



Australian Government

Department of Health,
Disability and Ageing

National Health Genomics Policy Framework and Implementation Plan 2026-2030

About this draft

This version of the National Health Genomics Policy Framework and Implementation Plan 2026-2030 is a working draft intended to support public consultation and feedback. Additional text boxes like this appear throughout to provide relevant context to readers.

Foreword

A brief introductory statement will be included to introduce the document and highlight its importance. It is anticipated this foreword will be written by Health Ministers and/or the Australian Health Genomics Commissioner. This will be developed following public consultation.

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Overview

A one-page graphical representation of the key elements of this document and a one-page written summary will be developed following public consultation.

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Terminology

The term genomics in this document refers to both [genetics](#) and [genomics](#), other than where it is necessary to distinguish between the two terms. This document considers genomics to be an extension of genetics, rather than a distinct field.

Purpose

The purpose of the *National Health Genomics Policy Framework and Implementation Plan 2026-2030* (this framework and implementation plan) is to provide a national, intergovernmental approach to embed genomics into the Australian health system. It seeks to maximise health and wellbeing outcomes and minimise fragmentation and duplication of effort. It sets out how Australian governments will collaborate and coordinate to embed genomics into the health system.

Vision

All people who access health care in Australia benefit from appropriately embedded genomics within the health system.

Mission

To establish and/or maintain processes that support [person- and family-centred](#), culturally safe, efficient, effective, ethical, and equitable implementation of [genomics-informed health care](#) and research in Australia.

Audience

This framework and implementation plan is primarily directed at decision and policy makers at the national, state and territory, and health service levels.

It is also intended to be a useful resource for members of the public, communities, advocacy organisations, industry, academics, researchers, clinicians and others. It provides visibility of the shared strategic priorities, desired outcomes, and activities of Australian governments. This may support the non-government-led development of policies, strategies, actions and health services that, where relevant, consider and ideally align with this framework and implementation plan.

Scope

The immediate focuses of this framework and implementation plan are health care and research involving the use of human [genomic information](#) to support the health and wellbeing of individuals and populations. This covers applications involving or informed by human [genomic testing](#) to prevent, diagnose, and treat genomic conditions, including [precision medicine](#). It is also inclusive of genomics in cancer care involving both [germline](#) and [somatic variants](#) (which is given heightened focus under the [National Framework for Genomics in Cancer Control](#)).

The scope of this framework and implementation plan is intended to be flexible to accommodate the rapid advancements in genomics and related fields, as was the case with the [National Health Genomics Policy Framework 2018–2021](#) (the lapsed framework).

Integrating genomic information with other emerging ‘omics’ technologies and information (including [proteomics](#), [transcriptomics](#), [metabolomics](#), [lipidomics](#), and [epigenomics](#)) is recognised as important to maximise health and wellbeing outcomes. Where relevant, strategic priorities, outcomes and activities of this framework and implementation plan will be progressed with consideration of omics and other emerging related technologies.

Other applications of genomic knowledge that contribute to improved individual and population health and wellbeing outcomes, such as the genomic testing of infectious [microbes](#) (which is the focus of the [National Microbial Genomics Framework for Public Health 2025–2027](#)) and other non-human genomics (such as human [microbiomics](#), the study of bacteria, viruses and fungi that live in and on the human body) are out of scope.

However, it is acknowledged there are close links and interdependencies between human, animal, plant and wider environmental health. [One Health](#) approaches to embedding genomics will likely have greater benefits for health and wellbeing than if the sectors independently consider genomics. Australian governments remain committed to pursuing a more unified, multi-sectoral approach over the longer term. It is anticipated that work will occur in parallel to this framework and implementation plan to support this commitment.

Strategic Context

Genomics is already improving health outcomes for individuals, families and communities across Australia. Rapid advancements in technology and evidence are supporting ongoing shifts in health care and research from genetics to genomics. This is accompanied by and necessitating shifts from care models that involve specialised [clinical genetics](#) services to those that integrate genomics into the routine practice of the [mainstream health workforce](#) including general practitioners (with this shift commonly referred to as [mainstreaming](#)). This supports specialised clinical genetics services to focus on complex patients.

Current applications of genomics in health care include:

- characterising, diagnosing and managing conditions (particularly rare inherited conditions and cancers, though increasingly relatively common conditions across a broad range of specialties) including reducing the time it takes to diagnose these conditions
- providing individuals and families with information on risk of developing conditions, and determining the need for ongoing surveillance, lifestyle changes and early interventions and treatment
- providing couples who are planning or actively trying to have a baby with information on their [carrier](#) status for inheritable conditions to inform their risk of having a child with one or more of these conditions
- supporting access to targeted treatments, such as [immunotherapies](#) and [cell and gene therapies](#), including those in clinical trials.

Other applications that are in the process of being embedded in health care or are a focus of current research include:

- [pharmacogenomics](#), the study of the role of the genome in the effectiveness and safety of medications
- the use of genomics for [population screening](#), health screening programs that look to identify genetic risk of certain conditions in target groups to facilitate prevention and early diagnosis and treatment
- integrating genomic information into electronic medical records and supporting linkages with familial and longitudinal information
- adopting [multi-omics](#) approaches that integrate genomic information with other emerging 'omics' technologies and information
- ensuring equity for populations that have historically and continue to face barriers to accessing and benefitting from advancements in genomics, such as Aboriginal and Torres Strait Islander people.

Background information on the development of this framework and implementation plan and the Australian health system are included in [Appendix 2](#) and [Appendix 3](#) respectively.

Implementation

This framework and implementation plan sets out high-level national activities. The activities reflect foundational work that will need to be undertaken to appropriately embed genomics within the health system in ways that are sustainable over a longer term than the life of this framework and implementation plan. Many of the outcomes are ambitious and go beyond what may be achievable over five years.

The Australian Government and state and territory governments have been identified to oversee and coordinate implementation of specific activities. Identified governments will be responsible for developing more detailed plans for activities in consultation with relevant stakeholders. However, all national activities will require Australian governments to work together. Adaptation will be needed across states and territories to account for differing government and local health system designs, legislation, priorities, and population needs.

The timeframes, identification of the government responsible for coordination, and prioritisation of activities will be developed following public consultation.

Background information on the Australian health system is included in [Appendix 3](#).

Priority Populations

In Australia, a number of groups disproportionately face challenges in accessing health care, taking part in research, and achieving good health and wellbeing outcomes. This includes access to and outcomes of genomics-informed health care and research.

In addition to Aboriginal and Torres Strait Islander people and communities, which are explicitly referenced throughout and are the focus of [Strategic Priority 4](#), other priority populations will need to be considered under this framework and implementation plan in accordance with government priorities.

