

Review of the Genomics Health Futures Mission

Genomics Health Futures Mission

6 August 2024



Nous Group acknowledges Aboriginal and Torres Strait Islander peoples as the First Australians and the Traditional Custodians of country throughout Australia. We pay our respect to Elders past, present and emerging, who maintain their culture, country and spiritual connection to the land, sea and community.

This artwork was developed by Marcus Lee Design to reflect Nous Group's Reconciliation Action Plan and our aspirations for respectful and productive engagement with Aboriginal and Torres Strait Islander peoples and communities.

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Table 1 | Glossary

Term or abbreviation	Definition
AGRD	Australian Genome Reference Database.
AMRAB	Australian Medical Research Advisory Board.
ARC	Australian Research Council
Awarding	Commitment to provide grant funding over a certain period made.
Cmwlth	Commonwealth of Australia.
The department	The Department of Health and Aged Care (Australian Government).
Disbursement	Expensing of monies from the MRFF.
DISR	The Department of Industry, Science and Resources, formerly the Department of Industry, Science, Energy and Resources (Australian Government).
EAP	Expert Advisory Panel.
ELSI	Ethical, Legal and Social Implications.
GAC	Grant Assessment Committee.
Genomics	The field of study focused on analysing and understanding an organism's complete set of Deoxyribonucleic acid (DNA).
GHFM	Genomics Health Futures Mission.
GHFM Review	Mid-term review of the Genomics Health Futures Mission, also referred to as 'the Review.'
Grants hubs	The National Health and Medical Research Council and the Department of Industry, Science and Resources each operate a grant hub which centralized operations to deliver grant administration services on behalf of other Australian Government agencies.
Group of Eight	A project whose host institution is a member of the Group of Eight Universities.
HMRO	Health and Medical Research Office, Department of Health and Aged Care.
MBS	Medicare Benefit Schedule.
MRFF	Medical Research Future Fund.
MRFF Evaluation Strategy	The MRFF Learning, Monitoring and Evaluation Strategy 2020-21 to 2023-24.
MRI	Medical Research Institution/s.
GHFM Review Panel	The Genomics Health Futures Mission Review Panel.
MSAC	Medicare Services Advisory Committee.
NHMRC	National Health and Medical Research Council.
Project lead	The chief or lead investigator of research project as defined by the grant agreement.
Translational research	The process of applying ideas, insights and discoveries generated through scientific inquiry to the treatment or prevention of human disease.

1 Executive Summary

Genomics, the field of study focused on analysing and understanding an organism's complete set of Deoxyribonucleic acid (DNA), is dynamic and rapidly evolving with transformative potential in health care. New applications and technologies are continually developed through basic and applied research, receiving significant attention and investment from the public and private sectors both nationally and internationally.

2 Background

The Medical Research Future Fund (MRFF) was established in 2015 by the Australian Government through the *Medical Research Future Fund Act 2015 (Cmwlth)* (MRFF Act). The MRFF aims to:

"Transform health and medical research and innovation to improve lives, build the economy and contribute to health system sustainability."

The MRFF provides an ongoing funding stream for medical research and innovation through a \$20 billion endowment to fund research projects via 21 initiatives. Progress towards the aim of the MRFF is articulated through the MRFF Monitoring, Evaluation and Learning Strategy 2020-21 to 2023-24, which establishes eight measures of success for MRFF initiatives. Since its inception the MRFF has awarded \$2.98 billion across 1,206 research projects (as of 31 December 2023).

The Genomics Health Futures Mission (GHFM) was established by the Australian Government in 2018, with a \$500.1 million commitment over 10 years, making it the MRFF's largest Research Mission. Its goal is to:

"Save or transform the lives of more than 200,000 Australians through genomic research to deliver better testing, diagnosis and treatment".

The GHFM is guided by the GHFM Roadmap, which establishes three aims and 13 priority areas (refer to Table 2, below), and the GHFM Implementation Plan which defines an evaluation approach and implementation considerations for each of the GHFM's aims.

Table 2 | Summary of GHFM aims and priority areas

GHFM Aim	Priority Areas
Aim 1 Faster and more effective disease diagnosis, prevention and earlier intervention	1.1 Rare disease 1.2 Cancer 1.3 Functional genomics 1.4 Infectious disease 1.5 Genomic screening
Aim 2 New targeted interventions that transform individual and population health	2.1 Pharmacogenomics 2.2 Common and complex disease 2.3 Gene-related therapies 2.4 Co-developing clinical capabilities
Aim 3 Increased community awareness and engagement, and better understanding of the societal and economic value of genomics in health care	3.1 Ethical, Legal, and Social Issues (ELSI) 3.2 Governance and technology 3.3 Aboriginal and/or Torres Strait Islander health 3.4 Australian Genome Reference Database

To achieve its goal the GHFM invests in research projects, and since its commencement via a \$20 million grant to Mackenzie's Mission, the GHFM has invested \$273.20 million in 88 genomics research projects (as at 31 December 2023).¹

The MRFF has also invested \$264.08 million in 82 genomics-related projects through 12 of the remaining 20 MRFF initiatives (refer to **Appendix B**).

3 About the Review

Due to the pace of change in the field of genomics, the landscape has shifted since the establishment of the GHFM. In light of the evolving environment, and with five years remaining for the initiative, a review of the GHFM's progress and impact is timely and appropriate to ensure the GHFM is well placed to reach its goal.

The department has engaged Nous Group (Nous) to prepare a mid-term review of the GHFM (the GHFM Review) with the intent to assess:

- I. How the MRFF has contributed to genomics research in Australia (**Contribution**).
- II. How MRFF-funded genomics research sits within the national and international genomics research funding landscape (**Reputation**).
- III. Alignment and progress of MRFF-funded genomics research (**Alignment and Progress**).
- IV. Opportunities (if any) to enhance MRFF funding and granting arrangements to improve the impact of MRFF funded genomics research (**Opportunities**).

The design and delivery of the GHFM Review was overseen by the department, who were supported by the advice of the Mission Review Panel. The GHFM Review is not an audit or evaluation of the administration of the GHFM, nor is it a scientific review of the value or impact of genomics.

To fulfill the intent of the GHFM Review, Nous, with the support of Australian Genomics, has used four methods of data collection to gather the views of the sector against each component of the intent:

- Grantee Survey
- Stakeholder Consultations
- Desktop Review²
- Document Review.

4 Findings

4.1 The genomics landscape

The GHFM's aims and priority areas are broad and comprehensive, with a focus on diagnosis, prevention and early intervention | There is general consensus from stakeholders from across the genomics research sector that the priorities of the GHFM are comprehensive and capture the breadth of applications and potential impact from genomics. However, the GHFM may benefit from a more targeted approach, further refining priorities to ensure the funding available delivers maximum impact.

Although the GHFM is a large funder it alone cannot fund the entire translation effort; the complexity of the Australian genomics landscape makes it difficult to coordinate cohesive investments with other funders | As the second largest funder of genomics research in Australia and focused on translation of genomics research, the GHFM seeks to complement investments in fundamental or basic genomics

¹ Department of Health and Aged Care, Medical Research Future Fund grant recipients, 31 December 2023. See MRFF initiative, Genomics Health Futures Mission.

² A Desktop Review report was prepared by Australian Genomics which was used as the primary evidence base to assess the **II. Reputation** component of the intent.

research made by the largest (NHMRC) and third largest (ARC) funders. The GHFM achieves this to some extent through greater focus on translational research, as well as its unique investments in cancer and rare disease diagnostics. The complexity and decentralised nature of the landscape makes it difficult to identify areas of unmet need and avoid overlaps, and there is some evidence of duplication of funding priorities, which reduces the complementarity.³

Australia boasts strong genomic research capabilities, but international counterparts have greater funding and broader remit to progress their agenda for advancing genomics | There is a significant focus internationally on integrating new technologies, clinical services advancement, and robust data linkage systems. In contrast, the GHFM primarily funds research projects without directly investing in complementary infrastructure or enablers, a gap highlighted when compared to the systemic prioritisation of genomics infrastructure in other nations' health care strategies.

4.2 Progress made towards the goals of the GHFM and MRFF

Project leads are optimistic about their progress towards outcomes, but are unaware of the successes and learning from other MRFF projects | Despite a minority of projects experiencing significant delays, MRFF projects are, on average, halfway towards completion, based on the progress reported by project leads towards their project milestones. While it is evident that some significant progress has been made by these projects in this time, the broader health sector is not aware of this progress.

Progress to the GHFM's aim to increase genomic diagnoses, preventions and early interventions has been most pronounced, reflecting the volume of funding provided to Aim 1 projects | Progress towards the GHFM aims has been mixed, with the most significant strides made in increasing genomic diagnoses, prevention, and early interventions (Aim 1). Despite fewer projects and a lower overall perception of progress, project leads of GHFM Aim 3 projects were optimistic about their contribution towards increasing community awareness and understanding of genomics, recognising this as a longer-term effort. Stakeholders acknowledge some groundbreaking projects in First Nations genomics but highlighted that projects not specifically researching First Nations genomics were inconsistent in their approach and level of engagement with First Nations communities.

Progress thus far reflects outcomes of early translation activities | Based on self-assessed progress by MRFF project leads, MRFF and GHFM genomics projects have made more progress in early-stage research activities such as increasing the focus on areas of unmet need and enhancing the research community's capacity for translational research. In contrast, less advancement has been observed in the later-stage outcomes, particularly embedding new health technologies into clinical practice and the commercialisation of health research outcomes.

4.3 Contribution of the MRFF to genomics in Australia

The MRFF, primarily through the GHFM, has bolstered and supported a consistent pipeline of activity | The sector views the GHFM as critical to enabling the volume of genomics research as the majority of projects would likely not have proceeded without the GHFM, underscoring the critical role the GHFM and the broader MRFF have played in advancing the field, especially in rare diseases. The consistent year-to-year funding of the GHFM has provided the sector with certainty and has encouraged separate non-governmental investment in genomics buoyed by consistent demand, though specific examples of direct investment in research infrastructure has been limited.

The GHFM is supporting some systemic benefits within the genomics sector, but longer-term and transition funding is required to sustain its impact | While there was optimism surrounding the projects supported by the GHFM, it was noted that the benefits of these projects were highly localised to the areas of the projects. The GHFM has fostered better collaboration within the genomics research community,

³ The establishment of Genomics Australia as an Australian Government entity is expected in 2025 and will impact the Australian genomics landscape.

although there is a need for more partnerships and collaborations with the broader healthcare sector, government agencies, and international collaborators. While there has been some co-investment and growing consumer engagement within genomics projects, efforts in these areas could be further progressed to maximise the impact of research.

The genomics sector is facing complex challenges particularly in overcoming persistent barriers to translation; the GHFM can play a stronger role to drive cohesive sector-wide efforts to overcome them |

The genomics sector in Australia requires stronger national coordination to effectively align research and health system translation, overcoming systemic obstacles such as gaps in the workforce, preparedness of the health system, infrastructure, data management, and addressing ethical, legal, and social implications. Stakeholders suggest the GHFM could better facilitate collaboration and guide genomic research towards nationally harmonised goals. Furthermore, the sector faces challenges transitioning from research to practical, sustainable health system applications, highlighting the necessity for a clear, coordinated investment strategy that includes infrastructure, regulatory understanding, and health system integration to ensure the longevity and impact of genomics projects.

6 Conclusion

The GHFM has played a significant role in the growth in the volume and profile of genomics research in Australia by investing in and securing a consistent pipeline of genomics research.

The goal of the GHFM to “save or transform the lives of more than 200,000 Australians through genomic research to deliver better testing, diagnosis and treatment” is ambitious. A range of structural and environmental factors have thus far slowed the impact of the GHFM. It is critical that the GHFM address these to realise the opportunities to enhance its ability to impact the sector over the remaining five years.

7 Opportunities

The GHFM Review has identified 12 opportunities across three opportunity themes that if addressed will increase the impact of the GHFM over the remaining five years. To realise these opportunities, the GHFM Review poses four strategic considerations for the future of the GHFM.

Opportunity themes

- Refine the investment strategy and associated priorities (five opportunities).
- Strengthen coordination and communication (three opportunities).
- Support collaboration across the sector (four opportunities).

The opportunities are influenced by a range of system and program factors, and will require a range of actions over a period of time to address.

Strategic considerations

The Review has identified four strategic considerations to address the opportunities. The review notes that some of these considerations cannot be achieved by the GHFM alone, and would require the participation of other stakeholders in the genomics landscape.

1. Consolidate priorities and investments strategies, in collaboration with other funders, behind a cohesive strategy.
2. Support genomics projects to overcome challenges to successful, sustainable translation of their project’s outputs.
3. Foster engagement, collaboration and coordination of activities between researchers, consumers and the health system.
4. Consider innovative funding models for future grant opportunities.

2 Background

This chapter provides an overview of genomics, the MRFF and the GHFM.

Genomics, the field of study focused on analysing and understanding an organism's complete set of deoxyribonucleic acid (DNA), is dynamic and rapidly evolving with transformative potential in health care. New applications and technologies are continually developed through basic and applied research, receiving significant attention and investment from the public and private sectors internationally.

2.1 Medical Research Future Fund

The MRFF was established in 2015 by the Australian Government through the *Medical Research Future Fund Act 2015 (Cth)* (MRFF Act). The MRFF aims to:

"Transform health and medical research and innovation to improve lives, build the economy and contribute to health system sustainability."

To achieve its aim, the MRFF acts as an endowment fund whose capital is preserved in perpetuity and earnings invested as grants to research projects and research infrastructure.⁴ MRFF grants provide secure, ongoing funding streams for medical research and innovation aimed at improving the health and wellbeing of Australians, building the economy, and contributing to the health system sustainability.⁵

The MRFF operates within the broader context of Australian Government support for health and medical research, which includes funding through the NHMRC and the Biomedical Translation Fund.⁶

2.1.1 MRFF governance

The MRFF Act sets out roles and responsibilities for:

- The Minister for Finance and the Treasurer, who are both responsible for oversight of the MRFF.
- The Minister for Health and Aged Care, who is responsible for administration of MRFF financial assistance to support medical research and innovation.
- The Australian Medical Research Advisory Board (AMRAB) who are responsible for setting the Australian Medical Research and Innovation Strategy and the Australian Medical Research and Innovation Priorities.

In addition to those mentioned in the MRFF Act, several other stakeholders are involved in the administration of the endowment fund and in the administration of grants from the MRFF (refer to Table 3, overleaf).

⁴ The Australian Government made regular contributions to the endowment fund until it reached \$20 billion in July 2020.

⁵ The MRFF Act defines medical innovation as including: *'the application and commercialisation of medical research for the purpose of improving the health and wellbeing of Australians; and the translation of medical research into new or better ways of improving the health and wellbeing of Australians.'*

⁶ The purpose of the MRFF and the historical approach of NHMRC are intended to be highly complementary. Broad-based NHMRC funding seeks to ensure Australia is actively engaged in research to address our diverse health needs, connected to international developments and ready to respond to emerging health challenges. By investing in priority areas, the MRFF seeks to deepen and build on the foundation of knowledge, capacity and capability established and maintained by NHMRC.

Table 3 | MRFF roles and responsibilities

Stakeholder	Roles and responsibilities
Responsibilities for the endowment fund	
Minister for Finance	<ul style="list-style-type: none"> Accountable to the Parliament for management of the endowment fund. Issue the investment mandate specifying the target rate of return on investments.
Treasurer	<ul style="list-style-type: none"> Credit funds to the MRFF.
Future Fund Board of Guardians	<ul style="list-style-type: none"> Determine the maximum annual distribution. Determine investment strategy for the MRFF endowment fund.
Responsibilities for disbursements from the MRFF	
Minister for Finance	<ul style="list-style-type: none"> Approves disbursements from the MRFF at the request of the Minister for Health and Aged Care.
Minister for Health and Aged Care	<ul style="list-style-type: none"> Accountable to the Parliament for administration of MRFF grants. Determines the allocation of disbursements from the MRFF via grants.
AMRAB	<ul style="list-style-type: none"> Determine the Australian Medical Research and Innovation Strategy and Priorities. Provide advice on matters referred by the Minister for Health and Aged Care.
Expert Advisory Panels	<ul style="list-style-type: none"> Provide advice to the Minister for Health and Aged Care on the strategic priorities for research investment through the MRFF Missions. Develop a Roadmap and Implementation Plan for each MRFF Mission.
Grant Assessment Committees (GAC)	<ul style="list-style-type: none"> Provide independent assessments of applications made in response to grant opportunities advertised under an MRFF initiative.
Department of Health and Aged Care	<ul style="list-style-type: none"> Provide reports and advice to the Minister for Health and Aged Care on MRFF policy. Responsible for the administration of the MRFF. Execute and administer grant agreements. Provide secretariat support to AMRAB and the Expert Advisory Panels.
NHMRC	
Department of Industry, Science and Resources (DISR)	<ul style="list-style-type: none"> Administer MRFF grants and grant review processes on behalf of the department, through their respective grants hubs.

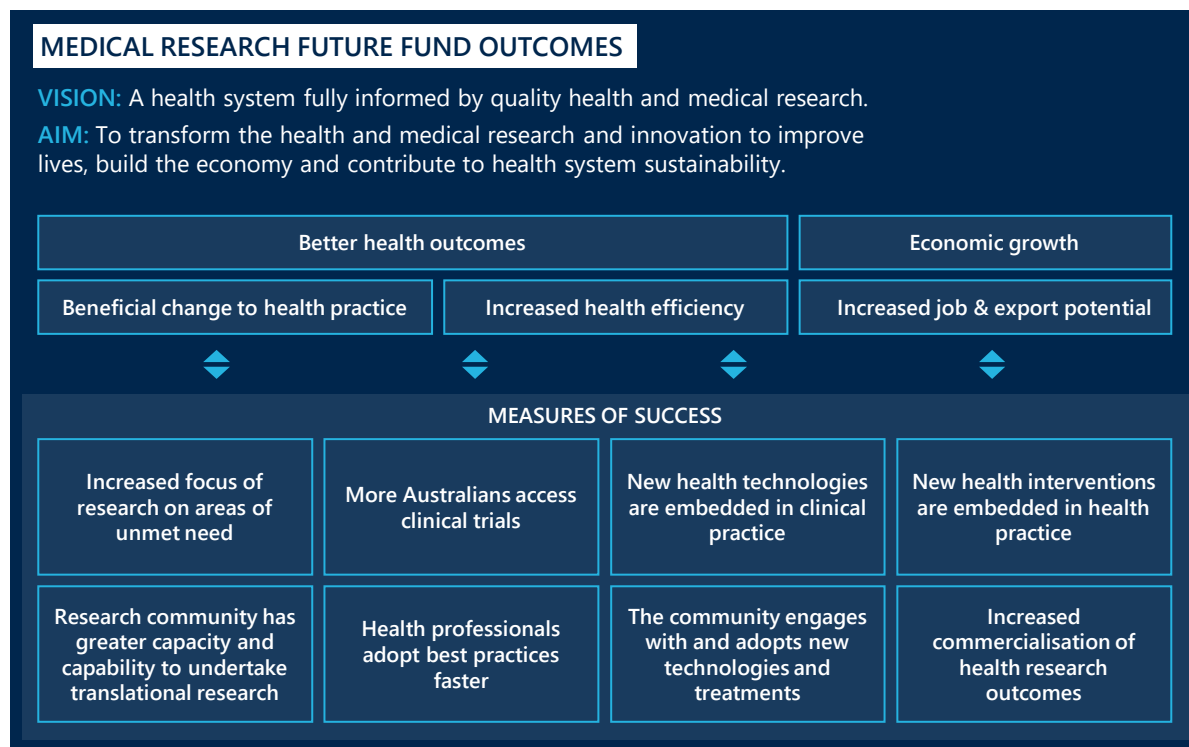
Source: *Medical Research Future Fund Act 2015 (Commonwealth of Australia).*

2.1.2 MRFF evaluation

The Australian Medical and Research Advisory Board first articulated the MRFF’s vision, aim, objectives and impact measures through the Australian Medical Research and Innovation Strategy.

In November 2020, the Australian Government published the [MRFF Monitoring, Evaluation and Learning Strategy 2020–21 to 2023–24 \(MRFF Evaluation Strategy\)](#), which provides an overarching framework for assessing the performance and impact of the MRFF.⁷ The MRFF Evaluation Strategy includes key measures of success, and links them to the achievement of five impact measures (refer to Figure 1, below).

Figure 1 | MRFF monitoring, evaluation and learning conceptual framework



Source: MRFF Monitoring, Evaluation and Learning Strategy 2020–21 to 2023–24.

The MRFF Evaluation Strategy proposes evaluation activities at the level of individual grants, grant opportunities, MRFF initiatives, and the MRFF holistically, with an emphasis on the need for evaluations to be independent, impartial, and transparent, ensuring credibility and objectivity. Additionally, the MRFF Evaluation Strategy states evaluations will be inclusive of consumers and stakeholders, a key feature of the design of the approach to the current GHFM Review.

In March 2023, the department published a set of [performance indicators](#) as part of the implementation of the MRFF Evaluation Strategy. The performance indicators provide a set of quantifiable metrics to help capture the outputs and outcomes from MRFF projects.

2.1.3 MRFF investments

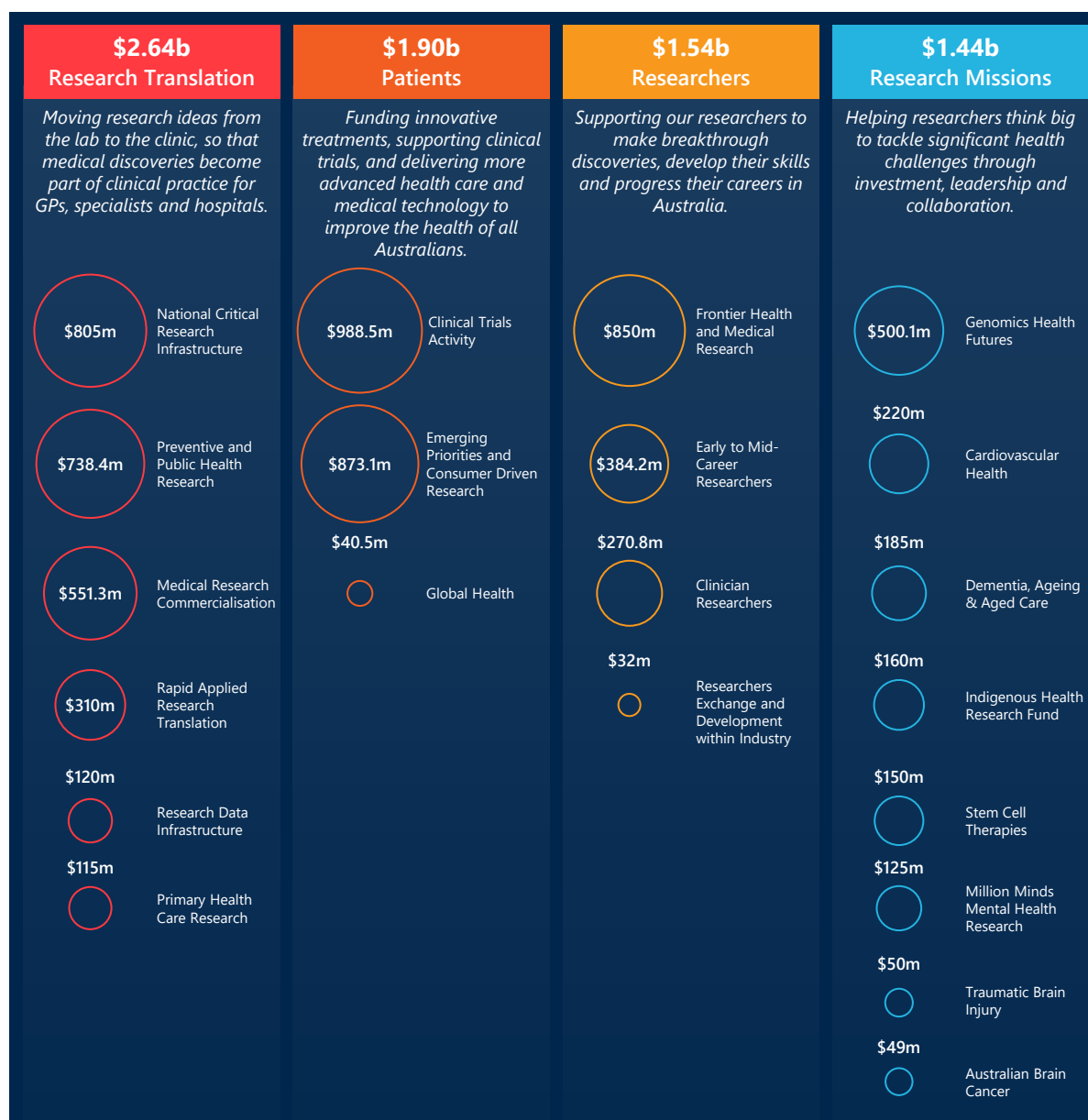
The Australian Government has allocated MRFF funding to research initiatives (which are organised into four funding themes) and typically announces these allocations through the MRFF's [10-year Investment Plan](#) (first published in 2019 and refreshed in September 2022 and May 2024).⁸ As of 31 December 2023,

⁷ 'Impact' is defined as the 'demonstrable contribution that excellent research makes to society and the economy'.

⁸ The Australian Government introduced the [Coronavirus Research Response initiative](#) between 2020 and 2021, that provided \$130 million to 80 projects, was announced as part of the COVID-19 National Health Plan.

the 1st and 2nd 10-year Investment Plans had allocated \$7.52 billion between 2018-19 and 2031-32 to 21 initiatives (refer to Figure 2, overleaf).⁹

Figure 2 | MRFF themes and initiatives



Source: MRFF 10-year Investment Plans, as at 31 December 2023.

