Genomics Acceptability

Societal Preferences, Public Opinion and Analysis

June 2025

Australian Genomics

Acknowledgement of Country

In the spirit of reconciliation Australian Genomics acknowledges the Traditional Custodians of country throughout Australia and their connections to land, sea, and community.

We pay our respect to their elders past and present and extend that respect to all Aboriginal and Torres Strait Islander peoples today.

Artwork by Yorta Yorta artist, Alkina Edwards, for Australian Genomics.

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CONTENTS

Author Acknowledgement

Project Leads:

Keri Finlay	Project Coordinator, Australian Genomics
Isabella Sherburn	Project Officer, Australian Genomics
Tiffany Boughtwood	Managing Director, Australian Genomics
A/Prof Ilias Goranitis	Principal Research Fellow, Health Economics, University of Melbourne

Working Group Members:

A/Prof Alison Archibald	Principal Research Fellow, Australian Genomics
A/Prof Stephanie Best	Principal Research Fellow, Implementation Science, University of Melbourne
Emma Bonser	Chief Executive Officer, Genetic Alliance Australia
Dr Marie-Jo Brion	Data Manager, Australian Genomics
John Cannings	Patient Advocate, Australian Genomics Community Advisory Group
Monica Ferrie	Chief Executive Officer, Genetic Support Network of Victoria
A/Prof Jodie Ingles	Head, Clinical Cardiac Genetics Group, Garvan Institute of Medical Research
Louise Lyons	Senior Manager, Australian Alliance for Indigenous Genomics
Bronwyn Terrill	Workforce Education Project Officer, Australian Genomics
Mary-Anne Young	Head of Clinical Engagement and Translational Platform, Garvan Institute of Medical Research

Executive Summary

Project Overview

Project Title: Genomics Acceptability – Societal Preferences, Public Opinion and Analysis

Background: The "Genomics Acceptability – Societal Preferences, Public Opinion and Analysis" project aimed to assess public attitudes toward the use of genomic technologies in healthcare. With the increasing integration of genomics into clinical practice, understanding societal perspectives is essential to ensure ethical, equitable, and effective implementation. This project focused on three key applications: reproductive carrier screening, adult health risk screening, and cancer treatment.

Aims and Objectives: The main aim of this project was to understand the public's acceptability, perceptions, and preferences of the applications of genomic technologies within the healthcare system and use this to identify priority areas for public health promotion, education and implementation.

The key objective was the creation and delivery of a concise, engaging public-facing survey and the subsequent data analysis and reporting.

Methods

This project undertook a quantitative public survey of societal preferences and opinions about genomic health interventions, utilising a market research company to ensure a representative sample of participants.

The project began in July 2024, with the public survey undertaken in February 2025. A scenario-based survey was devised by an expert working, including patient advocates, researchers and clinicians. The design included pilot testing and iterative refinement to ensure clarity and engagement. Responses were received from 1404 participants and covered topics such as the use of genomic technology in reproductive carrier screening, adult health risk screening and cancer treatment, and genomic data management.

Key Findings

- A majority of respondents expressed willingness to undergo genomic testing, particularly in the context of cancer treatment.
- Knowledge and confidence in understanding genetics were relatively high, though actual experience with testing was limited.
- Cost and data privacy were identified as key barriers to uptake.
- There was strong support for public institutions to manage genomic data, with limited trust in commercial entities.

- Ethical concerns, particularly around reproductive testing, were noted and warrant further public dialogue.

Recommendations

1. Further analysis of survey data to establish which populations may be reticent to undertake genomic testing.
2. Engage underrepresented communities through qualitative, culturally appropriate methods.
3. Prioritise transparent data governance frameworks that align with public trust.
4. Expand future research to include longitudinal studies and questions regarding additional genomic applications.

Conclusion

The project successfully met its objectives, providing critical insights into public perceptions of genomic healthcare. These findings will inform future policy, education, and implementation strategies to ensure that genomic technologies are introduced in a way that is both acceptable and beneficial to all Australians.

Plain Language Summary

This project explored how the Australian public feels about using genomic testing in healthcare. Genomics testing has a variety of benefits such as helping people understand their chance of developing certain health conditions, plan for future health needs or helping doctors select the right treatment. This study focused on three areas: testing before pregnancy to understand the chance of having a child with a genetic condition (reproductive carrier screening), testing to find out future health risks (adult health screening), and testing to guide cancer treatment.

A total of 1,404 people from across Australia took part in a survey. Most people were open to the idea of having a genomic test especially if it could lead to better treatment or help them plan for the future. Many said that they would like more knowledge about their health and felt that this testing should be available to everyone. However, there were some concerns with the use of this technology such as the cost, privacy of genomics data and the emotional impact of the test results.

The study also found that people generally trust public organisations, like government funded health organisations, to store their genomic data when compared to private companies.

These findings will help guide the introduction of genomic testing into the health system, future public education initiatives and policy decisions.

Introduction

Background

The ethical implementation of genomic technologies into mainstream healthcare practice must be informed by the societal acceptability of genomic technologies. This will become particularly relevant in the application of predictive genetic/genomic screening at population scale.

There is substantial evidence regarding the perspectives of research participants on genomic interventions. Among the group, public awareness and overall sentiment toward genomics are generally positive however detailed understanding remains limited, and attitudes or motivations are frequently shaped by perceived personal utility rather than informed comprehension. However, there is a notable gap in knowledge concerning the broader public, particularly individuals without direct experience in genomic care. Lack of information in this area was cited as a barrier in the Medical Services Advisory Committee (MSAC) response to the expanded reproductive carrier screening (ECRS) MSAC application.

It is imperative that efforts are made to understand the public's perspectives prior to wide scale implementation of genomics technologies as this will shape the prevailing narrative around genomic testing and influence the acceptability, and therefore uptake, of the technology. It is particularly important to understand the perceptions and views marginalised communities hold as genomic technology could further drive inequities in healthcare.

Aims

To understand the public's acceptability, perceptions, and preferences of the applications of genomic technologies within the health care system and use this to identify priority areas for public health promotion, education and implementation.

Objectives

1. Development of a concise, engaging survey for a general public audience
2. Deliver survey through Dynata, a market research company
3. Report on survey outcomes and their implications for health promotion, education, policy and implementation.

Inputs

Community Involvement: Three community members Emma Bonser, John Cannings and Monica Ferrie were involved as working group members throughout the entirety of the project. All three members have extensive experience in community involvement and an interest in public perceptions of genomic technology.

Engagement with First Nations Communities: Louise Lyons, a senior member of the Australian Alliance for Indigenous Genomics, was a working group member on this project.

Stakeholders: genomics researchers, research institutions, genetics services and health professionals, general public, HGSA, State/Territory and Federal Government genomics and health bodies, international genomics initiatives/bodies, patients, patient support/advocacy groups.

Milestones and Timeline

MILESTONE	TIMELINE	ACTIVITIES
Establish working group	May – July 2024	• Define scope • Identify stakeholders • Appoint chairperson and recruit members • Develop meeting schedule
Identify Government's key questions regarding public acceptability	July 2024	• Review MSAC response to the ECRS submission • Present findings to working group
Develop a scenario-based survey	August – November 2024	• Working group discussions • Review of literature • Scenario development • Scenario testing • Scenario testing with small group of general public (in-person)
Engage market research company to build and deploy survey	August 2024 – January 2025	• Source quotes from market research companies • Establish what different companies offer • Select most appropriate company to conduct the survey • Engage with project manager to discuss the requirements of the project • Test survey in Dynata software
Deliver survey	February 2025	• Pilot the survey with a small sample

		• Revise the survey based on pilot results • Carry out a second pilot to test amendments • Conduct the survey in full
Data analysis	March – May 2025	• Establish key points for analysis • Conduct broad analysis for presentation to working group • Meet with sub-groups interested in particular data sets within the larger cohort • Conduct sub-group analyses
Report development	April – June 2025	• Generate a high-level report which indicates the key findings and points to areas for further analysis

Frequency of meetings: From July 2024 meetings were held monthly. In 2025 one working group meeting was held to review the overall data analysis. Between April and May 2025 multiple sub-group meetings have been held with working group members interested in different aspects of the data.

Project Outcomes

Survey Development

Over the course of five months (July – November 2024), the working group established key topics for investigation which informed the development of survey scenarios.

The survey focused on three scenarios (detail described below):

- Reproductive carrier screening (Scenario A)
- Genomic testing for adult health risk screening (Scenario B)
- Use of genomic testing in cancer treatment (Scenario C)

A scenario-based survey was established as a method of gathering study data as it was agreed that a narrative would more effectively engage participants. Participants were directed to one of the three scenarios based on the market research company's algorithm, which enabled a representative sample to be recruited across each. Scenario information was delivered in line with how these initiatives would realistically be delivered by a public health initiative. Scenarios are detailed in Figure 1.

For each scenario, acceptability questions were adapted from[1,2]. Further questions investigating willingness to undergo testing, the impact of cost, number of conditions included in the test, the implications for society, data storage and security, and pharmacogenomics were developed by the working group (See Appendix 1 for survey).

The survey was first tested by six members of the general public in think-aloud sessions to establish if the scenarios or questions needed revision. This process also confirmed that respondents understood the questions enough to provide us with useful and relevant answers. As a result of the think-aloud sessions, the wording of scenarios and questions were refined where participants indicated some confusion.

When deployed, the following inclusion and exclusion criteria were used for the survey:

Table 1: Inclusion and exclusion criteria

Inclusion Criteria	Exclusion Criteria
Over 18 years of age	Under 18 years of age
Currently living in Australia	Not currently living in Australia
Proficient in English	Genetic health professional or those professionally involved in genetic testing
	International student

Genetic health professionals and those working professionally in genetic testing were excluded from survey participation as we wanted the views of the general public, and previously conducted research through the Australian *GeniOz* survey and the *Your DNA, Your Say* survey had an overrepresentation of genetic health professionals. We also excluded international students from completing this survey as there was concern that there may be an overrepresentation of this group in the sample which may not truly reflect the views of the broader Australian population.

Scenario A

"Imagine you are 30 years old, and you and your partner have decided you would like to start a family in the near future. You visit your doctor to discuss what you might need to do to prepare for this. They mention a screening test available called **genetic carrier screening**. This test can help work out the chances of you having children with a serious genetic condition.

The doctor directs you to a website which has information about the test, the possible results, and the testing process. You can order a testing kit through the website. The test uses a saliva sample from you and your partner to find out if you both are carriers of a serious genetic condition that may affect your children. About 2% of Australian couples will have an increased chance of having children with a genetic condition.

The website explains that carriers are people who are healthy but have a change in their DNA that means they can pass on a genetic condition to their children. If both you and your partner are carriers for the same condition, you have a 1 in 4 (or 25%) chance each pregnancy of having ~~a~~ children affected by the condition. Genetic conditions can be serious, reducing the child's quality of life, or shortening the life of the child. Most people who are carriers do not have a family history of the genetic condition they carry."

If you and your partner are found to be carriers, there are options available to help you have children without the condition.

You can choose whether or not you would like to have the test."

Scenario B

"Imagine you are 35 years old, and you are in a doctor's waiting room when you see a poster for a new **genetic health screening test**. You visit the website mentioned on the poster, which provides information about the test, the possible results and how the test is done.

The website explains that the test looks at specific genes in your body and can tell you if you have an increased risk of developing certain cancers (such as hereditary breast, ovarian, colon and bowel cancers) and hereditary (genetic) high cholesterol.

