Barriers to Genomic Implementation

Policy, Regulatory and Legislative Barriers to National and Jurisdictional Genomic Implementation

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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.

Policy, regulatory and legislative barriers to national and jurisdictional genomic implementation

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Project Overview

Genomic research, including testing and precision health care technologies, has advanced far more rapidly than the ability of Australia's health care systems to adopt these technologies into routine clinical care. The multi-tiered governmental frameworks that oversee the provision of health care in Australia add to the unique implementation challenges associated with genomics, including specific ethical, legal and social implications that require consideration. A multitude of policy, regulatory and legislative barriers affect national and jurisdictional genomic implementation. The experiences and opinions of stakeholders working in health genomics across different professions are critical to understanding the impact of these barriers and what can be done to address them.

In preparation for a coordinated national approach to the implementation of genomics in Australia, Australian Genomics sought to investigate stakeholder perspectives on the policy, regulatory and legislative barriers to genomic implementation into healthcare in Australia. This project serves as a point-in-time assessment of genomic implementation progress, from the perspective of those at the 'coal-face' of genomics health research and service delivery. The findings from this project can inform further work by the nascent Genomics Australia, as well as health policy and decision-makers in health systems across Australia.

Aims:

1: Provide an overview of stakeholder views and experiences of the policy, regulatory, and legislative barriers to the implementation of genomics into healthcare in Australia, and their views on how to address these barriers.
2: Inform further research, decision making, prioritisation, and resource allocation to further genomics implementation in Australian health systems via a coordinated national approach.

Methods

Australian Genomics conducted a survey in late 2024 to obtain stakeholder views on the policy, regulatory and legislative barriers to genomic implementation. We invited clinicians, laboratory scientists, researchers, consumer representatives, health professional body representatives and policy stakeholders to complete the survey. The survey asked respondents to nominate their top three policy, regulatory and legislative barriers to genomics implementation (including but not limited to a defined list of 15 options) and respond to four short answer questions about why they nominated the barrier, what they thought should be done about the barrier, evidence of the impact of the barrier, and current projects or actions to address these barriers. Respondents were asked specifically about policy, regulatory and legislative barriers, to preserve the scope of the project.

Survey data collected included the respondents' profession and jurisdiction. A thematic analysis of the free text responses to the first two short-answer questions was undertaken to identify common themes, interdependent issues and areas of contention.

As the project sought the professional opinions of expert stakeholders, human research ethics review was not obtained. Survey respondents are not identified, any potential identifying details have been removed, and no direct quotes are included in the analysis.

Key Findings

What did stakeholders think are the most significant policy, legislative and regulatory barriers to genomics implementation?

There were 104 valid survey responses. The most nominated barriers were 1) Equity of access to genomics in healthcare, and 2) genomic education and training for non-genomic healthcare professionals and 3) Standardised implementation pathways, with 68, 52, and 23 people nominating these respectively. Responses showed that barriers to genomic implementation are related and interdependent. For example, ***respondents related problems with inequity of access to inadequate staffing in clinical genomics services, insufficient genomic workforce capacity, inconsistent clinical practice across and within jurisdictions among genomics health professionals, and the low genomic education and training of non-genomic health professionals***. Respondents described how inequity of access to genomics informed healthcare is worse for those living in rural and remote areas, members of culturally and linguistically diverse (CALD) communities, those living on a low income, and Aboriginal and Torres Strait Islander people. Respondents pointed to ***the lack of national and jurisdictionally consistent policies and associated resourcing*** to address these barriers. Furthermore, respondents also raised the shortcomings of the ***processes to support the translation of research findings into standardised clinical practice*** as significant barriers to genomic implementation. Respondents noted the complex, obscure, lengthy, inefficient and jurisdictionally inconsistent regulatory and administrative approval processes required to adopt evidence-based technologies or changes to clinical practice.

How did stakeholders think we should address the barriers?

Of 104 individuals who selected their top three barriers, 60 provided recommendations on how to address at least one. Recommendations focused on the need for a ***nationally coordinated approach*** to genomics implementation to ***support equitable and effective integration of genomics*** into healthcare. Key recommendations include developing ***consistent, evidence-based clinical guidelines and referral criteria*** across health professions and jurisdictions, ***streamlining genetic testing and laboratory processes,*** and investing in ***secure, interoperable data infrastructure*** to support clinical care, research, and privacy. Respondents also emphasised the importance of a ***comprehensive education and workforce strategy*** that embeds genomics and ***cultural safety training*** at all levels. Active ***involvement of culturally diverse and priority populations*** in the ***design and delivery*** of genomic ***services*** was seen as essential. To enable implementation, stakeholders called for ***sustainable funding models, innovative service delivery approaches, and expanded training***

capacity. Finally, stronger ***policy leadership, legislative reform, and alignment of national service models*** were recommended to ensure genomics is accessible, trusted, and beneficial for all Australians.

Next Steps

This survey has provided a point-in-time snapshot of stakeholder perspectives on the policy, regulatory and legislative barriers to genomics implementation. Genomics Australia is well positioned to address the barriers described by the survey's respondents by forging a nationally consistent approach to many of these issues.

Due to the survey's limitations, further in-depth expert consultation is recommended to better understand stakeholder views on the priority actions needed to address the identified barriers. Almost all barriers raised had national implications and forging agreement between the Commonwealth and the states and territories on any proposed solutions will be important to address these barriers. Responses from some jurisdictions, including the Northern Territory and Queensland, as well as some stakeholder groups, such as laboratory workers, policy professionals and consumer advocates, were insufficient to draw strong conclusions from the data. Future targeted engagement may be beneficial to address these information gaps.

The refresh of the National Health Genomics Policy Framework, with a public consultation planned for mid-2025, will be an excellent opportunity to address knowledge gaps and ensure the updated Framework reflects the current issues identified by this survey's respondents and the needs of stakeholders involved in implementing genomics across Australia's health systems.

Background

Funding, policy and regulatory responsibilities within Australia's multitiered health systems are complex[1]. Public hospital service management and operation is the sole responsibility of state and territory governments, while funding is shared by both federal and state and territory governments, as agreed under the National Health Reform Agreement. Private hospitals are managed by the private sector which are overseen by government, and Medicare covers some of the costs of private patients in a private hospital. Policy direction and resourcing for primary care and funding for the Medicare Benefits Schedule (MBS) and Pharmaceutical Benefits Scheme (PBS) is provided by the Commonwealth, advised by several committees. The Medical Services Advisory Committee (MSAC), for example, assesses applications for subsidies of health services and technologies, particularly those that would attract MBS funding. Clinical practice guidelines can be developed by specialist colleges, jurisdictional health authorities and not-for-profit organisations with standardisation guidance from the National Health and Medical Research Council. Laboratory practice and accreditation is undertaken by the National Association of Testing Authorities under the National Pathology Accreditation Scheme. The Australian Commission on Safety and Quality in Healthcare develops national safety and quality standards and clinical standards.

Responsibility for education and training standards and professional competencies of health professionals is determined by professional clinical bodies and colleges and oversight bodies including the Australian Medical Council, the Medical Board of Australia and the Australian Health Practitioner Regulation Agency. Workforce planning is the responsibility of federal and state and territory governments in collaboration with universities, specialist colleges and professional bodies. Thus, the jurisdictional and national legislative, regulatory and policy frameworks supporting the delivery of healthcare are overlapping, complex and can be inconsistent, affecting the adoption of new health technologies, workforce development, and interjurisdictional cooperation. The complexity of these arrangements can also result in responsibility and policy gaps, which prevent critical planning processes and resourcing decisions from occurring.

Against this backdrop, genomic health care raises unique and complex social, ethical and legal issues, such as privacy and the ownership of genomic information. Inconsistent policy settings with regards to issues such as data sharing and use can create uncertainty for clinicians and researchers and impact patient care and access to the best available technology. Unaddressed ethical and regulatory issues can result in genetic discrimination, such as in life insurance underwriting, and data security is an ongoing concern. Timely public input into policy development on these matters is critical to ensure ethical issues are fully considered and addressed in regulatory and policy frameworks prior to clinical and research practice, but often does not occur.

The relatively small number of genomic technologies and interventions that have thus far been systematically implemented in Australia's health systems have contributed to a rapid growth in demand for genomic healthcare. The genomic health workforce cannot meet current consumer demand, the genomic education and training of non-genomic health professionals is insufficient to

[1] https://www.aihw.gov.au/reports/australias-health/health-system-overview

support patient access to such care or genomic implementation more broadly, and health systems are not keeping pace with supporting fit-for-purpose policies, processes and regulatory frameworks.

Furthermore, as research rapidly advances our knowledge of health genomics, there is an ongoing need for decision makers to understand the cost/benefit impacts of new technologies and interventions to make timely resourcing decisions, but the assessment of these aspects is not often built into the research cycle, where the focus tends to be on investigating clinical effectiveness. This issue, combined with obscure and complex processes for technology adoption, means that the time lag between research and implementation in public healthcare can take many years[2].

There have been various efforts to address some of the barriers raised in this report both in research and applied health contexts across Australian jurisdictions, such as the recently released culturally appropriate resources from the National Centre for Indigenous Genomics (NCIG)[3]. Additionally, while work to address genetic discrimination barriers has been successful with the Australian Government committing to legislating against genetic discrimination in insurance[4], draft legislation is yet to be presented to parliament. These efforts are valuable, however there are many other barriers remaining that require policy recognition and adoption on a national scale.

Introduction

From 1 July 2025, Genomics Australia will lead the development of a coordinated national approach to genomics implementation in Australia's health systems. This work will be guided by the refresh of the National Health Genomics Policy Framework presently being undertaken by the Australian Government's Department of Health, Disability and Aging, and further jurisdictional discussion and agreement on prioritisation and actions. For a national approach to succeed, however, there is a critical need to understand how barriers are affecting genomic implementation 'on the ground' and bridge the knowledge gap between decision- and policymakers and those who operate within the systems created by policy, regulatory and legislative frameworks guiding genomic healthcare implementation.

Therefore, this project sought to engage with stakeholders developing, implementing and delivering genomic healthcare, to understand their perspectives on policy, regulatory and legislative barriers to implementation, the impact of these barriers and their priorities for action. The findings of this report are intended to inform Genomics Australia in further work to investigate, prioritise and address these barriers. We also sought input from policy professionals to understand if there were significant differences in perceived barriers and actions for change between 'decision-makers' and 'implementers'.

[2] Morris, Z. S., Wooding, S., & Grant, J. (2011). The answer is 17 years, what is the question: understanding time lags in translational research. *Journal of the Royal Society of Medicine*, *104*(12), 510–520. https://doi.org/10.1258/jrsm.2011.110180
[3] https://ncig.anu.edu.au/news/culturally-appropriate-resources-expand-access-benefits-genetics-and-genomics
[4] https://ministers.treasury.gov.au/ministers/stephen-jones-2022/media-releases/total-ban-use-adverse-genetic-testing-results-life

Methods

An online stakeholder survey managed via the REDCap platform was used to gather the data for this project, conducted in the second half of 2024.

Survey design

The purpose of the survey was to explore different stakeholders' views about the most critical policy, regulatory and legislative barriers to genomics implementation, and proposed solutions to address these barriers. The survey also sought evidence of the impact of these barriers from respondents, and information on any projects or activities that had been undertaken or were underway to address the nominated barriers.

The project team sought to limit the time burden of responding to the survey to approximately 10 to 15 minutes, to encourage maximum participation. The project team developed a draft survey, which was reviewed and tested by project advisors and Australian Genomics staff. The survey was revised in response to feedback. The text of the final survey is at Appendix 1.

Minimum data for a valid survey response included the respondent's profession, the jurisdiction in which that person primarily operates (state/territory or national), and the respondent's 'top three' policy, regulatory and legislative barriers to genomics implementation. The respondent could choose three barriers from the example list of 15 provided in the survey or describe up to three of their own barriers. The barriers included in the list were identified through a desktop review of academic and grey literature and reviewed by the project leads and other experts at Australian Genomics. There were then four optional free-text questions relating to each barrier nominated by the respondent.

1. Why do you think this is a significant barrier?
2. What do you think should be done to address this barrier?
3. Do you know of any evidence relating to the significance or impact of this barrier to genomics?
4. Do you know of any projects underway that investigate and/or seek to address the barrier?

Responses to the free-text questions were not required for the minimum data to be included in the quantitative survey analysis below.

Target survey participants and recruitment

The stakeholders targeted by the survey included those developing, implementing and delivering genomics in healthcare, and those designing the policy and regulatory systems and processes within which implementation occurs. The targeted professions included:

a) genetic health clinicians (comprising clinical geneticists and genetic counsellors);
b) laboratory workers;
c) researchers (in any field related to health genomics);
d) consumer advocates;
e) health professional body representatives;
f) industry representatives; and

g) policy professionals

Any interested professional, however, could participate in the survey. All respondents were required to provide their profession. The survey was sent to several established stakeholder databases developed by Australian Genomics over the past eight years during its extensive research and stakeholder engagement activities. Survey links were sent directly by email to approximately 130 clinicians, clinical genomics and laboratory services, 40 researchers, professional bodies and industry and consumer advocates, and 40 genomics policy experts. Addressees were asked to share the survey link to any other individuals who may wish to participate. The survey was also promoted via Australian Genomics clinical, policy and consumer networks and follow up and reminders were provided to encourage participation. A reminder email was sent approximately one month prior to the survey's closure. The survey was open from 9 October 2024 to 13 December 2024.