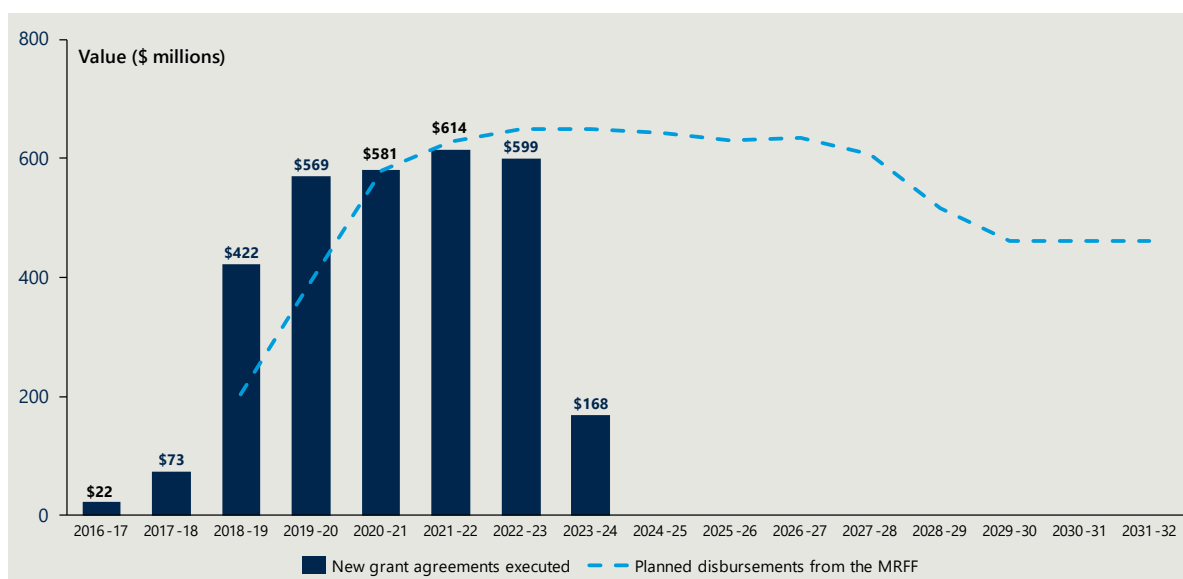
Figure note: The size (area) of the bubble indicates the funding allocated across both the 1st and 2nd 10-year Investment Plans to the initiative (i.e. the total allocation between 2018-19 and 2031-32). The area does not include: \$590.8 million that was allocated to initiatives under the Research Mission theme pending evaluations; and funding that was not allocated to a specific initiative.

Figure note: The figure excludes MRFF's initiatives not published in the MRFF's 1st or 2nd 10-year Investment Plan. Funding announced by the 3rd 10-year Investment Plan, published in May 2024, has not been presented.

As of 31 December 2023, the MRFF has awarded \$2.98 billion (35 per cent of the total commitment until 2031-32) to 1206 research projects (refer to Figure 3, overleaf).

⁹ In May 2024, the department published the 3rd 10-year Investment Plan which announced an additional allocation of \$1.3b, comprising \$194.4 million to establish two new initiatives under the Research Missions theme (the Low Survival Cancers Mission and Reducing Health Inequities Mission) and \$1.1 billion to continue 20 existing initiatives until 2033-34.

Figure 3 | Total Investments through the MRFF



Source: MRFF 10-year Investment Plans and the list of [MRFF grant recipients](#), as at 31 December 2023.
Figure note: Planned disbursements are as per the 1st and 2nd 10-year Investment Plans, and includes all funds allocated to an initiative. Additional funding is available under the 10-year Investment Plans that has not yet been allocated to an initiative.

2.2 Genomics funding through the MRFF

Since the MRFF began investing returns on the endowment fund it has awarded \$537.28 million to 170 genomics projects (as at 31 December 2023).¹⁰ Approximately half of the funds (\$273.20 million to 88 projects) awarded has been administered through the Genomics Health Futures Mission (GHFM) and the rest (\$264.08 million to 82 projects; refer to **Appendix B**) has been administered through 12 of the other 20 MRFF initiatives.

2.2.1 Genomics Health Futures Mission

The Australian Government established the GHFM in 2018, with a commitment of \$500.1 million over 10 years, the largest of the MRFF Research Missions. The GHFM’s goal is to:

“Save or transform the lives of more than 200,000 Australians through genomic research to deliver better testing, diagnosis and treatment”.

To achieve this, the GHFM is designed to fund research to integrate genomics knowledge and technology into clinical practice. By doing so the GHFM aims to:

- Ensure Australians live longer and healthier lives through access to genomics knowledge and technology.
- Position Australia as a global leader in genomics research.
- Deliver improved diagnostics and targeted treatments.
- Avoid unnecessary health costs.
- Improve patient experience and outcomes.

¹⁰ The department finalised the genomics projects within scope of the GHFM Review on 31 October 2023, refer to **Appendix B**.

GHFM governance, strategy and implementation

The administration of the GHFM is guided by the advice of an Expert Advisory Panel (EAP) on the priorities and considerations for the GHFM.¹¹ The EAP primarily provide this advice through the development of [GHFM Roadmap](#) and [GHFM Implementation Plan](#) which serve as foundational documents guiding the integration of genomics into Australian healthcare.¹²

The department develops grant opportunities and delivers other supporting activities to deliver the GHFM and support the priorities identified by the EAP. The department has Memorandums of Understanding with the NHMRC and DISR to administer the grant agreements through their respective Grants Hub. For each grant opportunity the Grants Hub appoints a GAC to assess eligible grant applications against the selection criteria outlined in the grant opportunity Guidelines.

The GHFM Roadmap sets the strategic direction, emphasising innovative research, fostering collaboration, and integrating genomics knowledge into practice. The GHFM Roadmap defines the goal, mission, considerations, funding principles and priorities for the GHFM. The three aims and corresponding priority areas for investment are outlined in Figure 4 (below).

Figure 4 | GHFM Aims and Priority Area for Investment

AIM 1	AIM 2	AIM 3
<p>Faster and more effective disease diagnosis, prevention and earlier intervention.</p>	<p>New targeted interventions that transform individual and population health.</p>	<p>Increased community awareness and engagement, and better understanding of the societal and economic value of genomics in health care.</p>
<p>1.1 Rare disease: Improving diagnostic rates for rare genetic diseases that present before birth, in childhood or in adults, and delivering the diagnosis as quickly as possible.</p> <p>1.2 Cancer: Improving early detection and targeted treatment for the most common cancers to reduce the burden of disease.</p> <p>1.3 Functional genomics: Promoting diagnostic effectiveness and efficiency through better understanding of the impact of genetic variants.</p> <p>1.4 Infectious disease: Developing novel methods to reduce the impact of infectious diseases on individual patients and on population.</p> <p>1.5 Genomic screening: Improving genomic screening to enable informed decision making for health.</p>	<p>2.1 Pharmacogenomics: Promoting precision medicine to improve medication efficacy and reduce harm.</p> <p>2.2 Common and complex disease: Deploying innovative methods to understand the genetic basis of complex diseases.</p> <p>2.3 Gene-related therapies: Developing novel therapeutics by investing in promising early-stage products.</p> <p>2.4 Co-developing clinical capabilities: for genomics applications that can be embedded in the primary health care sector.</p>	<p>3.1 ELSI: Developing a better understanding of the ethical, legal and social implications of genomics, and facilitating public trust and public engagement.</p> <p>3.2 Governance and technology: Developing innovative methods for the ethical and secure governance of genomics data for clinical and research purposes.</p> <p>3.3 Aboriginal and/or Torres Strait Islander health: Ensuring that Aboriginal and/or Torres Strait Islander people contribute to, and control the application of genomics research for, the health benefits to their communities.</p> <p>3.4 Australian Genome Reference Database: Enriching population cohorts to bring the benefits of genomics to all members of our multicultural nation.</p>

Source: GHFM Roadmap.

¹¹ The first EAP was appointed by the Minister for Health and Aged Care in 2019 and ceased on 30 June 2021. The second EAP was appointed by the Minister for Health and Aged Care in December 2023.

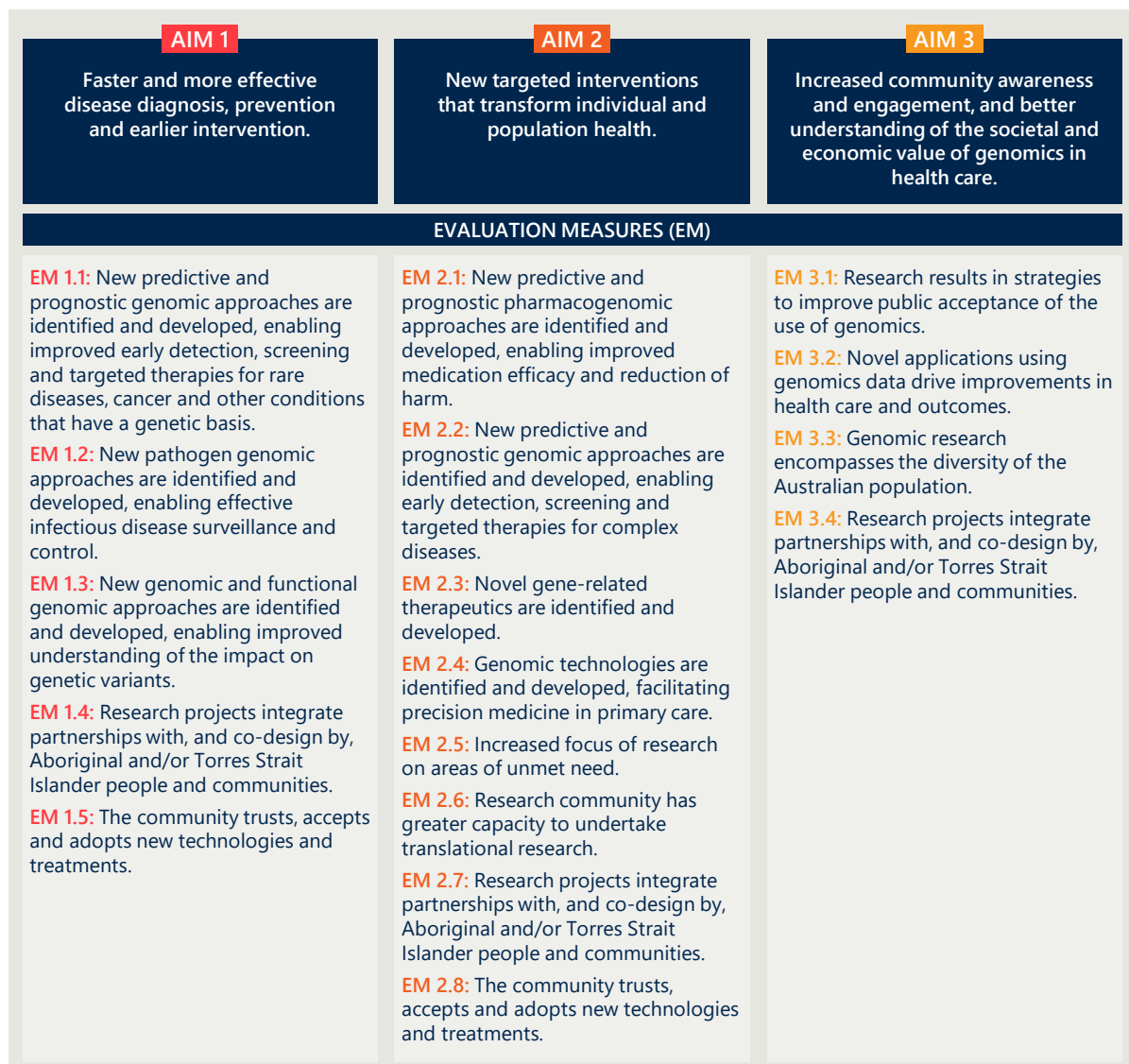
¹² The second EAP will develop a revised GHFM Roadmap and GHFM Implementation Plan following the GHFM Review. For the purposes of the GHFM Review, all references will be made to first GHFM Roadmap and GHFM Implementation Plan.

For each priority area the GHFM Implementation Plan identifies:

- The priority research questions and objectives in the short (1–2 years), medium (2–5 years) and long term (6–10 years).
- Opportunities to use additional investment and other research to support the priority area.
- Activities required to support the research and facilitate implementation.

The GHFM Implementation Plan supports the implementation of the GHFM Roadmap and establishes a strategic plan to address the GHFM aims within the context of the MRFF 10-year Plan. The Implementation Plan defines an evaluation approach and measures (EM) for each aim of the GHFM (refer to Figure 5, below).

Figure 5 | GHFM Aims and Evaluation Measures

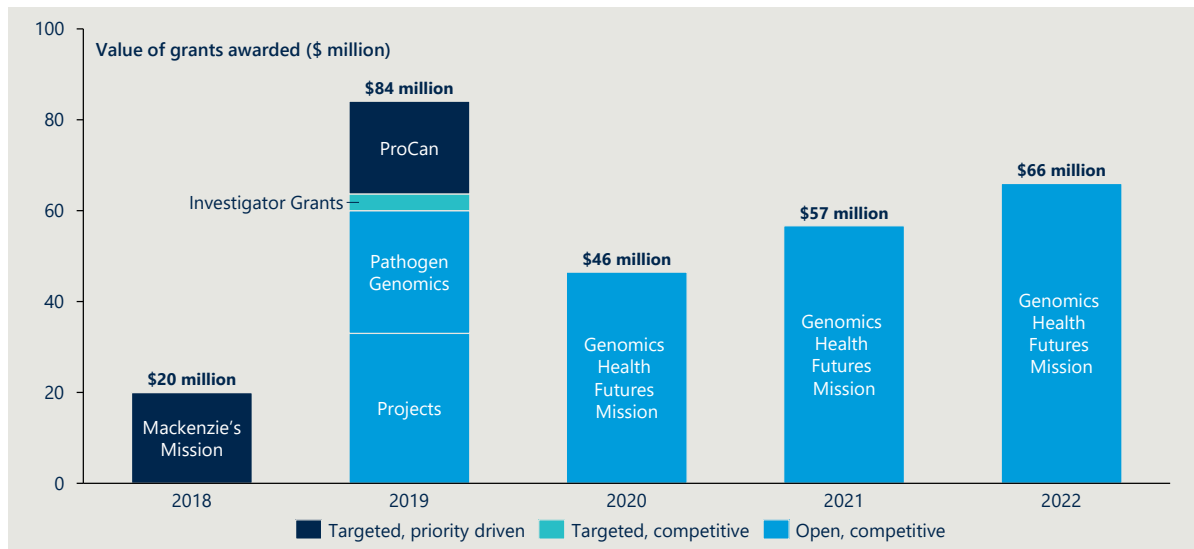


Source: GHFM Implementation Plan.

GHFM investments

Since its inception in 2018 the GHFM has had eight grant opportunities, five of which were open and competitive, resulting in funding of \$273.20 million being awarded to 88 projects (as at 31 December 2023). (Refer to Figure 6, overleaf).

Figure 6 | Grants awarded by GHFM Grant Opportunity

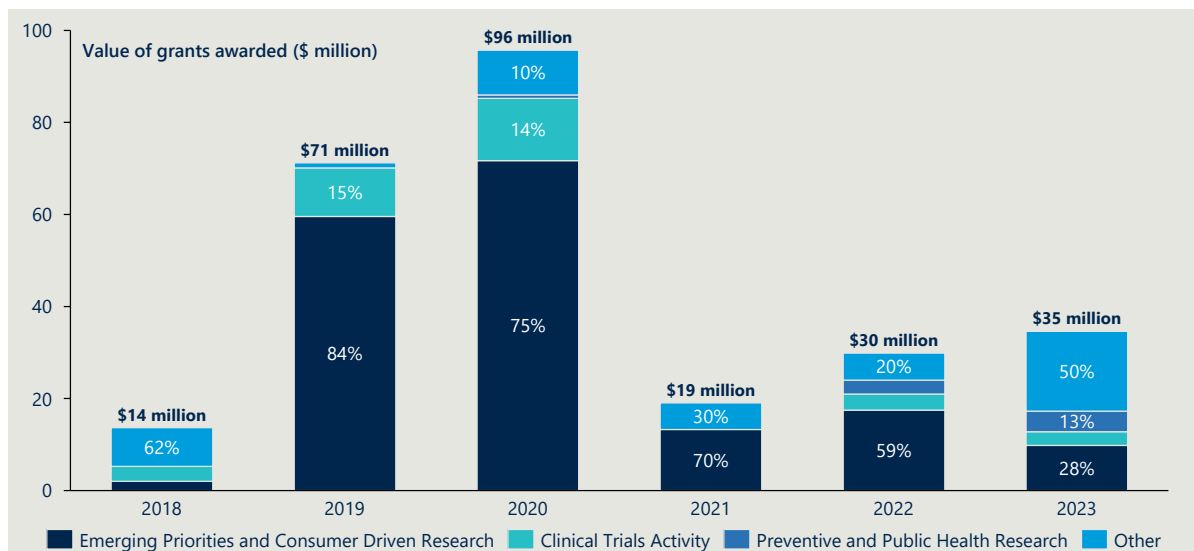


Source: Nous analysis of departmental records as at 31 December 2023.
Figure note: The *Targeted, priority driven grants* were 'one-off or ad-hoc'. The definition of the types of grant opportunities are as per the *Commonwealth Grant Rules and Guidelines 2017*.

2.2.2 Funding towards genomics research provided by other MRFF initiatives

Outside of the GHFM, 82 genomics projects have been funded by 13 of the remaining 20 MRFF initiatives (non-GHFM).¹³ Three quarters of identified genomics projects outside of the GHFM were funded by the Emerging Priorities and Consumer Driven Research Initiative (\$173.8 million; 66 per cent of all funding) and the Clinical Trials Activity Initiative (\$33.8 million; 13 per cent of all funding). The other nine initiatives have each provided between \$1 million and \$10 million to genomics projects (refer to Figure 7, below).

Figure 7 | Investments in genomics-related research projects by MRFF initiatives (excluding the GHFM)



Source: Nous analysis of departmental records as at 31 December 2023.

¹³ The department provided Nous the list of genomics projects funded by MRFF initiatives to be considered by the GHFM Review on 31 October 2023. For additional information on the MRFF projects not funded by the GHFM that have been included in the scope of the GHFM Review refer to **Appendix B**.

3 About the Review

This chapter describes the rationale, intent, and high-level approach of the mid-term review of the Genomics Health Futures Mission (GHFM Review).¹⁴

The Department of Health and Aged Care (the department) engaged Nous Group (Nous) to prepare a mid-term review of the Genomics Health Futures Mission (GHFM). The final report (the report) has been prepared for the department to fulfil the intent of the GHFM Review.

3.1 Rationale for the GHFM Review

The reviews, evaluations and other assessments of the MRFF and its initiatives are essential for providing the community, researchers, consumers, government and other stakeholders with¹⁵:

- Accountability.
- Policy and research benefit.
- Public engagement.
- Visibility.
- Lessons learned.

The pace of change in genomics—whilst exciting and engaging—presents a challenge for government when determining the best approach to investment in the field, whether this is through competitive grant programs, targeted investment in enabling infrastructure, or building capability to drive translation from the laboratory into health care. The genomics research landscape has evolved significantly since the establishment of the GHFM in 2018.

With five years remaining for the initiative, a review of the GHFM's progress and impact, in light of the evolving environment, is timely and appropriate to ensure the GHFM is well placed to reach its goal. The GHFM Review seeks to provide the evidence base to develop options for future investments and inform the refresh of the GHFM Roadmap and Implementation Plan as well as future funding and granting arrangements for GHFM. The GHFM Review is being undertaken as part of a series of mid-term reviews of MRFF initiatives.¹⁶

3.2 Intent of the GHFM Review

The intent of the GHFM Review was to assess:

- I. **How the MRFF has contributed to genomics research in Australia, via:**
 - All existing investments under the GHFM through the MRFF.
 - All other existing investments in genomics research made through the MRFF.

¹⁴ Also referred to as 'the Review'.

¹⁵ Department of Health and Aged Care, Performance indicators towards the impact of the Medical Research Future Fund, March 2023.

¹⁶ The GHFM Review is the sixth major evaluation activity of an MRFF initiative, and third review of a MRFF Research Mission.

- II. **How MRFF-funded genomics research sits within the national and international genomics research funding landscape.**¹⁷ This will involve:
 - Mapping MRFF-funded genomics projects according to the three GHFM Roadmap priority areas.
 - Assessing key genomics research funding priorities and outcomes nationally (outside of MRFF) and those set by appropriate international comparators.
 - Identification of national strengths and capacity to undertake the research and translate the findings, to better inform strategic allocation of future GHFM funding priorities.
- III. **Alignment and progress of MRFF-funded genomics research towards:**
 - The GHFM Roadmap and Implementation Plan.
 - The Australian Government’s 10-year MRFF investment plan and the MRFF’s Monitoring, Evaluation and Learning Strategy 2020-21 to 2023-24 (MRFF Evaluation Strategy).
- IV. **Opportunities (if any) to enhance MRFF funding and granting arrangements to improve the impact of MRFF funded genomics research.**

3.3 Scope of the GHFM Review

The GHFM Review examines all genomics investments made by the MRFF. The GHFM Review focuses on the progress towards the MRFF objectives; the administrative processes of the MRFF were out-of-scope.¹⁸ The GHFM Review is not an audit, evaluation or scientific review of literature.

Nous has relied upon the views as presented by stakeholders during consultations and the survey, as well as some provided through documentation. Nous has not sought to verify or provide assurance over the data collected. Verbatim transcripts of consultations were not prepared, therefore all quotes from stakeholders presented in the report are Nous’ interpretation of the sentiment expressed.

While the anticipated establishment of Genomics Australia is a topic of interest for the sector, its impact and implications for the MRFF have not be considered in detail.

3.4 Governance of the GHFM Review

The GHFM Review involves multiple individuals (responsible parties) who hold unique roles and responsibilities in the conduct of the GHFM Review. The roles and responsibilities of each responsible party are described in Table 4, below (for additional information refer to **Appendix A**).

Table 4 | Roles and responsibilities for the GHFM Review

Responsible parties	Responsibilities
Health and Medical Research Office	<ul style="list-style-type: none"> • Lead and oversee the activities of the GHFM Review. • Provide feedback on and approve reports prepared by Nous and Australian Genomics.
GHFM Review Panel	<ul style="list-style-type: none"> • Provide expert advice and guidance to all other responsible parties. • Provide feedback on deliverables prepared by Nous Group and Australian Genomics.

¹⁷Australian Genomics is leading the component II of the GHFM Review by conducting desktop analysis of the landscape. Nous has not sought to conduct additional analysis of, or validate, the findings of Australian Genomics Desktop Review Report.

¹⁸ Although the GHFM did not examine the effectiveness or efficiency of administrative practices of the department some of the opportunities identified have direct or indirect implications for administrative processes.

Responsible parties	Responsibilities
Nous Group	<ul style="list-style-type: none"> Consult key national and international stakeholders. Design and deliver a survey of MRFF genomics project leads. Review progress reports submitted by grantees and other key documentation. Collect and synthesise evidence collected through sector consultations, a grantee survey and the Desktop Review to prepare a review report.
Australian Genomics	<ul style="list-style-type: none"> Prepare a desktop review of the Australian and international genomics landscapes.

3.5 Methodology

To fulfill the intent of the GHFM Review, four methods were used (refer to Table 5, below):

- A survey of grantee project leads (*Grantee Survey*).
- A series of interviews and focus groups with Australian and international organisation with the genomics sector (*Stakeholder Consultations*).
- A desktop review of the Australian and international genomics research landscape (*Desktop Review*).
- A targeted review of internal and public GHFM and MRFF documentation (*Document Review*).

Table 5 | Data collection methodology

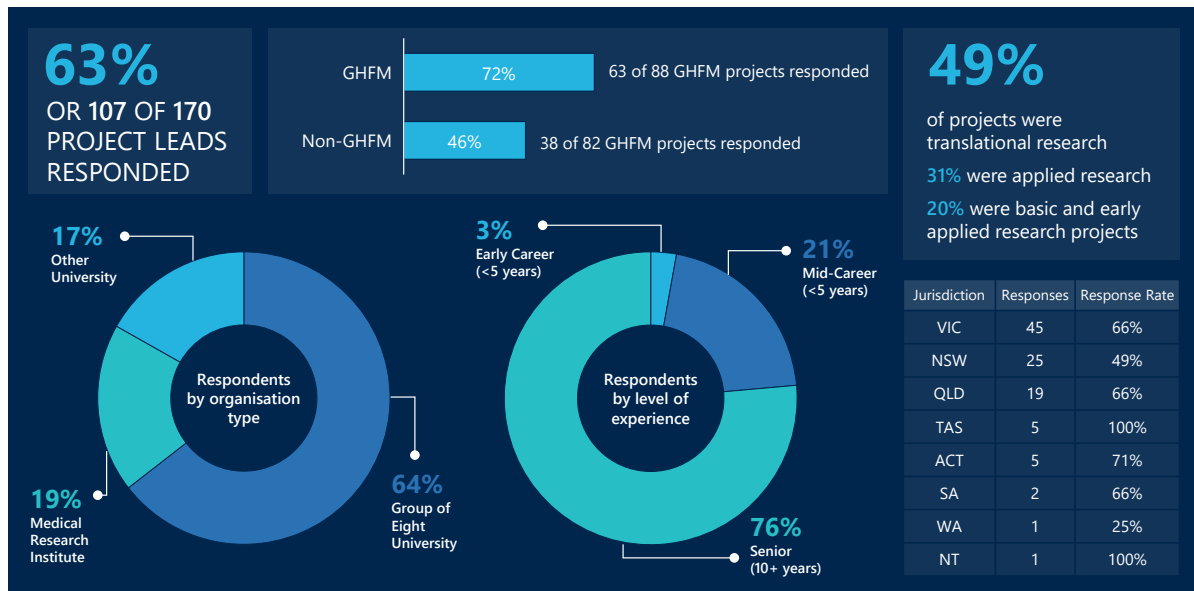
The GHFM Review assessed...	Grantee survey	Stakeholder Consultations	Desktop Review	Document review
I. Contribution <i>How the MRFF has contributed to genomics research in Australia.</i>				
II. Reputation <i>How MRFF-funded genomics research sits within the national and international genomics research funding landscape.</i>				
III. Progress <i>Alignment and progress of MRFF-funded genomics research.</i>				
IV. Opportunities <i>Opportunities (if any) to enhance MRFF funding and granting arrangements to improve the impact of MRFF funded genomics research.</i>				

Nous concluded data collection through the survey, document review and the majority of consultations in December 2023; a small number of consultations were facilitated in January 2024. The evidence was assessed against benchmarks in the MRFF Evaluation Strategy, GHFM Roadmap and Implementation Plan.

Grantee Survey

Between November and December 2023, Nous ran a survey to which 107 of the 170 project leads responded (63 per cent) which was broadly representative of the population of MRFF-funded genomics projects, except for a relatively lower response rate for non-GHFM projects (refer to Figure 8, below).

