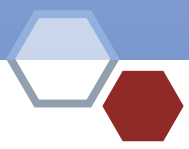
Australian Government
Department of Health

Medical Research Future Fund

Genomics Health Futures Mission

Implementation plan

November 2020



Background

The Genomics Health Futures Mission (GHFM) is investing $500 million over 10 years in genomics research under the Medical Research Future Fund (MRFF). It will improve testing and diagnosis for genetic diseases, 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 in partnership with Aboriginal and/or Torres Strait Islander people to deliver genomics research that is scientifically sound, culturally safe and competent to address inequity in research participation and outcomes.

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 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 areas for research investment 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 the most 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 innovative 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 the primary health care sector
3. Increased community awareness and engagement, and better understanding of the societal and economic value of genomics in health care	**3.1** ELSI: Developing a better understanding of the ethical, legal and social implications of genomics, and 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 people 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 help to clarify the intended outcome and facilitate tracking of the GHFM's progress towards its objectives.

Priority areas for investment are allocated across short, medium and long-term timeframes. There are currently some gaps in the long term to be informed by the outcomes of initial GHFM funding rounds, emerging health priorities, and a national and international landscape and gap analysis.

GHFM enablers

For each aim and priority area for investment, the implementation plan identifies non-research activities required to facilitate and support the MRFF-funded research and long-term implementation.

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 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

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, including 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.

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.'

Research to begin in the ...	Priorities for investment (research questions and objectives)
short term 1–2 years	Three rare disease research projects commenced in 2020: • Precision medicine for life-threatening infantile epilepsy (University of Melbourne, $4.1 million) • National rapid genomic diagnosis program for critically ill children (Murdoch Children's Research Institute, $4.8 million) • PreGen: Filling the Gap — antenatal genomics and newborn care (University of New South Wales, $4.9 million)
medium term 2–5 years	Identify and diagnose novel — primarily monogenic or oligogenic — rare diseases and increase the genomic diagnostic rate towards 70% by 2025.
long term 6–10 years	Identify and diagnose novel — primarily monogenic or oligogenic — rare diseases and increase the genomic diagnostic rate towards 90% by 2030.

Opportunities to use additional investment and other research to support the priority areas

Collaborative research with other MRFF initiatives and missions in the context of rare disease:

- Clinical Trials for Rare Cancers, Rare Diseases and Unmet Needs
- Indigenous Health Research Fund

Collaborate with:

- established rare disease cohort studies in Australia (eg state and national genomics alliances)
- international partnerships (eg Genomics England, GEM Japan, Care4Rare Canada)
- patient support and advocacy organisations
- industry (eg InGeNA)

Activities required to support the research and facilitate long-term implementation

- 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
- Early and ongoing engagement with relevant state and territory departments of health and health technology assessment processes to support implementation of new technologies
- Link into a functional genomics pipeline to identify therapeutic targets, develop novel therapies and facilitate clinical trials (see Priority area 1.3)

Priority area 1.2

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

The term ‘cancer’ refers to the common cancers that have the greatest burden of disease in Australia.

Research to begin in the …	Priorities for investment (research questions and objectives)
short term 1–2 years	2018 — ProCan: The Australian Cancer Research Foundation International Centre for the Proteome of Human Cancer ($20.4 million). Two genomics research projects in cancers with high mortality and low survivability commenced in June 2020: • SUPER-NEXT: Complete genome profiling for cancer of unknown primary (University of Melbourne, $4.9 million) • Whole genome sequencing in high-risk breast cancer patients (University of Queensland, $2.8 million)
medium term 2–5 years	Discover clinically relevant biomarkers to improve early detection of common cancers. Develop genomic tools and technologies to identify genetic predisposition to cancer and improve screening and targeted intervention. Develop genomic screening tools for predictive disease modelling and to guide therapy.
long term 6–10 years	Develop patient-friendly, noninvasive approaches to cancer screening and management. Explore methodologies for longitudinal cancer management using genomics and functional genomics.

