT report

A STARDIT Beta version 'living' report about this thesis can be found here:

https://wikispore.wmflabs.org/wiki/Genomics_research_and_involving_people:_PhD_Thesis

A machine-readable version can be found here: https://www.wikidata.org/wiki/Q113417439

Versions

This thesis was submitted for examination in December 2021, and this final version was published in September 2022. For transparency, the version of this thesis that was submitted for examination can be found here:

https://archive.org/download/genomics-research-and-involving-people-2021.12.17-for-examination/Genomics%20research%20and%20involving%20people%202021.12.17%20For%20Examination.pdf

Detailed table of contents

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586