Figure 8 | Response rates to grantee survey

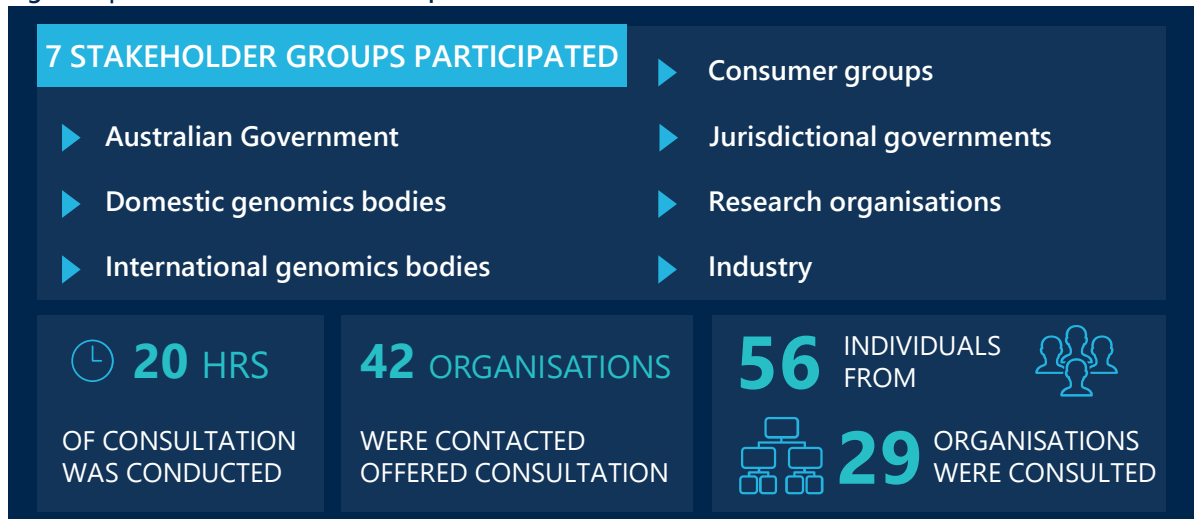


Source: Nous analysis of grantee survey results, as at 16 December 2023.

Stakeholder Consultations

Between November 2023 and January 2024, Nous conducted 20 hours of consultations with 56 individuals representing 29 of the Australian and international organisations contacted (refer to Figure 9, below).

Figure 9 | Stakeholders consulted as part of the GHFM Review



Source: Nous analysis of the consultation record, as at 31 January 2024.

Desktop Review

The department engaged Australian Genomics to undertake the Desktop Review, which aimed to contextualise MRFF-funded genomics research within the national and international genomics research

funding landscapes, which was primarily used to fulfil the second component of the intent of the GHFM Review. Australian Genomics' GHFM Desktop Review was conducted in three components¹⁹:

- Mapping MRFF-funded genomics projects according to the three priority areas defined by the GHFM Roadmap.
- Assessing key genomics research funding priorities and outcomes nationally and those set by international comparators.
- Identifying national strengths and capacity to undertake genomics research and translate its findings.

The final Desktop Review report was 144 pages and provided to Nous on 19 February 2024.

Document Review

Nous conducted a targeted review and data extraction of the 349 progress and 14 final reports submitted by grantees to the respective grant hubs. Nous reviewed the eight GHFM grant opportunities.²⁰ In addition to the foundational documentation of the GHFM and MRFF, Nous reviewed the following public documents:

- Australian Genomics: Outcomes of a 5-year national program to accelerate the integration of genomics in healthcare (The American Journal of Human Genetics, 2023).
- Valuing the impact of genomics on healthcare in Australia (Industry Genomics Network Alliance, 2021).
- GHFM Roadmap and Implementation Plan Consultation Report (Department of Health, 2021).
- Department of Health's Management of Financial Assistance under the Medical Research Future Fund (Australian National Audit Office, 2021).
- Genomics Health Futures Mission design, delivery and research priorities: summary of Scientific Strategy Committee recommendations (Department of Health, March 2021).
- A comparison of the distribution of Medical Research Future Fund grants with disease burden in Australia (Medical Journal of Australia, 2021).
- The International Review of the GHFM Roadmap and Implementation Plan (Department of Health, 2020).
- What does Australia's investment in genomics mean for public health? (Australian and New Zealand Journal of Public Health, 2019).
- National Health Genomics Policy Framework (Australian Health Ministers' Advisory Council, 2017).

¹⁹ Australian Genomics was separately engaged by the department to develop the Desktop Review. Nous reviewed and extracted insights from the Desktop Review and included them in the report. Nous did not validate or direct the Australian Genomics' analyses.

²⁰ Nous conducted a high-level review of the 48 grant opportunities that non-GHFM projects were funded through.

4 Findings

This chapter describes the key evidence gathered and findings synthesised from stakeholder consultations, the grantee survey, the desktop review, and the document review.

4.1 Genomics funding landscape

SCOPE

This section of the report describes the Review's findings in relation to the following components of the intent of the GHFM Review:

- The 'alignment' component of **III. Alignment and progress of MRFF-funded genomics research.**
- **II. How MRFF-funded genomics research sits within the national and international genomics research funding landscape.**

KEY FINDINGS

- The GHFM's aims and priority areas are broad and comprehensive, with a focus on diagnosis, prevention and early intervention.
- Although the GHFM is a large funder it alone cannot fund the entire translation effort; the complexity of the Australian genomics landscape makes it difficult to coordinate cohesive investments with other funders.
- Australia boasts strong genomic research capabilities, but international counterparts have greater funding and broader remits to progress their agenda for advancing genomics.

OPPORTUNITIES IDENTIFIED

- The review identified two opportunities for the GHFM to explore addressing in the next five years:
 - **Opportunity 1** | Refine and consolidate the GHFM's priorities.
 - **Opportunity 2** | Increase information sharing and partnership between members of the Australian genomics policy and funding landscape.

The genomics landscape in Australia is a complex, billion-dollar ecosystem that spans research innovation and capabilities, health system policy and frameworks, through to clinical genomics and diagnostic services. Australia is home to many world-class researchers, who are driving research discovery and innovation in human health genomics. The national genomics landscape is made up of a diverse network of consumers and stakeholders from Commonwealth, state and territory governments, academic and medical research institutes (MRI), industry, and not-for-profit organisations.

The MRFF funding Principles state that funding from the MRFF will²¹:

"Complement existing funding into health and medical research through a strategic top-down approach to investment."

The Review considered the following questions:

- Are the priorities of the GHFM, and funding patterns of the MRFF well-balanced and complete?

²¹ Department of Health and Aged Care, *Medical Research Future Funding Principles*, 2017.

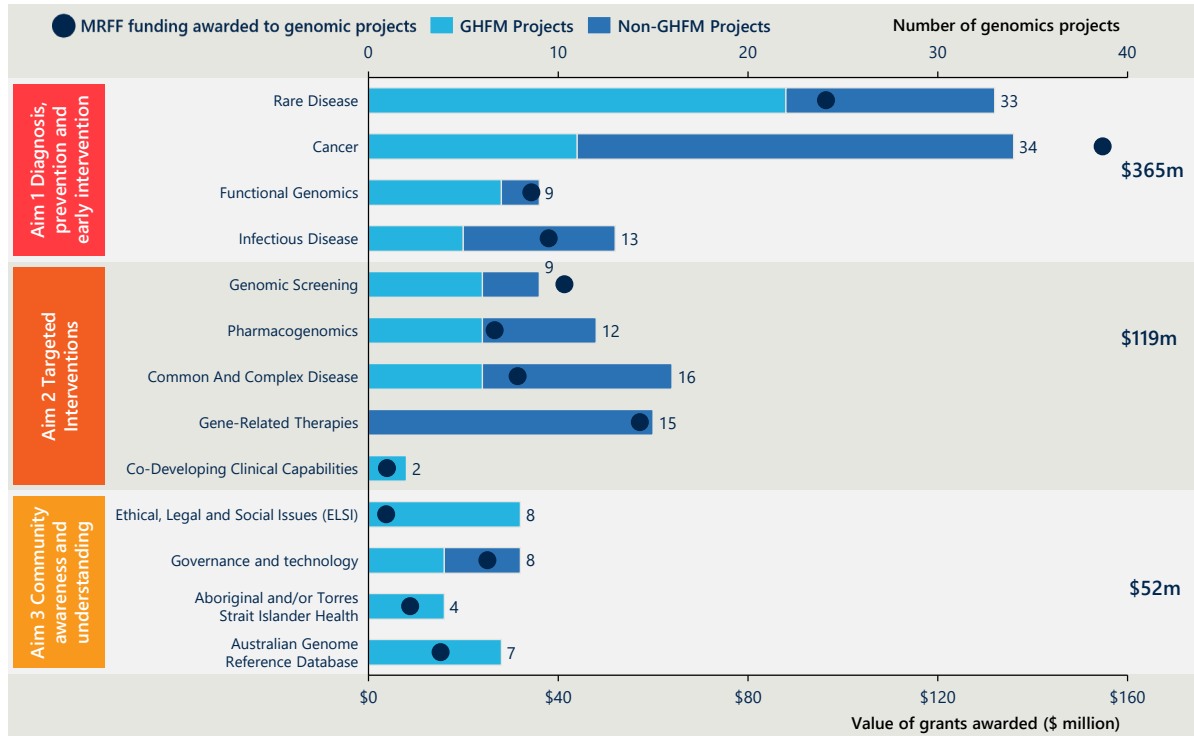
- Where does MRFF-funded genomics research sit within the national genomics research funding landscape?
- How does the GHFM, and the Australian landscape, compare to key international counterparts?

4.1.1 The GHFM’s aims and priority areas are broad and comprehensive, with a focus on diagnosis, prevention and early intervention

The GHFM has contributed a significant amount of funding towards genomics research to date, across a broad range of priority areas. In its first five years, GHFM investments have been largely centred around Aim 1 of the GHFM, ‘Diagnosis, Prevention and Early Intervention’, particularly in the priority areas of Rare Disease and Cancer. The least investment, when considering the primary priority of each project, was in gene-related therapies which has not had any investments by the GHFM in the first five years (however it did receive funding incidentally through other MRFF initiatives), and ethical, legal and social implications (ELSI) focused research (refer to Figure 10, below). The difference in the volume of investment to Aim 1 compared to Aims 2 and 3 is driven by:

- The number of projects funded (98 projects were supported under Aim 1, compared to 45 and 27 for Aim 2 and 3 respectively).
- The number of projects funded outside of the GHFM (47 projects related to Aim 1 were funded by non-GHFM initiatives compared to 31 and 4 for Aim 2 and 3 respectively).
- The average value of Aim 1 projects was higher (the average value of grants to Aim 1 projects was \$3.73m, compared to \$2.65m and \$1.96m for grants to Aim 2 and 3 projects respectively).²²

Figure 10 | GHFM Funded Projects by Primary Priority Area



Source: Australian Genomics’ GHFM Desktop Review, as at 16 February 2024.

²² The reasons for higher average grant value for Aim 1 projects were not explored by the Review, however a likely contributor is the relative cost of research activities associated with Aim 1 priority areas compared to Aim 2 and 3 projects.

GHFM funding pool is stretched across many priorities and could be better targeted

The GHFM Roadmap Funding principles state that activities funded under the GHFM:

“Should be, or contribute to, large national programs of work of strategic importance in key priority areas as outlined in the Implementation Plan. Research activities are expected to foster collaboration and harness resources across the system to deliver improved health outcomes for Australians.”

Prior to their finalisation in 2020 the GHFM Roadmap and Implementation Plan were reviewed by an international panel of experts, who found that²³:

“The GHFM scope and priorities were broad and ambitious, and the allocated funding may not be sufficient to achieve all identified aims.”

All stakeholders agreed that the aims and priorities have been broadly appropriate for the GHFM over the first five years. However, some stakeholders described the number of priorities as more of a ‘shopping list’ and did not represent a strong prioritisation of resources. A few stakeholders reflected that the dilution of funds across the large number of priorities had limited the ability of the GHFM to invest the sums needed to effectively translate genomics research.

The sector does not fully understand the reasons driving the distribution of GHFM funding

At an overall level, most stakeholders acknowledged they were not sufficiently informed on the spread of funding but generally reflected that most of the focus had been on GHFM Aim 1. Stakeholders generally recognised that the bias towards Aim 1 was partially explained by relative expense of conducting Aim 1 projects compared to social research projects funded in Aim 3.

Many stakeholders raised concerns that the reasons why and when grant opportunities were announced, or why particular research projects were funded, was not clear.²⁴ This made it more difficult for most stakeholders to understand the funding decisions or link the funding to the GHFM Roadmap, Implementation plan or towards the achievement of the GHFM aims and priorities.

There is a need to re-prioritise and balance investments, however the sector lacks consensus on how

Although all stakeholders agreed that the aims and priorities have been broadly appropriate for the GHFM over the first five years, many argued for minor adjustments in favour or against particular priorities. The views of stakeholders were often related to their research interests, areas of expertise, or place in the sector. Noting the breadth of views the core issues raised were:

- **The GHFM must consider how to balance burden of disease with addressing unmet needs.** There were mixed views across stakeholders on the GHFM’s current balance between rare diseases and high burden diseases such as cancer. Some stakeholders argued that given the scale of the GHFM’s goals, it was appropriate for its investments to focus on areas with a higher burden of disease which could impact the greatest number of Australians. While other stakeholders argued that given high burden diseases such as cancer already receive significant funding outside of the GHFM (via other countries, other research funding bodies and other MRFF initiatives), GHFM funding is better spent on rare diseases where funding could be targeted to rare diseases lacking investment elsewhere.
- **Increase the focus on First Nations Genomics.** There was a strong view in the sector that Australia does, and should continue to, lead in Indigenous genomics research, especially internationally. Until

²³ Department of Health, [International Review of the GHFM Roadmap and Implementation Plan](#), 2020.

²⁴ Stakeholders spoke generally at high level and did not refer to specific (or a subset of) grant opportunities or funding decisions.

the recent introduction of The Aboriginal and Torres Strait Islander Genomic Advisory Group, some stakeholders felt progress had been slow and unsympathetic to the distinct needs of Indigenous genomics.

- **Introduce more priorities linked to implementation.** A few stakeholders raised concerns that despite the MRFF’s intent to be more focused on translation, there has been insufficient investment in the implementation of genomics research. These stakeholders noted that there may be an argument to introduce new priorities to increase the focus (priorities for health economics and implementation science in the context of genomics were proposed).

Opportunity 1 | Refine and consolidate the GHFM’s priorities

With 13 priority areas, which are broad in scope, the current GHFM funding pool is spread thin across priority areas. This reduces the impact the GHFM can have in any one area of genomics, and limits the ability of the GHFM to attract further capital and capability to the priority area.

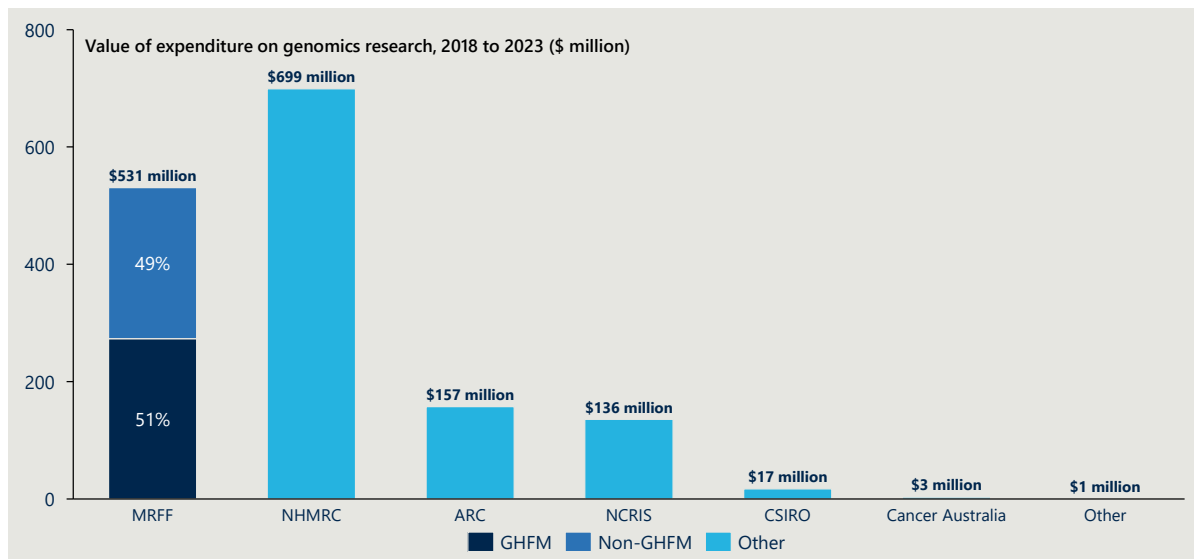
4.1.2 Although the GHFM is a large funder it alone cannot fund the entire translation effort; the complexity of the Australian genomics landscape makes it difficult to coordinate cohesive investments with other funders

As discussed in 2.1, the MRFF operates in tandem with other health and medical research funders.

The MRFF is the second biggest funder of genomics research in Australia

The MRFF, through the GHFM and other initiatives, has been the second largest funder of genomics research since the start of the GHFM in 2018 (refer to Figure 11, below).

Figure 11 | National genomics research funders



Source: Australian Genomics’ GHFM Desktop Review, as at 16 February 2024.

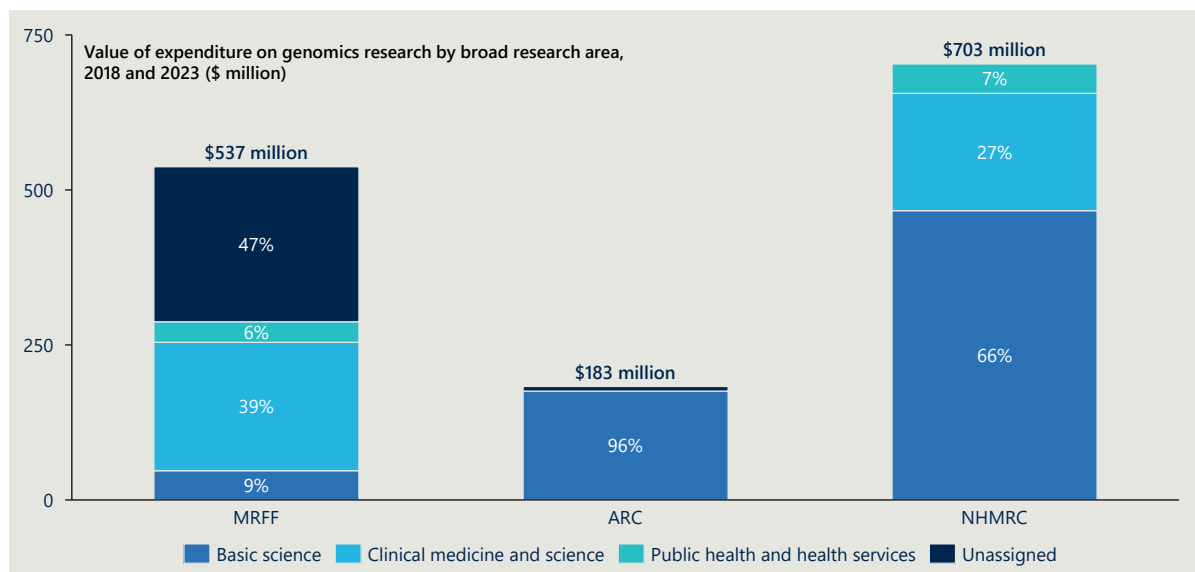
Figure note: Australian Research Council (ARC); National Collaborative Research Infrastructure Strategy (NCRIS); Commonwealth Scientific and Industrial Research Organisation (CSIRO).

The MRFF is more concentrated on later stage translation projects compared to NHMRC and ARC

The purpose of the MRFF and the historical approach of NHMRC are intended to be highly complementary. Broad-based NHMRC funding seeks to ensure Australia is actively engaged in research to address our diverse health needs, connected to international developments and ready to respond to emerging health challenges. By investing in priority areas, the MRFF seeks to deepen and build on the foundation of knowledge, capacity and capability established and maintained by NHMRC.

Analysis of self-assigned broad research areas for genomics projects funded by MRFF, ARC, and NHMRC shows a greater portion of MRFF funding is directed to clinical medicine and science compared to the NHMRC and ARC (refer to Figure 12, below).²⁵ While 'clinical medicine and science', 'public health' and 'health services' contain projects at various points of the translation pipeline, collectively they are generally considered to be later in the translation pipeline than 'basic science'.²⁶

Figure 12 | Distribution of Commonwealth genomics funding across broad research areas



Source: Australian Genomics' GHFM Desktop Review, as at 16 February 2024.

Figure note: The categories of health services and public health research are combined because they represent a relatively small quanta of the funding provided to genomics research across each of the organisations.

The Australian Government's [Science, research and innovation budget tables](#) reinforces that the GHFM is intended to have a greater focus on strategic coordination and direction setting for the sector.²⁷ It is notable that the tables indicate that the MRFF (whether via the GHFM or other initiatives) is intended to support research infrastructure.

²⁵ [Broad Research Area](#) is NHMRC's longest established research classification, the current version of the definitions was developed in 2022 to assist researchers to select the Broad Research Areas that best describes their research proposal/project.

²⁶ As a proportion of the funds that could be categorised. Forty-seven per cent of the funding could not be assigned to a broad research area because while the NHMRC provided the department with the Broad Research Areas nominated by researchers for most MRFF projects funded under grant opportunities administered by the NHMRC grants hub, no comparable data was available for those administered by DISR.

²⁷ The Science, research and innovation budget tables are published annually by DISR to report on the Whole-of-Australian-Government's investment in science, research and innovation.

The national genomics funding landscape is complex; the GHFM has some priorities common to other funders but tends to be more focused on translation within those areas

Australian Genomics found that the GHFM shares common priority areas with other national funders including translational research, rare disease, cancer, and First Nations research. The GHFM is unique in its funding of translational research for genetics and genomics. The GHFM is also focusing investment on research that investigates the diagnoses of rare disease and cancer, as opposed to treatments alone. The information collected by the Desktop Review demonstrates the complexity of the funding landscape and different approaches and priorities of genomics funders in Australia (refer to Table 6, overleaf).

The genomics sector is funded and supported by a variety of different organisations, each acting with different objectives and policy levers. The development of the GHFM Roadmap and Implementation plan relies on the experience and sector knowledge of the experts appointed to EAP in a way that minimises duplication with other funders. However, outside the Roadmap and Implementation plan process, the department and EAP does not receive insights on the evolving funding landscape to inform the timing or design of grant opportunities and funding decisions.²⁸

²⁸ The GHFM Review, in particular the Desktop Review, has been designed to update and inform the department and recently appointed EAP on the genomics funding landscape.

Table 6 | GHFM in the Australian genomics funding landscape

	GHFM	Australian Genomics	Other National Funders ^A	Industry	Private/NFP sectors
Est. Funding p.a. (proportion of research spend)	\$50m (100 per cent)	\$5 million (100 per cent)	> \$140.6 million (~ 10 per cent)	Not available	Not available
Priority areas	<p>Diagnosis, prevention, and early intervention</p> <p>Targeted interventions</p> <p>Community awareness and understanding</p>	<p>Improve efficiency, reach and timeliness of genomic research projects</p> <p>Support government health departments in implementation of genomics research outcomes</p>	<p>Basic science research</p> <p>Building research infrastructure</p> <p>Developing local and international research networks</p>	<p>Sponsorships, partnerships, and in-kind grant opportunities</p> <p>Align with corporate strategic priorities</p> <p>Support research capabilities (e.g.: sequencing services)</p> <p>Focus on investigator-led projects</p>	<p>Clinical and translational research to improve diagnosis and treatment for specific patient groups</p> <p>Educating, supporting, and empowering patients</p>
Consumer engagement	<p>Consumer involvement principles</p> <p>Grant assessment criterion</p> <p>Consumer representative on GHFM committees</p>	<p>Community Advisory Group</p> <p>Involve Australia: 'Guidelines for Community Involvement in Genomic Research'</p> <p>Ethical, Legal and Social Implications (ELSI) Network</p> <p>Summer Internship for Indigenous Genomics (SING) Australia</p> <p>Genomics in Schools</p> <p>DNA Dialogue</p> <p>Conferences and events</p> <p>Stakeholder consultations</p>	<p>Consumer advisory groups and consultations</p> <p>Peer review panels</p> <p>Toolkits and resources</p>	<p>Third party meetings and symposia sponsorship</p> <p>Therapeutic development</p>	<p>Community advisory groups and peer review panels</p> <p>Guides for researchers and participants</p> <p>Clinical trial registries</p> <p>Support services, networks, and resources</p>

	GHFM	Australian Genomics	Other National Funders ^A	Industry	Private/NFP sectors
Industry Partnerships	<p>Inclusion in grant assessment criteria and Implementation Plan</p> <p>Investigator-led industry partnerships</p>	Partnership with industry through the Industry Genomics Network Alliance	<p>Support research and development (R&D) collaborative projects (e.g.: NHMRC Partnership Projects)</p> <p>Facilitate research translation and commercialisation</p> <p>Bioplatforms Australia (BPA) is an InGeNA partner</p>	<p>Collaborations with other industry partners through sponsorships and InGeNA membership</p> <p>Industry expert webinars</p>	<p>Sponsorships</p> <p>Partnerships to facilitate research translation</p>
Clinical Partnerships	<p>Facilitating clinical translation of research advancements</p> <p>Funds Australian Functional Genomics Network (AFGN)</p> <p>Inclusion in grant assessment criteria and Implementation Plan</p>	<p>Clinical flagships</p> <p>Supports AFGN</p> <p>Partnerships with state/territory clinical genetic services and genomics alliances</p> <p>Clinical, Diagnostic and Research (CDR) Network</p> <p>National Implementation Committee (NIC)</p>	<p>NHMRC's Centres of Research Excellence</p> <p>Clinical trial funding and facilities</p> <p>Clinical partnerships embedded</p> <p>Commonwealth Scientific and Industrial Research Organisation</p> <p>Support from NCRIS for genomics infrastructure</p>	<p>Third party meetings and symposia sponsorship</p> <p>Therapeutic development</p>	<p>Connecting patients with clinical trial opportunities</p> <p>Building clinical research networks</p>
Priority Populations	Indigenous peoples	Indigenous peoples Culturally and linguistically diverse (CALD) communities	Indigenous peoples CALD communities Rural / remote communities	Indigenous peoples CALD communities	Indigenous peoples Rural / remote communities
Workforce Development	Investigator-led	<p>Workforce education including Genomics Education Network of Australasia (GENA)</p> <p>Reporting Item Standards for Education and its Evaluation in Genomics (RISE2 Genomics)</p> <p>CDR Network</p>	<p>Research fellowships</p> <p>Webinars hosted with InGeNA</p>	<p>Educational events</p> <p>Internship programs</p> <p>InGeNA Workforce Precision Medicine Competency Framework and webinars</p>	<p>Building capability and capacity of research workforce with fellowships and scholarships</p>

	GHFM	Australian Genomics	Other National Funders ^A	Industry	Private/NFP sectors
Innovative Funding Models		Policy Network			
	Incubator projects	Clinical flagships Co-funded projects Genomic implementation projects	Targeted Calls for Research and Ideas (NHMRC) Supporting people with cancer initiative (Cancer Australia) International Collaborations (NHMRC) International Relations (Department of Foreign Affairs and Trade)	Grant opportunities for provision of genomics sequencing services	Facilitating investment for innovative research with consumer involvement Innovative fundraising models to support research
Data and Infrastructure	Priority areas 3.2 and 3.4 in the Implementation Plan	Implementation recommendations for a National Approach to Genomic Information Management (NAGIM) Digital platforms: <ul style="list-style-type: none"> • Shariant • PanelApp Australia • Dynamic consent platform (CTRL) • Reimagining Health Genomics: Technology Summit 	Australian BioCommons Human Genome Informatics Initiative Phenomics Australia genome engineered cell, tissue, and animal models National Computing Infrastructure (NCI) computational and data resources BPA genomics platforms NAGIM Blueprint	Sequencing services to generate datasets and enable research platforms	Funding for technology, equipment, and infrastructure

Source: Australian Genomics' GHFM Desktop Review, February 2024.