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of cancer
  - Brain Cancer Mission
  - Clinical Trials for Rare Cancers, Rare Diseases and Unmet Needs
  - Indigenous Health Research Fund
- Zero Childhood Cancer Program: childhood brain cancer ($5 million); all childhood cancers ($54.8 million)
- Australian Genomic Cancer Medicine Program ($50 million)
- Collaboration with international cancer genomics initiatives (eg ICGC ARGO, Texas Medical Centre, Department of Health and Social Care UK) using the Department of Health's existing memorandums of understanding

Activities required to support the research and facilitate long-term implementation

- Established links aligning cancer research with the relevant cancer policy frameworks (Australian Government or state and territory governments)
- Early and ongoing engagement with relevant state or territory departments of health and health technology assessment processes to support implementation of new technologies
- Biobanking of tissues, and developing and cataloguing of patient-derived clinical models

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.

Research to begin in the …	Priorities for investment (research questions and objectives)
short term 1–2 years	Genomic autopsy of perinatal death (University of South Australia, $3.4 million). Funding of functional genomics projects through the GHFM cardiovascular genetics flagship ($1.2 million).
medium term 2–5 years	Develop disease-agnostic, high-throughput and scalable functional genomics platforms to increase the diagnostic rate of unresolved cases and accelerate the development of experimental therapeutics. Establish a national functional genomics approach to enhance novel gene discoveries, increase diagnostic rates and enable disease modelling to support development of targeted therapies.

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of functional genomics
  - Stem Cell Therapies Mission
  - Cardiovascular Mission
  - Indigenous Health Research Fund
- Working closely with GHFM Rare Disease networks and Cancer projects to resolve undiagnosed cases and variants of unknown significance
- Building on activity of the MRFF-funded Cardiovascular Genetic Disorders Flagship Functional Platforms
- Potential for industry partnership for expansion and industrialisation of functional platforms
- International collaboration — Canadian RDMM Network, with Solve-RD Europe and IRUD Japan

Activities required to support the research and facilitate long-term implementation

- A coordinated national functional genomics network, linked with other relevant networks, with the objective of validating novel gene discoveries, stimulating national activity in novel and known gene modelling, and increasing the efficacy of genomics to diagnostics

Priority area 1.4

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
short term 1–2 years	Host gene expression signatures to diagnose sepsis in children (University of Queensland, $2.4 million).
medium term 2–5 years	Four pathogen genomics research projects announced in October 2020: • Precision Public Health in Australia through Integrated Pathogen Genomics (University of Melbourne, up to $10.0 million) • H2Seq: Viral genomics for public health interventions in HIV and HCV (University of New South Wales, up to $6.6 million) • Genomics, Digital Health and Machine Learning: the SuperbugAi Flagship (Monash University, up to $3.4 million) • META-GP: Delivering a Clinical Metagenomics Platform for Australia (University of Melbourne, up to $6.9 million) Develop novel methods for infectious disease surveillance.

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of infectious disease, including the Indigenous Health Research Fund
- Tracking COVID-19 in Australia using genomics ($3.27 million in 2019–20)

Activities required to support the research and facilitate long-term implementation

- Alignment with the antimicrobial genomics policy framework
- Early and ongoing engagement with relevant state or territory departments of health and health technology assessment processes to support implementation of new technologies
- Establish links and information sharing with national and international infectious disease strategies and activities, such as
  - Communicable Diseases Network Australia
  - Communicable Diseases Genomics Network
  - US GenomeTrakr
  - Public Health England

Priority area 1.5

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
short term 1–2 years	Australian Reproductive Carrier Screening Project (Mackenzie's Mission, $20 million over 3 years).
medium term 2–5 years	Develop new models of genomic screening, including: • non-invasive prenatal testing • newborn screening

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of screening
  - Mackenzie's Mission
  - Rare Disease projects
  - Australian Genome Reference Database
  - Indigenous Health Research Fund
- International reproductive carrier screening programs, such as NSIGHT in the US

Activities required to support the research and facilitate long-term implementation

- Links with policy owners, Australian Government and state and territory governments
- Early and ongoing engagement with relevant health technology assessment processes to support implementation of new technologies
- Links with population genomics projects

Evaluation approach and measures

- 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 people and communities
- The community trusts, accepts and adopts new technologies and treatments

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.