To encourage frank responses, survey participation was anonymous, unless individuals wished to self-nominate for participation in a possible future interview. Participants were advised that a project report including anonymised survey data may be published on the Australian Genomics website and disseminated to stakeholders, including policy and decision-makers across Australia. Due to the significant amount of material provided by respondents to the survey's short answer questions and time required for analysis, interviews were not conducted.

Human research ethics review was not obtained given the survey sought to obtain the opinions of experts in the course of their professional duties. No quotes or identifying information in the analysis are provided here and survey respondents are not identified in order to preserve anonymity.

Survey analysis

The project team conducted a quantitative analysis of the survey data, and a thematic analysis of text-based responses to the first and second short answer questions. The thematic analysis sought to identify common themes within responses to the short answer questions for each barrier, common themes across barriers, illustrative examples of the significance or impact of a barrier, or clear recommendations for addressing the barriers. Areas of significant difference or contention were also identified. Some respondents nominated their own barriers. A summarised thematic analysis of the text-based responses to the first and second short answer questions is provided in the results section.

Significantly fewer participants provided a response to questions three and four than questions one and two. Many of the free-text responses to question three cited personal experiences or anecdotal evidence, while detail of the actual experience or evidence was not included. Responses to both questions three and four included links or references to ongoing studies, publications or projects that are underway. Due to the lower number and the nature of responses to questions three and four, these were summarised, edited to preserve anonymity and presented in tables in Appendix 2.

Results

Quantitative Survey Analysis

Of the 116 respondents, 104 provided minimum data for inclusion in the survey analysis. Of these, the primary occupational categories selected are represented in Figure 1.

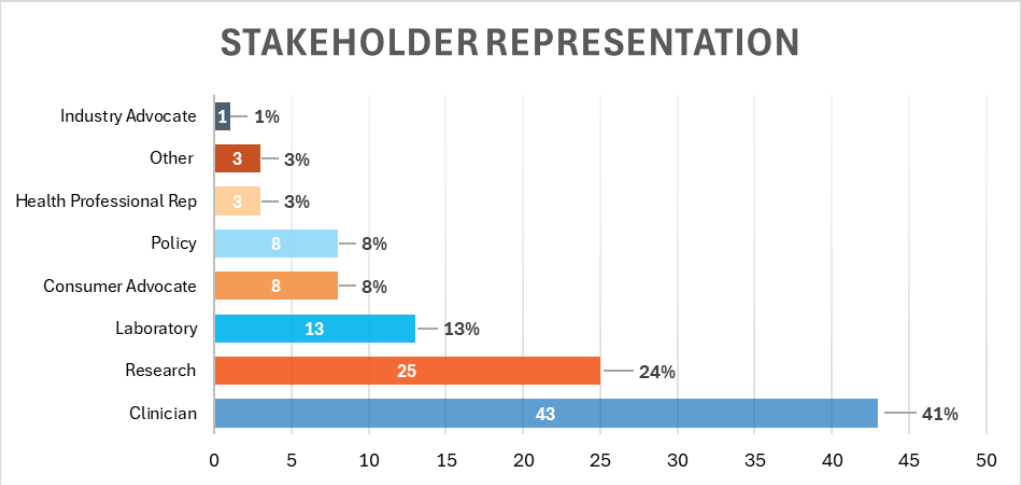

FIGURE 1: The number and percentage of respondents that selected each occupational category as their primary occupation. Of three respondents in the 'other' category, two specified they work in 'education' and the third specified their profession as a 'Genetic Counsellor'.

These individuals provided information about the jurisdiction in which their work is most relevant:

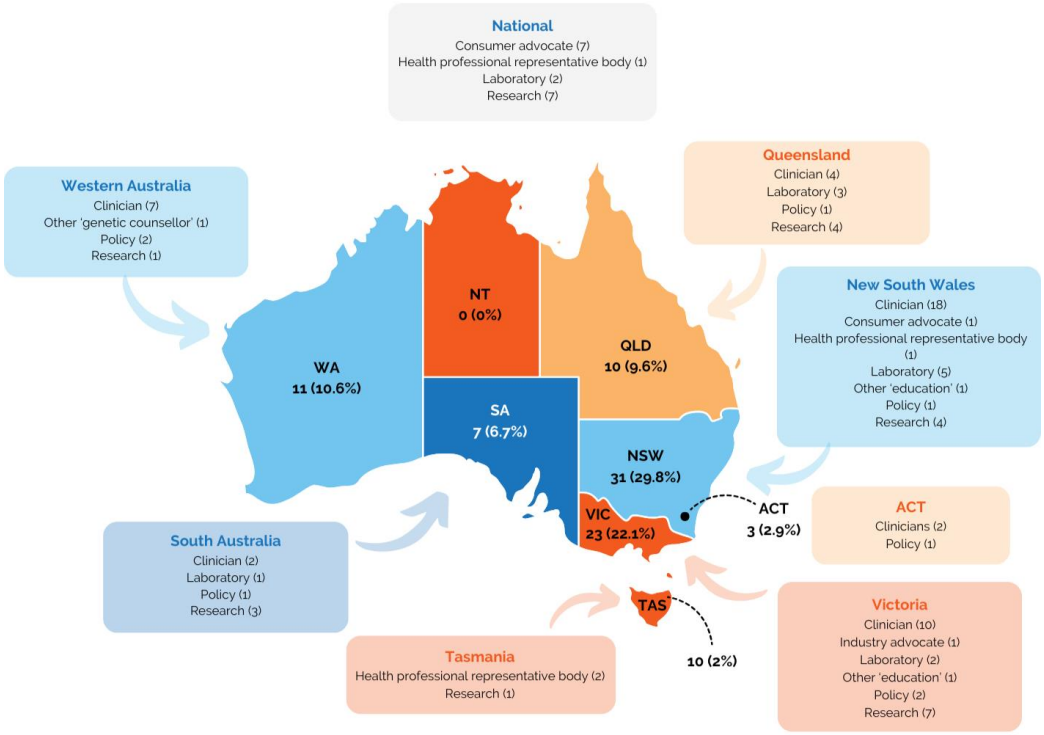

FIGURE 2: The number and percentage of respondents from each jurisdiction (state/territory or national), and a summary of stakeholder representation for each jurisdiction. 17 individuals indicated that they worked nationally.

Top Three Barriers

Of those who responded to the survey, **104 individuals** answered the question seeking their **top three legislative, regulatory or policy barriers** to genomic implementation**.** The category options and responses are summarised in Figure 3.

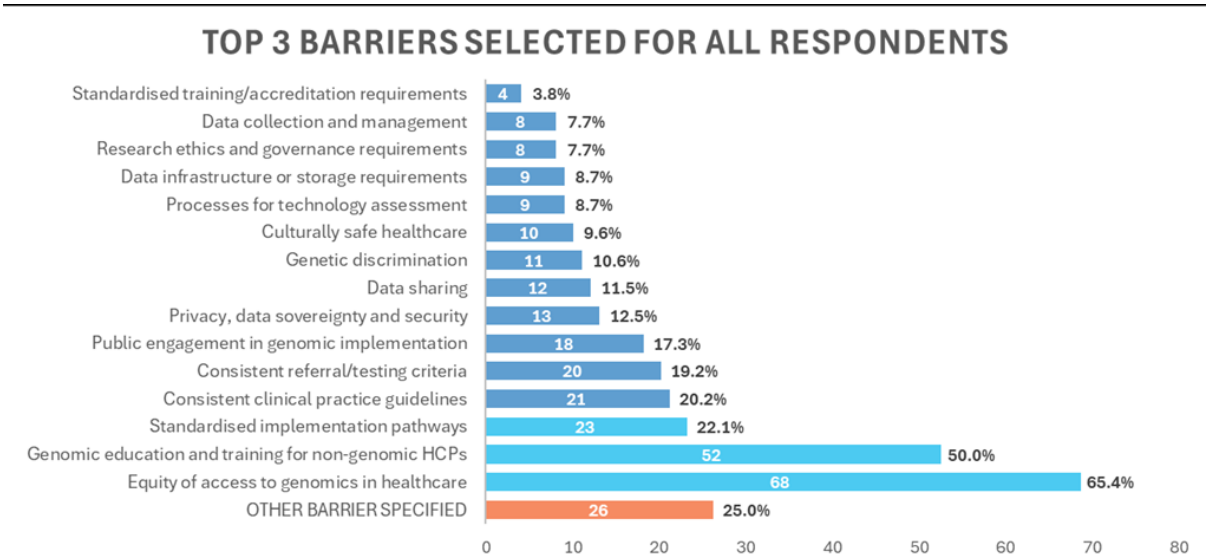

FIGURE 3: The number and percentage of respondents that selected each barrier as one of their top 3 barriers. Each respondent selected 3 barriers; therefore, the sum of barrier frequencies equals 300%.

The barriers most frequently included in the **top three** barriers nominated across all stakeholders were:

1. **'Equity of access to genomics in health care'** with 68 respondents, or 65.4% nominating this barrier.
2. **'Genomic education and training for non-genomic healthcare workers'** with 52 respondents, or 50% nominating this barrier.
3. **'Standardised implementation pathways'** with 23 respondents, or 22% nominating this barrier.

Other barriers

In addition to the defined barrier options, 26 additional barriers were specified by 22 respondents in the free text section. These are listed in Appendix 3.

Responses by Stakeholder Group

A breakdown of responses by stakeholder profession is below. Industry advocates and health professional representatives have not been included, as there were only 1 and 3 responses for these groups respectively.

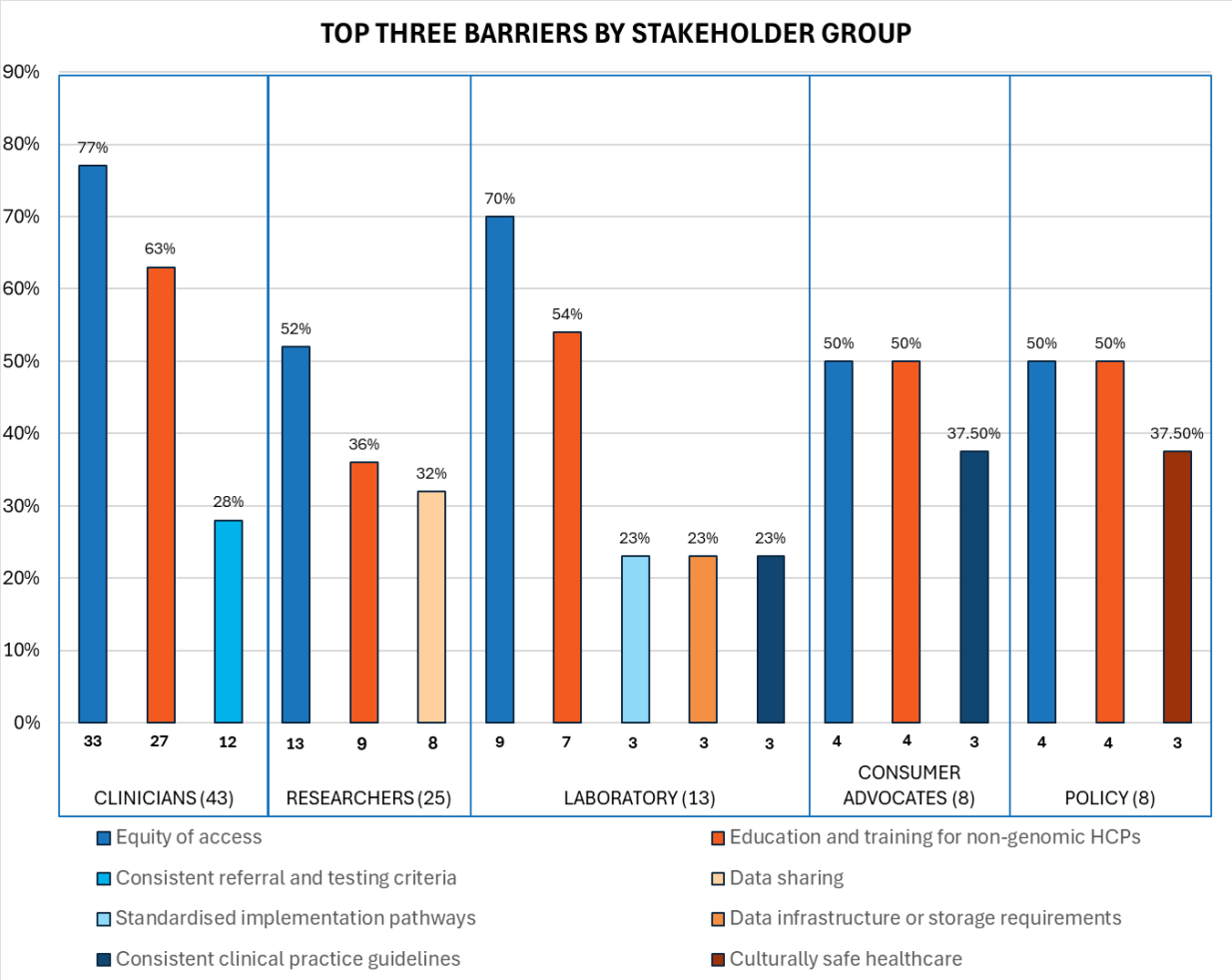

FIGURE 4: The top 3 barriers nominated for each stakeholder group. The percentages represent the proportion of respondents from within each stakeholder group that selected each barrier. The number of respondents that selected each barrier within stakeholder groups is stated below the bars in **bold,** as well as the total number of respondents for each stakeholder group in brackets. Only the top 3 barriers selected for each group are represented in this figure. For laboratory respondents, the 3rd most frequently selected barrier was equal between 3 barriers. Responses from the remaining stakeholder groups were too few to provide meaningful trend data.