All activities progressed under this framework and implementation plan will apply an equity lens. While the [social determinants of health](#) and avoidable inequities fall beyond the scope of this framework and implementation plan, it is important that consideration is given to how the planning and implementation of activities will affect priority populations to ensure equity is promoted. Individuals within priority population groups will have unique experiences and needs. In most instances, one-size-fits-all approaches are unlikely to be sufficient. Well considered engagement and partnerships will be required.

Guiding Principles

Five guiding principles have been identified to guide planning and implementation of all strategic priorities, outcomes and activities of this framework and implementation plan, and underpin the embedding of genomics into the Australian health system.

1. Equity

Equitable access to and outcomes of genomics-informed health care and research are effectively promoted, enabled and monitored. This includes addressing the needs of identified priority populations.

2. Empower individuals and communities

Individuals, their families and communities are central to their health care and are empowered to be aware of, involved in and benefit from genomics-informed health care and research.

Aboriginal and Torres Strait Islander people are empowered to determine efforts to embed genomics in ways that deliver benefits to their communities according to their own priorities and at their own pace and are supported by Australian governments.

3. Evidence-based and socially accepted

All clinical applications of genomics and genomics-informed health care are:

- ethically, legally and socially responsible and culturally appropriate
- supported by community trust and acceptance
- informed by research and evidence
- sustainably funded and embedded
- targeted to advance individual and population health and wellbeing.

4. Prevention-focused

Embedding clinical applications of genomics knowledge across Australia advance health and wellbeing outcomes and long-term sustainability by supporting a shift in the focus of health systems to:

- prevention
- early intervention
- effective and efficient precision medicine.

5. Whole-of-system focused

Efforts to embed genomics consider and account for the ways in which individuals interact with different parts of the Australian health system. This includes coordination across public and private health sectors, the research sector, and state and territory borders with consideration of services, workforce, and infrastructure. Efforts should support development of a nationally unified, locally controlled health system that supports and encourages integrated person- and family-centred care.

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Enablers of Success

Four key enablers have been identified to support successful planning and implementation of all strategic priorities, outcomes and activities of this framework and implementation plan, and the realisation of its vision and mission.

1. Collaborative and culturally appropriate governance, leadership, information-sharing and, where necessary, coordination across Australian governments

Federal, state and territory government leaders working together and in partnership with Aboriginal and Torres Strait Islander leaders is essential to a whole-of-governments and whole-of-system approach to equitably embedding genomics into the Australian health system.

This will involve fostering knowledge exchange, consideration of how the different parts of the Australian health system interact and working together to support health and wellbeing outcomes for all Australians.

2. Flexibility

Effective implementation will require flexibility over time. This is supported through the relative prioritisation of activities, and the establishment of appropriate governance structures and processes to allow adjustments during the timespan in response to external developments.

The timeframes, identification of the government responsible for coordination, and prioritisation of activities will be developed following public consultation.

3. Individual, community and stakeholder engagement and partnerships

All efforts under the framework and implementation plan must involve active engagement of individuals, families, communities, and public and private health sectors. This includes system managers, [clinicians and support staff working in specialised genomic](#) and mainstream health care, advocacy groups, and industry. It also includes researchers, universities and academics, regulators, relevant networks, and other stakeholders. This will build trust, and ensure all activities are progressed in ways that are consistent with values and meet expectations and needs.

4. International engagement and partnerships

Engagement and partnerships with overseas entities and genomics initiatives may complement the integration of genomics into health care and accelerate the pace of research in Australia.

Governance, Accountability, Monitoring and Evaluation

Effective governance and accountability arrangements are essential for driving coordinated implementation of this framework and implementation plan and supporting flexibility over time.

The Health Chief Executives Forum (HCEF) is responsible for governance of this framework and implementation plan and will have overall accountability for the success of its implementation. It will be assisted by supporting committees and subgroups it determines as necessary.

It will collaborate with the Australian Alliance for Indigenous Genomics (ALIGN) network and other peak bodies that represent Aboriginal and Torres Strait Islander people.

It is intended HCEF will prepare and publish an annual report on progress and challenges. The annual report will recommend amendments and updates to this framework and implementation plan during its five-year timeframe, as required.

A monitoring and evaluation plan has been developed to support this process. It commits Australian governments to share information to formally monitor progress and achievement of identified outcomes. It is intended to support independent evaluation throughout this framework and implementation plan's five-year timeframe. This independent evaluation will commence as soon as practicable following publication of this framework and implementation plan and will be managed by Genomics Australia.

All activities will require coordination across Australian governments and in consultation and partnership with relevant non-government stakeholders.

These governance and accountability arrangements will ensure implementation is monitored and stays on track by providing clear pathways for reporting, approval, and escalation and resolution of identified issues.

Strategic Priorities

This framework and implementation plan outlines four strategic priorities. The strategic priorities highlight key areas of focus for Australian governments and were developed to address identified opportunities and challenges.

Strategic Priority 1 — Person-, Family- and Community-Centred Approach

- Ensure people, their families and communities are central to, involved in and benefit from genomics-informed health care and research
- Increase equity of access to genomics-informed health care and research, particularly for priority populations and those who are currently underserved
- Strengthen the genomics knowledge of the public, to support informed decision-making about genomics-informed health care and research.

Strategic Priority 2 — Dynamic and Sustainable Genomics Ecosystem

- Improve understanding of the genomics landscape and patient journeys across health care and research nationally, to support development of the genomics ecosystem
- Support the specialised genomic and mainstream health workforce to deliver high-quality, appropriate genomics-informed health care
- Support efficient and effective genomics research innovation and translation.

Strategic Priority 3 — Genomic Samples, Data and Information Management

- Establish and enhance infrastructure, cyber security frameworks, workforce, standards and processes to support genomic information collection, sharing, use and management
- Promote ethical, safe, secure, trusted and equitable genomic information collection, sharing, use and management.

Strategic Priority 4 — Aboriginal and Torres Strait Islander-led Genomics

- Establish and strengthen partnerships between Australian governments and Aboriginal and Torres Strait Islander people, communities and stakeholders to improve equity of access and ensure that the benefits of genomics-informed health care are realised
- Empower Aboriginal and Torres Strait Islander people to lead efforts related to embedding genomics in accordance with their priorities and protocols, and at their own pace
- Support [Indigenous Data Sovereignty](#), the right of First Nations people to govern the collection, sharing, use and management of their own data, in Australia.

The timeframes, identification of the government responsible for coordination, and prioritisation of activities will be developed following public consultation.