Research to begin in the ...	**Priorities for investment (research questions and objectives)**
medium term 2–5 years	Conduct small-scale development projects to establish feasible, evidence-based approaches for pharmacogenomics research, including: • using the international landscape to inform an Australian approach • identifying potential cohorts for analysis • identifying opportunities for Australian leadership and contribution
medium term 2–5 years to **long term** 6–10 years	Develop pharmacogenomic approaches to enhance medication efficacy and reduce harm by preventing adverse side effects.

AIM 2

Opportunities to use additional investment and other research to support the priority areas

- Build on research outcomes from four projects funded under the MRFF 2019–20 Mental Health Pharmacogenomics grant opportunity
  - Effects of using pharmacogenomics to prescribe antidepressants on depression outcomes in patients with major depressive disorder in primary care (University of Melbourne, $1.39 million)
  - How we can improve the performance of pharmacogenomics in Australia? (Queensland Institute of Medical Research, $1.37 million)
  - Investigate the pharmacogenomic signatures of bipolar disorder for improving treatment outcomes (University of New South Wales, $1 million)
  - Genotype-guided versus standard psychotropic therapy in moderately-to-severely depressed patients (University of New South Wales, $2.95 million)
- Links with England's National Health System for a potential pilot research study in the context of pharmacogenomics
- Collaborative research with other MRFF initiatives and missions in the context of pharmacogenomics
  - Million Minds Mental Health Research Mission to predict medication responsiveness and compliance
  - Indigenous Health Research Fund
- Explore opportunities for pharmacogenomic interventions in context of the 10-year Primary Health Care Plan

AIM 2

Activities required to support the research and facilitate long-term implementation

- Links and information sharing with international pharmacogenomics initiatives to inform research investments and activities
- Early and ongoing engagement with relevant state or territory departments of health and health technology assessment processes to support implementation of new technologies

Priority area 2.2

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
medium term 2–5 years	Conduct small-scale development projects to establish feasible, evidence-based approaches for common and complex disease research, including: • using the international landscape to inform an Australian approach • identifying potential cohorts for analysis • identifying opportunities for Australian leadership and contribution
medium term 2–5 years to **long term** 6–10 years	Identify novel methods for using polygenic risk scores to: • identify subgroups of the population at high risk of common and complex diseases, including cardiovascular disease and diabetes • stratify people with common cancers for surveillance and treatment, including breast, colorectal and prostate cancer

AIM 2

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of common disease
  - Cardiovascular Health Mission
  - Million Minds Mental Health Research Mission
  - Indigenous Health Research Fund
- Collaborative research with the Australian Prevention Partnership Centre
- Linkages with Aboriginal and/or Torres Strait Islander genomics projects and population genomics activity to ensure equity of access for all Australians
- Links to the 10-Year Primary Health Care Plan

Activities required to support the research and facilitate long-term implementation

- Early and ongoing engagement with relevant state or territory departments of health and health technology assessment processes to support implementation of new technologies

Priority area 2.3

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
medium term 2–5 years	Conduct small-scale development projects to establish feasible, evidence-based approaches for research on gene-related therapies, including: • using the international landscape to inform an Australian approach • identifying opportunities for Australian leadership and contribution Develop pre-clinical evidence of organ-focused (eg eye, liver, central nervous system and haematopoietic system) gene-related therapies.
long term 6–10 years	Develop technologies to enable clinical use of gene-related therapies.