Responses by Jurisdiction

A breakdown of responses by jurisdiction is below. ACT and Tasmania have not been included, as there were only 3 responses for each jurisdiction.

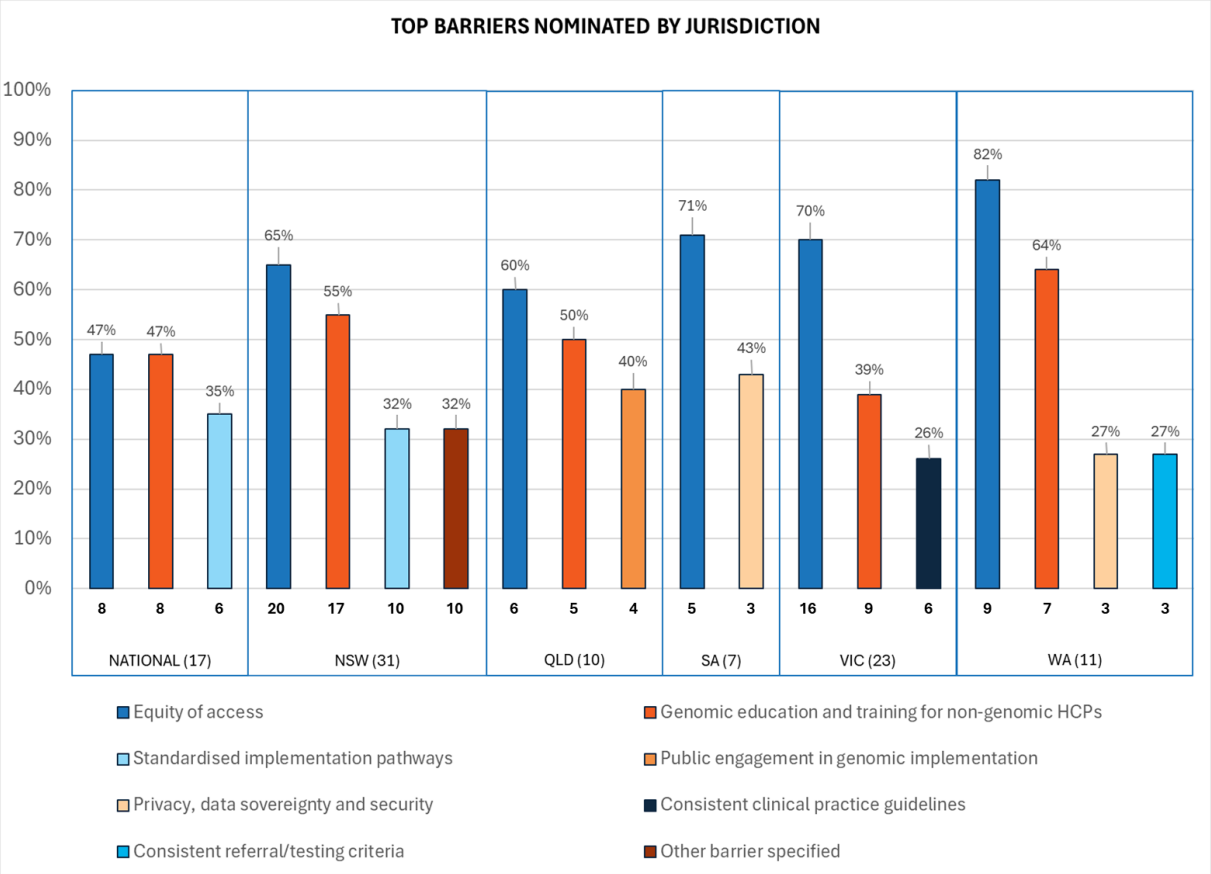

FIGURE 5: The top 3 barriers nominated for each jurisdiction. The percentages represent the proportion of respondents from within each jurisdiction that selected each barrier. The number of respondents that selected each barrier within jurisdictions is stated below the bars in **bold** and the total number of respondents for each jurisdiction are stated in brackets. Only the top 3 barriers selected for each group are represented in this figure. South Australia only had two barriers that were clearly the most frequently selected, while there were multiple barriers that were equal third, which are not represented here. For NSW and WA respondents, the 3rd most frequently selected barrier was equal between 2 barriers.

Qualitative Survey Analysis

A thematic analysis of the free-text answers to questions one and two are presented below in order of popularity of barrier nominated. Appendix 3 provides a summary of the responses. Appendix 4 provides a summary of responses to questions three and four.

Equity of access to genomics in healthcare

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	68	33	13	4	1	9	4	2	2
No. of people who provided responses to free text questions*									
Total	*40	20	5	3	1	5	4	1	1

* Not all respondents who answered short answer questions answered all four. Many of the comments covered multiple topics.

Responses to Question 1 - *Why do you think this is a significant barrier?*
The most commonly cited reason that equity of access was considered a barrier was because of **inconsistent or limited access in rural and remote areas**, raised by **15 respondents** representing all fields except health professional body representative. This was further supported by an additional **five people** who **said access and resourcing varies** based on the local health district and another **four who said genomics expertise and resourcing was concentrated in a few centres of expertise**, usually not in rural/remote areas.

Nine individuals commented on **access issues for Aboriginal and Torres Strait Islander people**, highlighting lack of representation, language or cultural barriers, that they are less likely to receive a referral and how these factors widen inequality. One consumer advocate also added that **Aboriginal and Torres Strait Islander people** are **underrepresented in research** along with **other culturally and linguistically diverse (CALD) people**. There were another nine comments on variable access for CALD people, again noting inaccessible models or challenges navigating healthcare, lack of representation, language barriers, limited health genomics literacy, and siting a need for culturally safe services. Similarly, **seven comments** discussed how **socioeconomic/social demographic factors were a reason for consumers to be less likely to get a referral or access care**. **Existing health inequities can be exacerbated in genomics** as raised by **three participants**, one clinician explaining that only highly active self-advocates are able to access the best care. Additionally, four people raised issues with **genomic data**, three saying it **lacked diversity** and one laboratory worker calling for better data sharing.

There were **eleven comments about funding issues**, five were about different mechanisms or models creating inconsistencies (e.g. between private and public), and six about a lack of or variable funding leading to insufficient resourcing to provide testing, appropriate resources (i.e. staff), and to integrate genomics. There were **eight comments directly related to workforce need**, mainly a shortage of genomic professionals, one clinician specifying a lack of administrative support, and

another clinician outlining that genetic counsellors are restricted to working in genetics departments. This was echoed by another **eight people who raised long wait times, increasing referrals and growing demand for genomics services**. Two clinicians also raised the inadequacy of current infrastructure. Two people also commented on the prioritisation of testing, in the context of public services having long wait lists, meaning critical cases are seen sooner, delaying access for less critical cases that still require care.

Eight people raised **inequity issues stemming from non-genomic health professionals**, including **not having** the **time or skills to provide genomic health care, and varying levels of expertise, motivation, integration or access to funds**. Separately, three people mentioned a **lack of genomics education for non-genomics health professionals** or that education could be improved. Four people cited **issues with referral pathways**, with patients not being referred or referral relying on non-genomic health professionals' variable understanding of health genomics. An industry representative and a laboratory worker said **bias or a clinician's training, or beliefs affects care**.

Four participants commented on a **lack of standardised approaches to genomic healthcare**, and **variations in the availability of genomic testing** across Australia. This was reflected in **four** other **comments about MBS testing**, indicating that the requirement that certain MBS items can only be ordered by (or in consultation with) clinical geneticists create barriers to accessing MBS subsidised testing, issues with providing MBS equivalent genomic services to public patients, and a rigid system that does not facilitate frequent revision of MBS item numbers and criteria. **Seven people** said the **cost of testing creates inequity in access to testing**.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Acknowledging that resourcing (e.g. funding) is often a barrier, please provide suggestions specifically relating to policy, regulation or legislation.

Over half of the individuals (27) that commented **mentioned funding**, even though the question included an acknowledgement that resourcing/funding is often a barrier and asked for suggestions specifically relating to policy, regulation, or legislation. Fourteen of the 27 comments called for **funding/investment** in a variety of areas including **education for healthcare professionals**, **implementation, clinical space, training positions or workforce or screening programs**. The other 13 comments about funding related to funding mechanisms including providing **funding to local directors** (suggested by a clinician), **increasing the number of genomics-related MBS item numbers and the** level of **subsidy they provide**, **expediting the MSAC process**, **consolidated and consistent funding models** across jurisdictions and healthcare settings (i.e. public vs private), influencing activity-based costing, and **exploring private funding options** such as private sector investment or public-private partnerships to expand access.

Another common theme, mentioned by **20 participants**, were **actions to improve access to genomic healthcare by culturally diverse, Indigenous, and priority populations**. A large portion of those comments, **14** out of 20, were about **targeting diverse communities through research and engagement**. There were **10 comments highlighting the need for engagement with representative groups to understand their needs**, **identify solutions** and **ensure involvement in decisions making** and genomics initiatives. The remaining four people commented on **improving equitable research**

participation for diverse populations, with some suggestions of mandating community involvement, including requirements in grant applications about sample databases and research partnerships with community. One consumer representative mentioned working with communities via both research and engagement regarding decision making. The other **six** of 20 participants specifically **called for culturally appropriate resources**, two of which mentioned appropriate **resourcing for translators or providing transport and accessibility options**.

Rural and remote access and **workforce related solutions** to **improve equity of access** to genomic healthcare were raised by **10 individuals**. Four proposed **digital solutions** including improved virtual/visiting mechanisms, virtual Multidisciplinary teams (MDTs) and telemedicine/telehealth solutions. The remaining six comments were more generalised recommendations to **improve rural services such as policy, infrastructure, workforce, centres of excellence, and education to support genomic champions in rural areas**. Separately there were **four** comments that **called for a general increase in workforce capacity and capability**.

Education and training for non-genomic health professionals was another common theme that **13 individuals** addressed in their comments, covering a variety of approaches. Eight comments highlighted **education and support needs for non-genetics health professionals**, one clinician saying this should be standardised and a consumer advocate stated health professionals should undertake training on the provision of culturally safe healthcare. Four other comments focused on training, three of which were about **increasing the genomics-trained workforce** by providing **training incentives/scholarships for genomics healthcare professionals**. One individual who worked in the education field suggested providing **scholarships for CALD students**. Another mentioned **incorporating more genomics into medical training**. Three individuals referred to **public education campaigns**, working with schools, community, and the public to improve the broader community's genomic health literacy.

Eleven individuals made comments about **improving access to genomic testing**. **Five** of these comments proposed making genomics available **at the primary or community health care level**, one individual suggested both. Mainstreaming was raised by three individuals, one clinician specifically recommending mainstreaming whereby clinicians caring for patients with a single system condition are empowered to order and interpret genomic tests and one representative from a health professional body suggesting mainstreaming needs to occur simultaneously with a focus on the genomics workforce. Three additional comments included suggestions for **alternative pathways to access genetic healthcare/testing outside tertiary hospitals**, two of which highlight **barriers that prevent genetic counsellors working in non-clinical roles or roles outside specialist genetics service**.

There were twelve comments regarding **standardising the provision of genomic healthcare** to improve equity of access. Six of these comments were about **consistency with regards to referral pathways/guidelines, approach to treatment, clear roles and responsibilities, clinical guidelines and improving state-based strategies**. Three individuals commented on the **need for a coordinated approach to policy** and collaborations and sharing of resources across Australia to prevent duplication. Another three comments called for a clear approach to **testing** to ensure **consistency and clarity across jurisdictions**, one laboratory worker proposing to adopt a **genetic test directory**

like that used in the NHS. Across all comments regarding standardisation, four specifically called for a **national approach**.