Strategic Priority 1 – Person-, Family- and Community-Centred Approach

Person- and family-centred care involves a collaborative approach to health care which allows patients and their families to be actively involved in shared decision-making and healthcare planning and delivery. It is a partnership between healthcare providers, patients and their families. It emphasises understanding and respecting the unique needs, values, and preferences of individuals and families. Making sure people are involved with and central to their health care and have trust in health services, systems and research, including those incorporating genomics, is critical to improving outcomes. Key components of building trust include:

- ensuring public knowledge and understanding of genomics is enhanced
- considering and addressing cultural, ethical, legal and social implications
- considering and addressing individual, family and community priorities and needs

In addition to building trust, ensuring all Australians have access to and benefit from genomics-informed health care and research is essential. This requires understanding and addressing barriers faced by different populations. It is dependent on enhancing the capacity and addressing the limitations of Australia's health system and research sector through partnership, targeted efforts and appropriate resourcing.

Outcome 1.1 — Empower informed decision-making and community engagement

- individuals, families and communities are supported to understand voluntary and informed consent processes for health care and, for Aboriginal and Torres Strait Islander people, in keeping with the notion of free and prior informed consent
- individuals, families and communities have visibility and access to their genomics-related health information to ensure they can make informed choices about their health care
- individuals, families, and communities are supported and empowered to participate and lead in public discourse on matters related to genomics-informed health care and research
- community engagement, understanding and trust in genomics-informed health care, health systems and research is built and maintained
- all genomics-informed health care and research are undertaken with a view to achieving benefit for the individual, their families and/or the broader public, in alignment with individual and community values and expectations
- access to and benefits of genomics-informed health care and research are known, understood and available to all Australians, particularly for priority population groups

National Activity	Timeframe	Responsible for Coordination	Priority
<p>1.1.1 — Establish and maintain processes to engage Australian governments with individuals who have already benefitted from or are likely to benefit from embedding genomics in the Australian health system, as well as patient, consumer advocacy and other stakeholder groups that represent the views of those individuals, to inform other activities of this framework and implementation plan by:</p> <ul style="list-style-type: none"> • understanding their priorities, values and expectations relevant to genomics • discussing identified policy concerns of national significance (including those that consider cultural, ethical, legal, social and funding implications) 			
<p>1.1.2 — Establish and maintain processes to engage Australian governments with representative samples of the Australian population (i.e., inclusive of those individuals who would not be engaged under Activity 1.1.1) to inform other activities of this framework and implementation plan by:</p> <ul style="list-style-type: none"> • understanding their priorities, values and expectations relevant to genomics • discussing identified policy concerns of national significance (including those that consider cultural, ethical, legal, social and funding implications) 			

1.1.3 — Evaluate and, if necessary, refine the existing [National Model of Consent for Clinical Genomic Testing](#) in line with community values and expectations and the needs of clinicians and health services, including consideration of how the model does or does not intersect with consent for secondary purposes and research.

1.1.4 — Develop a nationally cohesive strategy to promote adoption, adaptation and implementation of the [National Model of Consent for Clinical Genomic Testing](#) across the Australian health system.

1.1.5. — Consult with public and private pathology laboratories, health services, peak bodies and industry to determine how Australian governments can best support efforts to achieve nationally consistent genomic test result reporting.

1.1.6 — Consult with public and private pathology laboratories, health services, peak bodies and industry to determine how Australian governments can best support the consented uploading of accessible genomics test result reports to national and state and territory patient digital health records.

Outcome 1.2 — Improve public genomics knowledge

- individuals, families and communities have equitable access to evidence-based, culturally and linguistically appropriate educational resources to support improvement of genomics knowledge and trust, and decision-making about the use of genomics in health care

National Activity	Timeframe	Responsible for Coordination	Priority
1.2.1 — Engage with communities to understand what educational resources for the public are expected, valued and needed to improve knowledge and trust and support informed decision-making related to genomics, and identify currently available resources and what additional resources are desired or needed nationally and in states and territories.			
1.2.2 — Establish a process to: <ul style="list-style-type: none">• endorse existing or develop new accessible national educational resources for the public that improve knowledge and trust and support informed decision-making related to genomics• leverage existing or develop new state and territory specific genomics resources that support patient access to public hospital services in alignment with local population needs and health system design			

1.2.3— Establish a process or mechanism to make nationally endorsed educational resources available, and to direct the public to patient and community advocacy groups, information about clinical trials, and state and territory specific genomics resources, in line with public expectations and preferences.

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Strategic Priority 2 – Dynamic and Sustainable Genomics Ecosystem

Realising the full potential of genomics-informed health care is dependent on a dynamic and fit-for-purpose genomics ecosystem that fosters and embraces research and innovation. This will require an informed and capable multidisciplinary workforce and the necessary infrastructure to ensure that the health benefits of genomic knowledge and technology are harnessed and incorporated within the health system in an equitable, scalable and sustainable manner to benefit all Australians. This involves bolstering the specialised health genomics workforce, mainstream health workforce and infrastructure, and supporting service availability and provision across Australia. The outcomes and activities detailed under this Strategic Priority may support advice to Australian governments concerning the need for investments and alternative funding models.

Efforts should be backed by a strong research and innovation sector with the capacity and infrastructure to generate new knowledge, and the readiness of health systems to adopt evidence-based genomics research outcomes.

Outcome 2.1 — Understand and develop specialised workforce capacity, infrastructure, and health service design

- a capable, sufficiently sized, sustainable and culturally competent specialised health genomics workforce (including clinicians interacting directly with patients and those in laboratory settings) with clear roles and responsibilities built around core competencies to effectively and efficiently deliver a person- and family-centred approach and improve outcomes
- utilisation of existing and future investments in the specialised health genomics workforce and infrastructure that maximises effectiveness and efficiency
- all Australians have equitable, affordable and timely access to genomics-informed health care where there is evidence of benefit
- continuity of care is improved with clarity about the patient journey and navigation of the health system, to avoid unnecessary genomics-informed health care
- strategies that increase equity of access to genomics-informed health care and research, including clinical trials, are developed, implemented and evaluated, particularly for priority populations

National Activity	Timeframe	Responsible for Coordination	Priority
<p>2.1.1 — Develop a process for and conduct regular national analyses of:</p> <ul style="list-style-type: none"> • availability and provision of genomic testing and genomics-informed health care • workforce and infrastructure capacity, capability and optimisation • research being undertaken 			
<p>2.1.2 — Develop a process for and undertake the mapping of patient journeys through genomics-informed health care and research to inform appropriate referrals, and co-design of services and models of care, noting considerable variation between types of patients and across states and territories is expected</p>			
<p>2.1.3 — Informed by the national current landscape analysis (Activity 2.1.1) and patient journey mapping (Activity 2.1.2), identify relevant gaps and opportunities that would benefit from nationally coordinated approaches, and could be reflected in future additional activities under this framework.</p>			

2.1.4 — Determine the need for and, where required, update existing or develop new national best practice guidelines and standards to support the quality, evidenced-based and safe use of genomics-informed health care. For example, consideration of the need for national guidelines for managing secondary and additional findings.