AIM 2

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of gene-related therapies
- Interrogation of the ethical, legal and social implications of somatic gene therapy to relieve critical translational barriers
- Partnership with industry (eg InGeNa)

Activities required to support the research and facilitate long-term implementation

- National collaborative gene therapy network with specialised vector production facilities at a limited number of sites to avoid fragmentation and duplication
- National capability to deliver gene therapy in specialised facilities to increase patient access
- Industry partnership and engagement to maximise commercial and clinical benefits from genomics research outputs

Priority area 2.4

Co-developing clinical capabilities for genomics applications that can be embedded in the primary health care sector

Research to begin in the ...	Priorities for investment (research questions and objectives)
medium term 2–5 years	Conduct implementation research on strategies for improving clinical capacity to apply genomics in the primary health care sector.

Opportunities to use additional investment and other research to support the priority areas

- Leverage knowledge developed from the Australian Genomics Health Alliance research program and the Mackenzie's Mission pilot research project
- Pharmacogenomics projects of the GHFM
- Collaborative research with other MRFF initiatives and missions, including the Indigenous Health Research Fund, in the context of applying genomics in primary care

Activities required to support the research and facilitate long-term implementation

- Links to the 10-Year Primary Health Care Plan
- GHFM research outputs will inform the Project Reference Group for Health Genomics in driving the implementation of the National Health Genomics Policy Framework to harmonise translational research and provide a path for application of results

AIM 2

Evaluation approach and measures

- 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 primary care
- Increased focus of research on areas of unmet need
- Research community has greater capacity to undertake translational research
- 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

AIM 3

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

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
short term 1–2 years	Eight GHFM-funded research projects addressing the ethical, legal and social implications related to genomics in health care commenced in 2020 (~$3.7 million): • Preventing mitochondrial disease using genomics (Monash University) • Genome editing: formulating an Australian community response (University of Tasmania) • Towards a trusted genomic repository: tackling commercialisation fears (Swinburne University of Technology) • Cascade testing in intellectual disability: social and economic impact (Macquarie University) • Moratorium on genetic testing and life insurance: monitoring the impact (Monash University) • Centre for Ethics of Paediatric Genomics to Improve Paediatric Care (Murdoch Children's Research Institute) • Returning raw genomic data: patient autonomy or legal minefield? (University of Tasmania) • 'We need to talk': genomics and disability (University of Queensland)
medium term 2–5 years	Conduct research focused on ethical, legal and social implications relating to: • fostering trust and confidence within the community • engaging with Aboriginal and/or Torres Strait Islander peoples • engaging with industry • engaging with clinicians and health systems • developing data analytics capability

AIM 3

Opportunities to use additional investment and other research to support the priority areas

- National Health Genomics Policy Framework
- Collaborative research with other MRFF initiatives and missions, including the Indigenous Health Research Fund, in the context of ELSI related to the use of genomics data and information in health care

Activities required to support the research and facilitate long-term implementation

- Develop policies for patient and consumer involvement to guide and enrich GHFM-funded projects

Priority area 3.2

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
medium term 2–5 years	Conduct research addressing emerging ethical, legal and social issues associated with the governance of clinical and genomic datasets.
long term 6–10 years	Apply advanced analytics (eg artificial intelligence) to enhance the diagnostic utility of genomics.

Opportunities to use additional investment and other research to support the priority areas

- Existing small biobanks are 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: National Collaborative Research Infrastructure Strategy, BioPlatforms Australia, Australian Research Data Commons, Australian Institute of Health and Welfare
- International initiatives: European Genome-phenome Archive, ELIXIR
- Existing and piloted data pipeline developed by Australian Genomics

Activities required to support the research and facilitate long-term implementation

- Develop standardised governance frameworks for implementation across mission projects to support aggregation and management of research data
- Projects developing and supporting international collaboration on data sharing (governance, technological standards)
- Align with the blueprint for national genomics information management prepared by Queensland Health and the Queensland Genomics Health Alliance
- Align with international best practice and standards developed by the Global Alliance for Genomics and Health
- Development of a data plan outlining critical components and requirements to support GHFM research outputs, including the need to link research and clinical genomics data systems in Australia and internationally
- Evaluation of international best practice for standardised implementation across GHFM projects to support aggregation and management of genomics data from GHFM-funded projects
- Determining stakeholder needs across the genomics community (including research, clinical and vulnerable populations), developing a suite of strategies reflecting international best practice, and adopting standards to facilitate dissemination and consistency