There were four comments that provided specific recommendations for policy or government-focussed solutions. These covered policies that **specifically address inequities** in accessing genomics, policies/frameworks that ensure public health programs such as Medicare include comprehensive genomic testing and **ensuring Medicare covers genomics services**, and **state and territory government initiatives to integrate genomic services, particularly in rural areas**. Additionally, one researcher suggested there should be a focus on practical implementation instead of desktop review.

Three individuals had data related ideas including **ensuring data sovereignty** and **accurate data collection** about **demand** as well as to measure **disparity**. A policy professional suggested developing infrastructure to enable **system-wide linkage of genomics programs**, including multi-cultural and Aboriginal and Torres Strait Islander genomics projects and programs. Lastly, one consumer advocate suggested that **equity** should be **incorporated** as a **core principle in genomic research and healthcare**.

Education & training of non-genomic healthcare workers

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	53	27	9	4	0	7	4	1	1
No. of people who provided responses to free text questions*									
Total	26	11	4	3	0	4	3	0	0

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Of the 26 people who responded to this question, 12 stated that **non-genomic healthcare workers (NGHWs)** generally **do not have the genomic skills, knowledge and confidence needed** to **implement genomics-informed healthcare** in their practice, and six mentioned that **NGHWs** are **needed to mainstream genomics** throughout the health system. 10 mentioned **insufficient genomics education and training** for both existing and new NGHWs. Four individuals stated that the (lack) of education and training **of NGHPs restricts patient access** to genomics-informed healthcare. While one clinician thought that NGHWs should be empowered to order genetic testing, citing a culture of 'gate-keeping' holding back the implementation of genomics among NGHPs - another clinician thought that NGHPs should have to be credentialled before being **able to order testing**, to **ensure appropriate identification of suitable patients, interpretation of results** and **ability to discuss the ethical, legal and social implications of genomic testing with patients**. A policy professional noted the **shortage of qualified genomic health workers**, stating that there is **no policy for developing the genomics workforce**, and a clinician noted that neither governments, professional bodies nor educational institutions have taken **responsibility for coordinating or establishing standardised genomic education and training for NGHWs**. A clinician noted the

stresses placed on clinical genetics services, citing **long wait times for patients**, due to lack of genomics expertise among NGHWs.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
11 people stated that **genomics education and training should be provided to or required for all health professions**, both during **undergraduate** education, during **post-graduate specialisation**, as well as in **Continuous Professional Development** for already-practising health professionals, and seven mentioned that this should be **standardised across institutions** with required competencies. A policy professional argued that a government strategy is needed to integrate genomics across the health system, another that there should be a **national genomic workforce review** to understand **gaps and future needs**. One researcher thought that a **national public awareness campaign** may lead to patients seeking genomics-informed healthcare, which will encourage NGHPs to upskill on genomics. Two people argued that there needed to be **greater investment in the genomics health professional workforce**, while another argued for **resources** to **support genomic health professionals in educating NGHWs**. One clinician argued for the opportunity for NGHWs to be credentialled for ordering genetic testing. A researcher stated that **NGHWs should be engaged in any planning processes for genomics implementation**. Two individuals mentioned the importance of providing good quality, centralised **information resources on genomics for practising NGHPs**. One clinician argued for the **de-coupling of genetic counsellors from clinical geneticists**, stating the genetic counsellors are well-equipped to be embedded in other specialties, which **will expand patient access to genomic-informed health care** and **increase competencies of other health professionals**. One consumer advocate argued that **genetic counsellors** could also work in primary healthcare in **partnership with General Practitioners**, while another consumer advocate argued for genomic health professionals to be embedded into all **multi-disciplinary teams** in hospital settings.

Standardised implementation pathways

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	23	6	7	3		3	1	3	0
No. of people who provided responses to free text questions*									
Total	9	1	2	2	0	2	1	1	0

* Not all respondents who answered short answer questions answered all four questions.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Of the nine individuals who responded to this question, two consumer advocates stated that there are **no clear pathways to implement genomics into the health system**, while four individuals stated that despite many examples of **evidence-based innovations** developed in research contexts, the **mechanisms for implementation** of these innovations, including funding pathways, are **lacking**. Two individuals stated that **MSAC/PBAC processes** are **not always suitable** for **genomic innovations**. A health professional body representative stated that more **research needs** to **demonstrate** the **cost effectiveness of implementation as well as clinical utility**. A laboratory worker indicated that many

researchers do not understand the pathways to implementation and a policy worker indicated that a focus on **implementation of innovations for adults was lacking when compared with children**.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Of the nine individuals who answered this question, one clinician stated that **political leadership** is required to establish implementation pathways, and that **clinicians should not need** to **use research pathways to fund and support what should be standard clinical practice**. One clinician stated that **research participation** by clinicians **should be** properly **acknowledged in workloads and funded**. Three others discussed the need to establish **nationally consistent implementation pathways**, **developed collaboratively**, while another discussed the need for **strategic planning** for implementation that is accordingly **resourced** and another the need for a **single health technology assessment process**. One clinician stated that **clinical genomics services should be state-wide** to **support equity of access** and **workforce planning**, where this is not already the case. A laboratory worker discussed the need to **improve researchers' understanding of implementation pathways**, while another laboratory worker stated that aside from technology implementation pathways, implementation includes the need for implementation of **clinical practice guidelines**, **new models of care across specialties**, and a **nationally agreed genomic test directory** to guide funding support across jurisdictions in the public health system.

Consistent clinical practice guidelines

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	21	8	7	2	0	3	0	0	1
No. of people who provided responses to free text questions*									
Total	10	2	2	2	0	3	0	0	1

* Not all respondents who answered short answer questions answered all four questions.

Responses to Question 1 - Why do you think this is a significant barrier?
Of the ten individuals who provided a response to this question, five said that a lack of consistent clinical practice guidelines in public genetic testing results in **inequitable access for patients**. A consumer advocate raised problems **with inconsistent advice on management** given to patients with increased genetic risk of cancer. Two individuals stated this is a significant barrier because **non-genomic health professionals do not know when or how to refer patients for genetic testing, nor how to interpret results**, while a researcher said that this barrier was significant because it **hampers the implementation and mainstreaming of genomics throughout the health system**. One laboratory worker raised problems with the **significant length of time taken to obtain approvals for and implement innovations in laboratory settings**, processes for which are inconsistent across jurisdictions.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Six individuals stated that **nationally consistent clinical practice guidelines** for **genomics across all health professions should be established**, to ensure equity of access. One clinician suggested that

the **genetic testing process** should be **simplified and streamlined** and a laboratory worker suggested that **laboratory approvals processes** should also be **simplified**. An educator stated that consistent guidelines could then inform policy, infrastructure and workforce development. A researcher suggested that such **guidelines** should be **collaboratively developed** through **research and evidence-based**, rather than clinician-led, which may lead to the articulation of current practice rather than the identification of best practice. A consumer advocate called for **decision-aids for patients and clinicians**, to assist in management decisions where there is uncertain or no clinician consensus following the identification of increased disease risk following genetic testing.

Consistent referral and testing criteria

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	20	12	2	1	0	3	1	1	0
No. of people who provided responses to free text questions*									
Total	9	4	1	1	0	2	1	0	0

* Not all respondents who answered short answer questions answered all four questions.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Of the nine individuals who provided a response to this question, six individuals said that the lack of consistent referral and testing criteria **impacts** on the **equity of patient access** to genetic testing, and that **depending on patient location**, the clinician, and possible disease, individuals may or may not be offered genetic testing paid for through the public health system. Two individuals stated that a **lack of consistent criteria creates uncertainty for practising clinicians**, which **ultimately impacts** on **patient care**. One laboratory worker suggested that **lack of consistent testing criteria may mean** that the **choice of genetic testing is not evidence based** given the influence of private companies selling technologies.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Of the seven people who answered this question, six indicated that **consistent referral and testing criteria should be established**. One policy worker indicated that while these should be **consistent nationally**, they also need **some flexibility on a state-by-state basis, given dual funding sources** at state and national level and the limited resources of smaller jurisdictions. Two individuals suggested that **improved resources** should be provided to **both clinicians and consumers** about **testing, funding and cost options**. One clinician stated that along with testing and referral criteria for health professionals, **clinical genetics services** also **need consistent criteria to triage referrals and manage waitlists**. One laboratory worker indicated that **health professionals** need **improved education about the utility of different types of genetic tests** but that this should not be provided by private companies.

Public engagement in genomic implementation

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	18	7	4	2	1	1	1	1	1
No. of people who provided responses to free text questions*									
Total	8*	4	1	2	1	0	0	1	0

* Not all respondents who answered some short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*

Five of the responses emphasised the **low understanding of genomics** across the **community**, including the potential benefits and risks, and one clinician also mentioned the low understanding among **health professionals who are non-genetic specialists**. A consumer representative discussed how **lack of public knowledge of genomics worsened equity of access for some communities** due to low public engagement in how genomics is implemented in health systems. One researcher answered that **low public engagement resulted in insufficient public investment**, given the potential benefits of health genomics; an industry representative wrote that a lack of public engagement **increased the potential harm to consumers** from genomics implementation. One clinician felt that **some communities** affected by genetic conditions are **overburdened by engagement requests**.

Responses to Question 2 - *What do you think needs to be done to address this barrier?*

Five respondents wanted a **public engagement or awareness campaign**, two mentioned **increasing genomic education**. A consumer advocate also argued that a **diversity of consumers should be engaged in implementation** and that **those who collected genomic data should be more transparent on how data will be collected, stored and used** and should **ensure** that **consumer privacy and data security is protected**, thereby **building trust** with the public. The clinician who felt that some consumer groups are already overburdened with engagement requests answered that **engagement should be coordinated between specific groups**.

Privacy, data sovereignty & security

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	13	3	6	0	0	2	1	0	1
No. of people who provided responses to free text questions*									
Total	6*	2	2	0	0	1	1	0	0

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*

The **sensitive nature, familial implications and potential for reidentification of genomic data** were highlighted by one researcher as reasons why privacy, data sovereignty and security are barriers to

implementation. Two individuals (one clinician and one laboratory worker) commented that patients want **data security** and to feel comfortable consenting to data usage for personal benefit, healthcare or research; the clinician adding **consenting without fear of adverse consequences**. Another clinician pointed out that **patients are asking complicated questions about data access and storage or asking for access to their raw data**. One researcher raised that there is **awareness of how genomic data can be used to discriminate in terms of genetic testing information in life insurance underwriting and said that Indigenous data sovereignty (IDSov) principles are not embedded in healthcare or research**. A policy representative said there are **problems with individual interpretation of policy/legislation rather than the policy/legislation itself** and that current **systems protect data based on personal risk**. A laboratory representative discussed **inconsistencies in data privacy and security management across jurisdictions**.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Two respondents commented on the need for a **national, secure storage-system/facility**, a researcher stating this should be for samples and data, and a laboratory worker saying this should include a **consent option for both research and diagnostic purposes**, specifying a **digital and dynamic consent system with consumer control**. The same researcher also suggested setting up a **national registry and a national data management and sharing platform**. Three individuals raised legislation, a policy representative recommended **education around data legislation and policy, including appropriate data sharing**; a researcher proposed a **review of data privacy and security legislation**, in terms of **sensitivity of genomic data and jurisdictional differences**; and a clinician raised a need for genetic non-discrimination legislation with oversight at the Commonwealth level. The researcher that recommended review of legislation said IDSov principles should be respected in research and clinical settings. The policy representative also suggested de-risking decisions in terms of personal liability.

Data sharing

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	12		8	2		1	1		
No. of people who provided responses to free text questions*									
Total	*5		2	2		1			

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
All five individuals said that **data is siloed, within jurisdictions, institutions, health systems, state and territory based electronic medical records, services, research institutes, testing laboratories, private databases**. One consumer advocate expanded saying this makes it **difficult to share and access genomic data**. Two people pointed out that genetic **testing** often **relies on previous evidence**, and **siloed data** may **lead to missing diagnoses/misdiagnosis** or inconsistent interpretation due to not being able to access existing evidence; a laboratory worker adding that data, information, and interpretation is often never looked at after the initial test, which can lead to

an increased service cost. A consumer advocate explained that **fragmentation limits the potential for large and diverse datasets**, which are **critical for advancing genomic research and implementation**. The same consumer advocate had a detailed response outlining that due to the highly sensitive nature of genomic information, there are significant **concerns about patient privacy, data security and potential misuse of genetic information**, and that many individuals are hesitant to share their genomic data due to fears about confidentiality and discrimination. This individual raised the **inconsistent standards for collection, storage and sharing of genomic data** and how this lack of standardisation limits the ability to integrate data which limits collaboration and research. Additionally, the consumer advocate commented on how **data sharing is limited by strict data protection regulations that can create legal or bureaucratic obstacles**. Another consumer raised concerns about the lack of data being available or collected with regards to people with cancer risk undertaking preventative surgery, leading to a gap in understanding risk management outcomes.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Four respondents suggested **secure national data storage infrastructure**, with a researcher referring to NAGIM and one consumer also suggesting global genomic databases. A consumer advocate specifically mentioned **fostering public-private partnerships** to create **open-source data** systems that **facilitate collaboration while maintaining strong ethical oversight**. Another consumer advocate proposed a **national registry** for people with a genetic variant. A detailed response from a consumer advocate included **establishing national and international standards and agreements for the collections, storage and sharing of genomic data**, encouraging the development of **interoperable systems** allowing for **secure genomic data exchange between governments, healthcare providers and researchers**, developing **robust systems for securing genomic data** including encryption, secure storage and controlled access mechanisms and **simplifying regulatory processes that can delay or inhibit genomic data sharing**. This individual also suggested implementing **user-friendly consent processes** that clearly inform consumers about how their data will be used and shared, promoting initiatives that allow **patients to have greater control** over their genomic data and **fostering public trust** by engaging in discussions about benefits or and safeguards with regards to data sharing for genomic research.