The test is done using a saliva sample, and you can order the testing kit through the website.

A positive test result does not mean you will develop the condition, but you are at significantly increased risk. Around 174,000 Australians (or 2%) aged between 20-44 years will be at increased risk of the conditions mentioned above.

Knowing that you are at an increased risk means that you are able to take proven steps to reduce the risk of developing these conditions or manage the condition if it does develop.

If your test comes back negative, it does not mean that you will never develop cancer or heart disease. Other factors like environment and lifestyle can also increase your risk."

Scenario C

"Imagine that after a period of coughing, tiredness, chest pain and other symptoms, you are diagnosed with lung cancer. You undergo surgery and a section of the cancer is taken for testing. The testing confirms you have the most common type of lung cancer.

Your oncologist (a doctor that specialises in cancer) suggests that you could start a treatment that works for people that have the same type of cancer as you. However, they also tell you about a genetic test that may give you information about the cause of the cancer and that helps decide which treatment is better for you. You do not need to have more surgery. This test will be done on the section of cancer that has already been taken."

Figure 1: Survey scenarios

Survey Delivery

Once the survey was finalised it was piloted through Dynata, a market research company engaged to implement the survey. Dynata were directed to recruit a representative sample of the Australian population. It should be noted that the term genomics was not used within the survey as it was agreed that the meaning of genomics would not be clear for the general public. Instead, the testing was generally described within the scenarios.

The survey was initially piloted with 105 respondents distributed evenly across the three scenarios. This pilot enabled us to determine if respondents understood the questions being asked. Based on this initial pilot the following changes were made to the survey:

- Willingness to undergo the test was unexpectedly high, and we suspected this could be due to doctor bias. Scenario A and B were revised to slightly distance the testing from the doctor which more closely modelled how these tests would be provided should they form part of a public health initiative.
- To ensure clarity in the data analysis stage, we amended the responses to the willingness to do the test questions from “I need more information” to “Probably yes, but I need more information” and “Probably no, but I need more information”.
- Within the data questions, there was some confusion regarding the use of clinical data for research purposes. These questions were edited for clarity.

A second pilot of 107 people was carried out and results suggested no further edits were required and amendments made served their purpose.

The survey was launched in February 2025 and ran for 8 days. A total cohort of 1404 respondents (inclusive of the second pilot) completed the survey. Respondents were distributed across the three scenarios (A: n=469, B: n=468, C: n=467). There was a completion rate of 89%.

Survey Results

Demographics

A representative sample of the Australian population was achieved through Dynata. In the study cohort, there was an expected proportion across genders with 51% of respondents identifying as women, 48% as men and a further 0.6% choosing to self-describe (i.e. non-binary). As indicated in Figure 2 below, representation from states and territories, metropolitan and regional areas, age and educational attainment were relatively indicative of the Australia population. However, some discrepancies were seen such as slightly higher metropolitan sample in our study (73.9%) when compared to the 2021 census data (66.9%) and some variability in the educational attainment.

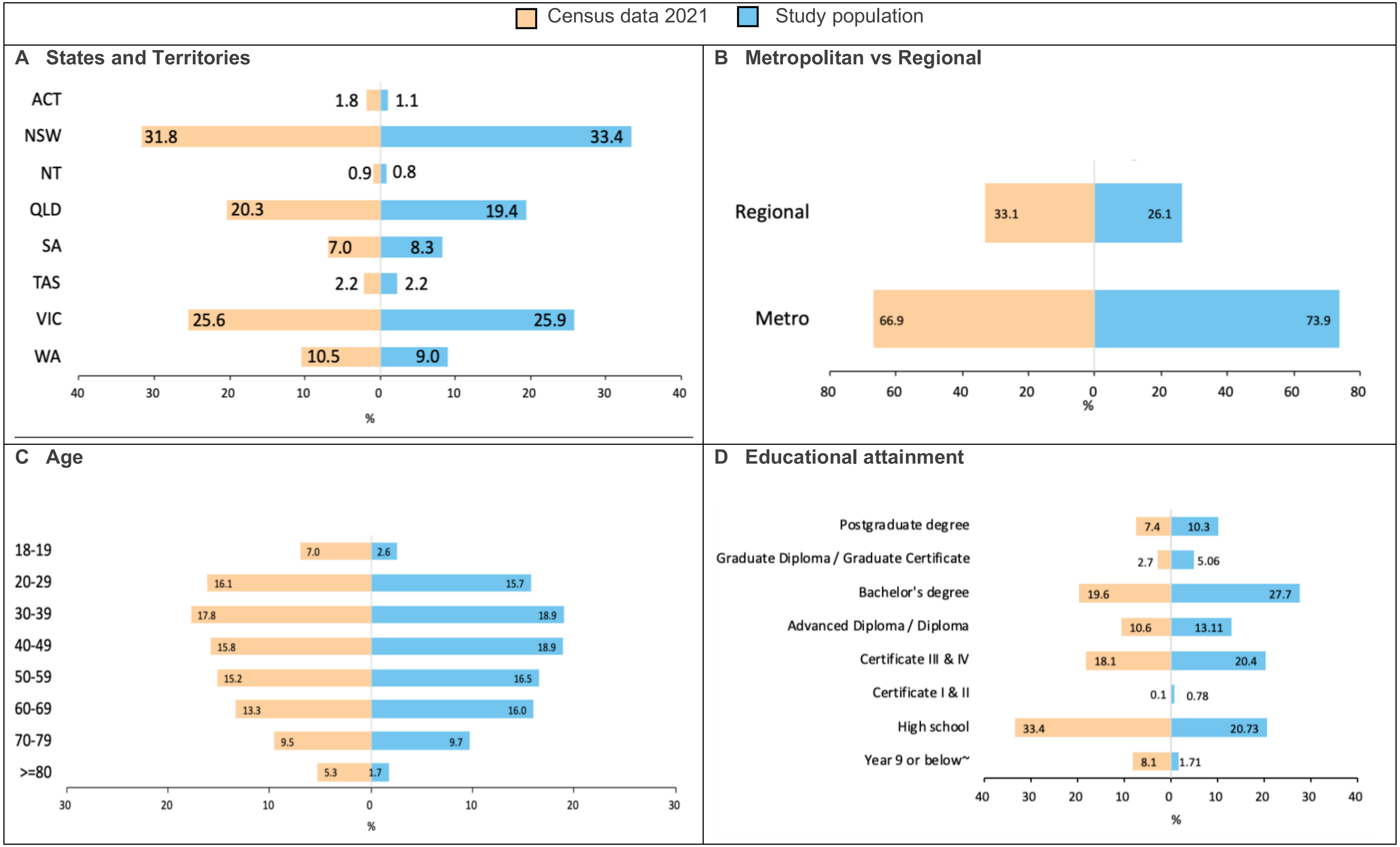


Figure 2: Demographic data comparison between study data and 2021 Census

Data from diverse communities

Of the respondents, 4.7% indicated that they were Aboriginal or Torres Strait Islander (Figure 3). This rate is higher than what was seen in the last census, with a more metropolitan sample than expected. This could indicate that views expressed in this survey cannot be generalised to the broader Aboriginal and Torres Strait Islander community, especially those living in rural or remote regions of Australia.

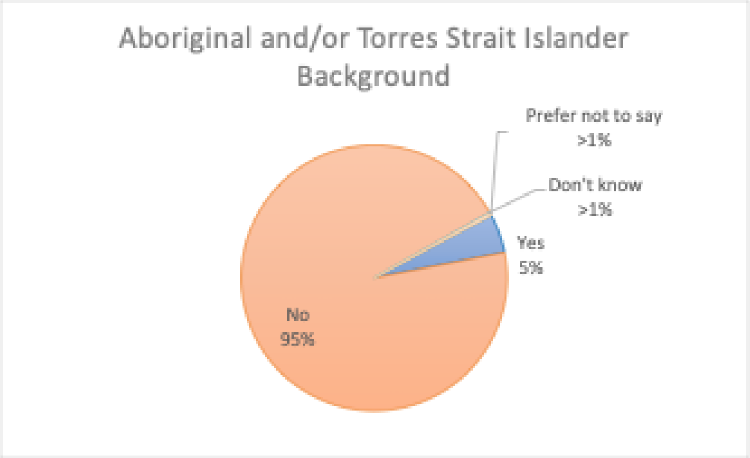


Figure 3: Aboriginal and/ or Torres Strait Islander Background

To establish Culturally and Linguistically Diverse (CALD) status, a standard set of questions were asked about birth country, languages spoken at home and ethnicity. Further to these questions, a self-identification was asked where the term CALD was explained. Based on the self-identification question, 28% of the cohort identified with the given definition. However, when looking at the corresponding responses to the standard set of questions there were some discrepancies. Using the definition given in Pham[3], respondents were considered CALD if they spoke a language other than English at home and/or were born in a non-English speaking country, two of the potential four ethnicities provided were non-English speaking, and they self-identified as CALD. Taking into consideration the factors above, 343 (24%) individuals were categorised as CALD. Of the 392 individuals that indicated they were part of the CALD community in the self-report question, 86 (22%) individuals did not meet the definition used in this study suggesting a clear definition may be needed.

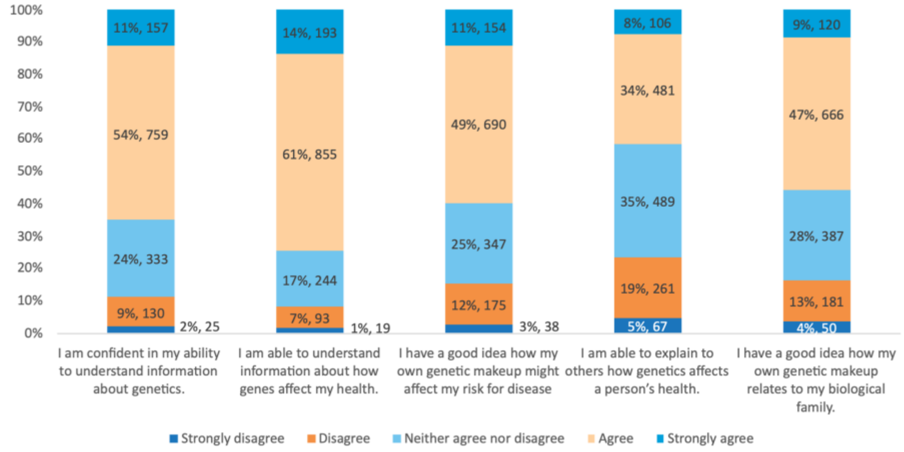
Knowledge and familiarity

In the subjective knowledge responses, the cohort showed high confidence in their genetic knowledge with over 55% of respondents indicating they felt they were able to understand genetics information, how it related to their health, how their own genes impact disease risk and relate to their family (Figure 4A). The one category where confidence fell slightly (42% agree or strongly agree) was if they had the ability to explain genetics to others.