2.1.5 — Establish a process for the regular review and updating of best practice guidelines and standards that incorporate or focus on genomics.

2.1.6 — Engage with and support the maintenance of national and international undiagnosed disease networks and improve links between these networks and the Australian health system.

Outcome 2.2 — Support the mainstream workforce

- a mainstream health workforce that is knowledgeable, skilled and culturally competent in genomics leading to an increase in appropriate referrals and support for individuals and families who should be offered, are considering, or have received genomics-informed health care

National Activity	Timeframe	Responsible for Coordination	Priority
2.2.1 — Evaluate the education and training needs of the existing and future mainstream workforce, determine the need to support integration of genomics into undergraduate and postgraduate curricula, clinical practice guidelines, capability frameworks and standards, and continuing professional development (CPD) training modules, and consider how to effectively support this integration.			
2.2.2 — Use the results of the evaluation (outlined in Activity 2.2.1) to inform the identification of existing or develop new educational materials and on-demand resources for the mainstream health workforce to improve genomic knowledge and establish a process to endorse and maintain these materials at a national level.			
2.2.3 — Engage with clinicians and health services to inform how the existing or newly developed educational materials and on-demand resources (outlined in Activity 2.2.2) should be distributed to support the mainstream workforce.			

2.2.4 — Use the results of the evaluation (outlined in [Activity 2.2.1](#)) to develop targeted activities and a long-term strategy to address gaps and support the existing and future mainstream health workforce to ensure it is operating at full scope within fit for purpose care models.

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Outcome 2.3 — Decision making processes, funding pathways and sustainable investment to enhance clinical translation

- genomics-informed health care is evidence based and underpinned by agreed national approaches to ethical considerations
- ensure high-value and appropriate genomics-informed health care, research and innovation receive targeted, cohesive, and sustainable investment, including funding pathways that support preventative, and early intervention models of health care
- active engagement, cooperation and collaboration with domestic and international stakeholders across the genomics ecosystem, in alignment with public values and expectations to ensure the Australian health system is well informed, facilitating the integration of genomics and accelerating the pace of research

National Activity	Timeframe	Responsible for Coordination	Priority
2.3.1 — Promote consideration of genomics as part of broader health technology assessment (HTA) improvement and reform processes, including the responses of the Australian Government and state and territory governments to the findings and recommendations of the Health Technology Assessment Policy and Methods Review report			
2.3.2 — Develop ethical models for partnerships, engagement, and co-investment with non-government stakeholders, including industry and academia, to enhance translational research and clinical adoption of innovations that prioritise broad public benefit and/or address the needs of under-served populations.			
2.3.3 — Establish and implement strategic partnerships consistent with the developed ethical models to enhance investment and clinical adoption of genomics.			

Outcome 2.4 — Support research and innovation

- support research and innovation programs targeted at improving access to and the evidence-base of genomics for priority populations underrepresented in genomics research
- support research and innovation programs to transition to longer-term healthcare models and avoid research substituting for health care
- ensure genomic and phenotypic databases are co-designed and appropriately built to reflect the ethnic diversity within Australia
- ensure appropriate investments are made in genomics-informed health care and research that service and support under-represented populations

National Activity	Timeframe	Responsible for Coordination	Priority
2.4.1 — Support the prioritisation of genomic and related research.			
2.4.2 — Support investments in research infrastructure to improve research outcomes and increase the likelihood of translation of research into standard of care.			
2.4.3 — Establish and maintain a clear, structured process to facilitate meaningful engagement between Australian Government and state and territory governments representatives and those undertaking genomics research, to ensure regulatory, HTA and health system requirements are integrated into project design, data collection and analysis.			
2.4.4 — Establish and maintain targeted horizon scanning for emerging genomics trends, research and technologies to inform prioritisation of research and investments, which are complementary to broader national and state- and territory-based horizon scanning activities.			

2.4.5 — Establish processes to formally share real-world evidence for new models of genomics-informed health care, enabling consideration of adoption in other state and territory health systems.

2.4.6 — Develop options for long-term, sustainable, and scalable integration of genomics-informed national pilot initiatives and research-funded programs into clinical practice where they are demonstrated to be clinically beneficial and cost-effective, and with consideration of alignment across federal and state and territory health systems.

2.4.7 — Develop culturally appropriate strategies, building on existing efforts of Australian governments and non-government entities, to ensure genomic and phenotypic databases reflect the ethnic diversity of the Australian population.

Strategic Priority 3 – Genomic Samples, Data and Information Management

Responsible, secure and appropriately governed collection, storage, analysis, use, sharing, and management of [biological specimens](#) and [genomic data](#) and genomic information generated in clinical and research settings is essential to driving improvements in health outcomes for Australians and supporting precision medicine. Wider sharing, greater accessibility, and greater integration of genomic information across health systems and the research sector will support further research and improved outcomes for patients.

At the same time, genomic information is often considered more complex, unique, and sensitive than other types of health information. Misuse of genomic information may have consequences for individuals, families, future generations, and entire populations that are impossible to make right again.

Effectively balancing the potential benefits and risks associated with genomic information will be contingent on the existence of appropriate infrastructure, cyber security frameworks, workforce, standards and processes that consider and address capacity, capability, interoperability, quality, cultural context, data sovereignty and governance. It will also require heightened consideration of consent, privacy and security.

To be truly responsible, approaches must reflect the priorities and concerns of the public, including those in priority populations, and be underpinned by consideration of cultural, ethical, legal and social implications. The outcomes and activities detailed under this Strategic Priority may support advice to Australian governments concerning the need for investments to support long-term, sustainable, and future-ready national approach to genomic information sharing between clinical and research settings, if such an approach aligns with community expectations.

Outcome 3.1 – Safe, secure and trusted data sharing, storage and management

- individuals, families and communities are confident that genomic and other related clinical and personal information is protected, securely stored, shared (where consented) and culturally safe
- preferences of individuals, families and communities regarding sharing or not sharing genomic information and the impacts of these preferences on genomics research are understood and embedded into approaches
- domestic and international genomic information sharing is facilitated in line with relevant legislation, policies and evolving community preferences, to increase knowledge and application of genomics in Australia
- national approaches and culturally appropriate stewardship models are co-developed that facilitate interoperable sharing of genomic information generated by health systems and research (such as utilisation of consistent identifiers) in line with relevant legislation, policies and evolving community preferences
- genomic information is used in an effective, efficient, ethical and timely manner

National Activity	Timeframe	Responsible for Coordination	Priority
<p>3.1.1 — Undertake a current state analysis and evaluation of genomic information management practices and infrastructure in Australia, considering the capability and capacity of Australian clinical and research laboratories and linked health systems to:</p> <ul style="list-style-type: none"> • obtain, record and support patient withdrawal of consent • collect, manage, govern, store, use and share biological specimens, genomic information, and other related data. <p>This will consider legal, regulatory and accreditation aspects that underpin practices in health care and research.</p>			

3.1.2 — Use the current state analysis of genomic information management practices and infrastructure in Australia ([Activity 3.1.1](#)) to develop culturally informed, future-proof and sustainable principles- and standards-based national approaches to genomic biological specimen and information management in clinical settings and for research funded by Australian governments, that include interoperability, security and privacy standards.