Genetic discrimination

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	11	6	3	0	0	1	0	0	1
No. of people who provided responses to free text questions*									
Total	8	4	3	0	0	1	0	0	0

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Of the eight individuals who provided a response to this question, three individuals specifically mentioned concerns with **insurance discrimination**, while three stated that **genetic discrimination** is a **barrier to genetic testing and research participation**. One researcher though that the public

remains unsure whether knowing one's genetic status is beneficial or not, while a laboratory worker was the only person to mention **social 'stigma' associated with genetic disorders**. One clinician thought that patients who are already unsure about genetic testing are able to use potential insurance discrimination as a reason to decline.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Five individuals mentioned the need to pass legislation, referring to the **Australian Government's commitment to legislate a ban on genetic discrimination in insurance**, legislation not yet introduced to Parliament. Two individuals mentioned the **need to** then **disseminate information to both the public and health professionals about the ban on genetic discrimination in insurance**, while one mentioned the **need to enforce the legislation when passed**. A researcher also mentioned the **need for insurance companies to be more transparent about the processes they use to make assessments.**

Culturally safe healthcare

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	10	2	3	0	1	0	4	0	0
No. of people who provided responses to free text questions*									
Total	6*	1	2	0	1	0	2	0	0

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Of the five individuals who provided a response to this question in relation to why they thought culturally safe healthcare was a barrier to genomic implementation, three mentioned a **lack of trust in researchers, health providers or science among some cultural communities** because of either **past or current racially discriminatory and exploitative practices**. Lack of trust, exacerbated by a **lack of culturally safe healthcare**, creates a **barrier to patient participation** in genomic research or genomics informed healthcare. One clinician argued that **most genomic datasets are based on individuals of European-descent**, stating that the absence of comprehensive genomic information from other communities **hampers the provision of safe and accurate healthcare**. This clinician also argued that **most research on consumer views** on genomics in healthcare and its acceptability is **also based on mostly 'white' or European-descent communities**, which has **skewed service delivery and implementation decisions**.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
Of the four individuals who provided a response to this question, three mentioned the **need to provide education and training on culturally safe practices to researchers and healthcare professionals**, and one suggested the establishment of a **national standard for cultural safety in healthcare** and assessment against standards. A researcher and a policy professional argued for the importance of having **individuals from affected cultural communities directly involved in**

governance and decision-making around genomics implementation. Two researchers argued that **culturally diverse communities need to be better informed** about genomics in healthcare, one stating that this could occur through the **production of culturally informed and safe information resources**. A researcher argued for the need to **embed cultural safety in all healthcare systems and processes**, including referral, access and treatment pathways. A policy professional argued for the need to **establish state and federal policies on culturally appropriate community engagement in genomics implementation**.

Processes for technology assessment

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	9	1	2	1	0	3	1	1	0
No. of people who provided responses to free text questions*									
Total	2	1	0	0	0	1	0	0	0

* Not all respondents who answered short answer questions answered all four questions.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Of the 2 individuals who provided a response to this question, both criticise **MSAC processes as being too slow**, with one clinician stating that the reviewers do not seem to understand genomic tests and that the **creation and wording of Medicare item numbers is impaired** and thus funding through the public system is inadequate, indicating that **critical tests are not supported** in some cases, while in others wording results in **inappropriate testing**. A laboratory worker states that due to TGA oversight of some companion diagnostics (CDx) assays and validation criteria, access to CDx and cognate therapies may not be accessible in Australia.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
The clinician stated that while MSAC has been advised of problems with genomic MBS item numbers, **greater advocacy from organisations such as the HGSA** or Australian Genomics could assist; the laboratory worker suggesting that **MSAC should have increased resourcing** while in reference to CDx assays, that the TGA should take a **pragmatic approach** based on test performance characteristics.

Data infrastructure or storage requirements

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	9	3	2			3			1
No. of people who provided responses to free text questions*									
Total	*5	1	1			3			

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Three comments were about issues with current data storage, two laboratory workers explaining that **current data** is **stored on external drives creating a risk of corruption** or losing data or that **data is in 'cold' storage, meaning it is not useful due to lack of aggregation at a national level and is expensive to store**. A laboratory worker mentioned that software requirements are not supported by qualified professionals. Three respondents discussed issues with addressing storage requirements, two laboratory workers noting **laboratories do not have cloud storage allocations** or that the **capacity for safe and cost-effective storage of genomic data does not exist**. A clinician acknowledged there is a significant amount of work required to meet cloud IT and storage requirements. A researcher described how, without a way to store genomic data, it is difficult to share it and advance research. The need to manage fear of genomic data being compromised was highlighted by a laboratory worker.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
The three laboratory workers suggested storage solutions, including **organised storage of genomic data, international cloud storage** (noting this is costly), and **national infrastructure** allowing ongoing use of data for clinical and research purposes. The clinician and researcher made a call for **government/federal funding**, the researcher specifically mentioning infrastructure funding. Laboratory representatives also proposed **overcoming jurisdictional barriers** with a **mandate at the national level** and support from qualified bioinformaticians/IT specialists at each laboratory.

Research ethics and governance requirements

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	8	0	5	0	0	1	2	0	0
No. of people who provided responses to free text questions*									
Total	4*	0	1	0	0	1	2	0	0

* Not all respondents who answered some short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
Two respondents highlighted the **complex, inefficient and lengthy** ethics and governance process that lacks harmonisation and mutual acceptance across sites and jurisdictions. The other two respondents from a policy background discussed the **inability of ethics and governance processes to keep up with the rapidly evolving space of genomics**, and the **lack of genomics expertise** in the **HRECs** assessing genomic research applications. They described this having further impacts on projects that include Indigenous genomics and longitudinal projects that might require access to retrospective patient data that was not originally obtained for the purpose of the project being proposed to HREC.

Responses to Question 2 - *What do you think needs to be done to address this barrier?*
Two of the respondents thought that more a **unified national approach** would help address this barrier, including suggestions of **cross jurisdictional and cross institutional agreements**, and a well-resourced **single national ethics and governance entity**. The other respondents advocated for **education of ethics, governance and data governance officers** in genomics research, and underserviced populations.

Data collection & management

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	8	4		2			1		1
No. of people who provided responses to free text questions*									
Total	3*		1	1					1

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
One representative selected this barrier because they felt it encompassed all aspects of data management, including consent. A researcher and consumer representative both stated that **data is siloed within jurisdictions, services, or institutions**. The researcher also highlighted the **exponential growth in the amount of genomic data being generated** and how genetic testing is often reliant on previous evidence. The consumer representative pointed out that there is **no central registry in Australia for germline variants**, and described the benefits of registries including follow-up, risk management, and identifying where there might be issues accessing testing. They also commented on there being no difference in MBS item numbers leading to inability to identify risk management or treatment decision for cancer patients.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
A consumer representative suggested **better sharing of data in Australia to address this barrier, recommending patient registries** to address the current lack of data for hereditary cancer and updating the granularity of MBS item numbers.

Standardised training/accreditation requirements

		Field of Work							
No. of people who chose this barrier in their top three		**Clinician**	**Researcher**	**Consumer Rep.**	**Industry Advocate**	**Laboratory worker**	**Policy**	**Health prof. rep body**	**Other**
Total	4	1	1	0	0	2	0	0	0
No. of people who provided responses to free text questions*									
Total	3*	1	0	0	0	2	0	0	0

* Not all respondents who answered short answer questions answered all four.

Responses to Question 1 - *Why do you think this is a significant barrier?*
The three respondents highlighted different issues for this barrier. One respondent described at length that recent **National Pathology Accreditation Advisory Council** (NPAAC) supervision guidelines have **given inexperienced laboratory clinicians more power** over key operational decisions than scientists in the diagnostic laboratory setting. Another respondent working in the laboratory setting described **inefficiencies and a lack of genetics knowledge within NATA, MSAC and TGA**. The third respondent from a clinical background describes how **lengthy and complex genetics dual training pathways decreases genetics knowledge** and the representation of genetics champions across other clinical and laboratory departments.

Responses to Question 2 – *What do you think needs to be done to address this barrier?*
One respondent recommended that **appropriately qualified clinical scientists should oversee technical and analytical decision-making in the laboratory** with **joint genomic data interpretations with medical colleagues**, and that there should be **supported training for scientists** to fill knowledge gaps. The other lab respondent called for a **Royal Commission into NATA, MSAC and TGA**. The third respondent advocated for **allowing sub-specialty pathways for genetics**, via fellowship, like other specialty programs, which would eliminate the need for an entire secondary specialty.

Other Barriers

1. Funding & Resourcing
Although the survey did not seek responses relating to funding but was focused on policy, regulatory or legislative barriers to genomic implementation in the health system, given that **funding is a major barrier to implementation**, affecting all aspects of implementation, 12 individuals nevertheless sought to raise this as an issue by nominating 'other' barriers and either directly or indirectly raising the issue of funding and resourcing, and 10 of these individuals provided responses to the free text questions.

1.1 Responses to Question 1 - *Why do you think this is a significant barrier?*
Seven individuals stated that the **demand on clinical genetics services is greater than what can be met with current staffing levels and workforce capacity**, in terms of both the availability of trained staff as well as the funded staffing resources provided to clinical genetics services and laboratories. One person mentioned **lack of funding for testing**. These individuals stated that the lack of **resourcing restricts patient access to testing, counselling and clinical advice, citing long patient wait times and overwhelming demands on staff**. One individual stated that the lack of resourcing results in **tightening the referral criteria** to clinical genetics services, meaning that **some patients who may benefit cannot access testing or specialist genomic advice at all**. One individual mentioned that while genomics is rapidly advancing, many of the **bureaucratic mechanisms** that support implementation are **slow to adapt** and that if these remain inflexible and overly prescriptive, implementation will continue to be hampered.

1.2 Responses to Question 2 - *What do you think needs to be done to address this barrier?*
Those who provided responses to this question in relation to funding as a barrier, either directly or indirectly, stated that **increased funding, improved service and funding models could address the**

barrier. Two individuals stated that **upskilling non-genomic health professionals** in genomics could assist, and one suggested that **increasing the training places for clinical geneticists, genetic counsellors and laboratory staff** would assist in addressing workforce shortages. One researcher suggested **better using researchers to evidence the value of implementation**, while a clinician stated that more genetic **clinicians could move into policy roles** to influence change, while researchers could continue to engage with media.

2. Navigating the red tape of the public system

A clinician nominated the red tape in the public system as a major policy, regulatory or legislative barrier to genomic implementation. The clinician cited **approvals processes** that involve **multiple committees, which are not motivated to progress genomic implementation**. This individual advocated for greater independence for local authorities rather than control by centralised bodies. As evidence the individual cited waiting over ten years for a service to be established following funding provision due to obstruction from a decision-making body.

3. A healthcare system that supports implementation across all levels, integrated & holistic care

A national consumer advocate that **overall reform of the health system is needed**. This person advocated for a **holistic approach** to genomic implementation in health care, including **health systems, population and preventative health and precision health care**, indicating the **need for a transition plan**. The individual argued that such a plan needs to **state the problems that genomics is seeking to solve in the health system**. The person stated that there is a large body of evidence showing the frustration of consumers with the pace of implementation, the health system overall and the available support for patients and families.

Discussion

The three barriers with the highest number of nominations in the survey were 1) Equity of access to genomics in healthcare, and 2) Genomic education and training for non-genomic healthcare professionals and 3) Standardised implementation pathways. Many of the free-text comments from participants about why they thought the barriers were important and ways to address them were interdependent, illustrating how barriers do not exist in isolation5 and efforts to address them will be multifaceted.

For example, genomics education for non-genomic healthcare professionals is a way to improve uptake of and access to genomics. Participants recommended education for medical students, for healthcare professionals already practicing and consumers, which included the provision of culturally safe healthcare and embedding genomics champions in regional/remote locations. Some participants suggested genomic competency standards should be required for health professionals to access some genetic tests or provide testing results interpretation and genomics advice to patients, which aligns with overall feedback about standardisation, but such competency standards could create other barriers. Due to the rapid changes in genomics, providing up-to-date education and training curricula will require ongoing effort to ensure new and already practising health professionals are able to deliver contemporary genomics-informed healthcare.