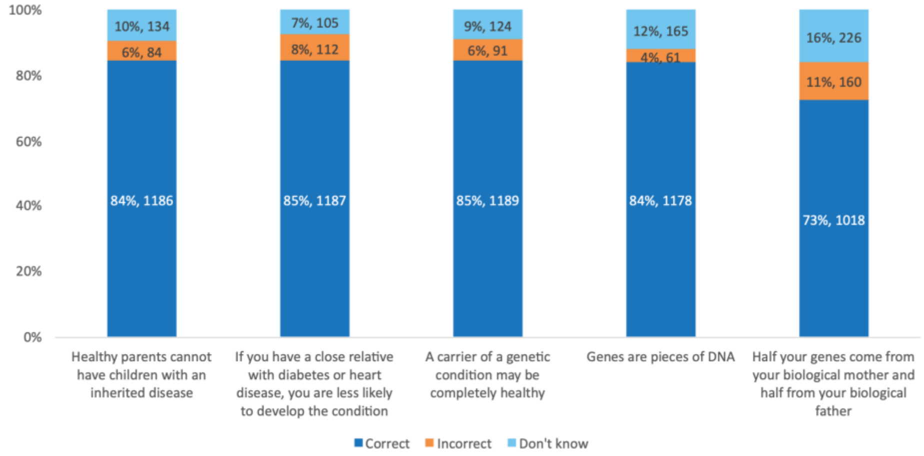
Results from the theoretical knowledge questions show this confidence was valid as four of the five questions were answered correctly by more than 80% of respondents (Figure 4B).

The genetic familiarity questions show 7.4% of respondents are living with a genetic condition and 16.1% know a family member or a friend with a genetic condition. Approximately 12% of respondents had undergone a genetic test with a further 20.8% previously considering having a test and 2.34% being offered a genetic test that they did not uptake. Despite having high genetic literacy, actual experience with genetic and genomic technologies is limited. Information regarding the type of genetic testing undertaken was not requested from participants.

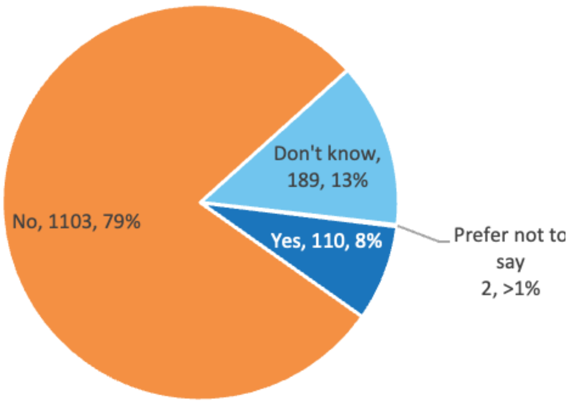
A. Subjective knowledge



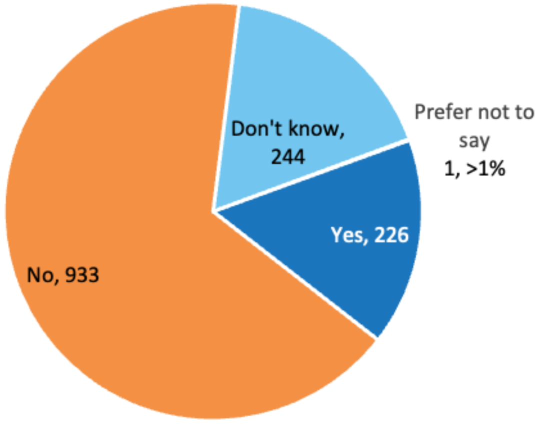
B. Theoretical knowledge (true or false) questions (percentage of respondents that answered correctly versus incorrectly)



C. Respondents with a genetic condition



D. Respondents with a friend or family member with a genetic condition



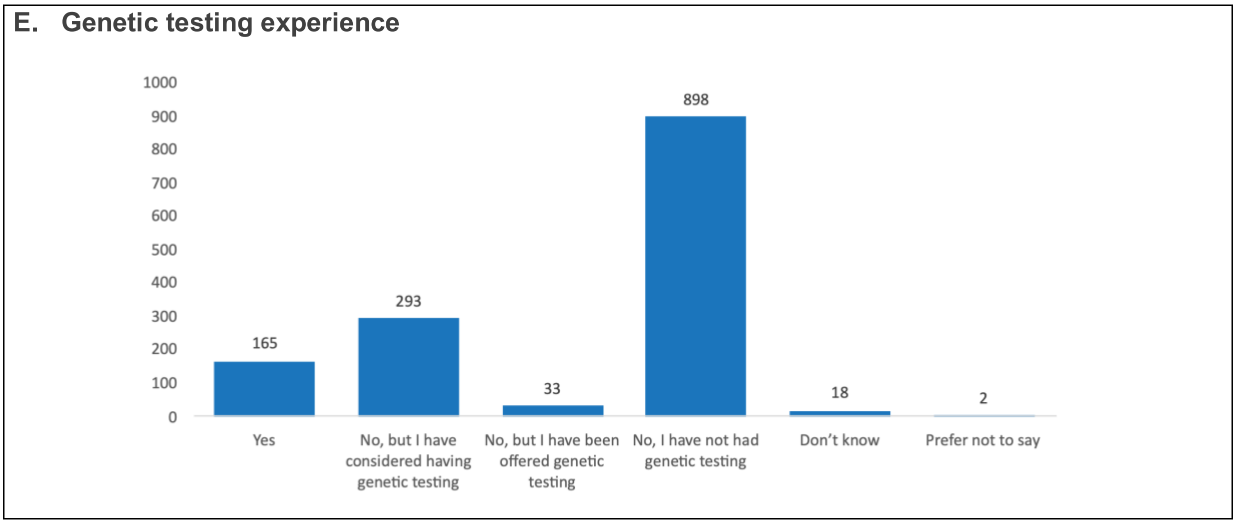

E. Genetic testing experience

Figure 4: Genetic knowledge and familiarity of participants

Willingness to undergo testing and it use in society

Across all three scenarios, a large majority of respondents were found to be willing to undergo testing (Figure 5).

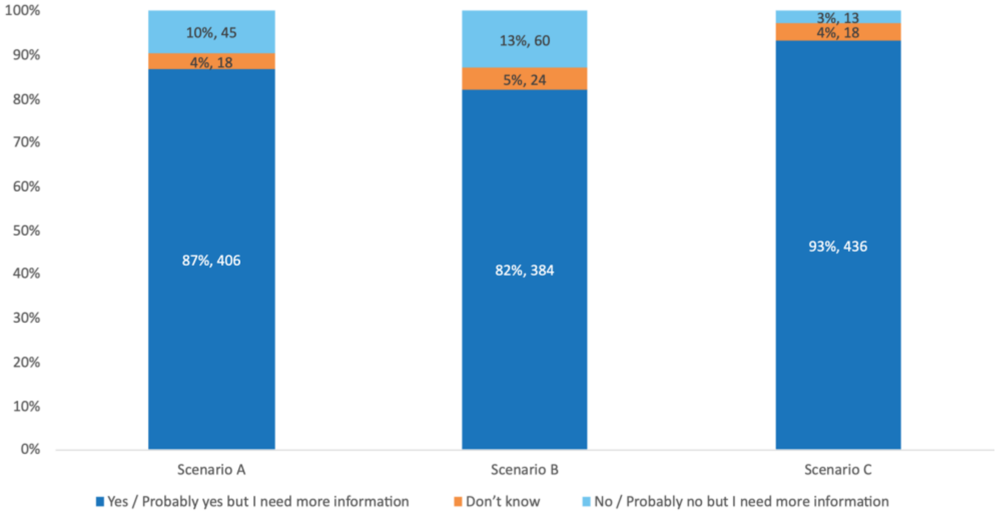


Figure 5: Participants' willingness to undergo testing

Analysis of respondent qualitative answers suggests that a key reason for wanting to undertake the testing was to acquire knowledge. Respondents wanted to plan ahead as indicated in this quote "*I need to know what may happen to my child. And be prepared if I'm willing to go through that"*. They would also like to be proactive about their health, access better treatment and hoped this could provide more information for their families and themselves.

The small percentage of respondents who were unlikely to undergo testing suggested increasing worries and impacts on mental health were reasons for this. Many individuals in this category also held the view that "*ignorance is bliss*" and suggested they *"cannot have anxiety about what [they] don't know about"*.

When asked if this type of genomic testing should be offered to all Australians most respondents felt that it should be, citing reasons such as the right to access appropriate healthcare and equity of access**:** *"We should all have the opportunity to create the best health outcomes for ourselves".*

However, some respondents seemed to misunderstand the question as many who selected 'don't know', 'need more information' or 'no' (Figure 6) indicated that they felt the test should be voluntary and not mandatory, despite mandated testing not being suggested. Other reasons for not believing the test should be offered to all Australians included affordability of testing and concerns about the abuse of data.