Table note A: Includes the Australian states and territory governments, however while collectively they provide a significant amount of funding to genomics, the exact amount could not be quantified by Australian Genomics, thus the 'Other National Funders' segment is understated. In addition to the funding provided to genomics research, state and territory governments also invest in the implementation of genomics through the health system and oversee the development of various frameworks and genomics alliances.

While state and territory investments in genomics research could not be estimated, some major ad-hoc investments in gene-related therapies in 2022-23 were identified²⁹:

- NSW Government allocated \$101.4 million to the development of a commercial scale viral vector manufacturing facility.
- NSW Government allocated \$119.0 million to the RNA Investment Fund.
- Victorian Government invested \$50m towards an mRNA vaccine and therapy manufacturing facility.

Opportunity 2 | Increase information sharing and partnership between members of the Australian genomics policy and funding landscape

The Australian genomics funding and policy landscape is complex. Without sufficient planning and knowledge sharing, the GHFM (and other genomics funders more broadly) risk duplicating the resources and efforts of other funders or overlooking under-served areas in genomics. Increasing the amount of information shared amongst funders would go some way to working through the complexity of the landscape.

4.1.3 Australia boasts strong genomic research capabilities, but international counterparts have greater funding and broader remit to progress their agenda for advancing genomics

Australian Genomics undertook a detailed benchmarking of the GHFM to key international comparators. The comparison provides a basis for considering the different approaches applied and the lessons they might have for the Australian landscape. The following criteria were applied by Australian Genomics to select the international comparators:

- The organisations should be comparable to the GHFM, to facilitate meaningful comparison with international investments in human genomic research:
 - Large-scale, national funders.
 - Demonstrable investment in human health genomics, preferably translational.
- The organisations should be active, ongoing funders of human genomics research.
- Other genomic research funders that represent edge-cases may be included as case studies.³⁰

The international comparators selected were Genomics England, Genome Canada, the National Human Genome Research Institute (NHGRI; USA) and Precision Health Research (PRECISE; Singapore).

The Australian landscape is organised differently to our closest comparators

As a research grant program attached to a broader fund, the GHFM has a significantly narrower scope and funding base relative to its comparators overseas, which are independent organisations with a range of policy, granting, workforce and regulatory functions. Some of the functions of these organisations are the responsibility of Australian actors such as the department and Australian Genomics, among others (refer to Table 7, overleaf).

²⁹ In addition to those investments most states and territories published other strategic plans or policy frameworks that address priorities in health genomics.

³⁰ Noteworthy examples of best practice approaches in key questions of interest, e.g. encouraging consumer, industry or health services involvement in priority setting or implementation; consideration of First Nations health or priority populations.

Table 7 | GHFM and Australian in the international genomics landscape

	Australia	England	Canada	USA	Singapore
Organisation/initiative	GHFM	Genomics England	Genome Canada	NHGRI	PRECISE
Estimated annual funding (approximate per capita)	\$50m (\$1.89 per capita)	\$242.18m (\$4.02 per capita)	\$76.16m (\$1.96 per capita)	\$801.02m (\$2.36 per capita)	Unavailable
Priority areas	Diagnosis, prevention, and early intervention Targeted interventions Community awareness and understanding	Genomic healthcare Research and partnerships Patients and participants 2023 priorities: newborn genomes; comprehensive cancer profiling; health inequity	Research and innovation Genomics in society Workforce	Genome structure, function Genomics and human disease Genomic medicine Genomic data science Genomics and society	Precision medicine program Population cohort Data linkage Clinical implementation
Consumer engagement	Consumer involvement principles Grant assessment criterion Consumer representative on GHFM committees	Participant Panel Ethics Advisory Committee Patient and participant representatives	Stakeholder roundtables Symposia and citizen science programs for school students	Strategy development Virtual roundtables Education working groups Community engagement Outreach partnership	Public consultations on PRECISE data collection and storage methods
Industry Partnerships	Investigator-led industry partnerships e.g.: AusPathoGen Program partnership with Illumina	Discovery Forum to share data with Industry partners	Public-private co-funding partnerships	Small and medium enterprise (SME) innovation research technology transfer Commercialising and licensing technologies	Project co-investment Industry consortia
Clinical Partnerships^B	Focus on facilitating translation of advancements to clinical practice	National Health Service (NHS) England Genomic Medicine Service (GMS)	Six provincial Genome Centres All for One and Canadian COVID-19 Genomics Network (CanCOGeN)	NIH, including NIH Clinical Centre and NIH Common Fund initiatives	Clinical Implementation Pilots (CIPs) Regional genomics initiatives

	Australia	England	Canada	USA	Singapore
	Inclusion in grant assessment criteria and Implementation Plan				
Priority Populations	Indigenous peoples	Minority and underrepresented groups	Indigenous peoples	Indigenous peoples LGBTQI+ Low- and middle-income countries	Singaporean population (Chinese, Indian, Malay)
Workforce Development	Investigator-led	Training and resources Health Education England's Genomics Education Program	Student engagement Industry experience Emphasis on Equity, Diversity, and Inclusion	Research training and career development Funding and partnerships to promote workforce diversity	Education/training in CIPs Optimising genetic counselling provision Industry internships
Innovative Funding Models	Incubator projects	'Infinity loop model': virtuous cycle between research and clinical care Fund patient and participant involvement representatives	COVID-19 pan-Canadian collaboration and impact relief funding Facilitate collaboration and co-funded grants on local and national scale Promote technology development and uptake	Intra- and extra-mural research Funding allocations for research centres and training (individual and institutional) Support technology commercialisation	Funding (\$3.4M AUD) for collaboration between shortlisted CIP applicants
Data and Infrastructure	Priority area 3.2 and 3.4 in the GHFM Implementation Plan	Research Environment: data sharing and linkage Bioinformatic products	Disruptive innovation, technologies, and data platforms	Openly available software and analysis tools Data resources Scientific Cores	Data sharing and linkage

Source: Australian Genomics' GHFM Desktop Review, February 2024.

Table note A: Direct comparison between GHFM and most international genomics research funders is complicated by both structural and organisational differences. The GHFM is a research funding mission under the broader MRFF investment whereas Genomics England, Genome Canada and PRECISE are nationally funded genomic initiatives delivering both research, translation, and infrastructure to embed genomics into mainstream healthcare.

Table note B: Clinical refers not to cohort studies alone, but to translation and implementation into healthcare.

International examples provide useful comparisons for other priority areas of funding for the GHFM

Australia boasts a wealth of internationally renowned genomic researchers. Australian Genomics found that the GHFM is unique nationally in its funding of translational research for genetics and genomics, particularly to improve the diagnosis of rare disease and cancer. Australian funding bodies are actively prioritising Indigenous genomics research where Australia has a unique capability and capacity to contribute to the global body of genomics knowledge. Australian Genomics identified examples of innovative strategies across Australia to involve consumers and underrepresented groups in genomics research and investment decision-making.

The GHFM shares priority areas, such as translational research, rare disease, cancer, and First Nations research, with other national and international funders of genomics research. Key priorities of other national and international funders that are not currently reflected in the GHFM include data infrastructure, industry partnerships and co-funding models, workforce development, and the development and translation of precision therapies. Other differences between the GHFM and international comparators identified are:

- There is an international trend to include priorities for the application of new technologies, translation into clinical services and development of therapeutics.
- There is a focus on applications for emerging genomic technologies where evidence is required for clinical implementation in both the national and international landscapes.
- Internationally, there is a trend of embedding an emphasis on linking data technology platforms, such as trusted research environments, and the use of multimodal data.
- International exemplars are striving to implement pharmacogenomic advancements.
- Enabling a nimble response to global health issues is being prioritised internationally, for example through substantial investment in COVID-19 programs in a clinically significant timeframe to the pandemic.
- Synergistic relationships between regional and national initiatives, particularly for the clinical implementation of precision medicine approaches at scale.
- The importance of actively engaging First Nations people and underrepresented communities in genomic research is being highlighted in the national and international genomics landscape, to develop global understandings of genomics in health. Enriching population cohorts and the diversity of genomic data is also a key priority.³¹

International comparators are taking a system-approach to their priorities

International comparators are taking a broader investment approach with genomics dedicated organisations investing directly in genomic technology development and enablers such as data platforms, workforce capability, and research infrastructure to support their funded research projects.

Australian Genomics found that an increasing proportion of international funding is being allocated towards developing and maintaining genomic technologies, platforms, and infrastructure, including the application of artificial intelligence (AI), to accelerate genomic discovery.

Unlike its international comparators, the GHFM is solely focused on research project-based funding and has not historically directly funded infrastructure and other enablers of genomics.³² The GHFM does fund

³¹ Australian Genomics, Desktop Review, February 2024.

³² While the Science, innovation, and research budget tables 2023-24 indicate that the MRFF is involved in supporting research infrastructure, it has been the position of the GHFM that it does not invest in infrastructure.

research on the implications and challenges of genomics technologies (Priority Area 3.2) and research projects contributing to the Australian Genome Reference Database (AGRD; Priority Area 3.4).

The GHFM Roadmap and Implementation Plan were informed by various inputs including Scientific Strategy Committee recommendations.³³ The Scientific Strategy Committee also noted significant investment in other countries into clinical genomics infrastructure:

“At least 14 countries have committed substantial government investment towards national research initiatives to drive the implementation of genomic medicine into health care. The overarching priority of these initiatives has been in developing infrastructure, which includes national frameworks, standards, and centres for testing and analysis, as well as platforms for collection, storage and sharing of data. However, the Scientific Strategy Committee notes that the development and investment in broader national clinical infrastructure is not the remit nor a strategic focus of the GHFM.”

³³ Department of Health, Summary of Scientific Strategy Committee recommendations, 2020.

4.2 Progress made towards the goals of the GHFM and MRFF

SCOPE

This section of the report describes the Review's evidence and findings in relation to following intent of the GHFM Review:

- The 'progress' component of III. **Alignment and progress of MRFF-funded genomics research.**

KEY FINDINGS

- Project leads are optimistic about their progress towards outcomes, but are unaware of the successes and learning from other MRFF projects.
- Progress to the GHFM's aim to increase genomic diagnoses, preventions and early interventions has been most pronounced, reflecting the volume of funding provided to Aim 1 projects.
- Progress thus far reflects outcomes of early translation activities.

OPPORTUNITIES IDENTIFIED

- Nous identified four opportunities for the GHFM to explore addressing in the next five years:
 - **Opportunity 3** | Improve the GHFM's communication and engagement with consumers, the community and the sector.
 - **Opportunity 4** | Strengthen linkages with other MRFF initiatives.
 - **Opportunity 5** | Ensure all projects consider the appropriate involvement of, and potential impact on, First Nations communities.
 - **Opportunity 6** | Concentrate investments in research projects further along the translation pipeline and enhance support to researchers to navigate translation activities such as clinical implementation and commercialisation of projects.

The goal of the GHFM is:

"To save or transform the lives of more than 200,000 Australians through genomic research to deliver better testing, diagnosis and treatment."

It does this *"by accelerating research that delivers more effective testing, diagnosis and treatment; facilitates the adoption of new interventions; and consolidates Australia's international leadership in genomics."*³⁴

Science, particularly translation of science, is a long-term endeavour. It often takes many years to take research into the clinic. Therefore, any assessment of the progress of the GHFM through its first five years should remain cognisant of this reality.

Without being able to directly measure progress to this goal, to understand the progress made by MRFF-funded genomics projects and, by extension Australia, the Review explored the following questions³⁵:

- How far progressed are individual MRFF-funded genomics projects towards their research objectives and outcomes?
- What progress has been made towards the aims and priorities of the GHFM?³⁶
- What progress has been made towards the MRFF measures of success and impact measures?

³⁴ GHFM Roadmap.

³⁵ There is insufficient data collection to determine progress to this goal. An international review of the GHFM Roadmap supported the ambition but stated that it was likely unachievable.

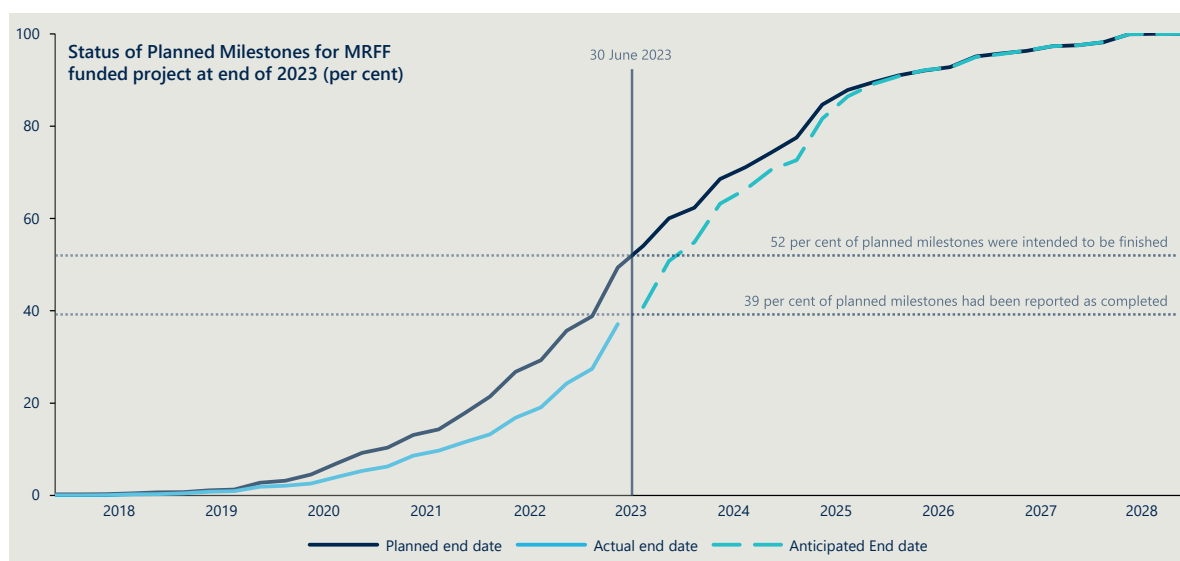
³⁶ The GHFM Evaluation Measures and Approaches resemble goals rather than metrics and do not include benchmarks.

4.2.1 Project leads are optimistic about their progress towards outcomes, but are unaware of the successes and learning from other MRFF projects

Although individual projects are at different stages, they are collectively around halfway towards completion of their objectives

It remains early days for genomics projects funded by the MRFF, particularly those through the GHFM, with 42 per cent of genomics projects in the first half of their original grant period (48 per cent for GHFM funded projects; refer to Figure 13, below).³⁷

Figure 13 | Completion of agreed milestones for MRFF genomics projects



Source: Nous Analysis progress and final reports submitted by grantees as at 30 June 2023.

Figure note: Nous has extracted data from progress reports, as provided by project leads, Nous has not been able to verify the accuracy or realism of the inputted data. There are also 16 projects (out of 170) not included in the data as they are yet to submit a progress report or the data they have submitted could not be interpreted by Nous. Therefore, this analysis is only indicative and should not be relied upon by others for other purposes.

Additionally, 52 projects have sought and received 60 variations to extend their grant period, primarily due to COVID-19 lockdowns but also caused from delays in the execution of various funding, multi-institutional and co-funding agreements. The median time of approved extensions was 12 months (with a mean of 9 months or 17 per cent).

The progress and achievements of, and lessons from, MRFF projects are not effectively communicated to the sector

Stakeholders frequently caveated their views on the GHFM's progress due to their lack of understanding of how projects are progressing (similar to Finding 4.1.1). Stakeholders often observed that it was difficult to find up-to-date and helpful information on the current status of many MRFF genomics projects. While it is not reasonable to expect stakeholders to have a strong view of the progress of all 170 projects, there is scope to improve the communication of progress or key milestones of more MRFF projects. This would provide the sector with up-to-date learnings or enable other stakeholders to identify where there may be

³⁷ The number of projects half-way through their grant period is not equivalent to the completion of project milestones as projects have different numbers of milestones.

emerging opportunities to support, collaborate or leverage the research progress of other MRFF genomics projects.

Opportunity 3 | Improve the GHFM’s communication and engagement with consumers, the community and the sector

The GHFM is making a positive contribution to the Australian and international genomics and health landscape. However stakeholders, including consumers, throughout the sector are largely unaware of many of these contributions and the various successes of the GHFM. Improving community and sector awareness would help facilitate communication of lessons learned and foster a strong authorising environment for genomics investments in Australia.

4.2.2 Progress to the GHFM’s aim to increase genomic diagnoses, preventions and early interventions has been most pronounced, reflecting the volume of funding provided to Aim 1 projects

Findings regarding the progress of the GHFM to date are based on analysis of feedback from stakeholder consultations and the survey of GHFM grantees.

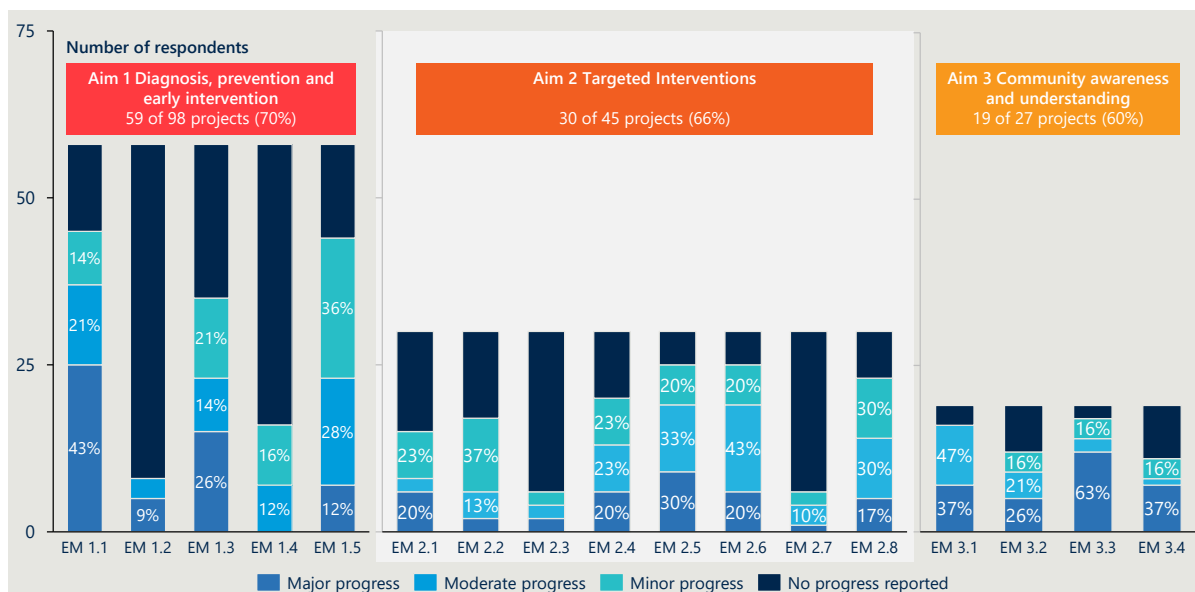
Progress towards the GHFM Roadmap and Implementation Plan were assessed primarily using:

- The GHFM aims (refer to Figure 4, page 14).
- The GHFM priority areas (refer to Figure 4, page 14).
- The GHFM evaluation measures and approaches (refer to Figure 5, page 15).

The sector is making the most tangible progress towards Aim 1 of the GHFM

Results from the survey indicated that the most progress has taken place towards Aim 1, which is consistent with the views of stakeholders. It appears the volume of grants is a key driver of the different levels of perceived progress, as Aim 3 project leads were proportionately the most positive when reporting on the progress of their project (refer to Figure 14, below).

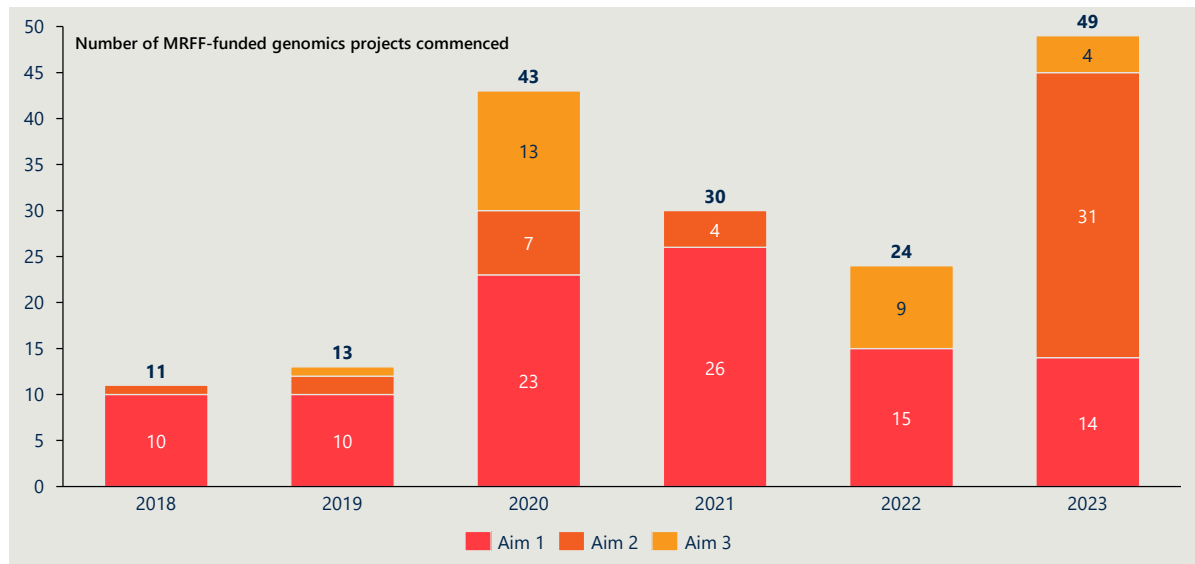
Figure 14 | Reported progress against GHFM Evaluation Measures



Source: Nous analysis of responses by project leads to the Grantee Survey, December 2023.

The difference in progress between GHFM Aims may be further exaggerated by the timing of those grants, with the majority of grant opportunities between 2018 and 2022 focusing on GHFM Aim 1 objectives (refer to Figure 15, below).

Figure 15 | Number of MRFF supported projects that commenced by GHFM Aim over time

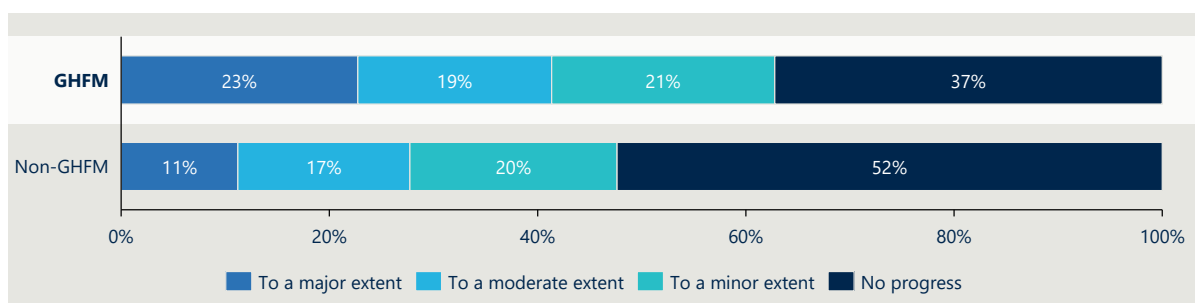


Source: Nous analysis of departmental records.

The GHFM Implementation plan acknowledged the importance of collaboration across MRFF initiatives that invest in genomics. Indeed, almost half of the total investment by the MRFF in genomics has occurred outside of the GHFM. This large proportion highlights the important role non-GHFM initiatives could play in achieving the GHFM’s goal.

GHFM project leads were more positive on their progress towards the GHFM evaluation measures (refer to Figure 16, below). As the GHFM aims and priorities are specific to the GHFM, it is unsurprising that this is the case.

Figure 16 | Average reported progress against evaluation measures by GHFM and non-GHFM projects



Source: Nous analysis of responses to the Grantee Survey, December 2023.

Opportunity 4 | Strengthen linkages with other MRFF initiatives

MRFF initiatives – including GHFM – should explore options to better align activities, recognising the separate objectives set out for each initiative. This opportunity is highlighted by the large proportion (almost 50 per cent) of genomics investment by the MRFF that occurs outside the GHFM which currently do not make the equivalent contribution to progressing the GHFM priorities.