Overall, a recurring theme across all free-text comments was an acknowledgement of inconsistent practices and implementation pathways and a call for standardisation to embed genomics across health care systems. Responses also recognised limitations to access resulting from demographic, cultural, or location-based factors and called for timely translation and implementation of genomic technologies.

Responses about why the barriers were important were generally consistent with existing evidence and understanding of barriers to genomic implementation6 7. For example, previous evidence shows disparity in access to genomics for Aboriginal and Torres Strait Islander people8 and that non-genomic healthcare professionals would like more education opportunities.9

While a lot of the responses were complimentary and aligned with expectations, some responses to the survey lacked clarity and raised some contradictions, highlighting challenges for policy makers when trying to develop solutions. For example, a stakeholders commented that data is siloed, and

[5] Gaff, C.L., M. Winship, I., M. Forrest, S. *et al.* Preparing for genomic medicine: a real world demonstration of health system change. *npj Genomic Med* **2**, 16 (2017). https://doi.org/10.1038/s41525-017-0017-4

[6] Friedrich B, Vindrola-Padros C, Lucassen AM, Patch C, Clarke A, Lakhanpaul M and Lewis C (2024) "A very big challenge": a qualitative study to explore the early barriers and enablers to implementing a national genomic medicine service in England. *Front. Genet.* 14:1282034. doi: 10.3389/fgene.2023.1282034

[7] Alarcón Garavito, G. A., Moniz, T., Déom, N., Redin, F., Pichini, A., & Vindrola-Padros, C. (2023). The implementation of large-scale genomic screening or diagnostic programmes: A rapid evidence review. *European journal of human genetics : EJHG*, *31*(3), 282–295. https://doi.org/10.1038/s41431-022-01259-8

[8] Luke, J., Dalach, P., Tuer, L., Savarirayan, R., Ferdinand, A., McGaughran, J., Kowal, E., Massey, L., Garvey, G., Dawkins, H., Jenkins, M., Paradies, Y., Pearson, G., Stutterd, C. A., Baynam, G., & Kelaher, M. (2022). Investigating disparity in access to Australian clinical genetic health services for Aboriginal and Torres Strait Islander people. *Nature communications*, *13*(1), 4966. https://doi.org/10.1038/s41467-022-32707-0

[9] Nisselle, A., King, E. A., McClaren, B., Janinski, M., Metcalfe, S., Gaff, C., & Australian Genomics Workforce & Education Working Group (2021). Measuring physician practice, preparedness and preferences for genomic medicine: a national survey. *BMJ open*, *11*(7), e044408. https://doi.org/10.1136/bmjopen-2020-044408

data sharing is limited by strict data protection regulations that can create obstacles, however the data protection regulations exist to ensure the safety and privacy of consumer data and information. This highlights the challenges for policy makers facing a delicate balance of standardising practices to encourage data sharing to improve accuracy and efficiency of genomic health care, while ensuring consumers and their data are not exposed to unnecessary risk, amongst layered national and jurisdictional regulatory frameworks.

The proposals for how to address barriers were wide ranging, indicating that there is never a simple, one-size-fits all solution for complex problems such as genomic implementation10. They also highlight the value in seeking a range of perspectives. Inequity of access to healthcare is a widespread problem across Australia and is not limited to genomics. To support improved equity of access and effective implementation of genomics in healthcare, a strong message from survey responses is that a national, coordinated approach is essential. Key recommendations include establishing nationally consistent clinical practice guidelines and referral criteria across all health professions, supported by streamlined genomic testing and laboratory approval processes. These guidelines should be research-informed and collaboratively developed to ensure they reflect best practice rather than just current practice. Investment in secure, interoperable data infrastructure, including dynamic consent models and national genomic registries, is also critical to facilitate data sharing, clinical care, and research while safeguarding consumer privacy. Many of the recommendations regarding data reflect the Blueprint for a National Approach to Genomic Information Management11, developed by Queensland Genomics, and the following Implementation Recommendations for a National Approach to Genomic Information Management12 developed by Australian Genomics.

A robust education and workforce development strategy is needed, integrating genomics into undergraduate, postgraduate, and continuous professional development for all health professionals, alongside cultural safety training. Culturally diverse and priority populations must be actively involved in the design, governance, and delivery of genomic services, with tailored resources, engagement strategies, and support mechanisms. Implementation planning must be underpinned by appropriate funding models—both public and private—that address system-wide needs, including service delivery innovation (e.g. mainstreaming, rural access solutions), increased training capacity, and sustainable workforce growth. An appropriately resourced and planned workforce development strategy requires making forward projections of workforce need and ensuring training and education places are matched to achieve these needs. Finally, stronger policy leadership, legislative reform (e.g. on genetic discrimination), and a shift toward nationally aligned service models will ensure genomics is accessible, trusted, and contributes to improved health outcomes for all Australians.

Limitations

The survey results should be interpreted while keeping in mind limitations in the breadth and depth of responses and the survey design. There were no responses from the Northern Territory and responses from Queensland were underrepresented given the size of the state's population and

[10]Gaff, C. L., M Winship, I., M Forrest, S., P Hansen, D., Clark, J., M Waring, P., South, M., & H Sinclair, A. (2017). Preparing for genomic medicine: a real world demonstration of health system change. *NPJ genomic medicine*, *2*, 16. https://doi.org/10.1038/s41525-017-0017-4

[11] NAGIM-Blueprint-v20201010-Final-v1.2.2.pdf

[12] https://www.australiangenomics.org.au/projects/progress-the-implementation-of-the-nagim-blueprint/

genomic workforce. Further investigation of stakeholder views from these jurisdictions is required before any aggregate conclusions could be drawn about where there are any consistent stakeholder experiences or perspectives at a national level. There was slight over-representation from Victoria and New South Wales, although more broadly, the proportion of responses from jurisdictions other than the Northern Territory and Queensland were reflective of population. The survey did not ask clinicians to differentiate between clinical geneticists, genetic counsellors or other roles, and therefore we cannot determine if there is significant difference in opinion and experience among types of clinicians from these results.

Furthermore, some professional groups were arguably underrepresented in the survey results, such as policy professionals, consumer advocates or laboratory workers/scientists. Therefore, we cannot draw any strong conclusions about profession-specific perspectives from these groups with limited data. Of the consumer advocates who responded, all but one operated at a national level. Further engagement with consumer advocates at a state or territory level is needed to better understand consumer perspectives.

Some respondents started the survey and selected their top three barriers, but did not complete the survey, suggesting the survey may have been longer than expected or included difficult questions.

Of those who articulated their own barriers, half of those respondents were from New South Wales. It is unclear to what extent this reflects a significantly different state-specific experience of genomic implementation.

Conclusion

The implementation of genomics into Australian healthcare is an ongoing challenge, due to the rapid changes in genomic technology and the complexity of Australia's multijurisdictional funding, policy and regulatory frameworks governing our health systems. This survey has provided a point-in-time snapshot of stakeholder perspectives on the policy, regulatory and legislative barriers to genomics implementation. Genomics Australia, to be established in July 2025, 'will provide national leadership and coordination on health genomics to improve the health and wellbeing of Australians'13 and is perfectly poised to address the barriers described by the survey's respondents. Almost all barriers and solutions raised in the survey had national implications and forging agreement between the Commonwealth and the states and territories on proposed actions will be a critical role for Genomics Australia.

Despite limitations, the results from this survey can be used to inform other work and as a platform from which to conduct more detailed investigations. Specifically, further work is required to understand stakeholder perspectives in the Northern Territory and Queensland and among certain professional groups (such as policy makers), as well as consumers. The survey asked for evidence about the impact of selected barriers and many answers provided anecdotal evidence with limited detail. Likewise, suggested solutions were often broad-based and lacking detail. Future stakeholder

[13] https://www.health.gov.au/our-work/establishing-genomics-australia

engagement may wish to delve deeper into the wealth of experience among stakeholders to ensure policy level solutions reflect the experience of those delivering and receiving genomic healthcare.

Longer term efforts to address policy, regulatory and legislative barriers to implementation should continue engagement with stakeholders to navigate an evolving landscape. Responses also seemed to indicate, however, that while service providers have a detailed understanding of the impact of these barriers, some of those delivering genomics healthcare may have limited knowledge of the policy, legislative and regulatory systems underlying service delivery, and efforts to address the barriers. It will be crucial to improve the communication and bridge gaps between policy makers, researchers and service providers to ensure implementation proposals and solutions are fit for purpose, meaningful and are adopted.

Furthermore, many respondents indicated that insufficient funding underlies many of the barriers raised. Even if policies are developed to address gaps, and are supported by legislation and regulatory processes, actions which in themselves require resourcing, the authors acknowledge that genomics implementation will not progress without increased funding for staffing, education and training, testing and equipment.

The Health Technology and Genomics Collaboration is currently coordinating a project reference group to update the National Health Genomics Policy Framework, with a public consultation planned for late June 2025. Encouraging wide dissemination of the public consultation opportunity will help to ensure the updated Framework reflects current issues identified by stakeholders surveyed here. The results of this survey demonstrate that many of the priorities from the 2018-2021 Framework - person-centred approach, workforce, financing, services, and data - are still highly relevant to addressing barriers to integration of genomics in 2025. The establishment of Genomics Australia and refresh of the National Health Genomics Policy Framework are timely opportunities to continue understanding current policy barriers and work to address these challenges from a national perspective.

Appendix 1

Text of survey hosted on REDCap:

Stakeholder Survey: Policy, Regulatory and Legislative Barriers to Genomic Implementation

In 2024-25, Australian Genomics is undertaking a project on the **policy, regulatory and legislative barriers to genomics implementation** in Australian healthcare.

In recent years, genomic technologies, including diagnostic testing and precision health care techniques, have advanced far more rapidly than the ability of Australia's health care systems to adopt these technologies into routine clinical care.

The policy and legislative frameworks supporting genomic health care across Australia are overlapping, complex, and inconsistent. There are multiple policy and regulatory barriers that prevent the effective and equitable implementation of and access to these technologies even when resourcing is available. Policy gaps can also prevent critical resourcing decisions from occurring.

This project seeks to inform policy makers of expert stakeholder views on these barriers and gather existing evidence of the impact, interdependencies and proposed solutions to support decision making and prioritisation.

We are seeking input from clinicians, consumer advocates, researchers and policy makers to ensure we obtain a broad view of these barriers and their impacts. In your professional capacity as an expert stakeholder in your field, we are inviting you to complete the following short survey. Completing the survey should take about **10 - 15 minutes.**

Survey findings will inform the next stage of stakeholder engagement later this year. Responses will remain **anonymous** and aggregate findings and analysis will be included in the project report. The report may be publicly available on the Australian Genomics website and circulated to stakeholders.

If you have any questions or concerns relating to this survey or the project, please contact australian.genomics@mcri.edu.au.

We greatly appreciate your time and input.

Questions

1. Please select which of the following represents your field of work (Compulsory)
If you work across several fields, please nominate the field that most closely aligns with your primary responsibilities.

- Research
  (select all that apply)
  - Laboratory or translational (such as data analysis or research coordination)
  - Clinical redesign and implementation science

    - Psychological or social research
    - Policy or regulatory research
    - Other (please specify)

- Clinical
    - Public
    - Private
    - Both

- Laboratory
    - Public
    - Private
    - Both

- Policy
  (This refers to individuals employed in the public service in any jurisdiction in roles related to policy or legislative development or reform, or planning, in health-related portfolios affected by genomics implementation)

- Consumer advocate
    - In a paid capacity
    - In an unpaid capacity

- Industry advocate

- Health professional representative body

- Other (please specify)

2. In which jurisdiction do you work or which jurisdiction is most relevant to the functions of your work? (Compulsory)

- Queensland
- New South Wales
- Victoria
- Western Australia
- Tasmania
- South Australia
- Northern Territory
- ACT
- National

3. Please select what you consider to be **the three most significant** policy, regulatory or legislative barriers to genomic implementation, or add others you believe are more significant (compulsory). *These barriers may include an absence of appropriate policies or legal frameworks, or there may be existing policies and legal frameworks that require reform.*

You will be able to provide more information on your selections on the following pages.

- Public engagement in genomic implementation
- Culturally safe healthcare
- Equity of access to genomics in healthcare
- Genetic discrimination
- Privacy, data sovereignty and security
- Consistent clinical practice guidelines
- Consistent referral/testing criteria
- Standardised training/accreditation requirements
- Genomic education and training for non-genomic healthcare workers
- Research ethics and governance processes or efficiency
- Processes for technology assessment
- Standardised implementation pathways
- Data collection and management
- Data sharing
- Data infrastructure or storage requirements
- Other (please specify)
- Other
- Other

(In Redcap, each barrier selected was then displayed on a page with the following questions)

(Barrier 1)

a) Please provide a brief description of why you think this is a significant barrier.

Free text

b) What do you think needs to be done to address this barrier?