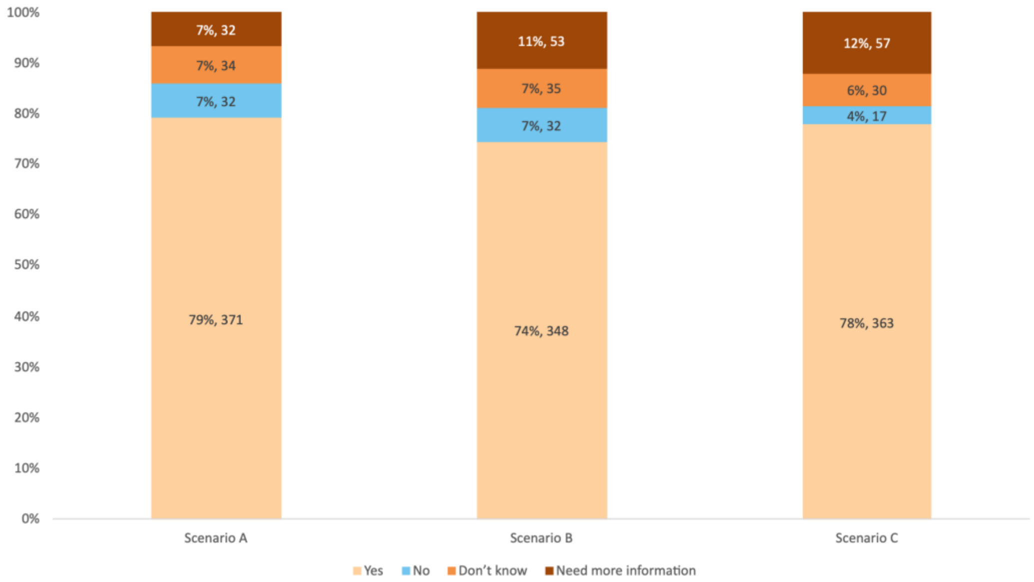


Figure 6: Societal access to genomic testing

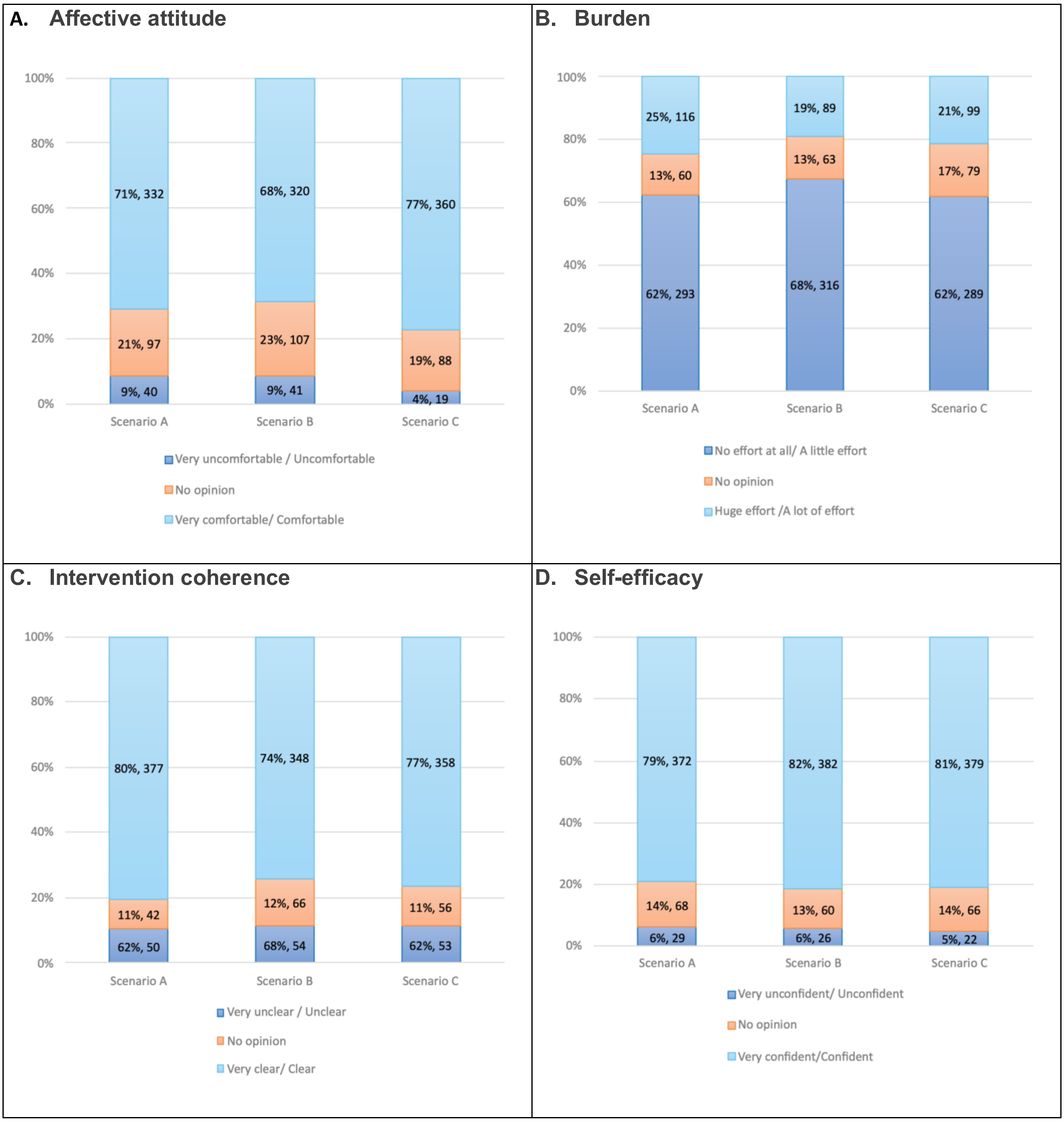
Theoretical Framework of Acceptability

In line with the Theoretical Framework of Acceptability[1,2] respondents were asked a set of questions to determine how appropriate genomic technologies would be as a healthcare intervention. See Table 2 for definitions of each of the domains associated with the Theoretical Framework of Acceptability. Responses in each domain across all scenarios were relatively positive, except for ethicality where responses were more evenly distributed (Figure 7).

Table 2: Theoretical Framework of Acceptability domains

Domain name	Definition (Sekhorn et al., 2017)	Question asked in Scenario A
Affective attitude	How an individual feels about the intervention	How do you feel about genetic carrier screening?
Burden	Perceived amount of effort required to participate	How much effort do you think genetic carrier screening would take?
Intervention coherence	The extent to which the individual understands the intervention and how it works	Is it clear to you how genetic carrier screening would help?

Self-efficacy	The individual's confidence that they can perform the behaviour(s) required to participate in the intervention	How confident are you in your ability to do the tasks needed to have genetic carrier screening?
Opportunity costs	The extent to which the benefits, profits or values must be given up to engage in the intervention	Do you think having genetic carrier screening would interfere with your other priorities?
Ethicality	The extent to which the intervention has good fit with an individual's value system	There are moral or ethical consequences of this type of genetic screening being available


A. Affective attitude

B. Burden

C. Intervention coherence

D. Self-efficacy

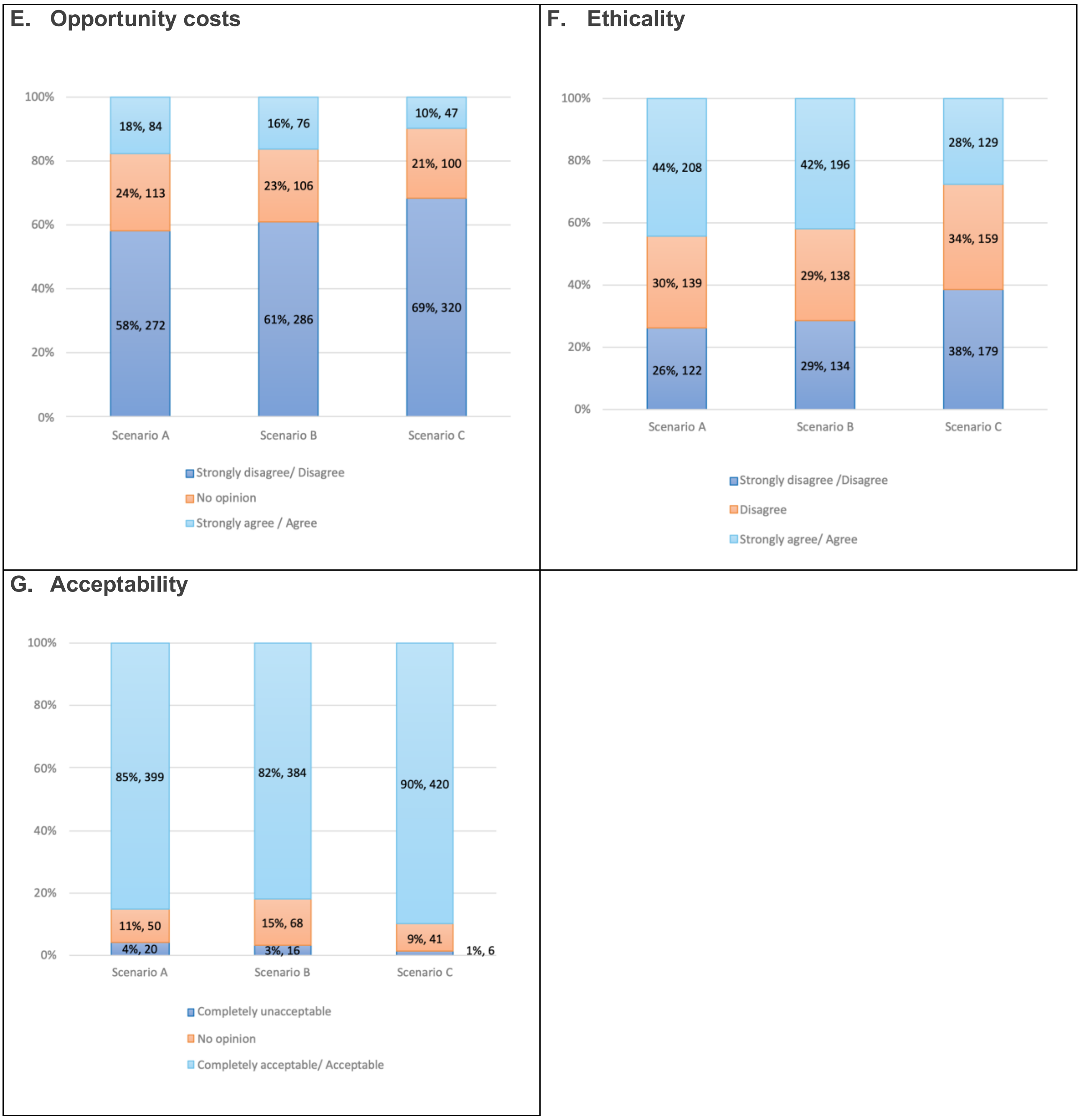


Figure 7: Theoretical Framework of Acceptability responses

Storage and use of genomics data

This survey also looked to understand respondents' perspectives on secondary data use and storage. They were asked how they felt about sharing data for clinical purposes (i.e. to aid the diagnosis of other patients), medical service improvements and research. Overall, these results show significantly high general acceptability of secondary data use for the reasons above with a vast majority (82%) suggesting it was acceptable to share data for clinical purposes. This fell slightly (79%) when asked about medical service use and further still (76%) when asked about sharing for research (Figure 8). This data also informed the Clinical NAGIM report and the Expanded Carrier Screening report.

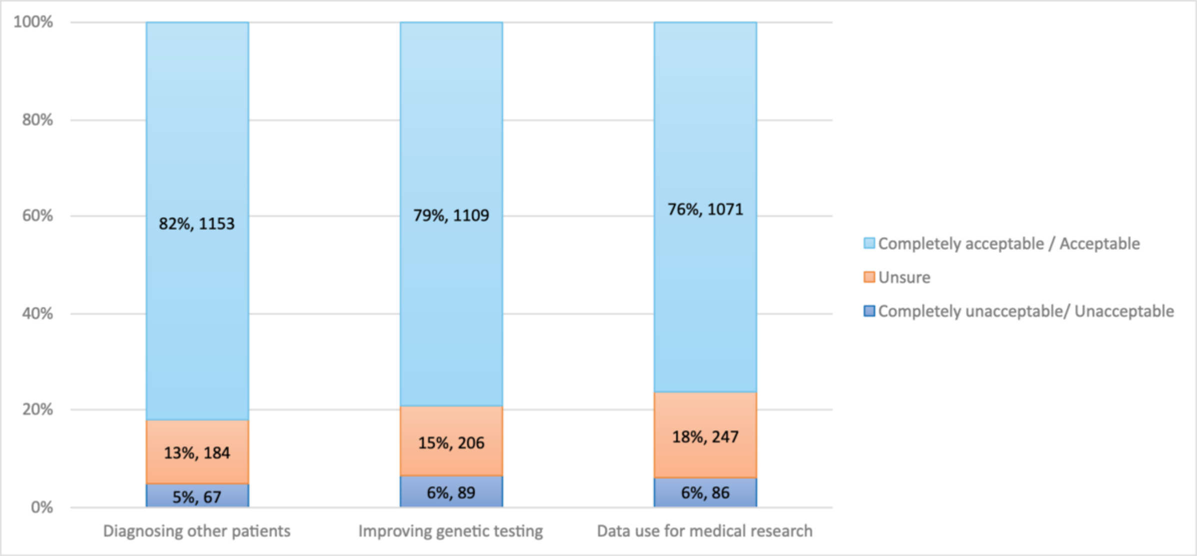


Figure 8: Preferences for secondary data use

Results from data storage questions show a strong preference for public institutions such as a government-funded national agency (77%), the Federal Government (61%) or local health services (72%) to act as data custodians. There is a clear lack of acceptability for for-profit commercial providers with only 41% of respondents indicating that this is an acceptable data custodian (Figure 9).