Reasonable progress has been made to the development of faster more effective disease diagnosis, prevention or earlier intervention (GHFM Aim 1)

Overall, there has been reasonable progress towards Aim 1 in the first five years of the GHFM. The perceived progress towards Aim 1 priorities from stakeholders has largely mimicked the levels of investment, with improvements in diagnostics for rare disease a notable effort. The Zero Childhood Cancer Project, whilst a non-GHFM project, was a notable example of progress made within the cancer priority. Additionally, the impact of COVID-19 has led to a stronger focus on infectious disease genomics.

At a project level there has been variable progress towards the Aim 1 evaluation measures and approaches. Noting that participants could self-select the measures to respond to, the most significant progress, represented as 'major progress', was reported by GHFM-funded projects regarding new predictive and prognostic approaches, and in both funding categories for new pathogen genomic approaches. The least progress, represented by a response of 'minor progress', was demonstrated in the non-GHFM group regarding partnerships with First Nations communities and community, trust, acceptance, and adoption.

New targeted interventions that transform individual and/or population health take time to work through and therefore it is too early to detect significant progress (GHFM Aim 2)

Overall progress towards Aim 2 priorities were more difficult to identify within the review, with fewer projects funded under these priority areas. Stakeholders also felt it was too early to comment on progress towards new interventions, at only five years into the GHFM.

The project leads, however, were generally positive around progress in terms of the evaluation measures under Aim 2. A large proportion of project leads saw major progress in 'increased focus on area of unmet need' and 'increasing the capacity of the research community.' There was the least perceived progress in novel gene-related therapeutics and partnerships with First Nations communities.

While there have been some notable projects, overall limited progress has occurred to increase community awareness and/or engagement, and/or better understanding of the societal and economic value of genomics in health care (GHFM Aim 3)

With fewer dedicated projects aligned to GHFM Aim 3 priority areas, there has been less perceived progress in this space. Under GHFM Aim 3, progress to date under ELSI, governance and Aboriginal and/or Torres Strait Islander health fell below expectations after five years into the GHFM. Many stakeholders indicated they anticipated the development of a national dataset to enable a more unified approach to GHFM Aim 3, however while the GHFM has supported projects to investigate governance and technology (via priority area 3.2) and contribute to the AGRD (via priority area 3.4), it has not directly supported the building of data infrastructure or assets.

Aim 3 project leads were proportionally the most optimistic about the progress made in their projects, however the small volume of projects and respondents compared to the other two aims likely caused a lower perception of progress overall.

There was mixed progress towards the shared evaluation measures

There were two measures shared by two or all three of the Aims. They were as follows:

- Research projects integrate partnerships with, and co-design by, Aboriginal and/or Torres Strait Islander people and communities.
- The community trusts, accepts and adopts new technologies and treatments.³⁸

The GHFM has enabled groundbreaking projects in First Nations genomics, GHFM projects as a whole have been inconsistent in their engagements with First Nations communities

Despite featuring across all three GHFM aims, the reported progress towards integrating partnerships with, and co-design by, Aboriginal and/or Torres Strait Islander people and communities was inconsistent. Many stakeholders in the sector argued that engagement with First Nations communities is a unique area that Australia can and should be leading. Recognising the importance of First Nations communities in the direction of the Australian genomics sector, the GHFM Roadmap included the underpinning consideration:

“Engagement and co-development with consumers and Aboriginal and Torres Strait Islander people will enrich genomics research design, delivery and implementation.”

While stakeholders valued the GHFM contribution to First Nations genomics with recent investment in dedicated projects, they argued that it was important for all projects to consider First Nations issues to ensure development of, and representation in, the body of genomics knowledge.

Opportunity 5 | Ensure all projects consider the appropriate involvement of, and potential impact on, First Nations communities

While there are a handful of notable projects seeking to improve Aboriginal and/or Torres Strait Islander health that are making great strides through engagement of First Nations communities, engagement by projects on other priority areas have inconsistently engaged First Nations communities. Engagement of First Nations communities offers a myriad of benefits for First Nations Australians as well as the project itself and should be considered in the conduct of every genomics research project.

Stakeholders were disappointed by the progress towards improving community understanding and engagement, but this may reflect the small number and early stages of relevant projects

Many stakeholders were generally pessimistic about the progress made towards improving the community’s understanding and engagement with genomics over the last five years. Comparatively, most project leads indicated making at least moderate progress towards the evaluation measures for Aim 3 in their project. The difference between data points is likely due to scale as:

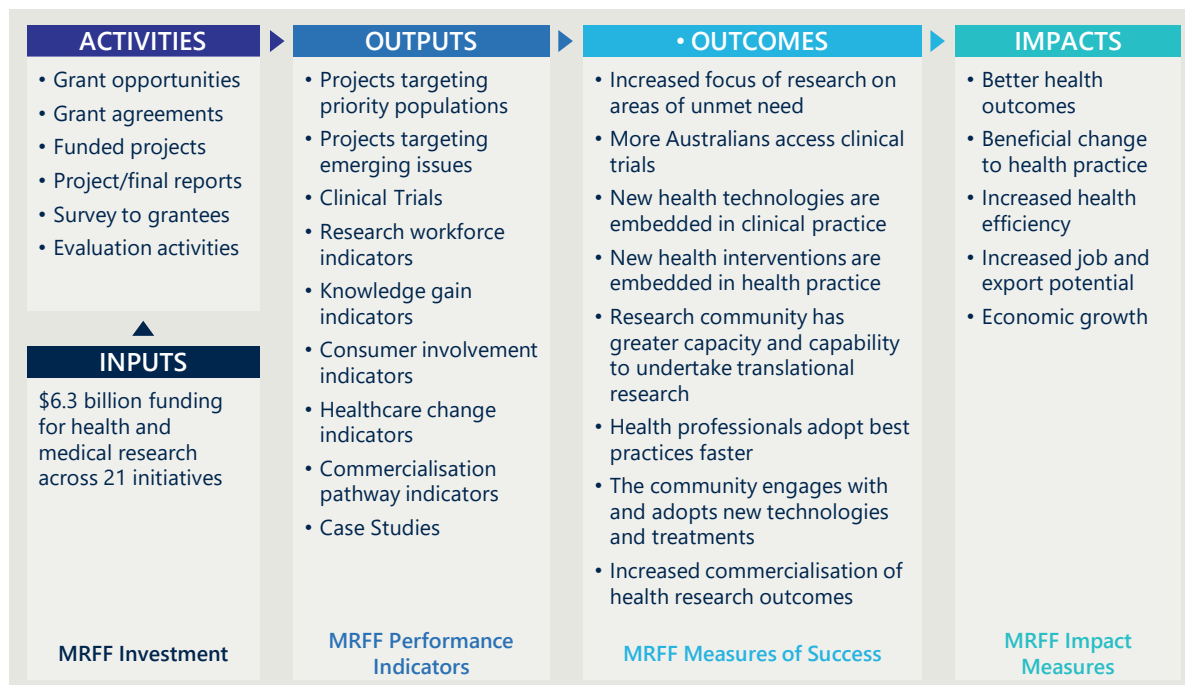
- As more projects draw closer to their finalisation there is likely to be an increase in communication with the public.
- Generating community awareness and understanding is a long-term venture requiring activities at scale, it will take consistent activity in the space to progress further.

³⁸ While GHFM Aim 3 did not include a measure of the exact same language the concept is similar in nature.

4.2.3 Progress thus far reflects outcomes of early translation activities

To assess progress towards impact under the MRFF, the MRFF Evaluation Strategy outlines eight measures of success (refer to Figure 17, below), which define outcomes to support assessment of impact.

Figure 17 | Program logic for the MRFF Learning and Evaluation Strategy

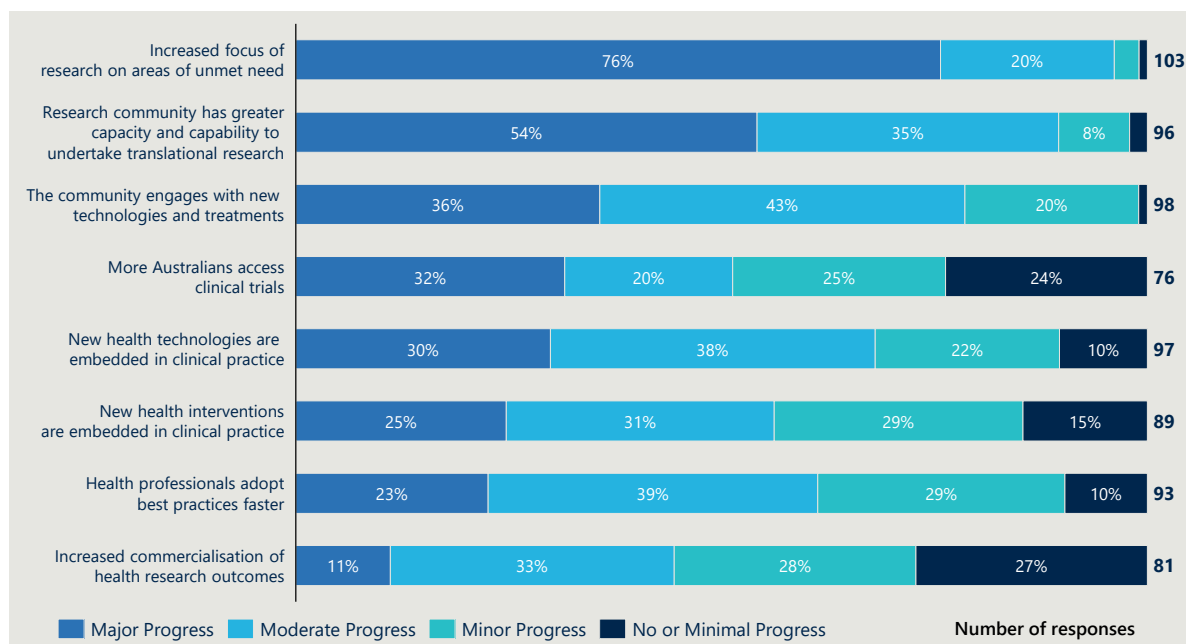


Source: Performance indicators towards the impact of the Medical Research Future Fund, March 2023.

Given the diversity in the measures of success, it was difficult for stakeholders to discuss overall progress against each of the measures of success. Through the survey, grant recipients were asked to comment on progress under all measures in the context of their projects, whereas through consultations, stakeholders spoke more generally about the progress of the genomics ecosystem over the last five years under the MRFF and GHFM.

Some measures of success relate to outcomes resulting from increased research activity, whereas the remainder relate more to changes in health practice. The former appears to have had more substantial progress made at this five-year mark. This theme was strong amongst stakeholders consulted, as well as grantees, with the most progress perceived in ‘increased focus on areas of unmet need,’ and the least in ‘increased commercialisation of health research outcomes’ (refer to Figure 18, overleaf).

Figure 18 | Progress reported by project projects leads to each of the MRFF Measures of Success



Source: Nous analysis of responses to the Grantee Survey, December 2023.

Figure note A: The priorities are ordered by highest proportion of 'Major Progress' responses to lowest.

Figure note B: The number of responses varies as a response to each measure of success was not mandatory and 'Not applicable' responses have been excluded from the Figure.

From this data and consultations with stakeholders there were strong signs that:

- Greater progress is being made in measures of success generally associated with early-stage research activities (Most notably: Increased focus of research on areas of unmet need. More Australians access clinical trials; and Research community has greater capacity and capability to undertake translational research).
- Less progress is being made in measures of success generally associated with late-stage translation activities and health system implementation.

This may be partially due to approximately half of the respondents to the survey indicating that their project was either basic or applied research rather than translation. Project leads of translation projects were far more positive in general on their progress against all measures of success.

There are strong signs of progress towards MRFF measures of success associated with early-stage translation activities

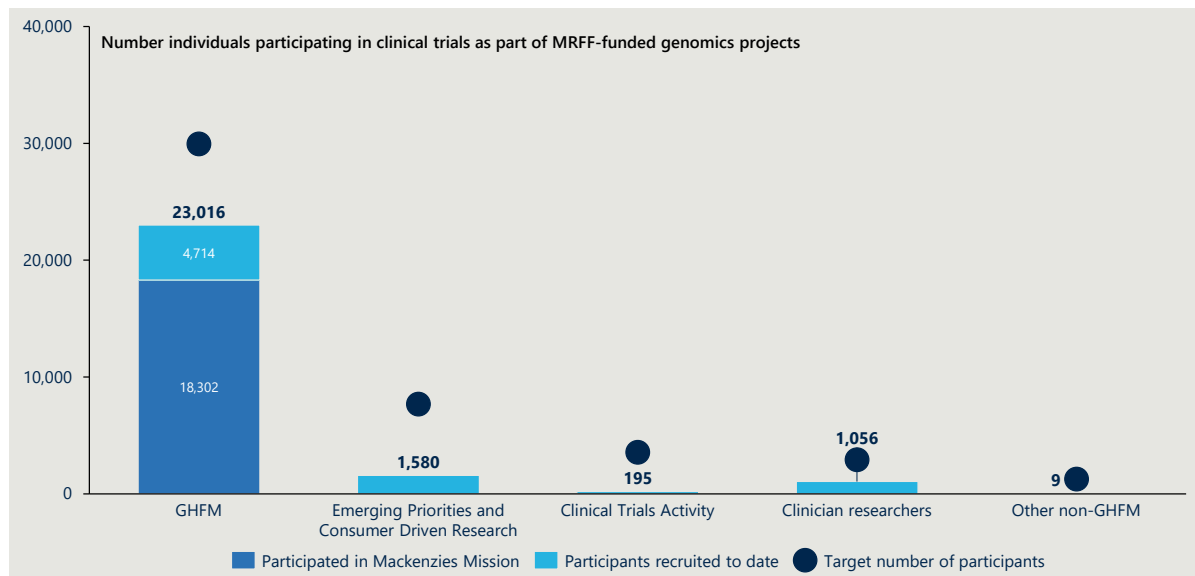
Increased focus of research on areas of unmet need

The GHFM is progressing well to increasing research in areas of unmet need. The MRFF Evaluation Strategy defines unmet need as 'serious health conditions whose diagnosis or treatment is not adequately addressed by existing options' and defines success where research identifies areas of unmet need, or leads to new health treatment, interventions, or diagnostics in these areas. The proportionately large investment towards rare diseases, cancer and infectious disease has evidenced progress in this area, with strong agreement among stakeholders that the MRFF and the GHFM have increased focus of research on areas of unmet need.

More Australians access clinical trials

Comparatively few projects have included a clinical trial, however many project leads felt their project was making progress towards them and would eventually provide more Australians access to clinical trials in Australia. Only six GHFM projects (15 per cent) have registered a clinical trial, compared to 20 amongst genomics projects funded by other MRFF initiatives (24 per cent).³⁹ As of the most recent progress reports (30 June 2023), over 25,000 Australians have been recruited to a clinical trial as part of an MRFF-funded genomics project, with a further 14,000 participants projected to be recruited by the end of these projects. Mackenzie's Mission has been the most significant recruiter of clinical trial participants, making up approximately 70 per cent of participants recruited to date (refer to Figure 19, below).

Figure 19 | Participants recruited through MRFF supported clinical trials



Source: Nous analysis of the [Australian New Zealand Clinical Trials Registry](#), extracted 28 February 2024.

Research community has greater capacity and capability to undertake translational research

The GHFM and MRFF have increased the scale and ability of the genomics community to undertake translational research.⁴⁰

With increased funding, naturally there has been an increase in researchers conducting genomics research, who have in turn developed their understanding of translational research. Additionally, experts are better placed to conduct high quality research.

Less progress is evident within late-stage translation activities and health system measures of success

Namely:

- New health technologies are embedded in clinical practice.
- New health interventions are embedded in health practice.
- The community engages with and adopts new technologies and treatments.

³⁹ This does not consider the broader context of research which can indirectly contribute to stronger participation of Australians in clinical trial.

⁴⁰ Translational research is defined as "the process of applying ideas, insights and discoveries generated through scientific inquiry to the treatment or prevention of human disease."

- Health professionals adopt best practices faster.
- Commercialisation of health outcomes.

While translation activities take time, these projects were on average 5 per cent closer to the completion of their planned milestones than the overall population. A few large projects were identified as coming close to clinical implementation, such as Acute Care Genomics and Zero Childhood Cancer. However, in general, stakeholders did not believe enough attention was being placed on embedding or commercialising interventions and technologies. As the Review will detail in later sections, several barriers have been identified that are impeding translational progress.

Opportunity 6 | Concentrate investments in research projects further along the translation pipeline and enhance support to researchers to navigate translation activities such as clinical implementation and commercialisation of projects

Within the genomics landscape, the GHFM is uniquely focused on translation research as compared to other major funders such as the NHMRC and ARC. However, despite this, nearly half of MRFF genomics projects have been in basic or applied research. While this balance has meant the GHFM has created a strong body of knowledge on which to build to reach the goal of the GHFM, there is a need to concentrate investments in later stage translation projects to increase the potential of translation within the GHFM's remaining five years.

4.3 Contribution of the MRFF to genomics in Australia

SCOPE

This section of the report describes the evidence and findings in relation to the following intent of the GHFM Review:

- I. How the MRFF has contributed to genomics research in Australia?

KEY FINDINGS

- The MRFF, primarily through the GHFM, has bolstered and supported a consistent pipeline of activity.
- The GHFM is supporting some systemic benefits within the genomics sector, but longer-term and transition funding is required to sustain its impact.
- The genomics sector is facing complex challenges particularly in overcoming persistent barriers to translation; the GHFM can play a stronger role to drive cohesive sector-wide efforts to overcome them.

OPPORTUNITIES IDENTIFIED

- Opportunity 7 | Support more collaborations between MRFF genomics research projects.
- Opportunity 8 | Strengthen the connections of health sector stakeholders (such as consumers, industry and health system) to design and delivery of MRFF genomics projects.
- Opportunity 9 | Consider greater investment in enabling research infrastructure to support effective translation.
- Opportunity 10 | Ensure policy research, including ethics and legal aspects are considered in every project.
- Opportunity 11 | Increase the degree to which MRFF genomics projects include realistic plans for implementation.
- Opportunity 12 | Improve cohesion of investments with those of other funding bodies to address translational and implementation challenges.

The GHFM's mission is:

"To improve the lives of Australians by accelerating research that delivers more effective testing, diagnosis and treatment; facilitates the adoption of new interventions; and consolidates Australia's international leadership in genomics."

The GHFM operates within the Australian genomics landscape described in section 4.1. This section describes stakeholder perspectives of the impact of the GHFM on the genomics landscape and the progress of genomics projects in Australia. The review has identified multiple areas or outcomes where the MRFF has:

- Made a significant impact or contribution to the realisation of the current state.
- Made a noticeable impact or contribution to the realisation of the outcome but there are opportunities to further enhance its contribution over the next five years.
- Struggled to make an impact or contribution but there is scope to do so in the next five years.

4.3.1 The MRFF, primarily through the GHFM, has bolstered and supported a consistent pipeline of activity

With the injection of over \$520 million into Australian genomics research in the last five years (\$273 million, 53 per cent, coming from the GHFM), there is little doubt that the GHFM and MRFF has had a valuable impact on the Australian genomics landscape.

The GHFM has provided a necessary injection of funding

The GHFM's \$500.1 million planned contribution to genomics research has been significant and an important addition to the genomics landscape in Australia. Many research initiatives may not have had funding to progress their project without the investment of the MRFF. For projects funded under the MRFF (including GHFM and non-GHFM funded projects), over 80 per cent of grantee survey respondents indicated their project was unlikely or very unlikely to proceed without the MRFF's support. Grantees from the survey were also overwhelmingly positive on the impact of the MRFF on achieving outcomes in their projects, with over 90 per cent signalling the MRFF had a major impact on their research.

With a little over half of the GHFM's funds invested to date (and complemented by \$264.08 million in genomics investments through other MRFF initiatives), there has undoubtedly been significant advancements in the number of projects investigating genomics-related research (examples of projects the MRFF has enabled are detailed in Appendix A). Many stakeholders felt the GHFM and the MRFF had made a 'transformative' and necessary contribution to genomics in Australia, particularly in the context of rare disease. A significant majority of survey respondents signalled the GHFM and, to a lesser extent, non-GHFM MRFF investment has had a major impact on advancing genomics research in Australia. Some stakeholders suggested this was a natural phenomenon as they found increased investment enabled opportunity growth and made resource expansion more affordable and accessible. Whereas other stakeholders felt advancement was due to a targeted focus on priority areas within genomics which contributed to highlighting opportunities for more collaboration between researchers.

The GHFM has created greater certainty for the genomics sector

The consistency of a ten-year fund has fostered greater certainty and a steady foundation for genomics research into the future. Stakeholders suggested the GHFM has provided an important 'ripple effect,' cementing Australia as a key country invested in genomics. Research organisations noted the increased activity in genomics has provided them the opportunity to scale up their operations and has provided incentive for non-government investment in genomics. Whilst the GHFM has not substantially invested in research infrastructure to date, the increased scale and consistency of funding has allowed research facilities to better justify investment in genomics facilities and capacity.

4.3.2 The GHFM is supporting some systemic benefits within the genomics sector, but longer-term and transition funding is required to sustain its impact

The GHFM has built capability and capacity, highly concentrated around funded projects

The dedicated investment towards genomics has created an increase in researchers and research institutions with a focus on genomics. Most stakeholders agreed the funding has supported a significant uplift in the number of research projects being conducted and the number of scientists working on them, which has accordingly grown the capability of the sector to perform genomics research. Additionally, a greater number of researchers and groups within the sector are seen to be focusing on genomics.

The capacity and capabilities fostered by genomics research, however, has been concentrated to the jurisdictions or research institutions in which GHFM or other MRFF genomics projects had been based.

Stakeholders were concerned the capability and capacity to deliver the services created and established by genomics research would remain only in the places they were piloted and therefore would not be accessible for most Australians. The reasons for the lack of diffusion of capabilities across the broader health system are complex, often specific to the projects and multifactorial, but namely include:

- Different ethical, legal and social frameworks and requirements across jurisdictions.
- Different funding arrangements across jurisdictions.
- Availability and capability of the workforce required across jurisdictions.

Without conscious effort to fund genomics researchers to plan and execute capability transfer activities there is a risk that the innovations and successes of MRFF will not be available to all Australians.⁴¹

Collaboration within the genomics research sector has improved

The GHFM funding principles state that funded activities should:

'enhance collaboration and translation across the research and health systems, particularly across other MRFF initiatives', and collaborate with "... international contributions".

The GHFM has organically strengthened collaboration within the research sector by providing sufficient capital to reduce competitive forces and providing the capital for projects to scale. Whilst grant programs require an element of competition, with greater funding to go around, stakeholders perceived there to be a more collaborative feel within the local sector, encouraging people to work together.⁴² To date, the MRFF has funded many projects that are complementary, however the MRFF has not provided the structures or incentives to facilitate these projects to identify synergies and collaborate. Where collaboration between MRFF projects has occurred, it has been through the individual efforts of the researchers or other organisations involved and the costs of the necessary coordination and governance activities have been borne within the relevant projects or by the organisations delivering them. The most formalised example of collaboration can be seen in the GenSCAN Consortium, involving five projects focused on advancing Newborn Screening in Australia (refer to Case-in-point 1, overleaf).

⁴¹ While it is not the GHFM's responsibility to fund health service delivery, the GHFM has an interest in funding sufficient capability transfer activities to secure the scaling of its successes and innovations.

⁴² It was noted that collaborations are complex to establish due to a number of reasons including legal and commercial challenges. Accordingly, many stakeholders felt that the time from being notified of a grant opportunity to application due date made it challenging to consider and organise collaborations.

Case-in-point 1 | Genomic Screening Consortium for Australian Newborns (GenSCAN)

Building a consortium to maximise collaboration

Stakeholders have referred to GenSCAN as a strong example of project collaboration enabled by the GHFM. The GenSCAN Consortium consists of five individual projects that individually received funding under the MRFF. With support from Australian Genomics, project lead investigators co-ordinated to create a consortium to leverage a common mission of researching the use of genomics in newborn screening

Grants and Institutions:

\$2,941,351, University of Adelaide

\$2,117,960, University of Sydney

\$2,999,919, Murdoch Childrens Research Institute (MCRI)

\$2,998,078.35, MCRI

\$2,954,189.32, University of Sydney

The GenSCAN consortium has developed a valuable system for collaboration and support. Project leads from each project make up a steering committee which meets monthly as a way of strengthening coordination and driving broader change. As a collaborative forum, GenSCAN members consider common research and policy considerations, including ethical, legal and social issues, health policy and economics, and stakeholder engagement.

The GenSCAN consortium demonstrates the value in bringing together projects and researchers sharing a common goal. Stakeholders have identified an opportunity for the GHFM to play a more involved part in facilitating networking for all funded projects so that strong collaboration, as demonstrated by GenSCAN, can exist across the board.

Table note: A full case study of the GenSCAN Consortium is detailed in **Chapter 5 Case studies**.

Some stakeholders were concerned that many projects were not aware of, or not engaging with, complementary or similar projects being undertaken by other organisations.

Improvement in collaboration could be further bolstered by the GHFM more formally, with more intentional focus on encouraging researchers to collaborate and leverage one another's experience. Feedback from grantees and research organisations highlighted that the governance requirements, such as multi-institutional agreements and the need to dedicate resources to coordinate activities across the grants, had made it burdensome to consider and organise consortia approaches to opportunities.⁴³ Some stakeholders noted that the GHFM's project-to-project based funding meant funding was not available to support coordination activities across MRFF projects funded separately and had to be borne by the grantees outside of the grant or by a third-party organisation.

The potential to leverage overseas research efforts is considerable. Countries such as Canada and United Kingdom invest significantly in genomics. Stakeholders demonstrated mixed views in terms of the GHFM and MRFF's impact on encouraging and connecting researchers with overseas collaborators, compared to local research collaborators. Two barriers to enhanced international partnerships were data privacy and policy hurdles. It was also noted that the lack of a 'single node' of scale for overseas entities to connect through was preventing identification and facilitation of collaboration opportunities.

⁴³ Stakeholders acknowledged that many governance challenges associated with consortia are not unique to the GHFM.

Opportunity 7 | Support more collaborations between MRFF genomics research projects

The benefits of collaborations between researchers are well documented. While there are important examples of successful collaborations between MRFF genomics researchers, there is scope to further increase the frequency and size of those collaborations.

Collaboration with the broader health sector is not formalised and consistent

The GHFM funding principles stated that activities funded under the GHFM should⁴⁴:

“promote engagement with service delivery, program and policy partners to facilitate adoption of safe and effective genomics technologies.”

