Australian Government
Department of Health and Aged Care

Medical Research
Future Fund

Implementation Plan

Genomics Health Futures Mission

Background

The Genomics Health Futures Mission (GHFM) is investing $500.1 million over 10 years in genomics research under the Medical Research Future Fund (MRFF). It will improve testing, diagnosis and treatment for genetic diseases, guide prevention and help personalise treatment options to better target and improve health outcomes and reduce unnecessary interventions and associated health costs for all Australians. The GHFM will also advance precision medicine for all Australians while keeping a focus on improving overall healthcare for Aboriginal and/or Torres Strait Islander peoples. This will be accomplished in partnership with Aboriginal and/or Torres Strait Islander peoples to deliver genomics research that is scientifically sound, culturally safe and competent to address inequity in research participation and outcomes.

To date, the GHFM has provided $234.3 million for 71 GHFM grants through eight grant opportunities (see Appendix A). The priority areas of these grant opportunities were informed by the first GHFM roadmap, which was published in October 2021.

Advances in genomics knowledge and technology have accelerated in the five years since the GHFM commenced. The GHFM's strategic priorities need to adapt to keep pace with this change. A review assessed the mission's progress against its goals. The GHFM Review Final Report details the findings of this evaluation and was published on 17 September 2024.

The second GHFM Expert Advisory Panel (the Panel) was appointed in December 2023. The Panel's role was to provide advice on the strategic priorities for the remainder of the mission, by refreshing the existing roadmap and implementation plan. As part of this process, the Panel considered the findings from the review and ensured that the priority-setting approach aligned with the approach used by the other MRFF missions. This mid-term refresh will enable the GHFM to leverage resources across the system and ensure advances in genomics research are translated into improved health and well-being for all Australians.

This plan supports the implementation of the GHFM roadmap and establishes a strategic plan to address the GHFM goals within the context of the MRFF 3rd 10-year plan. This implementation plan should be read in the context of the GHFM Roadmap, which describes the GHFM's scope, goals and principles.

Overview

To target activities to achieve the objectives of the GHFM within the 10-year plan, the following aims and priority research investment areas have been identified.

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

Implementation Strategy

The implementation strategy has been developed to guide research investment over the life of the GHFM. Investment aims to build capability and knowledge, as well as facilitate translation of advancements to clinical practice, to achieve the GHFM's objectives. The implementation strategy is intended to make the research purpose and direction transparent, and provide certainty to stakeholders. It also establishes how the outcomes of each focus area will be evaluated in terms of benefit to Australian patients, which will clarify the intended outcome and facilitate tracking of the GHFM's progress towards its objectives.

Research activities

Priority areas will be, or contribute to, large programs of work of national strategic importance that are informed by the key priority areas outlined in this implementation plan. The research activities are expected to foster collaboration and harness resources across the system to deliver improved health outcomes for Australians.

For all priority areas, applicants are required to propose research that:

- includes consideration of the ethical, legal and social implications (ELSI) of the genomics approach utilised and/or include research designed to further understanding of ELSI of genomics
- Aligns with the Department's position on consumer involvement in MRFF funded research, including principles and strategies that reflect a commitment to community engagement and involvement in research design, application and evaluation
- Is conducted in accordance with *Ethical conduct in research with Aboriginal and Torres Strait Islander Peoples and communities: Guidelines for researchers and stakeholders*
- adopts best practice data management which includes ensuring data is stored in a data repository, which complies with international genomics standards and all relevant legislation
- enables sharing of data in accordance with the Australian Data Strategy, best practice governance principles, participant preferences, and all relevant legislation

GHFM monitoring and evaluation

The MRFF Monitoring, evaluation and learning strategy is an overarching framework for assessing the performance of the MRFF, focused on individual grants, grant opportunities, initiatives (e.g., the GHFM) and the entire program.

The strategy sets out the principles and approach used to monitor and evaluate the MRFF. It outlines the need for evaluations to be independent and impartial. The strategy aims to be transparent in process and outcomes, and agile to the needs of the MRFF, its consumers and

stakeholders (such as the health and medical research industry). The GHFM and grants funded under this initiative will be evaluated against the strategy.