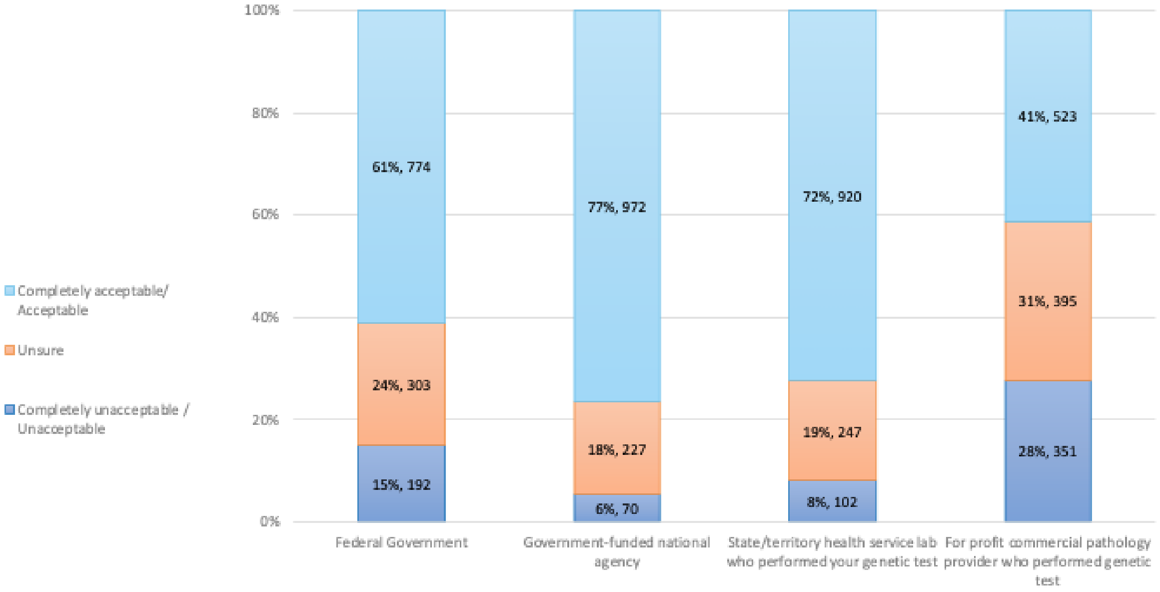


Figure 9: Preferences for data storage

Attitudes towards the survey

At the completion of the survey respondents were asked how difficult they found the survey to complete. Majority of respondents (72%) found the survey easy to complete (Figure 10).

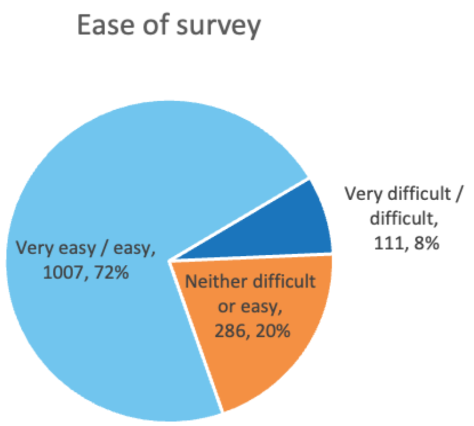


Figure 10: Easy of survey responses

Respondents were then given the opportunity to comment on their experience completing the survey. Across all ease of completion responses, participants found the survey "*very interesting and thought provoking*", with most comments from individuals who found the survey easy suggesting this. Some people within this category also raised the idea of an "ethical debate" that needs to be had and that the community should be involved in this: *"there should be more opportunities for the general public to have input to such scenarios- especially when there is the potential for debate about ethical and moral issues."*

Of the individuals that found the survey difficult, a large proportion indicated that the difficulty was due to a lack of information provided in the survey as suggest by this respondent: "*difficult subject to discuss with very little information to work on*." Others felt that this is a challenging topic to think about: "*I found some difficult to answer some questions as it made you question your moral and ethical judgement. You are totally responsible for bringing a child into a world where they may suffer from a condition. The benefits of research are fantastic though. I thoroughly enjoyed the survey. Very interesting."* However, for this participant as well as others, this didn't deter them from finding the survey enjoyable and interesting.

Discussion

The findings from this study suggest that the Australian public holds a strong interest in the uses and implications of genomic technologies in healthcare. Many respondents found the topic interesting and engaging and welcomed the opportunity to learn more and reflect on the broader ethical and societal implications. Participants highlighted the complexity of genomics, noting that it raises morally and ethically challenging questions that warrant thoughtful community deliberation. This underscores the importance of proactive public engagement to ensure that discourse remains accurate and constructive, thereby supporting informed decision-making and encouraging appropriate uptake of genomic testing.

Consistent with previous research in this area, the findings indicate a relatively high level of genetics knowledge across the general public. This suggests that Australians are well positioned to participate in meaningful discourse on the future use of genomic technologies in healthcare.

Beyond knowledge levels, the study also found a high degree of acceptability and willingness to undertake genomic testing across a range of scenarios. This suggests a broadly positive attitude toward testing, particularly when a clear health benefit, whether individual or societal, is evident.

Reponses to the scenario involving genomic testing in the context of cancer care, elicited the highest levels of willingness to participate. This is likely due to the public's relatively strong understanding of cancer and their ability to recognise both the potential harms of the disease and the tangible benefits of personalised, genomics-informed treatment options. In contrast, Scenario B prompted more ambivalence, possibly due to perceptions that the associated level of risk was too low to justify testing and the potential testing related anxiety. This highlights the importance of clear risk communication in shaping public support for genomic initiatives.

Despite overall support for genomic technologies, respondents expressed concerns that have also been identified in previous studies. This includes apprehension regarding data privacy, potential misuse or abuse of genetic information, the societal impact of reproductive impacts such as “designer babies,” and the possible mental health implications of receiving genomic test results. These issues must be taken into consideration in the design and implementation of public health initiatives with a genomics focus.

Across all scenarios, cost emerged as a significant barrier to testing. There was a clear preference for testing to be funded through Medicare, with concerns raised about equitable access.

Importantly, while there was strong public trust in public entities or government acting as data custodians, this level of trust did not extend to commercial providers. This notable difference in trust presents a challenge, given that private pathology services currently perform a large share of genetic testing in Australia. Addressing these concerns transparently will be critical to building and maintaining public confidence in the broader genomic testing landscape.

Limitations

A key limitation of this project is the lack of engagement with communities that are less likely to engage with digital survey technology. This includes CALD communities, First Nations peoples - particularly those living in remote or rural areas of Australia, young people or individuals without access to digital devices. These gaps likely contributed to an underrepresentation of certain voices in the data.

Additionally, the criteria used for analysing data from CALD participants posed challenges in capturing the full nuance of these groups’ experiences.

Time constraints also limited the ability to conduct a more in-depth qualitative analysis, which would have provided richer insight into the contextual factors influencing participant responses.

Recommendations and Future Directions

Areas for further analysis: Further analysis is needed to better understand the demographics of individuals who were hesitant to undergo genomic testing and to explore the underlying factors that may be driving this sentiment. Identifying these groups can inform targeted strategies aimed at increasing the understanding and acceptance of genomic testing as well as addressing specific concerns. Further to this, elucidating a deeper understanding of those who found clinical genomic data sharing “unacceptable” or who were “unsure” is also essential. Exploring the reasons behind these attitudes will inform the design of future implementation strategies to support data sharing for diagnostic purposes. These strategies may include improvements to consent processes, patient information materials, public engagement efforts, and education and training for the clinical workforce.

An in-depth investigation should be undertaken to explore how genomic knowledge varies across different demographic groups. Understanding who these individuals are will enable the development of tailored educational initiatives that can effectively bridge knowledge gaps and improve overall genomic literacy.

Targeted community engagement: Using qualitative methods, targeted community engagement is recommended to further explore these issues and to identify barriers specific to different populations that would not have access to online surveys. This should include direct conversations with particular communities and in collaboration with research projects and organisations that already have established relationships within these groups. Such efforts can also uncover nuanced insights that are often missed in broader surveys.

In addition, longitudinal studies should be conducted to track changes in public opinion over time, particularly in response to initiatives aimed at increasing public education and awareness around genomics. These studies will help evaluate the effectiveness of such efforts and inform future outreach strategies.

Future directions: Expanding the content of future surveys is recommended. This could include new scenarios with a focus on other genomic healthcare implementations such as genomic newborn screening and questions about where individuals acquire their knowledge of genomics, to better understand the sources and influence of public information.

Finally, replicating this research internationally can provide valuable insights into global perspectives on genomics. Comparing findings across countries will help identify both shared and unique challenges, informing the development of culturally and contextually appropriate approaches to public education and consent, and data sharing in genomics.

Conclusion

This project successfully achieved its aim of exploring public acceptability, perceptions, and preferences regarding the use of genomic technologies in healthcare. Through a carefully designed scenario-based survey, responses from a representative sample of 1,404 Australians provided valuable insights into societal attitudes toward genomic testing in reproductive carrier screening, adult health risk screening, and genomic testing in cancer treatment.

The findings reveal a generally high level of public interest and willingness to engage with genomic testing, particularly when clear health benefits are evident. However, concerns around cost, data privacy, and ethical implications, especially in reproductive contexts, highlight the need for thoughtful policy design and public involvement and engagement.

Limitations, including a lack of representation from particular communities and the constraints of digital survey methods, point to the importance of future qualitative and community-based research. Expanding the scope of future surveys and conducting longitudinal studies will be essential to track evolving public attitudes and inform equitable implementation strategies.

Overall, this project provides a strong foundation for shaping public health initiatives, education, and policy development in genomics. It underscores the importance of inclusive, transparent, and ethically robust approaches to integrating genomic technologies into healthcare systems.

References

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Appendices

Appendix 1: Genomics Public Acceptability Questionnaire

How should genetic testing be used in Australia?

The following sections will provide you with information about this survey so that you can decide if you would like to take part. Please take the time to read this information carefully. Your participation is voluntary. If you begin the survey, you can stop at any time before submitting your answers.

What is this research about?

Genetic testing analyses your DNA and generates a large amount of data. This data is unique to you and can impact decisions around your healthcare, now or in the future. With this research, we want to understand your views about how acceptable you find different types of genetic testing and any concerns you have with such testing.

What will I be asked to do?

If you agree to take part, you will be asked to complete one survey. The survey will take up to 15 minutes. Initially, the survey asks questions about you, your health and your understanding of genetics. The survey will then describe a scenario where genetic testing is used. You will be asked to imagine yourself in the position of the people in this scenario and answer a set of questions.

What are the benefits of taking part in this survey?

Aside from your survey completion payment, there will be no other direct benefits to you. However, this survey will inform researchers and policy makers about what Australians think of genetic testing and its use in healthcare. This will guide the future use of genetic testing in Australia.

What are the possible risks?

We do not anticipate any risks in participating in this research. It is possible that taking part may cause you to think about your or your family’s wellbeing. If you want more information about genetic testing, or if you have any concerns about your health, please seek advice from your doctor.

Do I have to take part?

No. Participation is completely voluntary, and you can withdraw at any time before submitting the survey without any consequences. However, if you do not complete the survey you will not receive your survey payment. Please note that once you complete the survey it is not possible to withdraw from the research because the data will be anonymised and not identifiable.

Will I hear about the results of this project?

A summary of the research findings will be published on the Australian Genomics website. We will also share the results of the research through academic journals and conferences, and through a report to share with patient advocacy groups, policy makers, and health providers. All results will be de-identified, and data will be combined so that you cannot be identified individually.

What will happen to information about me?

Although all survey responses will be anonymised and not linked to you, we will treat this data confidentially. Survey data will be held on Murdoch Children's Research Institute servers.

Who is funding this project?

This project is being funded by Australian Genomics, through a grant from the Australian Government (National Health and Medical Research Council Grant number GNT2035846). The funding agreement gives researchers independence in designing and conducting the research, interpreting the data, writing, and publishing the findings.

If you have any questions about this survey please contact Australian Genomics via email: australian.genomics@mcri.edu.au.

Consent

Do you consent to participate in this survey as outlined on the previous page?