Acknowledging that resourcing (e.g. funding) is often a barrier, please provide suggestions specifically relating to policy, regulation or legislation and specify if they are required at the State/Territory or Commonwealth level.

Free text

c) Do you know of any evidence relating to the significance or impact of this barrier on genomics implementation? Please take a few moments to provide a brief reference or description below.

(This may be an example from your professional practice, a report or research publication).

Free text

d) Do you know of any projects that are underway that investigate and/or seek to address the barrier you have identified?

Free text

Is there anything else that you would like to comment on relating to this topic?

Free text

9. Would you be willing to discuss this topic in more detail in a brief interview? (Compulsory)

- Yes (please provide contact email).
- No.

Thank you for completing the survey. **Please feel free to share the survey link with relevant professionals in your network that may be interested in participating.**

Appendix 2

Respondents were able to specify any combination of their top three policy, legislative and regulatory barriers from those listed examples in the survey and/or their own. The 'other' barriers specified are listed below.

Table 1. Other barriers specified by respondents.

Other barrier #	Other barriers specified (free text)
1	Safe/adequate staffing levels for genetic services
2	Inadequate funding
3	Appetite from Governments
4	Navigating the red tapes of public system is the biggest barrier
5	Medicare item numbers
6	Genetics not viewed as an essential part of life-long preventative health care at a state or federal level
7	Ordering genetics testing
8	Adequate, up to date, and responsive funding models for genetic/genomic tests and services (like genetic counsellors)
9	Lack of funding for genetics services
10	Funding of genomics clinics/genomic staff members in non-genomics clinics.
11	Genetics services cannot meet demands
12	Federal and state inconsistencies
13	Support requirements outside of the clinical setting
14	Workforce restraints leading to equity of access
15	Capacity/funded capacity to build implementation
16	Lack of engagement from research in policy areas
17	Clinical governance inconsistencies across LHD's.
18	Funding
19	Appropriate staffing for Clinical Genetics Units
20	Government engagement/funding for genomic healthcare
21	A healthcare 'system' that supports implementation across all levels of access, integrated and holistic care and appropriate resourcing to do that
22	Sufficient genomic workforce capacity
23	No MBS item number for genetic counsellors
24	Funding for genomic testing in all areas of health, particularly in adults.
25	Research driven agenda not taking into account health system challenges
26	Poor/inadequate study design to translate into clinical care

Appendix 3

The table below is a high-level summary of free-text survey responses to questions one and two. The barriers in the table are presented according to the number of nominations in the survey. The numbers in brackets represented the number of comments for each theme and the total number of comments for that barrier. Some participants provided comments that fit into more than one theme and not all participants responded to all questions.

Table 2. Summary of survey responses on policy, regulatory and legislative barriers and recommendations

Barrier	Why do you think this a significant barrier?	What do you think needs to be done to address this barrier?
Equity of access to genomics in healthcare	• Access is based on location, included inconsistent or limited access in rural and remote areas (15/40), variable access based on local health district (5/40), or genomics expertise concentrated in certain areas. (4/40) • Access issues for Aboriginal and Torres Strait Islander people, including lack of representation clinically and in research, language or cultural barriers, and being less likely to receive a referral. (9/40) • Access for CALD people, such inaccessible models or challenges navigating healthcare, lack of representation, language barriers, limited health genomics literacy, and culturally safe services. (9/40) • Socioeconomic/social demographic factors made it less likely consumers would get a referral or access care. (7/40) • Existing health inequities can be exacerbated in genomics as raised by three participants. (3/40) • Lack of diversity and sharing of genomic data. (4/40) • Funding issues (11/40), such as different mechanisms or models (e.g. between private and public) (5/40) and	• Funding based suggestions (27/40) such as increased funding or investment (14/40) or changes to existing funding mechanisms. (13/40) • Improve access to for culturally diverse, Indigenous, and priority populations (20/40) via targeting diverse communities through research and engagement. (14/40) • Engage representative groups to understand their needs, identify solutions and ensure involvement in decision making. (10/40) • Improve equitable research participation for diverse populations. (4/40) • Develop culturally appropriate resources. (6/40) • Improve rural and remote access (10/40) via digital solutions (4/40) or through policy, infrastructure, workforce, centres of excellence, and education to support genomic champions in rural areas. (6/40) • Increase in workforce capacity and capability. (4/40) • Deliver education and training for non-genomic health professionals (13/40), specifically for non-genetics health professionals (8/40) or through training or scholarships. (4/40) • Improve the broader community's genomic health literacy. (3/40)

	a lack of or variable funding leading to insufficient resourcing. (6/40) • Workforce issues such as a shortage of genomic professionals, including administrative support, or restrictions for where genetic counsellors can work. (8/40) • Long wait times, increasing referrals and growing demand for genomics services. (8/40) • Inadequate infrastructure. (2/40) • Prioritisation of testing exacerbating already long wait times. (2/40) • Lack of time, skills, or motivation for non-genomic health professionals to provide genomic health care. (8/40) • Lack of genomics education for non-genomics health professionals. (3/40) • Inconsistent referral pathways and practices. (4/40) • Bias or a clinician's training, or beliefs affects care. (2/40) • Lack of standardised approaches to genomic healthcare and variation in availability of genomic testing. (4/40) • Lack of flexibility and translatability (to public patients) regarding MBS items and criteria. (4/40) • Affordability of testing leading to out-of-pocket costs for many. (7/40)	• Improve access to genomic testing. (11/40) • Make genomics available at the primary or community health care level. (5/40) • Increase mainstreaming. (3/40) • Provide alternative pathways to access genetic healthcare/testing outside tertiary hospitals. (2/40) • Standardise the provision of genomic healthcare (12/40), via consistent referral pathways and clinical guidelines (6/40) • Coordinate the approach to policy and collaborations and sharing of resources across Australia to prevent duplication. (3/40) • Establish a consistent approach to genomic testing and its availability. (3/40) • Coordinated policy or government-focussed solutions to address inequities (4/40) • Ensure data sovereignty and accurate data collection regarding demand. (3/40) • Developing infrastructure to enable system-wide linkage of genomics programs. (1/40) • Incorporate equity as a core principle in genomic research and healthcare. (1/40)
Genomic education and training for non-genomic healthcare workers	• Non-genomic healthcare workers (NGHWs) generally do not have genomic skills, knowledge and confidence. (12/26) • NGHWs are needed to mainstream genomics throughout the health system. (6/26) • Insufficient genomics education and training for both existing and new NGHWs. (10/26) • Lack of education and training of NGHPs restricts patient access. (4/26) • Culture of 'gate-keeping' holding back the implementation of genomics among NGHPs. (1/26)	• Genomics education and training should be provided to or required for all health professions. (11/26) • Genomics education and training should be standardised across institutions with required competencies. (7/26) • Develop a government strategy to integrate genomics across the health system. (1/26) • Set-up a national genomic workforce review to understand gaps and future needs. (1/26) • Establish a national public awareness campaign. (1/26)

	• NGHPs should have to be credentialled before being able to order testing. (1/26) • Shortage of qualified genomic health workers. (1/26) • Governments, professional bodies nor educational institutions have taken responsibility for coordinating or establishing standardised genomic education and training for NGHWs. (1/26) • Stresses placed on clinical genetics services, citing long wait times for patients, due to lack of genomics expertise among NGHWs. (1/26)	• Increase investment in the genomics health professional workforce. (2/26) • Increase resources to support genomic health professionals in educating NGHWs. (1/26) • Provide opportunity for NGHWs to be credentialled for order genetic testing. (1/26) • Engage NGHWs in planning for genomics implementation. (1/26) • Provide good quality, centralised information resources for practising NGHPs. (2/26) • De-couple genetic counsellors from clinical geneticists and embed in other specialties. (1/26) • Allow genetic counsellors to work in primary healthcare. (1/26) • Embed genomic health professionals into all multi-disciplinary teams in hospitals. (1/26)
Standardised implementation pathways	• Implementation mechanisms that do exist for evidence-based innovations are lacking. (4/9) • No clear pathways for implementation. (2/9) • Existing processes are not always suitable for genomic innovations (i.e. MSAC/PBAC). (2/9) • Implementation of innovations for adults is lacking when compared with children. (1/9) • Research needs to demonstrate the cost effectiveness as well as clinical utility. (1/9) • Researchers lack understanding of pathways to implementation. (1/9)	• Collaboratively establish nationally consistent implementation pathways, and that is accordingly resourced. (3/9) • Political leadership is required to establish implementation pathways. (1/9) • Strategic planning for implementation. (1/9) • Recognition and funding for clinicians participating in research and acknowledged in workloads. (1/9) • A single health technology assessment process. (1/9) • Clinical genomics services should be state-wide services to support equity and workforce planning. (1/9) • Improve researchers' understanding of implementation pathways. (1/9) • Implementation includes implementation of clinical practice guidelines and models of care across specialties. (1/9)

Consistent clinical practice guidelines	• Lack of consistent clinical practice guidelines in public genetic testing results. (5/10) • Inconsistent management advice provided patients with increased genetic risk of cancer. (1/10) • Non-genomic health professionals do not know when or how to refer patients for genetic testing, nor how to interpret results. (2/10) • Lack of guidelines hampers implementation and mainstreaming of genomics throughout the health system. (1/10) • Significant time required to obtain approvals for and implement innovations in laboratory settings. (1/10)	• Establish nationally consistent clinical practice guidelines for genomics across all health professions. (6/10) • Simplify and streamline genetic testing processes. (1/10) • Laboratory approvals processes should be simplified. (1/10) • Consistent guidelines could inform policy, infrastructure and workforce development. (1/10) • Guidelines should be collaboratively developed through research and be evidence based. (1/10) • Develop decision-aids for patients and clinicians, to assist in management decisions. (1/10)
Consistent referral/testing criteria	• Impacts equity of access to genetic testing. (6/9) • Creates uncertainty for practising clinicians, which impacts patient care. (2/9) • Choice of genetic testing may not be evidence based. (1/9)	• Establish consistent referral and testing criteria. (6/7) • Criteria should be consistent nationally, with some flexibility on a state-by-state basis. (1/7) • Improved resources should be provided to both clinicians and consumers about testing, funding and cost options. (2/7) • Clinical genetics services need consistent criteria to triage referrals and manage waitlists. (1/7) • Improved education for health professionals about the utility of different types of genetic tests. (1/7)
Public engagement in genomic implementation	• Low understanding of genomics across the community. (5/8) • Lack of public knowledge of genomics worsens equity of access for some communities due to low public engagement. (1/8) • Low public engagement leads to insufficient public investment. (1/8) • Lack of public engagement increases the potential harm to consumers. (1/8)	• Develop a public engagement or awareness campaign. (5/8) • Increase genomic education. (2/8) • Engage a diversity of consumers in implementation. (1/8) • Improve transparency on how genomic data is collected, stored and used and ensure consumer privacy and data security is protected to building trust with the public. (1/8)

	• Some communities affected by genetic conditions are overburdened by engagement requests. (1/8)	• Coordinated consumer engagement between specific overburdened groups. (1/8)
Privacy, data sovereignty and security	• Sensitive nature, familial implications and potential for reidentification of genomic data. (1/6) • Patients want data security and to feel comfortable consenting to data usage without fear of adverse consequences. (2/6) • Patients are asking complicated questions about data access and storage or asking for access to their raw data. (1/6) • Awareness of how genomic data can be used to discriminate in terms of life insurance underwriting. (1/6) • Indigenous data sovereignty (IDSov) principles are not embedded in healthcare or research. (1/6) • Problems with individual interpretation of policy/legislation rather than the policy/legislation itself. (1/6) • The current systems protect data based on personal risk. (1/6) • Inconsistencies in data privacy and security management across jurisdictions. (1/6)	• Set-up a national, secure storage-system/facility. (2/6) • National storage should include a dynamic consent option for both research and diagnostic purposes. (1/6) • Set-up a national registry and a national data management and sharing platform. (1/6) • Implement legislation. (3/6) • Review existing data privacy and security legislation.(1/6) • Provide education around data legislation and policy, including appropriate data sharing. (1/6) • Genetic non-discrimination legislation with oversight at the Commonwealth level. (1/6) • IDSov principles should be respected in research and clinical settings. (1/6) • De-risk decisions in terms of personal liability. (1/6)
Data sharing	• Data is siloed within various organisations. (5/5) • Siloed data makes it difficult to share and access. (1/5) • Genetic testing often relies on previous evidence, and siloed data can impact analysis. (2/5) • Data, information, and interpretation is often never looked at after the initial test. (1/5) • Limited potential for large and diverse datasets. (1/5)	• Establish secure national data storage infrastructure. (4/5) • Set-up global genomic databases. (1/5) • Fostering public-private partnerships to create open-source data systems. (1/5) • Establish a national registry for people with a genetic variant. (1/5) • Establish national and international standards and agreements for the collection, storage and sharing of genomic data. (1/5) • Develop robust systems for securing genomic data including encryption, storage and access. (1/5)