Priority area 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

Research to begin in the ...	Priorities for investment (research questions and objectives)
short term 1–2 years	Two GHFM research projects commenced in June 2020: • Genomic architecture of chronic disease in Australia's First Peoples (University of Queensland, $1.4 million) • Achieving equity in genomic health for Indigenous Australians (University of Melbourne, $500,000)
medium term 2–5 years	Conduct research to develop effective approaches for responsible, culturally appropriate and nationally consistent involvement of Aboriginal and/or Torres Strait Islander people in genomics research and clinical practice. Conduct implementation research addressing inequalities of access and increasing the quality of clinical genetics services for Aboriginal and/or Torres Strait Islander people.
long term 6–10 years	Conduct research to optimise the diagnosis, treatment, monitoring and prevention of high-priority and high-burden diseases among Aboriginal and/or Torres Strait Islander people.

AIM 3

Opportunities to use additional investment and other research to support the priority areas

- Indigenous-led development of guiding principles for genomics research in Aboriginal and/or Torres Strait Islander communities
- Collaborative research with other MRFF initiatives and missions, including the Indigenous Health Research Fund, in the context of precision medicine for Aboriginal and/or Torres Strait Islander communities

Activities required to support the research and facilitate long-term implementation

- 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
- GenetiQs Project and SING Australia (QIMR Berghofer)
- National Centre for Indigenous Genomics
- Alignment with Aboriginal and/or Torres Strait Islander health strategies
- Consultation strategy to ensure GHFM activity is implemented in an ethical and culturally appropriate way
- Engage with the Aboriginal and Torres Strait Islander Advisory Group on Genomics to ensure co-design of research programs
- Capacity and capability building in
  - Indigenous genomics and bioinformatics researchers
  - enhancing the cultural competency of the genetics workforce
  - Aboriginal primary health care workforce
  - engagement with international consortiums and sources of expertise

Priority area 3.4

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

Research to begin in the ...	Priorities for investment (research questions and objectives)
medium term 2–5 years	Conduct small-scale development projects to establish feasible, evidence-based approaches for population cohort research, focusing on culturally and linguistically diverse groups, including: • using the international landscape to inform an Australian approach • identifying potential cohorts for analysis and integration • identifying opportunities for Australian leadership and contribution Conduct research to enrich genomics databases to support the inclusion of culturally and linguistically diverse groups in genomics research, and equitable benefit from genomics in health.

AIM 3

Opportunities to use additional investment and other research to support the priority areas

- Collaborative research with other MRFF initiatives and missions in the context of large cohort study development (eg Mackenzie's Mission)
- Engagement with existing Oceanic genomics research programs and the Aboriginal and Torres Strait Islander Advisory Group on Genomics

Activities required to support the research and facilitate long-term implementation

- A strategy for GHFM contribution to an expansion of genome reference databases is required to cover the full range of Australian diversity and ensure that the benefits of genomic medicine can be extended to all Australians
- Community consultation and scope definition: identifying communities of greatest need, determining existing Australian cohorts with representation of these groups and appropriate consent, community engagement protocols, and data-sharing permissions
- Alignment with international population reference databases (eg gnomAD)
- Alignment with the blueprint for national genomics information management prepared by Queensland Health and the Queensland Genomics Health Alliance

AIM 3

Evaluation approach and measures

- Research results in strategies to improve public acceptance of the use of genomics
- Novel applications using genomics data drive improvements in health care and outcomes
- Genomic research encompasses the diversity of the Australian population
- Research projects integrate partnerships with, and co-design by, Aboriginal and/or Torres Strait Islander people and communities