By clicking yes, you provide your consent to use survey responses for the purposes of our research.
Please remember that you can withdraw at any point before the survey is completed. As soon as the survey is completed, we are not able to identify your responses because the data will be anonymised and not identifiable.

☐ Yes, I consent to participate in this survey as outlined on the previous page.

☐ No, I do not wish to consent to participate in this survey. [Terminate]

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*Required fields for screening and randomisation to scenarios

Section 1: Questions about you

In this section, we will ask you questions about yourself (e.g., how old you are, your gender, your highest level of education). We are asking for this information to get an idea of who is completing the survey and to see if any of these factors may influence your responses to questions later in the survey.

1.1. My gender is*… [single selection]
☐ Man
☐ Woman
☐ Prefer to self-describe [please specify-*optional*]

☐ Prefer not to say

1.2. My age is*… [text box]
☐ 18-24 years old
☐ 25-34 years old
☐ 35-44 years old
☐ 45-54 years old
☐ 54-64 years old
☐ 65-74 years old
☐ 75 years old or older

1.3. My postcode is*… [4 digit number]

1.4. My highest level of education is* … (*If currently enrolled, please select the highest degree received to date)* [single selection]
☐ Year 9 and below
☐ Certificate I & II
☐ Year 10 and above
☐ Trade certificate
☐ Certificate III & IV
☐ Advanced Diploma / Diploma
☐ Bachelor's degree
☐ Graduate Diploma / Graduate Certificate
☐ Postgraduate degree
☐ Prefer not to say

1.5. What is your household's annual gross (before tax) income*?
☐ Lower than AU$40,000 per year
☐ AU$40,000 - AU$59,999 per year
☐ AU$60,000 - AU$79,999 per year
☐ AU$80,000 - AU$99,999 per year
☐ AU$100,000 - AU$119,999 per year
☐ AU$120,000 - AU$139,999 per year
☐ AU$140,000 - AU$160,000 per year
☐ Over AU$160,000 per year
☐ Prefer not to say

1.6. I am an international student* [single selection]
☐ Yes
    - If this is selected, thank participant for their time but they are not eligible to complete this survey.

☐ No

1.7. I work in the genetics field (i.e. clinical geneticist, genetic counsellor, genetic laboratory scientist) * [single selection]
☐ Yes
    - If this is selected, thank participant for their time but they are not eligible to complete this survey.

☐ No

1.8. I live with a … [select all that apply]
☐ Child/children
☐ Spouse/partner
☐ Parent/parents
☐ Other family [please specify] [answering in textbox is optional]

☐ I live alone [if selected, no other selection can be made]
☐ Prefer not to say [if selected, no other selection can be made]

1.9. I use a language other than English at home [single selection]

☐ Yes
  i. The other language/s I use are [select all that apply]
    ☐ Arabic
    ☐ Bengali
    ☐ Chinese – Cantonese
    ☐ Chinese – Mandarin
    ☐ Chinese – Other
    ☐ Dutch
    ☐ Farsi (Persian)
    ☐ French
    ☐ German
    ☐ Greek
    ☐ Hazaragi
    ☐ Hindi
    ☐ Indigenous Australian
    ☐ Italian
    ☐ Japanese
    ☐ Korean
    ☐ Nepali
    ☐ Portuguese
    ☐ Punjabi
    ☐ Russian
    ☐ Samoan
    ☐ Serbian
    ☐ Sinhala
    ☐ Spanish
    ☐ Tagalog (Filipino)
    ☐ Tamil
    ☐ Tongan
    ☐ Turkish
    ☐ Urdu
    ☐ Vietnamese
    ☐ Other (Please specify) [open text box] [answering in textbox is optional]
    ☐ Prefer not to answer
☐ No

1.10. I was born in [single selection]

☐ Country of birth [open text box/drop down]
☐ Don’t know
☐ Prefer not to say

1.11. Do you identify as Aboriginal and/or Torres Strait Islander?

☐ Yes
☐ No
☐ Don’t know
☐ Prefer not to say

1.12. My biological family originates from [multiple selections]
Please select all that apply.

☐ Adelie Land (France)

☐ Afghanistan
☐ Aland Islands
☐ Albania
☐ Algeria
☐ Andorra
☐ Angola
☐ Anguilla
☐ Antigua and Barbuda
☐ Argentina
☐ Argentinian Antarctic Territory
☐ Armenia
☐ Aruba
☐ Australia
☐ Australian Antarctic Territory
☐ Austria
☐ Azerbaijan
☐ Bahamas
☐ Bahrain
☐ Bangladesh
☐ Barbados
☐ Belarus
☐ Belgium
☐ Belize
☐ Benin
☐ Bermuda
☐ Bhutan
☐ Bolivia
☐ Bonaire, Sint Eustatius and Saba
☐ Bosnia and Herzegovina
☐ Botswana
☐ Brazil
☐ British Antarctic Territory
☐ Brunei Darussalam
☐ Bulgaria
☐ Burkina Faso
☐ Burundi
☐ Cabo Verde
☐ Cambodia
☐ Cameroon
☐ Canada
☐ Cayman Islands
☐ Central African Republic
☐ Chad

☐ Chile
☐ Chilean Antarctic Territory
☐ China (excludes SARs and Taiwan)
☐ Colombia
☐ Comoros
☐ Congo, Democratic Republic of
☐ Congo, Republic of
☐ Cook Islands
☐ Costa Rica
☐ Cote d'Ivoire
☐ Croatia
☐ Cuba
☐ Curacao
☐ Cyprus
☐ Czechia

☐ Denmark
☐ Djibouti
☐ Dominica
☐ Dominican Republic
☐ Ecuador
☐ Egypt
☐ El Salvador
☐ England
☐ Equatorial Guinea
☐ Eritrea
☐ Estonia
☐ Eswatini
☐ Ethiopia
☐ Falkland Islands
☐ Faroe Islands
☐ Fiji
☐ Finland
☐ France
☐ French Guiana
☐ French Polynesia
☐ Gabon
☐ Gambia
☐ Georgia
☐ Germany
☐ Ghana
☐ Gibraltar
☐ Greece

☐ Greenland
☐ Grenada
☐ Guadeloupe
☐ Guam
☐ Guatemala
☐ Guernsey
☐ Guinea
☐ Guinea-Bissau
☐ Guyana
☐ Haiti
☐ Holy See
☐ Honduras
☐ Hong Kong (SAR of China)
☐ Hungary
☐ Iceland
☐ India
☐ Indonesia
☐ Iran
☐ Iraq
☐ Ireland
☐ Isle of Man
☐ Israel
☐ Italy
☐ Jamaica
☐ Japan
☐ Jersey
☐ Jordan
☐ Kazakhstan
☐ Kenya
☐ Kiribati
☐ Korea, Democratic People's Republic of (North)
☐ Korea, Republic of (South)
☐ Kosovo
☐ Kuwait
☐ Kyrgyzstan
☐ Laos
☐ Latvia
☐ Lebanon
☐ Lesotho
☐ Liberia
☐ Libya
☐ Liechtenstein
☐ Lithuania

☐ Luxembourg
☐ Macau (SAR of China)
☐ Madagascar
☐ Malawi
☐ Malaysia
☐ Maldives
☐ Mali
☐ Malta
☐ Marshall Islands
☐ Martinique
☐ Mauritania
☐ Mauritius
☐ Mayotte
☐ Mexico
☐ Micronesia, Federated States of
☐ Moldova
☐ Monaco
☐ Mongolia
☐ Montenegro
☐ Montserrat
☐ Morocco
☐ Mozambique
☐ Myanmar
☐ Namibia
☐ Nauru
☐ Nepal
☐ Netherlands
☐ New Caledonia
☐ New Zealand
☐ Nicaragua
☐ Niger
☐ Nigeria
☐ Niue
☐ Norfolk Island
☐ North Macedonia
☐ Northern Ireland
☐ Northern Mariana Islands
☐ Norway
☐ Occupied Palestinian Territories
☐ Oman
☐ Pakistan
☐ Palau
☐ Panama

- ☐ Papua New Guinea
- ☐ Paraguay
- ☐ Peru
- ☐ Philippines
- ☐ Pitcairn Islands
- ☐ Poland
- ☐ Polynesia (excludes Hawaii), nec
- ☐ Portugal
- ☐ Puerto Rico
- ☐ Qatar
- ☐ Queen Maud Land (Norway)
- ☐ Reunion
- ☐ Romania
- ☐ Ross Dependency (New Zealand)
- ☐ Russian Federation
- ☐ Rwanda
- ☐ Samoa
- ☐ Samoa, American
- ☐ San Marino
- ☐ Sao Tome and Principe
- ☐ Saudi Arabia
- ☐ Scotland
- ☐ Senegal
- ☐ Serbia
- ☐ Seychelles
- ☐ Sierra Leone
- ☐ Singapore
- ☐ Sint Maarten (Dutch part)
- ☐ Slovakia
- ☐ Slovenia
- ☐ Solomon Islands
- ☐ Somalia
- ☐ South Africa
- ☐ South America, nec
- ☐ South Sudan
- ☐ Southern and East Africa, nec
- ☐ Spain
- ☐ Spanish North Africa
- ☐ Sri Lanka
- ☐ St Barthelemy
- ☐ St Helena
- ☐ St Kitts and Nevis
- ☐ St Lucia

☐ St Martin (French part)
☐ St Pierre and Miquelon
☐ St Vincent and the Grenadines
☐ Sudan
☐ Suriname
☐ Sweden
☐ Switzerland
☐ Syria
☐ Taiwan
☐ Tajikistan
☐ Tanzania
☐ Thailand
☐ Timor-Leste
☐ Togo
☐ Tokelau
☐ Tonga
☐ Trinidad and Tobago
☐ Tunisia
☐ Türkiye
☐ Turkmenistan
☐ Turks and Caicos Islands
☐ Tuvalu
☐ Uganda
☐ Ukraine
☐ United Arab Emirates
☐ United States of America
☐ Uruguay
☐ Uzbekistan
☐ Vanuatu
☐ Venezuela
☐ Vietnam
☐ Virgin Islands, British
☐ Virgin Islands, United States
☐ Wales
☐ Wallis and Futuna
☐ Western Sahara
☐ Yemen
☐ Zambia
☐ Zimbabwe
☐ Don’t know [Exclusive]
☐ Prefer not to say [Exclusive]

1.13. The term *Culturally and Linguistically Diverse* describes people from different cultural backgrounds and those who speak languages other than English. This includes immigrants and

refugees, as well as their families. It highlights the variety of traditions, languages, and experiences that make our community unique.
To help us better understand the diverse backgrounds of people completing this survey, we would like to know, do you identify as being from a culturally and linguistically diverse background? [single selection]

- ☐ Yes
- ☐ No
- ☐ Don’t know
- ☐ Prefer not to say

Section 2: What do you know about genetics?

In this section, we will ask you questions to see what you already know about genetics. Please answer these questions as best you can without searching the internet for answers.

2.1. Do you have a genetic condition (a health condition that is caused by changes to a person’s DNA)? [single selection]

- ☐ Yes
  - How much does this genetic condition impact your life?
    - ○ [sliding scale: 1. No impact; 2. Minor impact; 3. Moderate impact; 4. Major impact; 5. Severe impact]
    - ○ Prefer not to say
- ☐ No
- ☐ Don’t know
- ☐ Prefer not to say

2.2. Do any of your close family or friends have a genetic condition? [single selection]

- ☐ Yes
- ☐ No
- ☐ Don’t know
- ☐ Prefer not to say

2.3. Have you ever had genetic testing (a test that looks at your genetic information to find changes to your DNA that may be linked to a health condition or other traits)? This can include genetic testing ordered online.