3.1.3 — Identify community preferences on genomic information sharing between clinical and research settings, considering the ethical, legal, and policy barriers which would need to be overcome to support the development of a national approach to genomic information sharing.

3.1.4 — Encourage consideration of genomics as part of broader sector and Australian governments considerations of best practices and regulations related to cyber security frameworks and safeguards.

3.1.5 — Encourage consideration of genomics as part of broader sector and Australian governments considerations of best practices and regulations related to artificial intelligence.

3.1.6 — Evaluate existing approaches and digital platforms used within Australia and internationally that support the identification and resolution of variant interpretation discrepancies. and use of virtual gene panels and reference genomes in research and clinical settings

Strategic Priority 4 – Aboriginal and Torres Strait Islander-led Genomics

Australian governments, researchers and clinicians must work in partnerships with Aboriginal and Torres Strait Islander people to ensure trust, cultural safety and to improve equity of access to, and outcomes of, genomics-informed health care, health systems and research.

In the spirit of self-determination and to drive better outcomes, it is essential that Aboriginal and Torres Strait Islander people have agency and are empowered to lead efforts related to integrating genomics into the health system, to deliver benefits to First Nations communities according to their own priorities and protocols, and at their own pace.

Responsible use of data is central to advancing the use of genomics in health systems and research in Australia and, accordingly, ensuring Indigenous Data Sovereignty — that Aboriginal and Torres Strait Islander people have the right to exercise ownership and control over their biological specimens and data at all stages is critical.

It is intended that all activities under this Strategic Priority will be progressed in co-design with relevant Aboriginal and Torres Strait Islander stakeholder groups and established entities and networks, which may result in changes to outcomes to ensure they reflect those of communities.

Outcome 4.1 – Building Partnerships, Engagement and Trust

- health outcomes are improved for Aboriginal and Torres Strait Islander people
- leadership and engagement of Aboriginal and Torres Strait Islander communities in genomics-informed health services, research and public health initiatives is increased
- trust in genomics-informed health care and research is improved among Aboriginal and Torres Strait Islander people
- applications of genomic knowledge to health care and research are informed by Aboriginal and Torres Strait Islander people
- research that addresses health and wellbeing priorities for Aboriginal and Torres Strait Islander people provide outcomes that benefit Aboriginal and Torres Strait Islander people
- access to genomics-informed health care and research is improved among Aboriginal and Torres Strait Islander people
- all Aboriginal and Torres Strait Islander people who require genomic-informed health care and/or are eligible to participate in research can successfully access and engage with these services

National Activity	Timeframe	Responsible for Coordination	Priority
4.1.1 — Establish new or leverage existing culturally appropriate governance arrangements with Aboriginal and Torres Strait Islander people that support co-design and leadership of activities identified under this priority, and across other priorities.			
4.1.2 — Develop an implementation plan to support the implementation of the <i>Ensuring Culturally Safe Health Genomics with Aboriginal and Torres Strait Islander Peoples Guiding Principles</i> (the Guiding Principles) into health systems, service delivery and research, including supporting consideration of their integration into existing and new National Safety and Quality Health Service (NSQHS) Standards.			

4.1.3 — Develop opportunities and partner with Aboriginal and Torres Strait Islander people to ensure Indigenous Data Sovereignty is supported, and individuals and communities regain and maintain sovereignty over genetic material.

4.1.4 — Ensure Aboriginal and Torres Strait Islander governance and culturally aligned processes and mechanisms are developed with respect to biological specimens and genomic information including position statements on best practice in relation to biobanking, data sharing, data systems and infrastructure and Indigenous Data Sovereignty compliance.

4.1.5 — Develop strategies to understand and address the cultural, ethical, legal and social implications related to genomics-informed health care and research (consistent with the principles of the *National Cultural Respect Framework for Aboriginal and Torres Strait Islander Health 2016–2026*).

Outcome 4.2 – Improve access, outcomes and cultural safety

- accurate, sufficient and culturally appropriate information about genomics-informed health care, health systems and research support equity of access and outcomes for Aboriginal and Torres Strait Islander people
- changes in health systems and research that eliminate systemic discrimination of Aboriginal and Torres Strait Islander people are enabled
- cultural safety standards and training are embedded within health systems and research, and access and participation of Aboriginal and Torres Strait Islander people are improved
- the design of research studies that address the health and wellbeing priorities of Aboriginal and Torres Strait Islander people are culturally safe and empower the participants through co-design
- Aboriginal and Torres Strait Islander communities are supported to grow a workforce that supports their communities through health care and research

National Activity	Timeframe	Responsible for Coordination	Priority
4.2.1 — Develop pathways and activities for Aboriginal and Torres Strait Islander people to safely access and participate in genomics research through leading, identifying priorities, partnership, co-design of study methodology and defining outcomes and the benefits for Aboriginal and Torres Strait Islander people and communities.			
4.2.2 — Evaluate existing or emerging cultural competency resources for non-Aboriginal and Torres Strait Islander health professionals engaged in genomics and ensure these resources are considered as part of broader activities to improve the genomics knowledge and competency of the workforce (see Strategic Priority 2).			

4.2.3 — Conduct regular scoping activities to identify the current and ongoing need for the inclusion of cultural competency as it relates to genomics (and related disciplines) in undergraduate and postgraduate curricula in relevant courses and programs, clinical practice guidelines, capability frameworks and standards, and CPD training modules.

4.2.4 — Evaluate existing and develop new culturally informed and language appropriate educational resources on genomics-informed health care and research for Aboriginal and Torres Strait Islander people, and support individuals and communities to access these resources.

4.2.5 — Develop and implement strategies to build representation of Aboriginal and Torres Strait Islander people across the genomic health workforce (consistent with the [*National Aboriginal and Torres Strait Islander Health Workforce Strategic Framework and Implementation Plan 2021–2031*](#)), including defining the current and future needs of the workforce.

4.2.6 — Develop culturally informed strategies, building on existing ongoing research and efforts, to ensure genomic and phenotypic databases reflect the diversity of the Aboriginal and Torres Strait Islander population.