Collaboration can occur at multiple levels, the key types of collaboration explored by the Review included:

- Collaboration between the department and the sector on the development and implementation of the GHFM priorities.
- Collaboration between genomics sector stakeholders on sector-wide issues and developments.
- Collaboration between genomics sector stakeholders and project leads on project design, execution and/or reporting.

The GHFM can play a stronger leadership role in enabling collaboration more broadly across the genomics sector. Where individual projects were leveraging expertise of local and international contacts, this finding was not widespread and demonstrated as a core feature of the GHFM. Many stakeholders felt that there could be more formalised channels for collaboration, with specific stakeholder groups.

Key sources of collaboration⁴⁵:

- **With the clinical community and health services.** It is critical to involve the health care system throughout the life of a project and grant opportunities, to ensure feasibility, scalability, and real-world relevance. The review highlighted this was not actively encouraged under the GHFM, particularly with jurisdictional health departments and service providers. From grant selection to study design or implementation planning, there is significant untapped potential in involving those on the ground to facilitate implementation into health service delivery. An observation by many stakeholders was the need for broader stakeholder involvement within GHFM decision making. Implementation expertise was perceived to be a gap in the previous GHFM Expert Advisory Panel, which was seen to have very strong expertise in research, rather than a diverse membership of health system experts.
- **With government agencies.** Most stakeholders from Commonwealth and jurisdictional government agencies stated they had not been consulted on many projects. While the priority areas that GHFM grant opportunities address are pre-determined by the GHFM roadmap, the design of grant opportunities would benefit from consultation with relevant Commonwealth and jurisdictional government agencies to ensure appropriate consideration of the feasibility of health system integration and to encourage complementary investments in the health system. These stakeholders argued that the introduction of a policy, regulatory and health system funding lens would improve the design of projects and ensure they can be translated on completion.
- **With industry.** Most industry stakeholders and some non-industry stakeholders, raised concerns that the private sector had not been leveraged to the fullest extent possible through the GHFM.⁴⁶ Only 26

⁴⁴ GHFM Roadmap.

⁴⁵ In addition to the groups identified the GHFM Review identified strong examples of collaboration with consumers, however this is highlighted in a separate finding on page 48.

⁴⁶ Multiple representatives of non-industry stakeholders acknowledged the greater role industry could play in GHFM projects.

(of 170) MRFF genomics projects reported cash or in-kind contributions from private sector organisations, and not-for-profit organisations received a grant through the MRFF for genomics project.⁴⁷ Industry report challenges in meeting the eligibility criteria for MRFF funding, particularly early-stage companies.⁴⁸ Most industry stakeholders felt there had been a lack of consultation with industry on the agenda of the GHFM. Industry and some other stakeholders also argued there is potential for industry to complement research efforts and support the commercialisation of interventions and therapies.⁴⁹

- **With priority groups including First Nations.** Both international and national stakeholders generally perceive Australia as a leader in Indigenous genomics, though this view is largely driven by more concentrated efforts in a few dedicated projects. The Review has seen variable involvement of First Nations people and other priority groups within research projects. Some stakeholders reflected that while a small number of projects were directly focused on First Nations people, migrant groups or priority groups, engagement or consideration of these groups was not a requirement of study designs. Until the recent introduction of The Aboriginal and Torres Strait Islander Genomic Advisory Group, collaboration with Indigenous Australians was seen as infrequent, causing projects to lack consideration of the distinct needs of Indigenous people.

Opportunity 8 | Strengthen the connections of health sector stakeholders (such as consumers, industry and health system) to design and delivery of MRFF genomics projects

Non-researcher stakeholders in the health system bring a unique insight and capabilities that researchers should consider in the design and delivery of their research projects. While there are some examples of exemplary use of inter-disciplinary research teams, there is scope to improve the regularity of inter-disciplinary collaborations.

The GHFM has attracted some additional co-investment, but this has been primarily through in-kind support

Co-investment does not only bolster the available funds within projects but contributes to stronger partnerships and buy-in from stakeholders, to accelerate outcomes. This is recognised in the MRFF funding principles, which state:

“Encourage multi-government and agency, private sector and philanthropic co-investment to maximise program outcomes.”

Similarly, the GHFM funding principles state that activities funded under the GHFM should:

“leverage funding to maximise outcomes through collaboration with philanthropic, industry and international contributions, including the translation of genomics research into practice.”

There has been some degree of co-funding within MRFF genomics projects, with at least 64 of the 170 projects (38 per cent) receiving a total of \$55 million from non-MRFF sources in their first five years.⁵⁰

The majority of co-investment (71 per cent) was received via in-kind services being provided to the project, primarily by the institutions directly funded for the projects, which is reflected in Group of Eight

⁴⁷ Outside of MRIs and universities, only two not-for-profit organisations were successful in obtaining an MRFF grant for genomics project, Epilepsy Foundation (Victoria) and the Australian Genomics Cancer Medicine Centre.

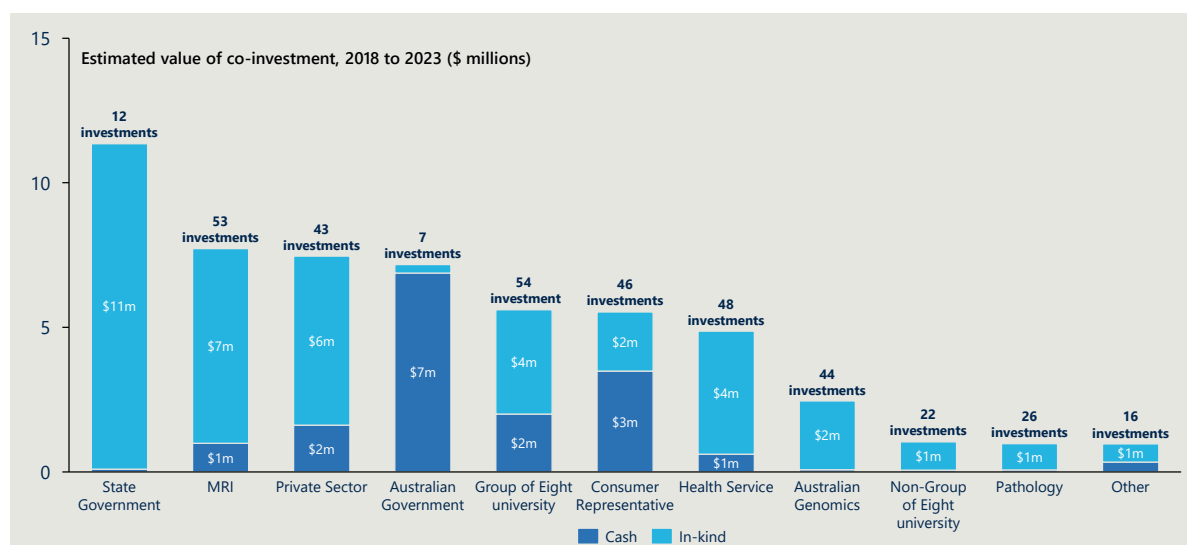
⁴⁸ The Review notes that private corporations are able to seek Eligible Organisation (EO) status under the MRFF, subject to meeting separation of duties requirements.

⁴⁹ The Review notes that a national consultation on the GHFM Roadmap and Implementation Plan was delivered from December 2020 to April 2021. The department sought engagement from 14 industry stakeholders for the national consultation and received written submissions from 4 industry stakeholders.

⁵⁰ There was a small difference in co-investment between GHFM (\$29m) and non-GHFM (\$26m) projects.

universities and MRI being the most frequent co-investment partners⁵¹ however state governments contributed the greatest value (refer to Figure 20, below). Stakeholders, both within and outside of industry, highlighted there could be more collaboration with the private sector.

Figure 20 | Co-investment in MRFF-funded genomics



Source: Nous analysis of progress reports submitted by grantees until 31 December 2023.

Figure note: Nous extracted reporting from progress reports as entered by grantees, Nous did not undertake any assurance or quality assurance of the data entered by grantees. It was clear there may have been different interpretations of the relevant period for which the grantee was reporting.

Consumer engagement in MRFF Genomics projects has seen improvement over time

Whilst there has been a trend towards greater consumer involvement, this could be more consistent across all MRFF and GHFM projects. Overall, 56 per cent of surveyed GHFM project leads identified that consumers were involved in their project (65 per cent for non-GHFM project leads). There has been growth in the frequency of consumer engagement with genomics projects growing from 55 per cent across 2018 and 2019 to 64 per cent in 2023, highlighting room for continued growth.

The MRFF recognise the importance of consumer involvement as “the ultimate funders, users and beneficiaries of health and medical research and innovation,” consumers have a right to be involved in research and valuable contributions to make.

The department published Principles for Consumer Involvement in Research Funded by the Medical Research Future Fund in March 2023 which represented advice from the MRFF Consumer Reference Panel that consumers should be involved:

- In every type of research.
- At all stages of research.
- In partnership with researchers.
- Effectively.
- Sensitively and safely.
- With broad diversity and equality.

⁵¹ State governments contributed the greatest value; however, this was almost entirely through in-kind contributions.

Many stakeholders were aware of this targeted focus, noting the MRFF had done a lot of work to ensure consumer voices were heard, but more could be done. Consumer representative groups were generally positive on the increased level of consumer involvement they had seen through the GHFM, but they brought two qualifying observations:

- GHFM project leads could show greater understanding of the difference between consumers who bring a lived experience compared to policy expertise through consumer and disease-specific representative groups.
- There was a need to better publicise research and generate visibility amongst a broad cohort of consumers, not only with a select few directly consulted by the project.

In survey responses there was evidence of engagement across all stages of research from design to dissemination. Thirty-two consumer representative groups actively supported 46 genomics projects through cash or in-kind contributions with an estimated value of \$5.5 million. Most consumer representative groups were associated with one to two projects each.

4.3.3 The genomics sector is facing complex challenges particularly in overcoming persistent barriers to translation; the GHFM can play a stronger role to drive cohesive sector-wide efforts to overcome them

The genomics sector needs stronger national coordination to align research and translation activities

The Science, Innovation and Research Budget papers set out the expectation that the MRFF will provide strategic coordination and direction for health and medical research.⁵² Further to this the GHFM implementation plan recognises the importance of a nationally coordinated approach to genomics investments for enabling the GHFM to succeed:

“A nationally coordinated approach to leverage core research capabilities can support all funded projects to drive activity and outcomes; harmonise project approaches; and develop, curate and manage a legacy dataset for future research use.”

The underpinning consideration of the GHFM affirms the GHFM’s intent to support a cohesive and collaborative approach to translation of genomics research:

“The GHFM will support a cohesive and collaborative national approach to the implementation of genomic medicine as standard of care...”

The genomics sector is complex and at times, operating in silos. The aims and objectives of the GHFM are bold and will require more intentional planning and prioritisation to achieve them over the remaining years of the program. While most stakeholders considered the GHFM’s funding fundamental to driving the sector towards unified priorities, some reflected that more sector coordination and leadership was needed to ensure the sector could grow with impact and presence. Some stakeholders considered that the GHFM had so far missed the opportunity to bring the sector in alignment behind key national priorities. With the number of aims and priorities of the GHFM (refer to Finding 4.1.1, page 24) the sector is generally continuing to work individually to meet their own goals, rather than moving towards nationally harmonised goals.

“Being able to leverage the resources of Australian Genomics was critically important”
- Project Lead

⁵² Department of Industry, Science and Resources, ‘Science, innovation and Research Budget Tables 2023-24’.

Most stakeholders were unable to distinguish between the activities of the GHFM versus Australian Genomics and demonstrated mixed perspectives on the role of each in providing national leadership. While many reflected that they had anticipated the GHFM would take on a greater sector coordination and leadership role, they generally saw Australian Genomics supporting elements of this coordinating role. Stakeholders also noted that Australian Genomics has provided significant support through supporting collaborations, public communications, and advocacy to governments (locally and internationally). Given the important role of Australian Genomics and the anticipated formation of Genomics Australia, greater clarity must be provided to the sector on the roles and responsibilities of genomics funders and leaders.⁵³

The accelerated activity in genomics has highlighted system gaps that are impeding translation, which if not addressed will undermine the sustainability of gains made

The MRFF's funding principles require that funding is used to:

"Fund transformative game-changing research that is balanced by investment in systemic sector improvements."

The GHFM Roadmap observes that:

"Coordination of core capabilities common to all GHFM-funded projects will achieve economies of scale, avoid duplication of effort, mitigate risk, and improve project implementation, evaluation and effectiveness."

Across both the research and health systems, there are a number of systemic factors that present a significant barrier to implementation and commercialisation of genomics research outputs. The GHFM thus far has funded some isolated research projects to investigate some of these barriers but has not directed funding or coordinated capabilities to overcome these research translation barriers. The most prevalent impediments to successful genomics research translation identified by stakeholders were:

- Workforce
- Health system preparedness
- Infrastructure
- Data storage and data sharing
- Ethical, Legal and Social Implications.

Mackenzie's Mission provides a demonstration of challenges in the wider system facing genomics projects in Australia and the complexities projects must navigate to embed sustainable innovations (refer to Case-in-point 2, page 56).

Workforce and Health system preparedness

The continued innovation in human genetics and genomics requires expanding our current workforce to keep up with the demand of genomic technology.⁵⁴ At present, there appears to have been insufficient planning and preparedness to implement many new genomics-related technologies in Australia. Genetic counsellors are a critical workforce element to consider, with the demand that new technologies may place on the profession likely to exceed supply and current capability.

Health services and jurisdictions commented throughout the review that greater engagement by the MRFF and funded projects is needed to facilitate planning upfront. Government stakeholders felt these issues

⁵³ The establishment of Genomics Australia was announced in 2022 for commencement in 2024. In 2023 the establishment of Genomics Australia was delayed until July 2025.

⁵⁴ Industry Genomics Network Alliance, *Valuing the impact of genomics on healthcare in Australia*, December 2021.

were only being considered once studies were nearing completion. The International Expert Review of the GHFM Roadmap and Implementation Plan recommended that:

“Research should include working with the Minister for Health, states and territories to develop a clinical service delivery system that is willing and able to embed genomics into health care.”

Infrastructure

Appropriate infrastructure is necessary to support genomics advancements. The GHFM Implementation Plan recognised this stating:

“It will be important to align the GHFM’s genomics research resources, tools and infrastructure to national endeavours; ... ensure that the needs of genomics in health are considered in the context of national infrastructure investment.”

“Australia is not keeping pace with resources available to some of our collaborators.”

- Project Lead

Unlike its international comparators, the GHFM has not invested in infrastructure directly (refer to Finding 4.1.3). However, indirectly the increased level of activity in the sector spurred by the GHFM has in turn increased the utilisation of existing infrastructure, providing strong business cases for expanding or establishing genomics infrastructure. These investments have been managed downstream of the MRFF without oversight or consideration by the department and other decision-makers.

Without centralised coordination of resources to invest in enabling infrastructure, the improvements to Australia’s genomics infrastructure will continue to be piecemeal and may be insufficient to implement national programs going forward.

Data storage and data sharing

The GHFM can play a greater role in ensuring appropriate data collection and sharing facilitates collaboration and progress of funded genomics projects. Given the ethical and regulatory challenges around genomic data capture and sharing, data policies are seen as a challenge within the international genomics sector. Many other countries have moved to provide more standardised approaches across new genomics projects, to allow for centralised data assets which enable more advanced research and provide greater insights.

“We met challenges with contracting between Australia and International groups, relating to the sharing of IP and the differences in governance and international regulatory requirements.”

- Project Lead

As part of the national leadership, data policies and coordination of a national data set were raised as key opportunities that would resolve some critical barriers to data sharing and collaboration. Some stakeholders conveyed that a consolidated data set may also increase the diversity of data available to researchers, ensuring inclusivity of minority genomes within Australia. Additionally, centralised consent policies or clear ethical guidelines were thought to allow for more standardisation within Australia and encourage broader collaboration locally and internationally.

Broader infrastructure, ELSI research and health sector capacity is needed to support sustainment of GHFM innovations and successes

Mackenzie's Mission, or the Reproductive Genetic Carrier Screening study was frequently cited as a key project under the GHFM and MRFF. Many stakeholders described the study as a key example of the transformative nature of the GHFM, however it does also provide insight into the challenges of implementing genomics outputs in the Australian clinical landscape.

Grant value: \$20,000,000

Lead Institution: MCRI

In 2018, the federal government announced \$20 million in funding towards 'Mackenzie's Mission', a study to garner evidence for a national reproductive genetic carrier screening program. The screening would give prospective parents information about the likelihood of having a child with a known severe, childhood-onset genetic condition.

A submission to MSAC failed to garner support for public funding of expanded reproductive carrier testing for over 1000 genes associated with autosomal recessive and X-linked conditions, largely due to challenges relating to implementation, ethics, social and system readiness.⁵⁵

Cited issues included:

- A lack of funding for genetic counselling in the private sector.
- Access issues for rural or remote patients.
- Lack of consideration of culturally appropriate approaches to testing, including for Aboriginal or Torres Strait Islander peoples.
- Storage and transfer processes for genomic data, including large-scale data infrastructure typically appropriate to support a national screening program.
- Concerns that the value-for-money of the testing proposed may be overestimated.

These challenges are rather indicative of the broader infrastructure and ethical elements that must be researched, considered, and overcome, prior to successful translation of genomics research. Without addressing them, appropriate GHFM projects risk finishing short of their original goal with additional effort required to solve implementation concerns which may not necessarily be funded.

In November 2023, new Medicare funded item numbers were made available for genetic carrier testing of three genetic conditions: cystic fibrosis, spinal muscular atrophy and fragile X syndrome. Despite positive momentum, implementation considerations remain a challenge for the sector.^{56,57}

⁵⁵ Medical Services Advisory Committee, Public Summary Document, Application No. 1636 – Expanded reproductive carrier testing of couples for joint carrier status of genes associated with autosomal recessive and X-linked conditions. July 2022.

⁵⁶ Department of Health and Aged Care, Reproductive carrier testing for cystic fibrosis, spinal muscular atrophy and fragile X syndrome. November 2023.

⁵⁷ In 2021, the Australian Government announced new Medicare rebate items for five pre-implantation genetic testing services relating to genetic disorders, dubbed 'Mackenzie's Gift'.

Opportunity 9 | Consider greater investment in enabling research infrastructure to support effective translation

Like the GHFM's international comparators (refer to Chapter 4.1), the GHFM could consider additional investment in enabling research infrastructure such as laboratories, equipment, digital and data infrastructure, or other facilitators of genomics projects.

Ethical, Legal and Social Implications

There are complex ethical, legal and social implications (ELSI) to be considered before many genomics technologies or interventions are implemented.⁵⁸ The development of genomics technologies has raised many ethical and legal concerns for society, and policy and regulation has not necessarily kept pace with these developments. While stakeholders applauded GHFM for including ELSI as a priority area, they noted it had received significantly less funding (refer to Finding 4.1.1, page 23). Several stakeholders were concerned that the findings and insights gathered from ELSI projects were not being effectively communicated to, and considered by, other MRFF projects.

Opportunity 10 | Ensure policy research, including ethics and legal aspects are considered in every project

For a research project to transition effectively into the health system, ELSI and policy issues are critical to resolve. And while the GHFM through several dedicated projects has developed great insights into many of these challenges, there is a need to ensure these insights are understood and applied by researchers leading MRFF genomics projects.

The transition from research funding to sustainable funding in the health system is challenging for genomics projects

The MRFF funding principles state that MRFF funding should:

Consider favourably proposals that have collaboration, translation and scalability features to ensure the MRFF is transformative, and effort is enduring.

Ultimately, to make a transformative and enduring impact on human health MRFF projects must transition from research-based funding through the MRFF to ongoing funding within the health system.

While it is important to acknowledge that translation of genomics technologies takes time and will always face significant challenges, there is a concern in the sector that MRFF-funded genomics projects are not sufficiently prepared for translation at their conclusion. If not addressed there is a risk that otherwise successful MRFF genomics research projects may be unable to secure an appropriate funding stream through the health system.

Many stakeholders were optimistic about some of the innovative and new genomics concepts being demonstrated through MRFF genomics projects. They argued it is a risk the current MRFF will serve only as a temporary solution if the fund isn't able to also support translational outputs for long term implementation. This risk has been

“Research studies need to be mindful of the way Government evaluates funding submissions ... Studies need to be designed with consideration of the translational and economic impact of the research.”

- Stakeholder

⁵⁸ Department of Health and Aged Care, International Expert Panel Review of the GHFM Roadmap and Implementation Plan, November 2020.

demonstrated with some projects being forced to seek alternatives to fund additional efforts to gather necessary data to file stronger regulatory and funding applications. The Zero Childhood Cancer Project (ZERO) is a key example of a project facing limbo across the research and health system funding divide (refer to Case-in-point 3, below).

Case-in-point 3 | Zero Childhood Cancer Project

A need for sustainable funding solutions

The Zero Childhood Cancer program (ZERO) is a sizeable and complex program that relies heavily on funding for sustainability. ZERO undertakes genomics research to inform, evaluate and measure the effectiveness of cancer treatments in patients. The program's scope has expanded to all Australian children, adolescents, and young adults affected by medium, high, and very high-risk cancers.

Grant value: \$54,800,000

Lead Institution: University of New South Wales

ZERO represents a significant change in the current treatment paradigm of children with cancer. As one of the world's largest precision medicine studies in paediatric oncology, the investment to date has been vital to accelerate progress. Children enrolled on ZERO, have a sample of their cancer analysed and where possible, assessed against treatment strategies.

Following a national clinical trial program, in 2020 the MRFF committed \$54.8 million to expanding ZERO's genetic testing platform. This contribution was bolstered by a \$12.2m commitment from the Minderoo Foundation, allowing the extension of the ZERO genetic testing program to all Australian children diagnosed with cancer, by 2023.

Stakeholders have noted the transformative nature of the ZERO program. Concern has been raised however, that without long term and sustainable funding through the health system the program may be at risk beyond the committed funding period, with one stakeholder commenting 'it is too important not to keep going'.

The Medical Services Advisory Committee (MSAC), as a health technology assessment pathway for new medical services under public funding, is a critical step to achieve funding and translation in many cases. Other processes such as through the Therapeutic Goods Administration and the Office of the Australian Gene Technology Regulator can be equally as critical for successful implementation. These processes require a very deliberate and careful approach to successfully complete, there are many professionals in industry focused specifically on facilitating this process.⁵⁹

Many stakeholders reflected that researchers are often unaware of how to navigate end-stage translation and commercialisation pathways. They noted that GHFM does not actively equip researchers to tailor their study designs and protocols to the requirements of regulatory and funding bodies. Nor does it sufficiently require researchers to consult the appropriate experts and plan a realistic path to implementation.

Opportunity 11 | Increase the degree to which MRFF genomics projects include realistic plans for implementation

The findings of the GHFM Review tend to indicate that many projects are designed without adequate sufficient consideration of implementation. As a result many stakeholders are concerned they will not be translatable upon conclusion. To avoid this scenario it is essential the GHFM encourage, support and require researchers to adequately prepare for, and allow time to work through, complex implementation challenges.

⁵⁹ Stakeholders argued these processes present unique challenges for genomic technologies that haven't been resolved. However, the regulatory design of these processes was not within scope.

The GHFM funds discrete clinical translation activities, but this approach is not sustainable long term

“While crucial for advancing genomic research, the scheme's success hasn't matched the rapid pace of technological advancements or been effective in expediting the transition into economically sustainable clinical practice change and in establishing pathways to commercialisation impact of the research.”

- Stakeholder

The MRFF's current model invests in singular translation projects without a pipeline of investment to support the full embedding of the projects. Most stakeholders reflected that to translate a new genomic technology would require many orders magnitude more funding than what is provided through the GHFM. Without coordinated commitments from the health sector to support a service in the health system after a proof-of-concept is demonstrated by a genomics research project, the GHFM's successes will face a funding cliff before being able to be implemented in the health system.

This concern in the sector was further reflected with an acknowledgement that the distinction between research funding, funding for enablers, and ongoing health sector funding, was not well defined. Currently there is a perception that with increasing costs of providing core health services, there may be little appetite from the health system to sustainably fund these programs.

Further to this, there is a need to coordinate multiple investments together to solve the various issues that need to be resolved to successfully embed a genomics service or technology in the health system.

Opportunity 12 | Improve cohesion of investments with those of other funding bodies to address translational and implementation challenges

The challenges that must be overcome to successfully implement a new genomics technology or service across the health system are multifactorial and large. In the Australian health system, no single funder or stakeholder has the resources or capability to resolve all these challenges. GHFM investments to translate research into the clinic should be complementary to the initiatives, investments and structures within health services to ensure sustainable implementation that is not reliant on research funding to deliver services.

5 Case studies

This chapter showcases some of the GHFM's projects and the progress they have made to this point.

Nous worked with the department and the GHFM Review Panel to identify relevant project case studies to demonstrate the activities enabled by MRFF and GHFM funding. Project leads nominated their interest through the Grantee Survey, and were engaged in the development of the case studies. Case studies were also informed by progress reports, public documents and websites.

Each case study, which can be read on the following pages, has been selected to highlight a unique lesson or highlight a positive impact of the GHFM.⁶⁰ The five selected projects were:

- Australian Pathogen Genomics Program (AusPathoGen, formerly Precision Public Health in Australia through Integrated Pathogen Genomics).
- Shining Light into the “unknown” on Indigenous and non-Indigenous Australians with Cancer of Unknown Primary (CUP).
- Pathways to benefit for Indigenous Australians in Genomic Medicine.
- The Genomic Screening Consortium for Australian Newborns (GenSCAN).
- Personalised medicine in the treatment of complex autoimmunity and autoinflammatory disease.

⁶⁰ The projects were not selected to be a representative sample of the 170 projects and are presented in chronological order.

Australian Pathogen Genomics Program (AusPathoGen)

PROJECT LEAD	GRANT PERIOD	MRFF VALUE	INSTITUTION	GHFM PRIORITY
Prof. Benjamin Howden	2021 - 2025	\$9,999,499	University of Melbourne	Infectious Disease ¹

Infectious diseases represent a significant threat to public health in Australia and globally. The vital role of integrated pathogen genomics data in Australian healthcare became evident during the COVID-19 pandemic, with outbreak surveillance critical to guide public health measures. With funding made possible under the GHFM's Pathogen Genomics grant opportunity, AusPathoGen is leveraging the successes from 2020 to integrate pathogen genomics into public health at a national level.