A note on the funding amounts: This plan represents $191.2 million in dedicated funding for the GHFM under the 3rd 10-Year Plan for MRFF Investment from 2024-25 to 2027-28. Additional unspent funds from previous years may also be utilised to ensure the GHFM meets the Government objective of investing $500.1 million over 10 years. The total funding amounts per priority are indicative and may change depending on the number and quality of applications submitted in response to the advertised grant opportunities.

GHFM Enablers

The GHFM will be delivered with clarity in structure and processes, guided by — and involving — international leaders, industry, patients and the public. Effective and extensive engagement across all levels of government will be established to ensure the outcomes of the GHFM are transformative to health care.

The GHFM enabling capabilities can also deliver:

- implementation research and health service engagement to realise the health benefits from genomic innovation.
- industry engagement to improve the uptake and implementation of genomics in health care, and translate genomics innovations into more effective treatments and better patient outcomes (noting that care with public perception and trust is critical).
- consumer engagement to develop and implement policies for involving patients and consumers to guide and enrich GHFM-funded projects (Involve Australia).
- international engagement and collaboration, which are critical to the GHFM's success, supported by a coordinated and strategic approach to fostering international partnerships and contributing to global genomics activity.
- Aboriginal and/or Torres Strait Islander leadership in developing and implementing policies for involving Aboriginal and/or Torres Strait Islander peoples to guide and enrich GHFM-funded projects.

The GHFM will also actively pursue opportunities of collaboration with other MRFF programs and missions, including the Indigenous Health Research Fund, and significant national and international genomics research endeavours, and genomics initiatives, including Genomics Australia, the Global Alliance for Genomics and Health and leading genomics initiatives in the region and globally.

The ethical and secure management of the GHFM genomic and health data resource, and access to these data in accordance with participant's preferences, will be a legacy of the GHFM for future research. It will be important to align the GHFM's genomics research resources, tools and infrastructure to national endeavours; harmonise investment where appropriate; and ensure that the needs of genomics in health are considered in the context of national infrastructure investment.

Activities required to support the research and facilitate implementation include, but are not limited to, the following:

- Early and ongoing engagement with relevant state and territory health departments and health technology assessment processes to support implementation of new technologies
- Harmonised collection of detailed phenotypic information and genomics data from both clinical and research genomes, adhering to international standards; ensuring these data are accessible for clinical and research use.
- Projects developing and supporting international collaboration on data sharing (governance, technological standards).
- Investment in and commitment to Aboriginal and/or Torres Strait Islander control and ownership over research, sample curation, decision making, interpretation and sovereignty — this is paramount.
- Industry partnership and engagement to maximise commercial and clinical benefits from genomics research outputs.
- Links, alignment with and information sharing with relevant policy frameworks, strategies and activities including international best practice and standards.
- Capacity and capability building in enhancing the cultural competency of the genetics workforce; capability to deliver gene therapy in specialised facilities to increase patient access to Aboriginal and/or Torres Strait Islander genomics and bioinformatics researchers and engagement with international consortiums and sources of expertise.

Opportunities to use additional investment and other research to support the priority areas include, but are not limited to, the following:

Collaboration with:

- other MRFF initiatives and missions
- patient support and advocacy organisations (including by utilising the *Principles for Consumer Involvement in Research Funded by the Medical Research Future Fund*)
- industry
- established cohort studies in Australia (e.g. state and national genomics alliances)
- international partnerships, for example:
    - in the context of rare disease: Genomics England, GEM Japan, Care4Rare Canada
    - in the context of international cancer genomics initiatives - ICGC ARGO, Texas Medical Centre, Department of Health and Social Care UK
    - in the context of functional genomics: Canadian RDMM Network, Solve-RD Europe and IRUD Japan
    - International reproductive carrier screening programs, such as NSIGHT in the US
    - in the context of pharmacogenomics England's National Health System

- in the context of governance and technology - European Genome-phenome Archive, ELIXIR
- existing Oceania and regional genomics research programs