Examples of a genetic test may include ancestry testing through an online company, paternity or sibling testing, testing for a genetic condition, testing for conditions that are potentially serious and can be prevented or treated (e.g., cancer, diabetes, iron overload, fitness/athletic ability, or nutrition/wellness genetic testing). [Multiple response]

- ☐ Yes, I have had genetic testing [Exclusive]
- ☐ No, but I have considered having genetic testing
- ☐ No, but I have been offered genetic testing
- ☐ No, I have not had genetic testing [Exclusive]
- ☐ Don’t know [Exclusive]
- ☐ Prefer not to say [Exclusive]

2.4. Please read the following statements and select how much you agree with them*.

	Strongly disagree	Disagree	Neither agree nor disagree	Agree	Strongly agree
I am confident in my ability to understand information about genetics.	1	2	3	4	5
I am able to understand information about how genes affect my health.	1	2	3	4	5
I have a good idea how my own genetic makeup might affect my risk for disease.	1	2	3	4	5
I am able to explain to others how genetics affects a person's health.	1	2	3	4	5
I have a good idea how my own genetic makeup relates to my biological family.	1	2	3	4	5

2.5. Please read the following statements about genetics and select whether you think they are true or false*. [True/False/Don't know]

☐ Healthy parents cannot have children with an inherited disease (Correct answer: False)
☐ If you have a close relative with diabetes or heart disease, you are less likely to develop the condition (Correct answer: False)
☐ A carrier of a genetic condition may be completely healthy (Correct answer: True)
☐ Genes are pieces of DNA (Correct answer: True)
☐ Half your genes come from your biological mother and half from your biological father (Correct answer: True)

Section 3: Specific scenarios

In this section, we will describe a scenario where genetic testing is offered. Please imagine yourself in the position of the people in this scenario when answering the questions.

If this scenario causes you any distress or if you have any concerns about your health, please seek advice from your doctor.

Scenario A. Genetic carrier screening

Imagine you are 30 years old, and you and your partner have decided you would like to start a family in the near future. You visit your doctor to discuss what you might need to do to prepare for this. They mention a screening test available called **genetic carrier screening**. This test can help work out the chances of you having children with a serious genetic condition.

The doctor directs you to a website which has information about the test, the possible results, and the testing process. You can order a testing kit through the website. The test uses a saliva sample from you and your partner to find out if you both are carriers of a serious genetic condition that may affect your children. About 2% of Australian couples will have an increased chance of having children with a genetic condition.

The website explains that carriers are people who are healthy but have a change in their DNA that means they can pass on a genetic condition to their children. If both you and your partner are carriers for the same condition, you have a 1 in 4 (or 25%) chance each pregnancy of having ~~a~~ children affected by the condition. Genetic conditions can be serious, reducing the child's quality of life, or shortening the life of the child. Most people who are carriers do not have a family history of the genetic condition they carry.

If you and your partner are found to be carriers, there are options available to help you have children without the condition.

You can choose whether or not you would like to have the test.

When you first hear about this type of test:

1) Would you, personally, be willing to have genetic carrier screening? [Yes, No, Don't know, Probably yes but I need more information, Probably no but I need more information]
    i. Why did you select this response? [open text box] [question is optional]
2) How do you feel about genetic carrier screening? [very uncomfortable, uncomfortable, no opinion, comfortable, very comfortable]
3) How much effort do you think genetic carrier screening would take? [No effort at all, a little effort, no opinion, a lot of effort, huge effort]
4) Is it clear to you how genetic carrier screening would help? [very unclear, unclear, no opinion, clear, very clear]
5) How confident are you in your ability to do the tasks needed to have genetic carrier screening? [very unconfident, unconfident, no opinion, confident, very confident]
6) Do you think having genetic carrier screening would interfere with your other priorities? [Strongly disagree, disagree, no opinion, agree, strongly agree]
7) How acceptable is genetic carrier screening to you? [completely unacceptable, unacceptable, no opinion, acceptable, completely acceptable]
8) There are moral or ethical consequences of this type of genetic screening being available [strongly disagree, disagree, no opinion, agree, strongly agree]
    i. Would you like to comment further? [open text box] [question is optional]
9) Do you think genetic carrier screening is something all Australians of reproductive age should have access to? [Yes, No, Don't know, Need more information]
    i. Why did you select this response? [open text box] [question is optional]

There are two types of genetic carrier screening tests available.

- **Option A:** One is a Medicare-funded test where there is no cost to you. This test looks at three of the more common serious genetic conditions that affect children in Australia.

- **Option B:** The second test covers more genetic conditions. This expanded test looks at 700 serious conditions in childhood (including the conditions covered in Option A). There is currently no Medicare funding for this test, and it will cost AU$1500.

10) Which genetic carrier screening test would you do?

☐ Option A
  i. Why did you choose this answer? [question is optional]
☐ Option B
  i. Why did you choose this answer? [question is optional]
☐ Neither, I don't want to do this testing
  i. Why did you choose this answer? [question is optional]
☐ Don't know
  i. Why did you choose this answer? [question is optional]
☐ Need more information
  i. What additional information would you need? [question is optional]

Imagine there is now funding available for the expanded carrier screening (**Option B**) and you are able to test for over 700 serious conditions in childhood at no cost to you.

11) Would you do the testing?

☐ Yes
☐ No
☐ Don't know
☐ Need more information
  i. Would you like to comment further? [question is optional]

Imagine you decided to have the expanded test (**Option B**) at no cost to you. The screening has given you and your partner a low chance result. This means it is unlikely that you and your partner will have children with the genetic conditions screened.

12) Would you be reassured by this result? [Yes, No, Don't know]
  a. Why did you choose this answer? [question is optional]

Imagine the screening shows you and your partner have an increased chance of a serious genetic condition in your children. This means there is up to a 1 in 4 (25%) chance of the condition for each pregnancy you have.

13) Which of the following options would you most likely do? We understand that these are personal decisions. Please think about which option you would most likely consider. [single selection only]

☐ Access fertility services and genetic testing of embryos to avoid having children with the condition
☐ Access fertility services and use donor sperm/egg/embryo to avoid having children with the condition
☐ Adopt a child/children
☐ Conceive and then test during pregnancy
  i. You test during pregnancy and the result shows the developing baby has a severe genetic condition that will significantly shorten the baby's life, would you:
    ☐ Continue with the pregnancy
    ☐ End the pregnancy
    ☐ Don't know
    ☐ Prefer not to say
☐ Not have children

- ☐ Don’t know
- ☐ Need more information

14) What do you think are the impacts of offering genetic health screening to society? [open text] [question is optional]

Scenario B. Genetic screening for disease prevention

Imagine you are 35 years old, and you are in a doctor’s waiting room when you see a poster for a new **genetic health screening test**. You visit the website mentioned on the poster, which provides information about the test, the possible results and how the test is done.

The website explains that the test looks at specific genes in your body and can tell you if you have an increased risk of developing certain cancers (such as hereditary breast, ovarian, colon and bowel cancers) and hereditary (genetic) high cholesterol.

The test is done using a saliva sample, and you can order the testing kit through the website.

A positive test result does not mean you will develop the condition, but you are at significantly increased risk. Around 174,000 Australians (or 2%) aged between 20-44 years will be at increased risk of the conditions mentioned above.

Knowing that you are at an increased risk means that you are able to take proven steps to reduce the risk of developing these conditions or manage the condition if it does develop.

If your test comes back negative, it does not mean that you will never develop cancer or heart disease. Other factors like environment and lifestyle can also increase your risk.

When you hear about this type of test:

1) Would you, personally, be willing to have genetic health screening?
[Yes, No, Don’t know, Probably yes but I need more information, Probably no but I need more information]

  i. Why did you select this response? [open text box] [question is optional]

2) How do you feel about genetic health screening?
[very uncomfortable, uncomfortable, no opinion, comfortable, very comfortable]

3) How much effort do you think genetic health screening would take?
[no effort at all, a little effort, no opinion, a lot of effort, huge effort]

4) Is it clear to you how genetic health screening would help?
[very unclear, unclear, no opinion, clear, very clear]

5) How confident are you in your ability to do the tasks needed to have genetic health screening done?
[very unconfident, unconfident, no opinion, confident, very confident]

6) Do you think having genetic health screening would interfere with your other priorities?
[strongly disagree, disagree, no opinion, agree, strongly agree]

7) How acceptable is genetic health screening to you?
[completely unacceptable, unacceptable, no opinion, acceptable, completely acceptable]

8) There are moral or ethical consequences of this type of genetic screening being available.
[strongly disagree, disagree, no opinion, agree, strongly agree]
  i. Would you like to comment further? [question is optional]

9) Do you think genetic health screening is something all Australians should have access to?
[Yes, No, Don't know, Need more information]
  i. Why did you select this response? [open text box] [question is optional]

This test is not currently covered by Medicare, you would have to pay AU$400 for the test.

10) Does this impact your decision on whether to have genetic health screening?

☐ Yes
  1. If yes, how does it impact your decision? [question is optional]
  2. What would be the maximum you would be willing to pay?
     Enter a number between 0 and 9999.

☐ No
  i. If no, why not? [question is optional]
  ii. What would be the maximum you would be willing to pay?
     Enter a number between 0 and 9999.

11) You have no family history of the conditions included in the genetic health screen. Does this impact your decision whether to have the test?

☐ Yes
  - If yes, how does it impact your decision? [question is optional]

☐ No
  - If no, why not? [question is optional]

12) The conditions being tested for are generally diagnosed later in life. If a screening test is to be offered before symptoms appear, what age group do you think is most appropriate to offer this testing to? [only select one]

☐ From birth
☐ Over 18 years old
☐ Over 30 years old
☐ Over 40 years
☐ Over 45 years

The website also offers **additional genetic testing**, which looks at how well your body uses some medicines, such as antidepressants, pain killers and cholesterol-lowering medications. It explains that by having this test, you can find out what medications will work best for you, and which might make you feel sick. This test costs AU$200 and is not covered by Medicare.

13) Would you do the testing?

☐ Yes
  - Why did you choose this answer? [question is optional]

☐ No
  - Why did you choose this answer? [question is optional]

- ☐ Don't know
    - Why did you choose this answer? [question is optional]
- ☐ Need more information
    - What additional information would you need? [question is optional]
- ☐ Not relevant to me
    - Why did you choose this answer? [question is optional]

14) Even if you, personally, do not want to have this additional testing about medications, do you think this is something all Australians should have access to, if they want it?
[Yes, No, Don't know, Need more information]

  i. Why did you select this response? [open text box] [question is optional]

15) What do you think are the impacts of offering genetic health screening to society? [open text] [question is optional]

Scenario C. Genetic testing for cancer treatment

Imagine that after a period of coughing, tiredness, chest pain and other symptoms, you are diagnosed with lung cancer. You undergo surgery and a section of the cancer is taken for testing. The testing confirms you have the most common type of lung cancer.

Your oncologist (a doctor that specialises in cancer) suggests that you could start a treatment that works for people that have the same type of cancer as you. However, they also tell you about a genetic test that may give you information about the cause of the cancer and that helps decide which treatment is better for you. You do not need to have more surgery. This test will be done on the section of cancer that has already been taken.