Combating the rise of infectious diseases in Australia

AusPathoGen's research focuses on pathogens that cause human diseases, positioning it uniquely under the GHFM. This research is vital for developing a deeper understanding of pathogens, to support the diagnosis, surveillance and control of infectious diseases. AusPathoGen will implement national genomics-based responses to key pathogens of public concern, initially focussing on:

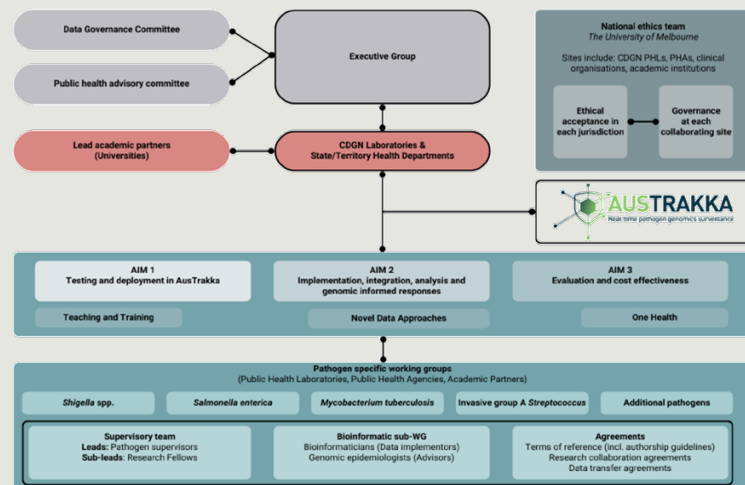
- *Shigella* resistance
- *Salmonella enterica*
- *Mycobacterium tuberculosis*
- Invasive group A *Streptococcus*
- Antimicrobial resistance

Research supported by strong governance and co-design

AusPathoGen has a clear goal to implement national genomics-based responses, underpinned by a strong foundation of governance and collaboration (see Figure 1 below).

To ensure implementation success, the project's governance structure included representation from a wide variety of stakeholders, ranging from policy and governance experts, public health stakeholders and academics. To maintain a national view, projects under AusPathoGen have been co-designed with jurisdictions and public health laboratories across the country. A national and coordinated approach is critical, as Professor Benjamin Howden pointed out; diseases don't respect borders.

Figure 1 | AusPathoGen governance and relationships



National Surveillance using AusTrakka

AusTrakka underpins the AusPathoGen program. The national surveillance platform has enabled equitable data sharing, analysis and reporting. A national ethics approach has bolstered the data sharing capabilities, as has a community of practice around analysis and reporting.

¹ Priority Area | 1.4: Infectious Disease

Shining Light into the “unknown” on Indigenous and non-Indigenous Australians with Cancer of Unknown Primary (CUP)

PROJECT LEAD

Prof. Christos Karapetis

GRANT PERIOD

2021 - 2025

MRFF VALUE

\$2.40 million

INSTITUTION

Flinders University

GHFM PRIORITY

EPCD Research Initiative¹

Cancers of Unknown Primary (CUP) are a diverse group of cancers without an original source cancer. Patients affected by CUP face poor prognoses, with many surviving less than one year from diagnosis. Around 2,600 Australians have been diagnosed with CUP, with Indigenous Australian's twice as likely as other Australians. The project seeks to improve survival rates for CUP and reduce the proportion of Australians diagnosed with it, using novel genomic based strategies to determine the primary site of the cancer.

The project aims to fill knowledge gaps in research and treatment for CUP Indigenous and non-Indigenous Australians

Like many other types of cancer, CUP has been under researched in the past, especially in relation to Indigenous Australians. For all Australian demographics, there are goals to improve identifying cancer sites of origin through tissue of origin testing and genomic profiling. Specifically for Indigenous Australians, MRFF funding has enabled this project to initiate world-first research into the genomic profile of CUPs. This includes data collection on tumour biology, which may help to inform how patients respond to cancer treatments. Another area of focus is ensuring that research is translational. The clinical trial protocol has led the development, and promotion, of a statewide molecular tumour board meeting, representing an important advance in the framework and infrastructure for cancer patient management in SA.

MRFF funding is boosting patients in trials and generating optimism

Currently eight patients for clinical trials have been recruited over a 12-week period. Funding under the MRFF is facilitating patient recruitment, with a goal of 200 patients (including 20 indigenous Australians). There is growing hope that the research will break new insights into unique dynamics of CUP in Indigenous Australians.

Safety nets have been established through research collaborations

The project has worked closely with the National Indigenous Genomics Network (also funded through the MRFF). Leading researchers in Indigenous health are chief investigators on this study. They lead engagement with Indigenous Australians with the intention to ensure that research is conducted with cultural safety. The project has engaged with Australian researchers that have pioneered tissue of origin testing for patients with CUP. This research will help to establish that this new technology can also be an effective test in Indigenous Australians.

1: Emerging Priorities and Consumer Driven Research initiative

Pathways to benefit for Indigenous Australians in Genomic Medicine

PROJECT LEAD	GRANT PERIOD	MRFF VALUE	INSTITUTION	GHFM PRIORITY
Prof. Alex Brown	2022 – 2027	\$4.99 million	Australian National University	Indigenous Australian Health ¹

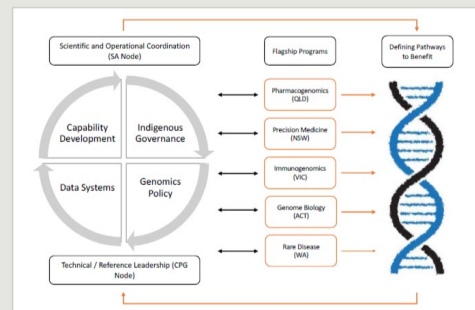
Indigenous Australians represent the world’s longest continuous surviving culture, having inhabited the Australian continent for over 60,000 years. From a global perspective, it is likely that Indigenous Australians contain genetic diversity seen in few (if any) other population groups. Indigenous Australians remain virtually absent from databases of human genetic variation. This hinders the complete understanding of health, disease and human development. The prioritisation, involvement, conduct, analysis and sharing of genomic data for the benefit of Indigenous Australians is overdue.

Indigenous governance both underpins and leads The Australian Alliance for Indigenous Genomics’ work (ALIGN)

Under the GHFM, this program sought to establish a national Indigenous genomics network now known as ALIGN - The Australian Alliance for Indigenous Genomics. ALIGN aims to build and extend Indigenous leadership and participation in genomic sciences to deliver benefit from its application in health systems, and ultimately reduce inequity in health and wellbeing among Australia’s First Peoples. ALIGN’s four pillars, “Indigenous Governance, Genomics Policy, Data Systems and Capability Develop”, will be delivered through 6 established nodes. Five nodes will also develop a national flagship program that will define a roadmap to benefit for Aboriginal and Torres Strait Islander peoples (Figure 1 below). To achieve this, ALIGN has established a comprehensive Indigenous governance structure that recognises local diversity and priorities while appreciating and informing the national agenda.

Each ALIGN node has established multi-jurisdictional Indigenous Governance Committees (IGCs) comprising Aboriginal and Torres Strait Islander people with connections to many of the Indigenous communities, health services and peak bodies within each node. The IGCs work with the relevant node investigators and the ALIGN team to address key research projects and ALIGN priorities. Along with independent advisors, the IGCs make up membership of ALIGN’s National Indigenous Governance Council (the “Council”) which provides cultural guidance, advice and leadership to nodes, research projects and key stakeholders.

Figure | ALIGN’s governance structure



Strong governance is supporting national change for Indigenous Australians

The governance model facilitates and ensures local Aboriginal and Torres Strait Islander peoples’ voices. Leadership and priorities are embedded within the network’s many different research projects, programs, protocols and structures. Defined governance and engagement has enabled the project to establish a growing presence in national health and genomics spaces. Through ALIGN’s growth, the program has made progress towards its goals for ensuring Indigenous empowerment and benefit through:

- Self-determining research and governance structures.
- Indigenous leadership, participation and voice that will impact key changes in government policy.
- Driving national and international genetic and genomic health partnerships and governance models.
- Developing frameworks for improving access to precision medicine research and clinical trials for Indigenous Australians.

¹ Priority Areas | 3.3 (Primary): Aboriginal and/or Torres Strait Islander Health & 3.2 (Secondary): Governance and Technology

The Genomic Screening Consortium for Australian Newborns (GenSCAN)

PROJECT LEAD	GRANT PERIOD	MRFF VALUE	INSTITUTION	GHFM PRIORITY
Multiple	2022 – 2027	\$14,011,498 Across 5 Projects	Multiple	Genomic Screening ¹

The Genomic Screening Consortium for Australian Newborns (GenSCAN) is an investigator led initiative (see Figure 1 below), supported by Australian Genomics, to maximise GHFM funding investment whilst enhancing the national coordination of genomic newborn bloodspot screening. Maximising extensive expertise in the use of genomics, the consortium brings together five Australian projects funded under the GHFM and NHMRC, with representatives from governments, community and Australian Genomics.

Exploring the potential of genomics in newborn screening

GenSCAN was developed for the purpose of enabling improved efficiency and impact of the MRFF GHFM investment through complementary and collaborative research. The consortium enables a cohesive national approach to the exploration of genomics into newborn screening. Cognisant of the national expansion efforts to align Newborn Bloodspot Screening Programmes nationally, the consortium includes representation from state and federal departments of health as well as patient advocacy representatives.

Figure 1 | Chief Investigators of GenSCAN projects



Leveraging a community of practice

GenSCAN acknowledges the significance of the Newborn Bloodspot Screening Programmes as public health initiatives. Initial collaboration included exploration of potential impacts that would disrupt the current uptake of 99.8 per cent participation in standard newborn screening. Through a community of practice, the consortium has allowed significant cross project support through common challenges, such as extensive delays in project agreements and restrictions in the recruitment of resources in an agile manner to meet tight timeframes for grant funded projects. As a consortium, GenSCAN is finding the ethical, social and political factors associated with the exploration of genomics in newborn screening significantly more complex than initially planned. A community of collaborators has been invaluable, specifically relevant to:

- Shared knowledge of technical and bioinformatic challenges.
- Exploration of data sharing models through collaboration with the National Approach to Genomic, Information Management and Law, sociology and ethics in data governance for genomics.
- A united voice and shared vision in the exploration of the potential use of genomics into the public health newborn screening programmes.
- An understanding of what is (and what isn't) an acceptable model of consent for genomic-enhanced newborn bloodspot screening.

¹ Priority Areas | 1.5 (Primary): Genomic Screening (including newborn screening), & 1.1 (Secondary): Rare Disease

Personalised medicine in the treatment of complex autoimmunity and autoinflammatory disease

PROJECT LEAD	GRANT PERIOD	MRFF VALUE	INSTITUTION	GHFM PRIORITY
SIMON JIANG	2023 - 2027	\$1.55 million	Australian National University	Early to Mid-Career Researchers initiative

Autoimmune and autoinflammatory diseases impact between five and 10 per cent of Australians causing a range of illness from fatigue to arthritis and organ failure. It is increasingly recognised that genetic variation is central to the development of autoimmune disease, with each variant causing disease in a unique way. However, conventional treatments are not tailored to each patient's genes.

Since 2014 the Personalised Medicine and Autoimmunity Laboratory at the Australian National University has been leading a program to identify the genetic causes of autoimmune diseases in specific patients to develop personalised therapies that improve efficacy and outcomes for patients.

Cross-disciplinary capabilities and collaboration has accelerated the patient journey

Over the last decade the laboratory has combined the expertise of members across clinical and research settings to develop and deliver personalised medicine. The lab collaborate extensively with rheumatology, nephrology, gastroenterology and other clinical professionals to recruit, diagnose and treat patients from across Australia.

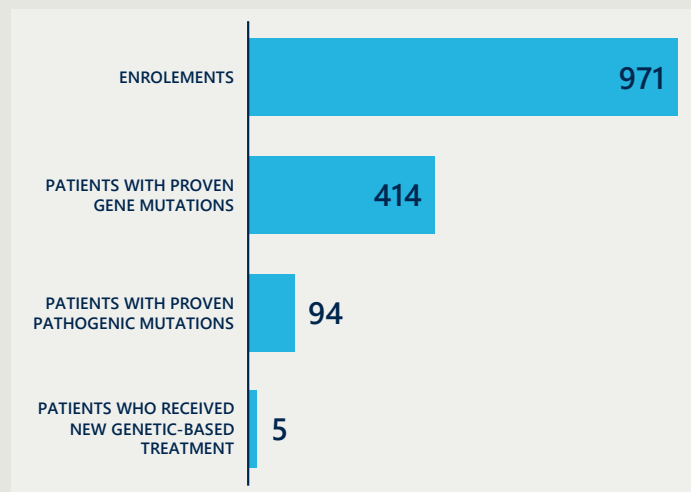
Using this approach setting, the lab has accelerated a 10-year gene discovery-to-treatments journey from into a 6 to 12 month timeframe. This genomics approach has started translating into treatment successes for study participants.

The MRFF has facilitated access to capabilities and increased the patient pipeline

MRFF funding has enabled the lab to significantly expand capacity to test patients from around 10 patients at one time to now being able to analyse more than 100 patients through their program at any one time.

To support these large volumes of patients the lab has used MRFF funding to increase scientific staff as well as clinician positions to support patients through recruitment, diagnosis and treatment.

Figure 1 | Number of patients through the program between 2014 and 2023



The MRFF funding has expanded genomic medicine footprint

This grant has extended the input of genomic medicine into hospitals around Australia. 20 doctors from 16 hospitals around Australia are recruiting into the program, advancing the utilisation and incorporation of genomic medicine in the clinical setting.

1: Early to Mid-Career Researchers initiative

6 Conclusion

This chapter outlines the conclusions of the GHFM Review against the original intent.

SCOPE

This section of the report synthesises the findings into conclusion against the intent of the Review:

- **How the MRFF has contributed to genomics research in Australia (Contribution)**
- **How MRFF-funded genomics research sits within the national and international genomics research funding landscape (Reputation)**
- **Alignment and progress of MRFF-funded genomics research (Progress)**
- **Opportunities (if any) to enhance MRFF funding and granting arrangements to improve the impact of MRFF funded genomics research (Opportunities).**

CONCLUSION

- The GHFM has played a significant role in the growth in the volume and profile of genomics research in Australia by investing in and securing a consistent pipeline of genomics research.
- The goal of the GHFM to *“save or transform the lives of more than 200,000 Australians through genomic research to deliver better testing, diagnosis and treatment”* is ambitious. A range of sector-wide challenges have limited the impact of the GHFM during its first five years.
- It is critical that the GHFM address these factors and the opportunities to enhance its ability to impact the sector over the remaining five years.

The intent of the GHFM Review was to assess the I. Contribution, II. Reputation, III. Progress and IV. Opportunities for the GHFM.

I. Contribution

The MRFF, most notably through the GHFM, has driven a step-change increase in the volume of genomics research activity in Australia and directed the focus of the research community to a range of challenges in genomics. The impacts of the GHFM are largely driven by the volume of activity it is enabling rather than as a result of targeted investments in key priority areas.

II. Reputation

Led by its flagship genomics investment, as the second largest funder of genomics in Australia, the MRFF stands out as uniquely focused on the translation of genomics. The GHFM is narrower in size and scope than its closest international comparators who also have a range of policy and sector wide functions.

III. Progress

Progress in genomics in Australia, particularly through the MRFF, has been closely linked to the volume of projects and investment. GHFM Aim 1 has received the majority of the focus in the first five years of the GHFM and accordingly has seen the greatest progress.

IV. Opportunities

The GHFM Review has identified 12 opportunities to improve the impact of the GHFM in the final five years of its remit (the GHFM Review details these opportunities and strategic considerations in **Chapter 7**).

7 Opportunities

This chapter describes in detail the opportunities identified through the synthesis of the evidence gathered and findings of the GHFM Review.

SCOPE

This section of the report describes the Review's evidence and findings in relation to the following component of the intent of the GHFM Review:

- **Opportunities (if any) to enhance MRFF funding and granting arrangements to improve the impact of MRFF funded genomics research.**

The opportunities identified in the Review do not constitute direct recommendations but should be considered by stakeholders (such as the GHFM Expert Advisory Panel, the department and other leading genomics organisations) in their administration of their relevant responsibilities to the genomics landscape in the future.

KEY FINDINGS

- The GHFM Review has identified 12 opportunities across three themes that, if addressed, will increase the impact of the GHFM over the remaining five years:
 - Refine investment strategy, approach and associated priorities (five opportunities)
 - Strengthen coordination and communication (three opportunities)
 - Support greater collaboration across the sector (four opportunities).
- The overlapping responsibilities in the Australian health system, genomics and research sectors, mean that to fully address many of the opportunities, the department will require the support of other stakeholders in the sector.
- The GHFM Review has identified four strategic considerations for the department and the EAP's consideration to realise the 10-year vision of the GHFM.

7.1 There are a range of opportunities to enhance the success and impact of the GHFM

Engagement with researchers, government, industry, consumers, and genomics bodies, both locally and internationally throughout the Review has uncovered a range of views on how the GHFM can provide maximum impact over the next five years.

The GHFM Review has identified 12 distinct opportunities, that if addressed can enhance the impact of the GHFM going forward. The opportunities can be grouped into three distinct themes:

1. Refine the investment strategy, approach and associated priorities
2. Strengthen coordination and communication
3. Support greater collaboration across the sector.

The extent to which the GHFM can realise the opportunities will be partially dependent on the actions of other stakeholders. The opportunities should be considered by not only the EAP in the refresh of the GHFM Roadmap and Implementation Plan, but also by:

- The department when formulating other genomics sector policies and supports for the GHFM.
- Other genomics funders in Australia when considering their genomics funding initiatives.
- Other organisations working to support genomics research and its translation in the health system.

7.1.1 Refine investment strategy, approach and associated priorities

The GHFM's approach to investments in its first five years has been beneficial for the development of a strong, broad base of knowledge. However, the findings of the GHFM Review demonstrate that there is need for the investment approach to evolve to be more targeted and cohesive. Five opportunities were identified to evolve the GHFM's investment approach:

- Opportunity 1 | Refine and consolidate the GHFM's priorities.
- Opportunity 6 | Concentrate investments in research projects further along the translation pipeline and enhance support to researchers to navigate translation activities such as clinical implementation and commercialisation of projects.
- Opportunity 9 | Consider greater investment in enabling research infrastructure to support effective translation.
- Opportunity 10 | Ensure policy research, including ethics and legal aspects are considered in every project.
- Opportunity 11 | Increase the degree to which MRFF genomics projects include realistic plans for implementation.

7.1.2 Strengthen coordination and communication

The Australian genomics sector is highly capable, however the size of the challenges to genomics translations combined with complex, disaggregated funding makes it challenging for the GHFM to support its priority areas on its own. Accordingly, there is scope to improve the way Australia's genomics funders and stakeholders work together and coordinate their objectives and actions, to ensure their investments are cohesive and effectively contribute to key national priorities in genomics.

The GHFM Review identified three opportunities to improve the coordination of the genomics sector:

- Opportunity 2 | Increase information sharing and partnership between members of the Australian genomics policy and funding landscape.
- Opportunity 4 | Strengthen linkages with other MRFF initiatives.
- Opportunity 12 | Improve cohesion of investments with those of other funding bodies to address translational and implementation challenges.

7.1.3 Support greater collaboration across the sector

Collaboration is key to successful research and translation outcomes. While the GHFM Review highlighted good examples of collaboration by MRFF genomics projects with other researchers and health sector stakeholders, there is potential to make collaborations and engagement by researchers with genomics sector stakeholders' business-as-usual practice for genomics researchers. The GHFM Review identified four opportunities to ensure MRFF genomics consistently involve adequate collaborations with other researchers and stakeholders:

- Opportunity 3 | Improve the GHFM's communication and engagement with consumers, the community and the sector.
- Opportunity 5 | Ensure all projects consider the appropriate involvement of, and potential impact on, First Nations communities.
- Opportunity 7 | Support more collaborations between MRFF genomics research projects.
- Opportunity 8 | Strengthen the connections of health sector stakeholders (such as consumers, industry and health system) to design and delivery of MRFF genomics projects.

7.2 Strategic considerations

The GHFM Review has identified four strategic considerations for the department and the EAP's consideration to realise the 10-year vision of the GHFM⁶¹:

1. Consolidate priorities and investment strategies, in collaboration with other funders, behind a cohesive strategy.
2. Support genomics projects to overcome challenges to successful, sustainable translation of their project's outputs.
3. Foster engagement, collaboration and coordination of activities between researchers, consumers and the health system.
4. Consider innovative funding models for future grant opportunities.

7.2.1 Consolidate priorities and investment strategies, in collaboration with other funders, behind a cohesive strategy

The findings of GHFM Review indicate that the number of priority areas is not proportional to the amount of funding available. To effectively reach the objectives in priority areas, the GHFM must align its investments with the investments of other genomics and health sector funders, such that they cohesively contribute to the same objectives. This would reduce the potential for duplication of investments and the likelihood that GHFM investments are not able to be translated due to insufficient capacity of enablers or funding in the health system.

While achieving this alignment of investments is dependent on the cooperation of other stakeholders who the GHFM does not oversee or control, there are a number of actions which the GHFM could take that would contribute to the alignment:

- Establish information sharing practices with the other genomics funders.
- Establish cross-organisation coordination or working groups with other genomics funders.
- Consolidate funding behind a smaller number of priority areas to concentrate investments.

This strategic consideration addresses:

- Opportunity 1 | Refine and consolidate the GHFM's priorities.
- Opportunity 2 | Increase information sharing and partnership between members of the Australian genomics policy and funding landscape.

⁶¹ The primary audience for the strategic considerations is the EAP and department. However, to action them effectively may require the department to seek the support of other organisations.

- Opportunity 10 | Ensure policy research, including ethics and legal aspects are considered in every project.
- Opportunity 12 | Improve cohesion of investments with those of other funding bodies to address translational and implementation challenges.

7.2.2 Support genomics projects to overcome challenges to successful, sustainable translation of their project's outputs

The GHFM Review found that while the GHFM has enabled and supported many important genomics research projects, there are significant barriers to the sustainable implementation of the project's outputs. If these barriers are not addressed, the important findings and outcomes of GHFM projects may not be embedded or sustained in the health system. The EAP and department should consider how the GHFM can do its part to address these barriers.

This may involve:

- Requiring and supporting researchers to prepare robust and realistic plans for implementation from early on in their project.
- Requiring and supporting researchers to address ELSI aspects in their project planning.
- Aligning GHFM priority areas or investments to areas where there is likely to be sufficient capacity in the health system to support sustainable translation of projects.

This strategic consideration addresses:

- Opportunity 8 | Strengthen the connections of health sector stakeholders (such as consumers, industry and health system) to design and delivery of MRFF genomics projects.
- Opportunity 9 | Consider greater investment in enabling research infrastructure to support effective translation.
- Opportunity 11 | Increase the degree to which MRFF genomics projects include realistic plans for implementation.
- Opportunity 12 | Improve cohesion of investments with those of other funding bodies to address translational and implementation challenges.

7.2.3 Foster engagement, collaboration and coordination of activities between researchers, consumers and the health system

Genomics, like all scientific endeavours, benefits significantly from regular and effective collaboration between researchers. The GHFM Review found that many organisations and individuals in the genomics research landscape are operating in silos. While there have been some positive examples of cross-researcher and inter-disciplinary collaborations (refer to **Chapter 5 Case Studies**), there is scope to expand upon this and improve the connection between members of the sector.

This may involve⁶²:

⁶² The GHFM Review acknowledges that a number of similar activities are already taking place, however where this is the case the department could consider how to enhance the impact of these activities.

- Establish or working with networking forums and providing opportunities for MRFF researchers to attend conferences which may focus on their research area or may be more broadly related to translation issues.
- Requiring and supporting researchers to prepare robust and realistic plans for implementation from early on in their project.
- Maintain and distribute among MRFF project leads and relevant stakeholders an up-to-date registry with information on funded projects and their progress to date.
- Encourage more applications by different types of organisations (outside of universities and MRIs).⁶³
- Supporting or facilitating collaboration and coordination activities between complementary research projects identified after commencement of the grant agreement.⁶⁴

This strategic consideration addresses:

- Opportunity 3 | Improve the GHFM's communication and engagement with consumers, the community and the sector.
- Opportunity 7 | Support more collaborations between MRFF genomics research projects.
- Opportunity 9 | Consider greater investment in enabling research infrastructure to support effective translation.

7.2.4 Consider innovative funding models for future grant opportunities

As a grant program, the GHFM seeks to achieve its policy objectives through the allocation of funding to genomics research projects. There are many funding models used in grant design, each with their own benefits and limitations. During consultations, many stakeholders made observations about the current project funding model and provided some limited detail on how to improve it. These contributions have been synthesised into three potential innovative funding models⁶⁵:

- Stage-gated funding.
- Funding the establishment of consortia.
- Funding foundational research infrastructure and other key research enablers.

The innovative funding models could be used to achieve a variety of opportunities.

Stage-gated funding

A stage-gated funding model, commonly used in product development, is a model that requires projects to progress through milestones or 'gates', before continuing investment. Connecting the outcome of stage gates to the release of funding aims to manage the risk of investments, and more closely monitor the progress of projects.

The recently launched Australia's Economic Accelerator (AEA) from the Department of Education, provides an example of a research funding program that utilises a series of stages to manage investments. Designed to generate a 'research commercialisation ecosystem' within the higher education sector, the

⁶³ The GHFM Review notes that private sector organisations are able to seek Eligible Organisation status under the MRFF. However, the GHFM has only provided two grants to organisations that were not a university or a MRI.

⁶⁴ The GHFM Review acknowledges that a number of similar activities are already taking place, however where this is the case the department could consider how to enhance the impact of these activities.

⁶⁵ Each funding model has its own strengths and weaknesses and present unique opportunities and/or risks. Nous has synthesised a high-level articulation of each funding model however further investigation and testing is required to conclude on the overall utility (to the outcome or objective sought) of each of the funding models.

AEA has three phases, a pilot AEA Seed, and two further phases (AEA Ignite and AEA Innovate), to support projects at different levels of their commercialisation journey.

A stage-gated funding model may allow for the HMRO to have the ability to support projects through the commercialisation journey, by engaging with them during the stage-gates, and identify risks and potential problems more readily to prevent unnecessary investment. On the other hand, a stage-gate process if not well designed and implemented, may:

- Make projects less agile to evolving findings and research and health priorities.
- Increase administrative burden on researchers.
- Increase the amount of uncertainty and time-pressure on researchers, particularly junior researchers.