To ensure equity of access for all Australians, ensure linkages with:

- Aboriginal and/or Torres Strait Islander genomics projects
- the Aboriginal and/or Torres Strait Islander Advisory Group on Genomics
- Aboriginal and/or Torres Strait Islander-led development of guiding principles for genomics research in Aboriginal and/or Torres Strait Islander people

Leverage knowledge and build on activity and research outcomes of:

- Previously funded GHFM grants see Appendix A
- Other MRFF funded genomics research
- the Australian Genomics Health Alliance research program
- Biobanks including:
  - Existing small biobanks which could be consolidated into a federated network of state and territory facilities
  - Existing or planned biobank infrastructure to incorporate data and physical samples from existing cohorts
  - Federally funded health data networks including: National Collaborative Research Infrastructure Strategy, BioPlatforms Australia, Australian Research Data Commons, Australian Institute of Health and Welfare
  - Existing and piloted data pipeline developed by Australian Genomics

Please note, the above-named organisations, linkages and research areas do not constitute an exhaustive list and are only provided as possible examples.

AIM 1

Faster and more effective disease diagnosis, prevention and earlier intervention

Priority area 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

The term 'rare disease' refers to a presumed monogenic disorder. The focus of this priority area for investment is 'providing a specific molecular diagnosis in rare and undiagnosed diseases presumed to be monogenic.'

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2028	**Objective:** develop new diagnostics to increase rare disease diagnostic rates and address unmet needs for patients. Priority for funding: • Rare diseases in Aboriginal and/or Torres Strait Islander people. • Rare disease where needs have not been addressed and are not being addressed internationally. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by improving patient outcomes and support for clinical/therapeutic care. **Funding:** $7 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years

[1] 'Common cancer' is defined as those with age-standardised rates of 12 or more cases per 100,000 people.
Source: Australian Institute of Health and Welfare

Priority area 1.2

Cancer: Improving early detection and targeted treatment for common cancers[1] to reduce the burden of disease

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2027	**Objective**: develop genomic tools and technologies to identify genetic predisposition to cancer and improve screening and targeted intervention. Priority for funding: **Focus** on increased access to genomic testing and on liquid biopsy. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by increasing access to genomic testing. **Funding:** $5 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years
2028	**Objective:** develop patient-centred, non-invasive approaches to cancer screening and management. Target areas relevant to Australia such as skin cancers or where Aboriginal and/or Torres Strait Islander people have high incidence. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by increasing access to genomic testing. **Funding:** $5.1 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years

Priority area 1.3

Functional genomics: Promoting diagnostic effectiveness and efficiency through better understanding of the impact of genetic variants

The term 'functional genomics' refers to the use of genomics data to understand the impact of genetic variants on gene and protein function and the relationship to disease phenotype.

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2027	**Objective:** conduct scalable research to enhance novel gene discoveries, increase diagnostic rates and enable disease modelling to support development of targeted therapies or prevention strategies. Research is required to: - stratify ways to elucidate the impact of variants of unknown significance in known and novel genes. - develop the application of gene-specific platforms (including multi-omics) to new and/or established cohorts. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by building capacity for turning genomic knowledge into effective diagnosis, prevention and targeted therapies. **Funding:** $28 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years

Priority area 1.4

Infectious disease: Developing novel methods to reduce the impact of infectious diseases and antimicrobial resistance on individual patients and on populations

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2025	**Objective:** develop and/or implement novel genomic methods to improve diagnosis, therapeutic intervention, and/or surveillance of new, emerging, and/or re-emerging infectious diseases, and antimicrobial resistance. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by reducing the impact of infectious diseases and antimicrobial resistance on individual patients and populations. **Funding:** $15 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years

Priority area 1.5

Genomic screening: Improving genomic screening to enable informed decision making for health

Refer to Appendix A for research already funded in this priority.

AIM 2

New, targeted interventions that transform individual and population health

Priority area 2.1

Pharmacogenomics: Promoting precision medicine to improve medication efficacy and reduce harm

The term 'pharmacogenomics' refers to the use of an individual's genomic information to select the best medicine and the best dose to reduce preventable medication harm and improve quality of care.