When you hear about this genetic test:

1) Would you, personally, be willing to have this genetic test?
[Yes, No, Don't know, Probably yes but I need more information, Probably no but I need more information]

  i. Why did you select this response? [open text box] [question is optional]

2) How do you feel about this genetic test?
[very uncomfortable, uncomfortable, no opinion, comfortable, very comfortable]

3) How much effort do you think this genetic test would take?
[no effort at all, a little effort, no opinion, a lot of effort, huge effort]

4) Is it clear to you how this genetic test would help?
[very unclear, unclear, no opinion, clear, very clear]

5) How confident are you in your ability to do the tasks needed to have this genetic test done?
[very unconfident, unconfident, no opinion, confident, very confident]

6) Do you think having this genetic test would interfere with your other priorities?
[strongly disagree, disagree, no opinion, agree, strongly agree]

7) How acceptable is this genetic test to you?
[completely unacceptable, unacceptable, no opinion, acceptable, completely acceptable]

8) There are moral or ethical consequences of this type of genetic testing being available.
[strongly disagree, disagree, no opinion, agree, strongly agree]

  i. Would you like to comment further? [open text box] [question is optional]

9) Do you think this genetic test is something all Australians should have access to? [Yes, No, Don't know, Need more information]
  i. Why did you select this response? [open text box] [question is optional]

There are two types of genetic tests available.

- **Option A:** A small genetic test is available that tests for 10 genes (genes are the instructions that tell our body how to grow and function/work) and is offered through Medicare with no cost to you. If one of these 10 genes are causing the cancer, you can get publicly-funded treatments.
- **Option B:** A large genetic test is available that tests 500 genes known to cause various cancer types, including the 10 genes in Option A. This test could provide information that identifies more treatments that could be used to treat the cancer, avoid the use of drugs that will not stop the cancer's progression, and might give you access to a clinical trial for new cancer drugs. This test is not publicly-funded and will cost you AU$3000.

10) Which test would you take?
☐ Option A
  - Why did you choose this answer? [question is optional]
☐ Option B
  - Why did you choose this answer? [question is optional]
☐ Neither, I don't want to do this testing
  - Why did you choose this answer? [question is optional]
☐ Don't know
  - Why did you choose this answer? [question is optional]
☐ Need more information
  - What additional information would you need? [question is optional]

Imagine, there is now funding available for the larger test (**Option B**) and you are able to test for 500 genes at no cost to you.

11) Would you do the testing?
☐ Yes
  - Why did you choose this answer? [question is optional]
☐ No
  - Why did you choose this answer? [question is optional]
☐ Don't know
  - Why did you choose this answer? [question is optional]
☐ Need more information
  - What additional information would you need? [question is optional]

You have been placed on a chemotherapy drug that is showing some control of the cancer, and the tumour is shrinking. However, you are getting side effects such as mouth ulcers, diarrhoea and nausea. You are offered a genetic test that looks at how your body reacts to different cancer drug treatments. The results of this test can help find treatments with less side effects for you. This test costs AU$200 and is not covered by Medicare.

12) Would you do the testing?
☐ Yes
  - Why did you choose this answer? [question is optional]
☐ No
  - Why did you choose this answer? [question is optional]

- ☐ Don't know
    - Why did you choose this answer? [question is optional]
- ☐ Need more information
    - What additional information would you need? [question is optional]

13) Even if you personally do not want to have this additional testing, do you think this is something all Australians should have access to if they want it?
[Yes, No, Don't know, Need more information]
   i. Why did you select this response? [open text box] [question is optional]

14) What do you think are the impacts of offering genetic testing for cancer treatment to society? [open text] [question is optional]

Section 4: Your Genetic Data

If you have a genetic test it can look at all of your DNA or only parts of it, which creates a lot of **genetic data**.

A medical scientist or doctor will examine your genetic data to identify which changes in your DNA are important to your health or your family's health.

You will receive a **genetic test result** which tells you information about what these changes mean for-you and your family.

Genetic data is the full set of information from your DNA. This looks like a giant code (Image 1).

Genetic test results summarise the important information about your health that were found by looking at your DNA. They are often medical reports (Image 2).

Image 1: Your genetic data

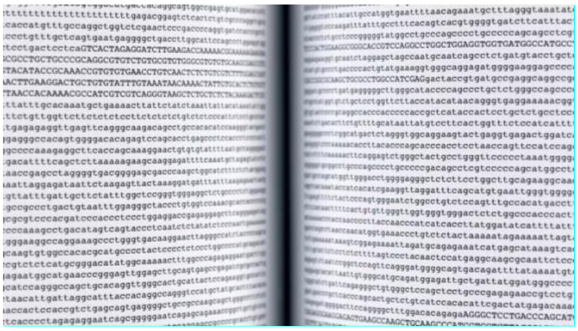

Image 2: Your genetic test result

Office on the web Frame

p2: *De novo* dominant

Australian Genomics

Patient name's Genomic Test Results	**Family Report Issued: 27/5/22**
Parents' names	**Parents' names** Study ID: A Testing Laboratory: Sample IDs: Patient – 22W, Mum – 22W, Dad – 22W
Reason for test	Unexplained seizures in Patient
About the test	We performed a **'trio whole genome sequencing' (trio WGS)** test. This test examines your and patient's genetic information to try and find a cause for patient's condition. You can find links to more information about this test at the bottom of this document.
Patient's result	**KCNQ2-related epileptic encephalopathy** **Gene:** *KCNQ2* **Variant:** NM_ : c.369C>G, p.Arg123Cys
Inheritance and recurrence	Parents Child Genetic variant **Inheritance pattern:** Patient has not inherited the *KCNQ2* gene variant, it has occurred in him/her for the first time (it is *de novo*).

In these questions we are talking about using and storing your **genetic data** (your genetic code), not the genetic test result.

Use of your medical genetic data for research

A patient's genetic data can be useful for health and medical research.

In health and medical research, scientists study what makes people healthy and why they might have health conditions or disabilities. Findings from this research can help people by leading to better treatments, better ways to identify a health condition, or better ways to deliver health services.

By looking at genetic data in many different people, scientists can learn more about the genetic causes of a range of health conditions.

The research may not directly benefit the person providing the data, but could help other patients in the future.

If you are offered a genetic test, your doctor/health professional could also ask you if your genetic data (not your personal details or your results) can be **used in medical research.**

4.1. How acceptable is sharing your genetic data for medical research, with your consent?

completely unacceptable, unacceptable, unsure, acceptable, completely acceptable

4.2. How acceptable would it be to have a **<u>discussion</u>** about research when you are having a medical test? ~~be to you?~~

[Completely unacceptable, unacceptable, unsure, acceptable, completely acceptable]

i. Would you like to comment further? [question is optional]

4.3. Your health professional could ask you about using your data (not your personal details e.g., name and address) in research at different times. When would it be acceptable to you?

For each option rate: completely unacceptable, unacceptable, unsure, acceptable, completely acceptable

☐ In the first discussion with my doctor/healthcare professional when I consent to do the test
☐ When I receive my genetic test result
☐ At another time (after my appointment where I have received my test result)
☐ Never - I do not want my doctor to ask me about the use of my genetic data for research when I am getting a medical test

i. Would you like to comment further? [question is optional]

Use of your genetic data for medical care

When your doctor orders a genetic test, your genetic data is compared with data from other people to get a diagnosis.

This helps experts (medical scientists or doctors) figure out which changes in your DNA might be important for your health or linked to certain conditions.

4.4. Your genetic data (not your personal details e.g., name and address) could be made available to doctors **to help diagnose other patients**.

How acceptable is this to you?
completely unacceptable, unacceptable, unsure, acceptable, completely acceptable.

i. Would you like to comment further? [question is optional]

4.5. Your genetic data (not your personal details e.g., name and address) could be made available to medical services **to improve their genetic testing**.

How acceptable is this to you?
completely unacceptable, unacceptable, unsure, acceptable, completely acceptable

i. Would you like to comment further? [question is optional]

Storing your genetic data after a genetic test

After a genetic test, a patient's **genetic data** is often securely stored by the lab who did the test, or it may be deleted after a few years.

Storing genetic testing data in a **secure database** for a long period of time could allow experts (e.g., medical scientists or doctors) to use your genetic data for healthcare purposes or for medical research, to help other people.

Your genetic data would usually be stored in a secure way without personal details attached.

It could be stored and managed by some different organisations or agencies.

4.6. How acceptable would you find a database/storehouse that is managed by:

For each option rate:
completely unacceptable, unacceptable, unsure, acceptable, completely acceptable

- ☐ Federal Government [grid]
- ☐ Government-funded national agency e.g. CSIRO, Australian Digital Health Agency, a national genetics agency[grid]
- ☐ Your State/Territory health service lab who performed your genetic test[grid]
- ☐ For-profit commercial pathology provider who performed your genetic test[grid]
- ☐ None of these - my data should be deleted after the test [Exclusive]
- ☐ Other [open text box]

4.7. Genetic data is stored in a secure way without personal details attached. However, as with all data, there is always a small chance it could be lost or accessed without permission. Would this change your decision to have the test?

- ☐ Would definitely change
- ☐ likely to change
- ☐ uncertain
- ☐ not likely to change
- ☐ Would definitely not change

Section 5: Your health

We would like to ask the following questions to understand if living with a health condition impacts your thoughts about the type of genetic technology in this survey.

5.1. In general, would you say your physical and/or mental health is [sliding scale]

- ☐ Excellent
- ☐ Very good
- ☐ Good
- ☐ Fair
- ☐ Poor
- ☐ Prefer not to say [Exclusive]

5.2. Do you have a health condition?

- ☐ Yes:
  - **i.** What kind of health condition do you have? [select all that apply]
    - ☐ Autoimmune condition
    - ☐ Cancer
    - ☐ Cardiovascular disease
    - ☐ Diabetes
    - ☐ Kidney disease
    - ☐ Sensory condition (for example, sight and/or hearing)
    - ☐ Respiratory condition
    - ☐ Musculoskeletal condition
    - ☐ Mental health condition or cognitive condition
    - ☐ Neurological condition
    - ☐ Other long term
    - ☐ Prefer not to say [if selected, no other selection can be made]
  - **ii.** What is the impact of this/these conditions on your day-to-day life
    - ▪ [sliding scale: 1. No impact; 2. Minor impact; 3. Moderate impact; 4. Major impact; 5. Severe impact]
    - ▪ Prefer not to say [Exclusive]
- ☐ No
- ☐ Prefer not to say

5.3. Have you ever cared for someone with a chronic condition? (e.g. child, parent, spouse, other family member)

- ☐ Yes
  - **i.** What kind of health condition do they have?
    - ☐ Autoimmune condition
    - ☐ Cancer
    - ☐ Cardiovascular disease
    - ☐ Diabetes
    - ☐ Kidney disease

    - ☐ Sensory condition (for example, sight and/or hearing)
    - ☐ Respiratory condition
    - ☐ Musculoskeletal condition
    - ☐ Mental health condition or cognitive condition
    - ☐ Neurological condition
    - ☐ Other long term
    - ☐ Prefer not to say [if selected, no other selection can be made]
- ☐ No
- ☐ Prefer not to say [if selected, no other selection can be made]

Thank you for completing this survey. We would now like to ask you a couple of questions about the difficulty of the survey and give you the opportunity to provide us with any feedback you may have.

How difficult did you find answering these questions*: (very difficult, difficult, neither difficult or easy, easy, very easy]

Do you have any comments that you would like to make about this survey in general or about the specific scenarios?

Thank you for completing this survey.