Appendix 1: Abbreviations and glossary

Abbreviations and Acronyms

ACSQHC	Australian Commission on Safety and Quality in Health Care
AHMAC	Australian Health Ministers' Advisory Council
ALIGN	Australian Alliance for Indigenous Genomics
APS	Australian Public Service
Cancer Control Framework	National Framework for Genomics in Cancer Control
CAPS Committee	Cancer and Population Screening Committee
COVID-19	Coronavirus disease 2019
CPD	Continuing professional development
Framework and Implementation Plan	National Health Genomics Policy Framework and Implementation Plan 2026–2030
GHFM	Genomic Health Futures Mission
GMO	Genetically modified organism
Guiding Principles	Ensuring Culturally Safe Health Genomics with Aboriginal and Torres Strait Islander Peoples Guiding Principles
HCEF	Health Chief Executives Forum
HMRO	Health and Medical Research Office
HST	High cost, highly specialised therapy
HTA	Health Technology Assessment
HTGC	Health Technology and Genomics Collaboration
Lapsed Framework	National Health Genomics Policy Framework 2018–2021
MBS	Medicare Benefits Schedule
Microbial Framework	National Microbial Genomics Framework for Public Health 2025–2027
MRFF	Medical Research Future Fund
MSAC	Medical Services Advisory Committee
NGO	Non-government organisation
NHMRC	National Health and Medical Research Council
NHRA	National Health Reform Agreement
NSQHS	National Safety and Quality Health Service Standards
PBAC	Pharmaceutical Benefits Advisory Committee
PBS	Pharmaceutical Benefits Scheme
PHN	Primary Health Networks
PICO	population, intervention, comparator and outcome
PRG	National Health Genomics Policy Framework Project Reference Group
TGA	Therapeutic Goods Administration

Key technical and defined terms

Biological specimen	A sample collected from a living thing to support its analysis for health care or research. In human genomics, common types of biological specimens collected include blood, saliva, a swab rubbed against the inside of the cheek (buccal swab), and tissue (for example, a biopsy of a tumour).
Carrier	A person who has an inheritable genetic variant associated with a condition who is typically not affected by that condition and may pass the genetic variant to their children.
Cell and gene therapies	Advanced treatments that target conditions at the cellular or genetic level through introducing, removing or modifying cells or genetic material.
Clinical genetics	A field of speciality practice in medicine focussed on the assessment, management and treatment of patients who are at risk, suspected or known to have a genetic or genomic condition. Clinical geneticists typically interact directly with patients, request genomic testing, and interpret its results.
Epigenomics	Study of how external factors (such as diet, stress and environmental factors) can change the ways genes are expressed without altering the genes themselves.
Genetics	Study of genes and how they are inherited.
Genomic data	Data generated from genomic testing part or all of an organism's genome.
Genomic medicine	<p>The use of genomic information as part of assessment, management and treatment of patients who are at risk, suspected or known to have a genetic or genomic condition.</p> <p>This framework and implementation plan primarily uses the largely synonymous but broader term 'genomics-informed health care' to recognise the growing importance of genomics as a routine part of many areas of clinical practice as well as public health initiatives.</p>
Genomic testing	<p>The technical process performed in laboratories by genetic pathologists, medical scientists with appropriate qualifications in genetics, and other support staff, to analyse the genome to support diagnosis, prognosis or treatment of a condition.</p> <p>Involves the collection of a biological specimen and the extraction of DNA, sequencing, analysis, interpretation and reporting.</p>
Genomics	Study of an organism's complete set of genetic information.
Genomic information	Data generated from genomic testing all or part or all of an organism's genome that has been analysed, interpreted and provides meaningful insights for health care or research.
Genomic information management	Processes and/or systems that support the storage, management and sharing of genomic information and related health information.
Genomics-informed health care	The use of genomic information as part of individual clinical care or public health initiatives.

Germline variants	A permanent change in a gene that exists in reproductive cells (sperm and eggs) which is usually inherited from parents or may, rarely, present spontaneously during conception or early gestation. Germline variants can be passed to a person's children.
Horizon scanning	A systematic process to explore potential or emerging trends, technologies, and other developments that could impact an organisation, industry, or specific area of interest with the goal of taking full advantage of opportunities and mitigating risks.
Health technology assessment (HTA)	Multidisciplinary evaluation of health technologies to assess evidence of benefits, harms and costs to determine its value and inform its use in health systems and/or potential subsidy under government programs.
Immunotherapies	Cancer treatments that utilise a patient's immune system to more effectively target and attack cancerous cells.
Indigenous Data Sovereignty	Right of First Nations people to govern the collection, access and use of data derived from or about First Nations people and their communities.
Lipidomics	Comprehensive study of lipids, the fatty substances that are essential components of cellular structures and functions.
Mainstreaming	Process of integrating genomics-informed health care as a routine part of many areas of clinical practice, such that the ' mainstream health workforce ' can request genomic testing, interpret results and effectively manage patients as opposed to referring patients to be managed by the ' specialised health genomics workforce '.
Mainstream health workforce	Professionals and support staff working to provide healthcare services (for example, medical doctors and specialists, nurses and midwives, and allied health professionals) who are not formally qualified in human genetics, genomics or related disciplines (that is, the ' specialised health genomics workforce ').
Medicare Benefits Schedule (MBS)	A listing of medical services subsidised by the Australian Government. In the context of genomics-informed health care, this includes consultations with general practitioners and specialists that involve the clinical management of suspected or known genomic conditions, genomic testing referred to pathology providers, and other non-pharmaceutical treatments to manage genomic conditions.
Metabolomics	Comprehensive study of substances involved in metabolism, aiming to identify, quantify, and understand their roles in biological processes and how they change under different conditions.
Microbes	Microorganisms, such as bacteria, virus, or fungi, that can be seen only through a microscope.
Microbiomics	Study of microbes that naturally live in a given environment (for example, on or in the human body), their structure, function, and dynamics.
Multi-omics	The use of information from multiple "-omics" disciplines (like genomics, transcriptomics, proteomics, and metabolomics) at the same time.
My Health Record	Australia's national digital health record system, managed by the Australian Digital Health Agency (ADHA).