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2025	**Objective:** Develop and/or implement pharmacogenomic approaches to reduce medication harm and adverse side effects. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by improving precision medicine to enhance medication efficacy and safety. **Funding:** $18 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years
2026	**Objective:** Develop and implement pharmacogenomic approaches to identify new medicines and enhance medication efficacy. Research will focus on one of the following topics: - Cancer therapies - Antimicrobials - Medicines for other infectious or chronic diseases **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by improving precision medicine to enhance medication efficacy and safety.

Starting around	Priorities for investment (objective, outcome, and funding)
	Funding: $15 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years

Priority area 2.2

Common and complex disease: Deploying novel methods to understand the genetic basis of complex diseases

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2025 ($5m) **2027** ($13m)	**Objective:** Develop novel methods for using polygenic risk scores to identify subgroups of the population at high risk of common and complex diseases. Research will focus on one of the following topics: - Cardiovascular disease - Diabetes - Other common and/or complex diseases **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by enhancing risk prediction to support early intervention for people with common and complex diseases. **Funding:** $18 million **Grant Model:** Incubator (approximately 18 grants across 2025 and 2027) **Grant Duration:** up to 2 years
2025 ($5m) **2027** ($14m)	**Objective:** Develop novel methods for improving accuracy and usefulness of polygenic risk scores to stratify people with common cancers for surveillance and treatment. Research will focus on one of the following topics: - Breast cancer - Colorectal cancer - Prostate cancer - Other common cancers **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by enhancing risk prediction to support early intervention for people with cancer. **Funding:** $19 million **Grant Model:** Incubator (approximately 19 grants across 2025 and 2027) **Grant Duration:** up to 2 years

Priority area 2.3

Gene-related therapies: Developing novel therapeutics by investing in promising early-stage products

Starting around	Priorities for investment (objective, outcome, and funding)
2028	**Objective:** fund a large-scale interdisciplinary research program that translates promising novel therapeutics arising out of genomics research through to clinical implementation or commercialisation. Research projects are required to enhance partnerships with clinical services, state/territory and federal government areas and/or industry. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by increasing adoption and implementation of genomics into clinical practice and the commercialisation of genomics research outcomes. **Funding:** $12 million **Grant Model:** Accelerator **Grant Duration:** up to 5 years

Priority area 2.4

Co-developing clinical capabilities for genomics applications that can be embedded in practice

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2025 ($3m) **2027** ($6m) **2028** ($6m)	**Objective:** Undertake research to enhance or streamline uptake of clinical genomics into practice. Research Projects will: - Use best practice implementation science to identify barriers and system changes required for effective implementation. - Conduct research focused on identified populations, geographical locations or health care settings where the proposed research could make most difference. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by improving access to effective, genomics-based diagnostic testing and therapies in health care. **Funding:** $15 million **Grant Model:** Targeted Call for Research **Grant Duration:** up to 5 years

AIM 3

Increased community awareness and engagement, and better understanding of the societal and economic value of genomics in health care and practice

Priority area 3.1

ELSI: Developing a better understanding of the ethical, legal and social implications of genomics, and facilitating public trust and public engagement.

Refer to Appendix A for research already funded in this priority.

Ethical, legal and social implications are to be embedded across all priorities, and all research projects funded under the Genomics Health Futures Mission.

Priority area 3.2

Governance and technology: Developing innovative methods for the ethical and secure governance of genomics data for clinical and research purposes.

Starting around	Priorities for investment (objective, outcome, and funding)
2027	**Objective:** Develop and maintain infrastructure to support research collaboration by enabling data sharing, portability, longevity and connectivity of analysis across Australia. Conduct research addressing emerging ethical, legal and social issues associated with the governance of clinical and genomic datasets with particular focus on the application of advanced analytics (e.g. artificial intelligence) to enhance the diagnostic utility of genomics. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by improving the diagnostic utility of genomic data through enhanced data sharing. **Funding:** $8 million **Grant Model:** Accelerator **Grant Duration:** 3 years

Priority area 3.3

Aboriginal and/or Torres Strait Islander health: Ensuring that Aboriginal and/or Torres Strait Islander peoples contribute to, and control the application of genomics research for, the health benefits to their communities.