While stage-gated funding could be designed to address several of the opportunities, it could be a particularly useful mechanism to support researchers to undertake preparatory activities to enhance the impact of their core project, for example by funding them to appropriately plan implementation pathways or engage various stakeholder groups (particularly consumers and First Nations communities) in detail before commencing the core project.

Funding the establishment of consortia

While the GHFM currently supports consortium approaches or networks, such as KidGen and ALIGN (refer to **Chapter 5 Case Studies**, see *'Pathways to benefit for Indigenous Australians in Genomic Medicine'*) there are many projects which apply and receive funding outside of established consortia that may benefit from being connected to other projects.

As demonstrated by GenSCAN (refer to Case-in-point 2, page 49 and **Chapter 5 Case Studies**, see *'The Genomic Screening Consortium for Australian Newborns'*), a consortium lets its partners share relevant skills, experience and expertise in such a way that every member complements each other. This approach may also allow for cost sharing or the sharing of critical infrastructure and technology. A consortium of this nature, however, can take time to develop. There are set-up costs, governance considerations and membership to consider.

When considering consortium approaches as an innovative funding model, the GHFM would need to determine how they support researchers to identify complementary projects as well as support in the initial phases of establishment and reduce the feeling of competition amongst grantees. Funding may be necessary to support individuals or groups to take on the coordination role across complementary projects.⁶⁶

Funding foundational research infrastructure and other key research enablers

The GHFM scope (or the departments actions to support the GHFM) may be broadened beyond research project grants to provide funding for more national genomics research infrastructure. In particular, the Review identified investment towards technology and data in many comparable countries, including within Genome Canada and the National Human Genome Research Institute (NHGRI) in the United States.

Funding beyond research grants would require the GHFM to redefine its scope and consider how such investment would be structured, however a national approach to key enablers of implementation, such as technology and data, could bolster the impact of individual projects. It is noted that any change in scope and investment must be in line with legislation set out in the *Medical Research Future Fund Act 2015* and could not include clinical infrastructure. The department, or other funders, may seek to use other funding

⁶⁶ The funding provided for coordination activities would need to be appropriately connected to research activities to be eligible for funding.

mechanisms to make investments in the clinical genomics infrastructure and enablers to complement the research investment.

Similarly, the GHFM Review's findings tend to indicate that current MRFF genomics projects are not developing robust plans for implementation of their innovations or consistently involving appropriate consideration of policy, ELSI, consumer and First Nations community issues. This may indicate a gap in whether project leads are utilising advice or the latest evidence to design their project engagement, manage ELSI and develop realistic implementation plans.

There is a need for mechanisms, or resources, to be available to researchers to provide them with reliable and consistent advice and support on issues that commonly affect genomics projects in particular on:

- Commercialisation planning and pathways
- Consumer communication and engagement
- First Nations community engagement
- ELSI.

This may be achieved by leveraging existing capabilities or resources developed and maintained by genomics organisations such as those from industry, state and territory governments, genomics health alliances, Aboriginal Community Controlled Health Organisations or consumer representative organisations.

Appendix A Contributors to the GHFM Review

The department engaged Nous to:

“conduct a review of the MRFF’s GHFM to assess its progress in line with the Mission Roadmap and Implementation Plan, the Australian Government’s 10-year MRFF investment plan and the Evaluation Strategy, and to guide investment from 2024-25.”

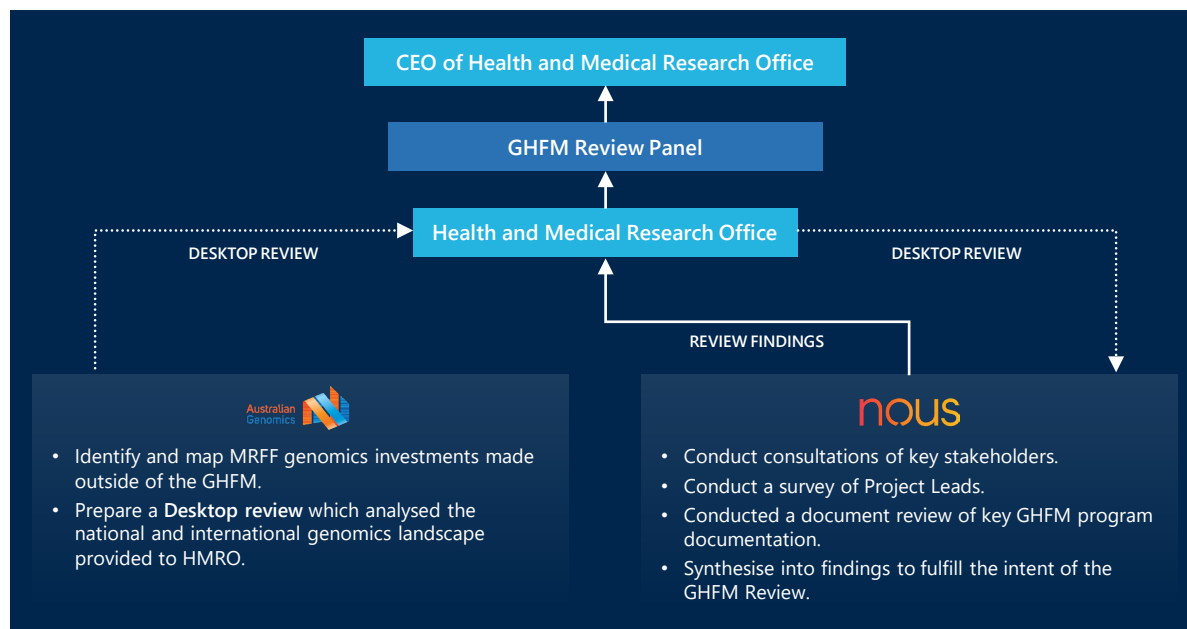
To conduct the GHFM Review Nous conducted: broad consultations with stakeholders across the health and medical research sector; a survey of MRFF genomics project leads; analysed GHFM and MRFF program documentation and departmental data.

Australian Genomics was separately engaged by the department to provide a Desktop Review analysing the national and international genomics landscape as well as identify and map genomics investments outside the GHFM within the MRFF. This was principally to support answering the second component of the intent of the GHFM Review.⁶⁷ The Desktop Review was overseen by the HMRO and provided to Nous to inform the development of findings and opportunities.

The HMRO oversaw and guided the activities of Australian Genomics and Nous. The HMRO established a GHFM Review Panel to provide expert oversight and advice on the collection, analysis, and interpretation of information supporting the work of both Australian Genomics and Nous. HMRO coordinated the activities of the GHFM Review Panel who were ultimately accountable to the CEO of the HMRO for the GHFM Review.

The governance relationships between the various contributors are displayed in Figure 21, below.

Figure 21 | Governance of the GHFM Review



The GHFM Review Panel comprised of a number of genomics experts within Australia and overseas (refer to Table 8, overleaf).

⁶⁷ Due to the potential of an actual or perceived conflict of interest, the department limited Australian Genomics contribution to the GHFM to the development of the Desktop Review. While Australian Genomics were interviewed by Nous Group as part of the stakeholder consultations, they had no direct input on the Final Report or other Nous deliverables.

Table 8 | Members of the GHFM Review Panel

Name	Affiliation(s)	Research focus, or experience	Conflicts of Interest
Prof. Ken Smith Chair	<ul style="list-style-type: none"> University of Cambridge WEHI (formerly Walter and Eliza Hall Institute of Medical Research) 	The Smith laboratory combines genetics, genomics, immunology and clinical medicine. Integrating detailed laboratory analysis of mechanisms of immune regulation with a prospective translational medicine programme in major autoimmune and inflammatory diseases. The main focus of the group is investigation of the biology underlying clinical outcome in immune-mediated disease.	None ^A
Prof. Doug Hilton AO AMRAB Representative	<ul style="list-style-type: none"> WEHI University of Melbourne 	He is best known for his discoveries in the area of cytokine signalling, particularly the isolation and cloning of an entirely novel family of negative regulators of cytokine signalling, the SOCS proteins.	None ^A
Prof. Denise Doolan AMRAB Representative	<ul style="list-style-type: none"> University of Queensland 	She is a molecular immunologist with expertise in malaria immunology, vaccinology, and omic-based approaches for therapeutic and diagnostic development. A particular focus is the discipline of systems immunology which integrates immunology with cutting-edge omics-based technologies, bioinformatics and computational sciences to identify pathogen antigens and host immune molecules that can be targeted for novel interventions against disease.	None
Ms Merryn Carter Consumer Representative	<ul style="list-style-type: none"> WEHI 	Merryn is a consumer advocate with the Walter and Eliza Hall Institute's Breast Cancer Laboratory. Merryn is a member of the MRFF Consumer Reference Panel and the Breast Cancer Trials Australia New Zealand's Consumer Advisory Panel.	None
Assoc. Prof. Glenn Pearson	<ul style="list-style-type: none"> Telethon Kids Institute and National Centre for Indigenous Genomics 	Glenn Pearson is a Noongar man, who has a state-wide mandate to ensure that the Institute's research reflects the needs of Aboriginal families, and that research is conducted in accord with Aboriginal community ethical and cultural protocols. Mr Pearson was a member of the Consultative Committee on the Indigenous Collection (CCIC) convened by the ANU in 2011, whose recommendations led to the establishment of National Centre for Indigenous Genomics.	None
Prof. Sir Jim Smith	<ul style="list-style-type: none"> Francis Crick Institute Zoological Society of London Medical Research Council Wellcome Trust 	During his time at the National Institute for Medical Research (UK) he served variously as Director of Research of the Francis Crick Institute, Deputy CEO and Chief of Strategy at the Medical Research Council, and a Trustee of the Crick. He was Director of Science at the Wellcome Trust and led the development of its new strategy.	None

Table note A: During the course of the GHFM Review, four conflicts of interest were identified and considered by the GHFM Review Panel and department but were deemed not to be actual conflicts of interest.

Members of the GHFM Review Panel met on five occasions between September 2023, and April 2024 to consider and discuss GHFM Review and provide guidance to Nous and Australian Genomics⁶⁸:

- **September 2023** | To consider and discuss the project plan for the Desktop Review prepared by Australian Genomics.
- **October 2023** | To consider and discuss the project plan for the GHFM Review prepared by Nous.
- **December 2023** | To consider and discuss the Desktop Review prepared by Australian Genomics.
- **March 2024** | To consider and discuss the outcomes of the survey and stakeholder consultations conducted by Nous.
- **April 2024** | To consider and discuss the Key Findings report prepared by Nous.

⁶⁸ The GHFM Review Panel also considered and approved the questions to be answered by the survey and consultations out-of-session.

Appendix B List of in-scope non-GHFM projects

According to the department, the assignment of a genomics related project was based on coding entered by the relevant policy section within the department when executing a grant agreement. The definition of genomics research used was *'research that focuses on genomics knowledge and/or technologies to improve testing, diagnosis, treatment and intervention, excluding research on diseases that are known to have a genetic cause.'* Nous has not sought to verify the accuracy or completeness of the list as part of the GHFM Review (refer to Table 9, below).

Table 9 | Non-GHFM genomics projects included in the GHFM Report

Project title	Grant value (millions)	Grant period	Project lead	Grantee organisation ^A
Emerging Priorities and Consumer Driven Research (29 projects)	\$173.84			
Role of the NKp44-PDGF-DD axis in Glioblastoma	\$0.57	2019-2023	Dr. Alexander Barrow	UoM
The Australian Genomics Cardiovascular Genetic Disorders Flagship	\$6.00	2019-2023	Not applicable	MCRI
The Australian Epilepsy Research Fund	\$2.00	2018-2023	Not applicable	Epilepsy Foundation
Massimo's Mission	\$3.00	2019-2023	Not applicable	MCRI
Australian National Phenome Centre (Murdoch University): Support for Establishment and Sustainability of Critical Infrastructure to Provide Transformational Phenomics Capacity for Australian Medical Research	\$10.00	2019-2020	Not applicable	MU
The Australian Parkinson's Mission: Integrating genomics, biomarkers and patient cell phenotyping into disease modifying clinical trials to identify therapeutics to slow or stop disease progression	\$30.00	2019-2025	Not applicable	UNSW
Phenomics Capability (Australian National University): The Phenomics Translation Initiative	\$10.00	2019-2024	Not applicable	ANU
ASPiRATION: Assessing the impact of genomic profiling in lung cancer	\$5.00	2020-2024	Not applicable	AGCMC
Genetic variants, early life exposures, and longitudinal endometriosis symptoms study (GELLES)	\$1.86	2020-2024	Prof. Gita Mishra	UQ

Australian Pharmacogenomics Diversity Project: Examining the evidence and improving the performance of pharmacogenomics in the Australian context	\$1.37	2020-2025	Prof. Sarah Medland	QIMR Berghofer
The PRESIDE (Pharmacogenomics in depression) Trial: An RCT of pharmacogenomically-informed prescribing of antidepressants on depression outcomes in patients with major depressive disorder in primary care	\$1.39	2020-2024	Prof. Jon Emery	UoM
A multifaceted approach to the pharmacogenomic signatures of bipolar disorder for improving treatment outcomes	\$1.01	2020-2025	Assoc. Prof. Janic Fullerton	UNSW
An Australian Multicentre Double-Blinded Randomised Controlled Trial of Genotype-guided versus Standard Psychotropic Therapy in Moderately-to-Severely Depressed Patients Initiating Pharmacotherapy	\$2.95	2020-2024	Dr. Kathy Wu	UNSW
Towards A New Era in Granulosa Cell Tumour Research: Patient Driven Outcomes, Genomics, Diagnostics & Therapeutics	\$2.22	2020-2025	Prof. Peter Fuller	Monash
High throughput discovery of synergistic drug combinations for patients with low-grade serous ovarian cancer	\$1.11	2020-2024	Dr. Dane Cheasley	UoM
Zero Childhood Cancer National Precision Medicine Program	\$54.80	2020-2025	Not applicable	UNSW
The Victoria Paediatric Cancer Consortium: A Multi-institutional Partnership to Catalyze Advances in Childhood Cancer Research and Clinical Implementation	\$9.60	2021-2024	Assoc. Prof. Ron Firestein	Monash
Shining Light into the “unknown” on Indigenous and non-Indigenous Australians with Cancer of Unknown Primary	\$2.40	2021-2026	Prof. Christos Karapetis	Flinders
Predicting and Preventing Ovarian Cancer: a machine learning approach	\$1.26	2021-2025	Prof. Elina Hypponen	UniSA
Advancing congenital and childhood-onset muscle disease diagnosis and treatment - a cross-disciplinary Australian collaboration	\$2.50	2022-2026	Dr. Emily Oates	UNSW
RTTomics: Towards developing new treatments and therapies for Rett syndrome individuals using cortical brain organoids	\$0.60	2023-2026	Assoc. Prof. Wendy Gold	USYD
A new substrate reduction strategy to treat childhood dementias: Glucosylceramide synthase-targeting antisense oligonucleotides	\$0.60	2023-2025	Assoc. Prof. Anthony Cook	UTAS
Developing Nanoparticle Mediated Gene Transfer for Childhood Dementia	\$0.30	2023-2025	Dr. Nicholas Smith	UoA

Developing an mRNA-based gene therapy strategy for Niemann-Pick Disease Type C1: a blueprint to treat childhood dementia	\$0.60	2023-2025	Dr. Ya Hui Hung	UoM
Introducing Mitochondrial Donation into Australia: The mitoHOPE (Healthy Outcomes Pilot and Evaluation) Program	\$15.00	2023-2028	Prof. John Carroll	Monash
Applying OCCAMS molecular razor to study the role of EBV in MS pathogenesis	\$2.00	2023-2027	Prof. Tri Phan	UNSW
Unravelling the interplay between EBV genomics and host T cell immune regulation in multiple sclerosis	\$2.00	2023-2027	Dr. Yuan Zhou	UTAS
Understanding how Epstein-Barr virus and other factors program multiple sclerosis onset and progression through epigenetic pathways to inform prevention and treatment with risk stratification	\$1.78	2023-2027	Prof. Anne-Louise Ponsonby	UoM
How does Epstein-Barr virus infection lead to multiple sclerosis?	\$1.92	2023-2027	Prof. Pamela McCombe	UQ
Clinical Trials Activity (18 projects)	\$33.84			
A registry-linked national platform trial to improve precision-based outcomes using novel therapies in acute myeloid leukaemia (AML)	\$1.51	2018-2023	Assoc. Prof. Andrew Wei	Monash
The DIAAMOND study: D iagnostics of a plastic a naemia, m anagement, and o utcomes utilising a n ational d ataset	\$1.75	2018-2024	Assoc. Prof. Erica Wood	Monash
Improving outcomes of children and young adults with primary ciliary dyskinesia (PCD): A multi-centre, double-blind, double-dummy, 2x2 factorial, randomised controlled trial (RCT)	\$2.38	2019-2025	Prof. Anne Chang	CDU
FaR-RMS: F rontline and r elapse study in r habdomyosarcoma	\$1.35	2019-2025	Dr. Martin Campbell	Monash
Novel Venetoclax Combinations to Improve Outcomes in Unfit Older Patients with Acute Myeloid Leukaemia	\$1.38	2019-2024	Assoc. Prof. Martin Wei	WEHI
The AIM2 Study: Genomically Guided Novel Combination Treatment of Mantle Cell Lymphoma	\$2.01	2019-2025	Dr. Constantine Tam	UoM
ALS Trials Australia (ALSTA) - to develop precision medicine	\$1.70	2019-2024	Prof. Matthew Kiernan	USYD
Improving survival in myelofibrosis	\$1.73	2019-2023	Prof. Andrew Perkins	Monash
Circulating tumour DNA guided therapy for stage IIB/C BRAF mutant positive m elanoma after surgical r esection (DETECTION)	\$3.23	2020-2025	Dr. Shahneen Sandhu	UoM

CONNECT 1903: A Pilot and Surgical Study of Larotrectinib for Disease Control in Children with Newly-Diagnosed High-Grade Glioma with NTRK Fusion	\$0.32	2020-2025	Assoc. Prof. Nicholas Gottardo	Monash
LOGGIC: A phase III, randomised international multi-centre trial for Low Grade Glioma In Children and adolescents	\$1.13	2020-2026	Assoc. Prof. David Ziegler	UNSW
PARAGON-II: Phase 2 basket study of an ARomatase inhibitor plus PI3KCA inhibitor or CDK4/6 inhibitor in women with hormone receptor positive recurrent/metastatic Gynaecological Neoplasms	\$2.00	2020-2025	Dr. Chee Khoon Lee	USYD
The IMPEDE-PKD trial: Implementation of Metformin therapy to ease decline of kidney function in PKD	\$2.57	2020-2027	Assoc. Prof. Andrew Mallett	UQ
The TELO-SCOPE study: Attenuating Telomere Attrition with Danazol. Is there Scope to Dramatically Improve Health Outcomes for Adults and Children with Pulmonary Fibrosis?	\$1.83	2020-2025	Prof. Daniel Chambers	UQ
Ataxia-telangiectasia: Treating mitochondrial dysfunction with a novel form of anaplerosis	\$2.46	2020-2024	Prof. David Coman	UQ
A Platform trial of combination precision guided therapies for high-risk childhood cancer	\$1.52	2022-2027	Assoc. Prof. David Ziegler	UNSW
LUMOS: Low and Anaplastic Grade Glioma Umbrella Study of Molecular Guided Therapies	\$1.98	2022-2027	Prof. Hui Gan	USYD
Targeted, Adaptive Genomics for Ethical, Evidence-based Expansion of Newborn Screening: a type II hybrid effectiveness-implementation trial	\$2.99	2023-2026	Assoc. Prof. Natalie Taylor	UNSW
Preventative and Public Health Research (six projects)	\$8.22			
Prenatal environments, offspring neurodevelopment and epigenetic programming	\$0.75	2020-2024	Prof. Ann-Louise Ponsonby	UoM
PRECISION- PhaRmacogEnomiC medIcines optimiSatIion for peOple with caNcer	\$1.50	2023-2027	Dr. Marliese Alexander	UoM
Pharmacogenomics for better treatment of fungal infections in cancer	\$1.50	2023-2026	Prof. Jason Roberts	UQ
Personalising the management of obesity-associated asthma using medical nutrition therapy and physical activity prescription: The IDEAL Study	\$1.47	2023-2027	Dr. Hayley Scott	UoN
Treatable Traits in Interstitial Lung Disease (TTRILD) Study: The New Frontier	\$2.00	2023-2028	Prof. Yuben Moodley	UWA
Development of a generalisable evaluation framework for high upfront-cost therapies: clinical, economic, ethico-legal, social and cultural considerations	\$1.00	2023-2026	Prof. Kirsten Howard	USYD

Early to Mid-Career Researchers (four projects)		\$7.89		
Developing a promoter-less gene therapy approach for haemophilia A	\$0.51	2023-2025	Dr. Marti Cabanes Creus	USYD
Harnessing nanopore sequencing technology to improve diagnosis of human disease	\$0.95	2023-2025	Dr. Ira Deveson	UNSW
The missing heritability of human disease: discovery to implementation	\$4.88	2023-2028	Dr. Gianina Ravenscroft	UWA
Personalised medicine in the treatment of complex autoimmunity and autoinflammatory disease	\$1.55	2023-2027	Dr. Simon Jiang	ANU
Cardiovascular Health Mission (four projects)		\$7.09		
Gene Expression to Predict Long-Term Outcome in Infants After Heart Surgery	\$3.07	2020-2025	Assoc. Prof. Luregn Schlapbach	UQ
Using Polygenic Risk Scores to Target Statin Therapy in Primary Prevention	\$1.42	2020-2024	Prof. Stephen Nicholls	Monash
The Elusive Hearts Study: Using genomics to diagnose and manage inherited cardiovascular diseases	\$1.50	2023-2027	Assoc. Prof. Jodie Ingles	UNSW
Gap Junction Modulation: A Novel Molecular Target in the Management of Ventricular Arrhythmia in Ischaemic Cardiomyopathy	\$1.10	2023-2027	Assoc. Prof. Eddy Kizana	USYD
Rapid Applied Research Translation (one project)		\$5.87		
P-OMICs-flow: Integrating precision oncology into clinical programs	\$5.87	2022-2027	Assoc. Prof. Natalie Taylor	UNSW
National Critical Research Infrastructure (two projects)		\$5.86		
Building an A ustralian C ardiovascular disease D ata C ommons (ACDC)	\$2.93	2023-2027	Prof. Peter John Meikle	Baker Heart and Diabetes Institute
Drug Target Identification Platform	\$2.93	2023-2028	Assoc. Prof. Darren John Creek	Monash
Frontier Health and Medical Research (three projects)		\$5.27		
c-FIND: C RISPR F rontier I nfection D iagnostics to Detect Infection	\$1.07	2019-2020	Prof. Marc Pellegrini	WEHI
Tracking COVID-19 using genomics	\$3.27	2020-2022	Prof. William Rawlinson	UNSW

Earlier Diagnosis and Personalised Treatments for Endometriosis (EndoAIMM)	\$0.93	2021-2022	Prof. Grant Montgomery	UQ
Australian Brain Cancer Mission (one project)	\$5.00			
Zero Childhood Brain Cancer program	\$5.00	2018-2024	Prof. Michelle Haber	UNSW
Clinician Researchers (six projects)	\$4.14			
Precision medicine for epilepsy and beyond: From discovery to implementation and evaluation	\$0.48	2018-2022	Prof. Patrick Kwan	Monash
Personalised early detection of melanoma	\$0.58	2018-2022	Prof. H. Peter Soyer	UQ
Optimising interventions for Staphylococcus aureus and skin infections	\$0.33	2018-2021	Assoc. Prof. Steven Tong	UoM
Sepsis Outcomes Research	\$0.28	2018-2022	Prof. Bala Venkatesh	UNSW
Advancing Personalised Treatment in Colorectal Cancer with Tissue and Liquid Biomarkers	\$1.19	2021-2025	Assoc. Prof. Jeanne Tie	WEHI
Closing the critical knowledge gaps in perinatal genomics	\$1.28	2021-2025	Assoc. Prof. Lisa Hui	UoM
Stem Cell Therapies Mission (four projects)	\$3.39			
Novel SMART AAV vectors for gene therapy for Friedreich's Ataxia	\$0.98	2021-2024	Assoc. Prof. Mirella Dottori	UOW
Stem Cell Derived-Retinal Organoids to Test Novel Genetic Therapies	\$0.50	2021-2023	Dr. Anai Gonzalez Cordero	USYD
Stem cell models of glomerular kidney disease for understanding disease and developing treatments	\$0.93	2021-2024	Prof. Melissa Little	MCRI
PAGETURNA: Pioneering Application of Gene Editing in Transplant Using RNA	\$0.98	2023-2025	Assoc. Prof. Andrew Deans	St Vincent's Institute of Medical Research
Global Health (three projects)	\$3.33			
Using metagenomics and the Registry of Ageing South Australians to understand carriage and transmission of antimicrobial resistance in the elderly	\$1.73	2018-2021	Assoc. Prof. Geraint Rogers	SAHMRI

Rapid detection of drug resistant tuberculosis using real-time sequencing	\$0.78	2020-2025	Prof. Lachlan Coin	UoM
Preparing Fiji for Pathogens with Critical Antimicrobial Resistance	\$0.82	2020-2023	Prof. Kirsty Busing	UoM
Dementia, Ageing and Aged Care Mission (one project)	\$0.32			
Targeting SARS-CoV-2 using Stealth nanoparticles loaded with gene silencing siRNAs	\$0.32	2020-2021	Prof. Nigel McMillan	GU

Table note A: The following acronyms were used: Australian Genomic Cancer Medicine Centre Limited (AGCMC); Australian National University (ANU); Charles Darwin University (CDU); Flinders University (Flinders); Griffith University (Griffith); Monash University (Monash); Murdoch University (Murdoch); Murdoch Childrens Research institute (MCRI); QIMR Berghofer Medical Research Institute or The Council of the Queensland Institute of Medical Research (QIMR Berghofer); South Australian Health and Medical Research Institute (SAHMRI); University of Adelaide (UA); University of Melbourne (UoM); University of Newcastle (UoN); University of New South Wales (UNSW); University of Queensland (UQ); University of Sydney (USYD); University of Tasmania (UTAS); University of Western Australia (UWA).