	• Significant concerns about patient privacy, data security and potential misuse of genetic information. (1/5) • Inconsistent standards for collection, storage and sharing of genomic data. (1/5) • Data sharing is limited by strict data protection regulations that can create legal or bureaucratic obstacles. (1/5) • Lack of data available or collected with regards to people with cancer risk undertaking preventative surgery. (1/5)	• Simplify regulatory processes that can delay or inhibit genomic data sharing. (1/5) • Implement user-friendly consent processes. (1/5) • Foster public trust by engaging in discussions about benefits and safeguards regarding data sharing. (1/5)
Genetic discrimination	• Concern regarding insurance discrimination. (3/8) • Impacts genetic testing and research participation. (3/8) • The public remains unsure whether knowing one's genetic status is beneficial or not. (1/8) • Social 'stigma' associated with genetic disorders. (1/8) • Patients who are already unsure about genetic testing can use potential insurance discrimination as a reason to decline. (1/8)	• Pass legislation, referring to the Australian Government's commitment to legislate a ban on genetic discrimination in insurance. (5/8) • Disseminate information to both the public and health professionals about the ban on genetic discrimination with regards to insurance. (2/8) • Enforce the genetic discrimination legislation when passed. (1/8) • Insurance companies need to be more transparent about the processes they use to make assessments. (1/8)
Culturally safe healthcare	• Lack of trust in researchers, health providers or science among some communities because of either past or current racially discrimination. (3/5) • Lack of trust is exacerbated by a lack of culturally safe healthcare. (1/5) • Most genomic datasets are based on individuals of European descent. (1/5) • Most research on consumer views and acceptability on genomics in healthcare is mostly based on 'white' or European descent communities, skewing service delivery. (1/5)	• Provide education and training on culturally safe practices to researchers and healthcare professionals. (3/4) • Establish of national standard for cultural safety in healthcare. (1/4) • Include individuals from affected cultural communities directly in governance and decision-making around genomics implementation. (2/4) • Improve education and resources for culturally diverse communities. (2/4) • Embed cultural safety in all healthcare systems and processes, including referral, access and treatment pathways. (1/4) • Establish state and federal policies on culturally appropriate community

		engagement in genomics implementation. (1/4)
Processes for technology assessment	• MSAC processes are slow or do not lead to support for genetic tests and MBS items are not clear leading to inappropriate testing. (2/2) • Due to TGA oversight of some companion diagnostics (CDx) assays and validation criteria, access to CDx and cognate therapies may not be accessible in Australia. (1/2)	• Advocacy from organisations such as HGSA or Australian Genomics to assist in advising MSAC of problems with genomic MBS item numbers. (1/2) • Increased resourcing for MSAC. (1/2) • The TGA should take a more realistic approach to oversight of some companion diagnostics. (1/2)
Data infrastructure or storage requirements	• Issues with current data storage, such as date being external or in cold storage. (3/5) • Challenges addressing storage requirements and capacity, such as cloud storage. (3/5) • Significant amount of work required to meet IT and storage requirements. (1/5) • Software requirements are not supported by qualified professionals. (1/5) • Challenges to share data and advance research without effective storage. (1/5) • Fear of genomic data being compromised. (1/5)	• Improved storage solutions such as organised storage of genomic data, international cloud storage, and national infrastructure allowing ongoing use of data for clinical and research purposes. (3/5) • Need for government/federal infrastructure funding. (2/5) • Overcome jurisdictional barriers with mandated support from qualified bioinformaticians/IT specialists at each laboratory. (1/5)
Research ethics and governance requirements	• Complex, inefficient and lengthy ethics and governance process that lacking jurisdictional harmonisation. (2/4) • Inability of ethics and governance processes to keep up with the rapidly evolving space of genomics. (2/4) • Lack of genomics expertise in the HRECs assessing genomic research applications. (2/4)	• A unified national approach and a well-resourced single national ethics and governance entity (2/4) • Education of ethics, governance and data governance officers in genomics research and underserviced populations. (2/4)
Data collection and management	• Data is siloed within jurisdictions, services, or institutions. 2/3 • Exponential growth in the amount of genomic data being generated while	• Improve sharing of data in Australia to address this barrier (1/3) • Establish patient registries to capture data about hereditary cancer. (1/3)

	genetic testing is often reliant on previous evidence. (1/3) • no central registry in Australia for germline variants to capture follow-up, risk management, and issues accessing testing. (1/3) • No difference in MBS item numbers allowing a record for risk management or treatment decision for cancer patients. (1/3)	• Updating the granularity of MBS item numbers to capture risk management and treatment. (1/3)
Standardised training/accreditation requirements	• NPAAC supervision guidelines have shifted power for operational decisions from scientists to inexperienced laboratory clinicians in diagnostic laboratory settings. (1/3) • Inefficiencies and a lack of genetics knowledge within national bodies (e.g. NATA, MSAC and TGA). (1/3) • Dual clinical training pathways are lengthy and complex decreasing genetics knowledge and representation. (1/3)	• Appropriately qualified clinical scientists should oversee technical and analytical decision-making in the laboratory. (1/3) • Provide supported training for scientists to fill knowledge gaps. (1/3) • Call for a royal commission into NATA, MSAC and TGA. (1/3) • Allow sub-specialty pathways for genetics, via fellowship. (1/3)

Appendix 4

Responses to questions 3 and 4 from the survey are combined and summarised in the table below. The information has been edited to preserve the anonymity of respondents and only includes specific examples of publicly available evidence provided by respondents. Numerous respondents listed their own professional experiences as evidence, but this information has not been included.

Table 3. Combined, summarised and anonymised responses to survey questions 3 (*Do you know of any evidence relating to the significance or impact of this barrier to genomics?*) and question 4 (*Do you know of any projects underway that investigate and/or seek to address the barrier?*)

Barrier	Summary of Responses
Public engagement in genomic implementation	Evidence of Impact: Magic Study; MAGENTA study. Current projects: All-of-US; SAHMRI; Involve Australia; Australian Alliance for Indigenous Genomics; Genetic Health Western Australia Service Plan; Consortium for National Indigenous Genomics Capacity.
Culturally safe healthcare	Evidence of Impact:

	• Australian Department of Home Affairs (2024). Multicultural Framework Review 'Towards fairness: a multicultural Australia for all'. Commonwealth of Australia. Towards Fairness • Croy, S., Ambegaokar, M., & MacArthur, D. (2022). Towards an Inclusive Genomics (1.0). Zenodo. https://doi.org/10.5281/zenodo.6070378 Current projects: ALIGN; GHWA; CONNECTS; Our DNA Program at the Centre for Population Genomics; NCIG; VACCHO; NACCHO
Equity of access to genomics in healthcare	Evidence of Impact: Reports: • 2024 HGSA Clinical Genetic Services Framework, • HGSA Victorian Genetic Counsellor Enterprise Agreement • Working Group Victorian Genetic Counsellor Survey • Accelerating genomic medicine in the NHS: A strategy for embedding genomics in the NHS over the next 5 years (2022) https://www.england.nhs.uk/long-read/accelerating-genomic-medicine-in-the-nhs/ Literature: • Croy, S., Ambegaokar, M., & MacArthur, D. (2022). Towards an Inclusive Genomics (1.0). Zenodo. https://doi.org/10.5281/zenodo.6070378 • Caron et al (2020). Indigenous Genomic Databases: Pragmatic Consideration and Cultural Contexts. *Front. Public Health* **8**. https://www.frontiersin.org/articles/10.3389/fpubh.2020.00111/full • Lewis, D.A., Mitchell, T. & Kowal, E. (2024). Precision medicine in Australia: Indigenous health professionals are needed to improve equity for Aboriginal and Torres Strait Islanders. *Int J Equity Health* **23**, 134. https://doi.org/10.1186/s12939-024-02202-7 • Kanga-Parabia et al (2024). Genetic counselling workforce diversity, inclusion and capacity in Australia and New Zealand. *Genetics in Medicine Open* 2(4). https://www.sciencedirect.com/science/article/pii/S2949774424009944 • Alarcón Garavito, G.A., Moniz, T., Déom, N. *et al.* The implementation of large-scale genomic screening or diagnostic programmes: A rapid evidence review. *Eur J Hum Genet* **31**, 282–295 (2023). https://doi.org/10.1038/s41431-022-01259-8 • Long et al. (2021). The long and winding road: perspectives of people and parents of children with mitochondrial conditions negotiating management after diagnosis. *Orphanet J Rare Dis*. **16**(1). doi: 10.1186/s13023-021-01939-6. PMID: 34256797; PMCID: PMC8276535.

	• Friedrich et al. (2024). "A very big challenge": a qualitative study to explore the early barriers and enablers to implementing a national genomic medicine service in England. *Front. Genet.* **14**. https://www.frontiersin.org/journals/genetics/articles/10.3389/fgene.2023.1282034/full#B18 • White, S., Jacobs, C. & Phillips, J. Mainstreaming genetics and genomics: a systematic review of the barriers and facilitators for nurses and physicians in secondary and tertiary care. *Genet Med* **22**, 1149–1155 (2020). https://doi.org/10.1038/s41436-020-0785-6 • Gleeson et al. (2020). The development and evaluation of a nationwide training program for oncology health professionals in the provision of genetic testing for ovarian cancer patients. *Gynecologic Oncology* **158**(2): 431-439. Current projects: OurDNA Program, ALIGN, NCIG, NACCHO, VACCHO, Australian Genomics projects, NSW Genomics Strategy, PEACH, DNA Screen, Genomic Literacy Network (Australia), HGSA census, WA Genomics Strategy 2022-2032, Melbourne Uni priority placements for Indigenous MGC/MGH students: https://medicine.unimelb.edu.au/school-structure/paediatrics/news-and-events/MGC-and-MGH-indigenous-scholarship
Genetic discrimination	Evidence of impact: • Tiller et.al. (2023). Final stakeholder report of the Australian Geneitics and Life Insurance Moratorium: Monitoring the Effectiveness and Response (A-GLIMMER) Project. Final Stakeholder Report of the Australian Genetics and Life Insurance Moratorium: Monitoring the Effectiveness and Response (A-GLIMMER) Project.
Privacy, data sovereignty and security	Current projects: National Approach to Genomics Information Management (NAGIM); National Cancer Plan for Genomics
Consistent clinical practice guidelines	Evidence of impact: • Sachdev et al. 2021, 'Paediatric genomic testing: Navigating medicare rebatable genomic testing', Jounal of Paediatrics and Child Health, DOI: 10.1111/jpc.15382 • Garavito et al. 2023, 'The implementation of large-scale genomic screening or diagnostic programmes: A rapid evidence review', European Journal of Human Genetics, 31: 282-295. • Shen et al. 2022, 'Barriers and Facilitators for Population Genetic Screening in Healthy Populations: A Systematic Review', Frontiers in Genetics, DOI: 10.3389
Consistent referral/testing criteria	Current projects: MRFF 2025 Embedding Genomics in Primary Care; Draft Genetic Health Western Australia Service Plan for 2024-2029.
Standardised training/accredi	

tation requirements	
Genomic education and training for non-genomic healthcare workers	Evidence of impact: • Chen LS, Goodson P. (2009). Barriers to adopting genomics into public health education: a mixed methods study. *Genet Med*. **11**(2). doi: 10.1097/GIM.0b013e31818fa2c7. • Nisselle et al. (2021). Ensuring best practice in genomics education and evaluation: reporting item standards for education and its evaluation in genomics. *Genetics in Medicine*, **23**(7). https://doi.org/10.1038/s41436-021-01140-x. • Dunlop et al. (2025). Building capacity for genomics in primary care: a scoping review of practitioner attitudes, education needs, and enablers. *Frontiers in Medicine* 12. http://dx.doi.org/10.3389/fmed.2025.1577958 Current projects: Genomics Education Network Australasia; Australian Genomics; PRECISE MRFF project; Draft Genomics Health WA Service Plan; Mito Foundation; Inherited Cancers Australia;
Research ethics and governance requirements	Current projects: ALIGN project, National Cancer Genomics Plan
Processes for technology assessment	Evidence of impact: • Health Technology Assessment Policy and Methods Review Report. Health Technology Assessment Policy and Methods Review | Australian Government Department of Health, Disability and Ageing • Norris S, Belcher A, Howard K, Ward RL. (2022). Evaluating genetic and genomic tests for heritable conditions in Australia: lessons learnt from health technology assessments. *J Community Genet*. **13**(5):503-522. doi: 10.1007/s12687-021-00551-2.
Standardised implementation pathways	Evidence of impact: • Mackenzie's Mission; DNA Screen; OMICO; ZERO childhood cancer mission; Genomics autopsy • Stark Z, Ellard S. Rapid genomic testing for critically ill children: time to become standard of care? Eur J Hum Genet. 2022 Feb;30(2):142-149. doi: 10.1038/s41431-021-00990-y.
Data collection and management	Current projects: ICCon (Peter Mac) databases.
Data sharing	Current projects: NAGIM
Data infrastructure or storage requirements	Current projects: NAGIM, Australian Genomics projects.