One Health	An approach to designing and implementing programs, policies, legislation, and research in which animal, human and environmental sectors communicate and work together to achieve better public health outcomes.
Pharmaceutical Benefits Scheme (PBS)	A listing of medicines subsidised by the Australian Government. In the context of genomics-informed health care, this includes common medicines used to symptomatically treat genomic conditions and those that address the underlying causes, including ‘immunotherapies’ and ‘cell and gene therapies’ .
Person- and family-centred care	An approach to health care that prioritises the preferences, needs and values of patients, and where appropriate their families, and actively involves them in decisions about their care.
Pharmacogenomics	Study of the way in which the genome influences responses to certain drugs. In a clinical context, pharmacogenomics can be used to tailor treatment to maximise effectiveness and avoid adverse reactions.
Primary Health Networks (PHN)	Independent not-for-profit organisations funded by the Australian Government to coordinate primary health care in their respective regions
Population screening	The systematic offering of testing to everyone within a defined target group to support early detection of risk factors or signs of specified conditions, with the goal of reducing the impact of those conditions on individuals and the target group as a whole.
Precision medicine	The use of genomic, environmental and lifestyle information to guide prevention, diagnosis, and treatment (typically by separately patients into subgroups, but may conceptually involve tailoring approaches to individual patients) in a more targeted way than can be achieved by conventional medicine, which focuses on diagnosing patients based on their symptoms and applying the same treatment regardless of individual differences.
Proteomics	Study of all proteins present in a cell, tissue, organ, or organism, known as the proteome.
Social determinants of health	Non-medical factors that influence an individual's health outcomes.
Somatic variant	A permanent change in a gene that occurs after conception and is not present in reproductive cells (sperm or eggs). Somatic variants are the common cause of most cancers. Somatic variants cannot be passed to a person's children.
Specialised health genomics workforce	Professionals and support staff working to provide healthcare services who are formally qualified in human genetics, genomics or related disciplines, for example, clinical geneticists, genetic pathologists, and genetic counsellors.
Transcriptomics	The study of the structure and function of the transcriptome, the complete set of RNA transcripts produced by a genome.
Translational research	Research aimed at converting scientific ideas and novel discoveries into findings that have practical, feasible and scalable applications in clinical practice.

Appendix 2: Background

In 2016, the Health Chief Executives Forum (HCEF) — then known as the Australian Health Ministers' Advisory Council (AHMAC) — agreed on the need to better integrate genomics into the Australian health system.

This resulted in publication of the [National Health Genomics Policy Framework 2018–2021](#) (the lapsed national framework). The lapsed national framework presented a shared commitment and coordinated intergovernmental approach to embedding genomics in the Australian health system. It identified activities to enable collaboration, information sharing, and avoid duplication of effort across the federal, state and territory governments. It also outlined actions to be taken nationally to maximise health outcomes for all Australians.

In 2020, coordinated effort to progress the lapsed national framework concluded as Australian governments focused on the coronavirus disease 2019 (COVID-19) pandemic response. Australian governments and non-government stakeholders all continued to support the integration of genomics into the Australian health system in the following years. These efforts have improved outcomes for individuals, families and communities, and have reshaped clinical practice. However, the extent of coordination and formal collaboration across federal, state and territory governments has varied.

In 2023, the Health Technology and Genomics Collaboration (HTGC) committed to review and update the lapsed national framework, which led to the development of this framework and implementation plan.

This work has been led by the Australian Government Department of Health and Aged Care in close collaboration with the National Health Genomics Policy Framework Project Reference Group (PRG). The PRG is composed of representatives nominated by the federal, state and territory health departments and the Aboriginal and Torres Strait Islander Advisory Group on Health Genomics.

This section will be further developed following public consultation to include additional details of:

- this public consultation process
- targeted consultation with Aboriginal and Torres Strait Islander stakeholders

Appendix 3: Australian health system overview

Australia's health system is a federated system, under which responsibility for health care is divided and shared between the federal, state and territory governments. It is a complex mix of separately funded and operated health services and programs that, when functioning effectively, form a cohesive network to meet the health and wellbeing needs of Australians.

In the context of genomics and the scope of this framework and implementation plan, key Australian Government responsibilities include:

- funding and managing the [Medicare Benefits Schedule \(MBS\)](#) and the [Pharmaceutical Benefits Scheme \(PBS\)](#)
- co-funding public hospitals, with state and territory governments, through the National Health Reform Agreement (NHRA)
- funding Primary Health Networks (PHN)
- supporting the Medical Services Advisory Committee (MSAC) and Pharmaceutical Benefits Advisory Committee (PBAC) to provide advice concerning the assessment and funding of high cost, highly specialised therapies (HST)
- funding national health and medical research through the Medical Research Future Fund (MRFF) and the National Health and Medical Research Council (NHMRC)
- regulating medical devices and medicines through the Therapeutic Goods Administration (TGA), and gene technology through the Gene Technology Regulator

Key state and territory government responsibilities include:

- co-funding and managing public hospitals including emergency departments, admitted settings in hospitals, and other non-admitted settings managed by public hospitals
- health service and public health planning
- developing models of care and pathways responsive to local needs, legislation and structures of state and territory health systems
- employing clinical, administrative, support and technical staff, and supporting their ongoing education and training
- community education
- setting operational guidelines for how patient information is collected, used and stored in public hospital settings
- funding health and medical research

Commercial businesses deliver healthcare services, including those subsidised by the Australian Government or contracted by federal, state or territory governments, and develop, manufacture and supply medical devices, pharmaceuticals, equipment and consumables.

Educational and research institutions, peak clinical and industry bodies, and non-government organisations (NGO) also make vital contributions to the Australian health system. These stakeholders advocate to Australian Government and state and territory government decision makers for policy development, inform communities and other stakeholders about new policy developments, research or other work being undertaken, fund and conduct research, contribute to the training and development of health professionals, and, in some instances, deliver health services.

There is a need for close engagement and partnerships across the health system to support coordination and, where appropriate, collaboration. This ensures the entire system functions as seamlessly as possible so that individuals, their families and communities can equitably benefit from genomics-informed health care and access genomics research, regardless of where they live.

Genomics Australia

This section is written in past tense, as publication of this framework and implementation plan will occur after 1 July 2025.

On 1 July 2025, the Australian Government established Genomics Australia, a new national body, to provide leadership, coordination and expertise so all Australians can reap the benefits of cutting-edge genomic technologies and research.

Genomics Australia has been established within the Department of Health and Aged Care, with the flexibility to evolve its model to meet changing technology and policy needs over time. It is supported by a Commissioner who advises the Australian Government based on broad engagement with the sector and community.

Its remit includes conducting projects to give effect to this framework and implementation plan. It has, accordingly, been assigned to lead activities. It is also expected to support the Australian Government in other national genomics policy work.

Appendix 4: Policies, strategies and plans relevant to the framework and implementation plan

The framework and implementation plan sits within a broader policy context at the national and state and territory levels. It was developed with consideration of related policies, strategies and plans and the responsibilities of existing government entities and committees. This approach was taken to ensure alignment with Australian governments' objectives and to avoid duplication of effort.