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2026	**Objective:** Conduct genomics research to optimise the diagnosis, treatment, monitoring and prevention of high-priority and high-burden diseases among Aboriginal and/or Torres Strait Islander peoples. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by targeting genomic solutions to diseases prevalent among Aboriginal and/or Torres Strait Islander peoples. **Funding:** $29.8 million **Grant Model:** Targeted Call for Research **Grant Duration:** 3-5 years

Priority area 3.4

Australian Genome Reference Database: Enriching population cohorts to bring the benefits of genomics to all members of our multicultural nation.

Refer to Appendix A for research already funded in this priority.

Starting around	Priorities for investment (objective, outcome, and funding)
2025	**Objective:** Conduct small-scale developmental projects to establish feasible, evidence-based genomics approaches for population cohort research that focus on culturally and linguistically diverse communities. Applicants should propose research that: - uses the international landscape to inform an Australian approach. - identifies potential cohorts for analysis and integration. - identifies opportunities for Australian leadership and contribution. **The intended outcome** of the research funded by this grant opportunity is to improve the health and wellbeing of Australians by improving health outcomes in culturally and linguistically diverse communities through the use of genomics. **Funding:** $4 million **Grant Model:** Incubator **Grant Duration:** up to 2 years

Evaluation approach and measures

Aim	Evaluation approach and measures
Aim 1: Faster and more effective disease diagnosis, prevention and earlier intervention	• New predictive and prognostic genomic approaches are identified and developed, enabling improved early detection, screening and targeted therapies for rare diseases, cancer and other conditions that have a genetic basis. • New pathogen genomic approaches are identified and developed, enabling effective infectious disease surveillance and control. • New genomic and functional genomic approaches are identified and developed, enabling improved understanding of the impact on genetic variants. • Research projects integrate partnerships with, and co-design by, Aboriginal and/or Torres Strait Islander peoples and communities. • The community trusts, accepts and adopts new technologies and treatments, as facilitated and evidenced by increased outreach to consumers and healthcare providers, and increased consumer involvement in research design and delivery. • Increased diagnostic rates for rare diseases, cancer and other conditions that have a genetic basis.
Aim 2: New, targeted interventions that transform individual and population health	• New predictive and prognostic pharmacogenomic approaches are identified and developed, enabling improved medication efficacy and reduction of harm. • New predictive and prognostic genomic approaches are identified and developed, enabling early detection, screening and targeted therapies for complex diseases. • Novel gene-related therapeutics are identified and developed. • Genomic technologies are identified and developed, facilitating precision medicine in health care and practice. • Increased focus of research on areas of unmet need as evidenced by increased investment and improved health outcomes in Australians living with unmet need. • Research community has greater capacity to undertake translational research, as evidenced by a health professional workforce trained in integrating genomics in practice, and projects that involve partnerships across regions, nationally, and globally.

Aim	Evaluation approach and measures
	• Research projects integrate partnerships with, and co-design by, Aboriginal and/or Torres Strait Islander peoples and communities. • The community trusts, accepts and adopts new technologies and treatments, as facilitated and evidenced by increased outreach to consumers and healthcare providers, and increased consumer involvement in research design and delivery.
Aim 3: Increased community awareness and engagement, and better understanding of the societal and economic value of genomics in health care and practice	• Research results in strategies to improve public acceptance of the use of genomics. • Novel applications using genomics data drive improvements in health care, practice and outcomes. • The community trusts, accepts and adopts new technologies and treatments, as facilitated and evidenced by increased outreach to consumers and healthcare providers, and increased consumer involvement in research design and delivery. • Genomic research encompasses the diversity of the Australian population and includes underrepresented multicultural populations. • Research projects integrate partnerships with, and co-design by, Aboriginal and/or Torres Strait Islander peoples and communities.