Related national policy documents

National Strategies, Policy Documents and Reviews
<p>Digital Health Blueprint and Action Plan 2023–2033</p> <p>The Digital Health Blueprint 2023-2033 is a ten-year roadmap that lays out the Australian Government's vision for the role digital health capabilities will continue to play in delivering a more tailored, integrated, efficient and contemporary health system. The Action Plan highlights the initiatives the Australian Government is investing in to meet the target outcomes identified in the Blueprint and outlines key delivery partners and progress. The Action Plan will be refreshed regularly to outline progress and to include new digital and data investments over time.</p> <p>This framework and implementation plan aligns with the key outcomes of the Digital Health Blueprint, focusing on giving Australians choice in how they manage their health and wellbeing through patient digital health records that follow them through the system, and data and information that is shared and reused securely.</p>
<p>Ensuring Culturally Safe Health Genomics with Aboriginal and Torres Strait Islander Peoples: Guiding Principles (the Guiding Principles)</p> <p>The Guiding Principles were developed by the Aboriginal and Torres Strait Islander Advisory Group on Health Genomics to guide delivery of culturally safe and equitable access to genomic health care and research. It provides information for policymakers, researchers, clinicians, ethics committees and Aboriginal and Torres Strait Islander communities on integrating genomics into the health system and research through partnerships with and at a pace determined by Aboriginal and Torres Strait Islander people.</p> <p>This framework and implementation plan provides an opportunity to partner with Aboriginal and Torres Strait Islander people to implement the Guiding Principles.</p> <p>The Guiding Principles will also be considered in the implementation of all activities under this framework and implementation plan.</p>

Framework for Governance of Indigenous Data

The [Framework for Governance of Indigenous Data](#) provides practical guidance for the Australian Public Service (APS) on how to practically support the principles of Indigenous Data Sovereignty for data governed by the Australian Government.

Indigenous Data Sovereignty is an important element of Strategic Priority 4 of this framework and implementation plan.

Framework for the assessment, funding and implementation of high cost, highly specialised therapies and services

The [Framework for the assessment, funding and implementation of high cost, highly specialised therapies and services](#) describes the steps agreed by the Australian, state and territory governments to assess and implement high cost, highly specialised therapies (HST) in public hospitals.

The Framework applies to TGA approved medicines and biologicals delivered in public hospitals where the average annual treatment costs exceed \$200,000 per patient (including ancillary services). HSTs may target rare diseases and/or involve new technologies, like gene and cell therapies.

Health Technology Assessment (HTA) Policy and Methods Review

A review of Australia's health technology assessment (HTA) policies and methods (HTA Review) was conducted in 2023-2024. The Australian Government has established an Implementation Advisory Group to provide advice on developing a roadmap for sequencing the Australian Government's response to the recommendations of the [HTA Review final report](#).

National Aboriginal and Torres Strait Islander Health Plan 2021-2031

The vision for the [National Aboriginal and Torres Strait Islander Health Plan 2021–2031](#) (Health Plan) is that Aboriginal and Torres Strait Islander people enjoy long, healthy lives that are centred in culture, with access to services that are prevention-focused, culturally safe and responsive, equitable and free of racism.

Consistent with this vision, this Health Plan acknowledges Aboriginal and Torres Strait Islander people's right to self-determination by ensuring that they are leading the decisions that impact their health and wellbeing. The Health Plan sets out a holistic approach that considers cultural and social determinants across the whole life course. It also highlights the need for mainstream health services to provide culturally safe and responsive care, in partnership with First Nations people and communities.

National Aboriginal and Torres Strait Islander Health Workforce Strategic Framework and Implementation Plan 2021-2031

The [National Aboriginal and Torres Strait Islander Health Workforce Strategic Framework and Implementation Plan 2021–2031](#) (Workforce Plan) aims to increase Aboriginal and Torres Strait Islander representation in all health roles and locations across the Australian health system, to improve health, mental health and social and emotional wellbeing of Aboriginal and Torres Strait Islander peoples. The Workforce Plan also aims to strengthen the health system to create and sustain its cultural and professional capabilities, increase access to services and improve the attraction, retention and career development of Aboriginal and Torres Strait Islander staff.

National Strategic Action Plan for Rare Diseases

The [National Strategic Action Plan for Rare Diseases](#) (the Action Plan) is the first nationally coordinated effort to address rare diseases in Australia. The Action Plan outlines the priorities and actions required to improve the health of those impacted by a rare disease.

Genomics has great potential for rare diseases. Approximately 80 per cent of rare diseases are of genetic origin and while statistically rare, it is estimated that 8 per cent of Australians live with a rare disease – this equates to around 2 million people. It is therefore important to consider the impacts this framework and implementation plan will have on people living with rare diseases.

National Digital Health Strategy (2023 – 2028)

The [National Digital Health Strategy \(2023 - 2028\)](#), developed by the ADHA on behalf of all Australian Governments, identifies opportunities for digital health to support planned national health system reforms and address emerging contemporary health system challenges to deliver a sustainable, interoperable and inclusive health system now and into the future.

The National Digital Health Strategy and this framework and implementation plan both strive to develop safe and secure genomic data and information sharing capabilities across the Australian health system.

National Framework for Genomics in Cancer Control

The [National Framework for Genomics in Cancer Control](#) (the Cancer Control Framework) focuses on genomics across the cancer care continuum. It is intended to guide policy to enable equitable access to genomic medicine for all Australians affected by cancer.

This framework and implementation plan addresses activities and priorities detailed in the Cancer Control Framework, including equity, workforce, and access, that are relevant not only to cancer care but also to the wider health landscape.

National Microbial Genomics Framework for Public Health 2025-2027

The [National Microbial Genomics Framework for Public Health 2025–2027](#) (the Microbial Framework) provides a nationally consistent approach to integrate microbial genomics into the Australian public health system.

The Microbial Framework and this framework and implementation plan are complementary. It is acknowledged that there are close links and interdependencies between human, animal, plant and wider environmental health. There may be opportunities for cooperation across sectors to embed genomics to advance health under a more integrated (i.e., One Health) approach in the future.

Population Based Screening Framework

The [Population-based Screening Framework](#) provides guidance for decision makers when considering potential population-based screening programs in Australia. The Framework includes details about the criteria which should be used to assess whether screening should be offered, or a screening program introduced for diseases or conditions, and the key principles for the implementation and management of screening programs. The [Genomic tests in population-based screening programs – position statement](#) sets out further criteria to be considered on proposals for genomic specific tests in population screening.

Review of the Medical Research Future Fund’s (MRFF) Genomics Health Futures Mission (GHFM)

The independent [Review of the Genomics Health Futures Mission](#) was conducted to assess how the MRFF has contributed to genomics research, its place in the genomics research funding landscape, the alignment and progress of MRFF-funded genomics research, and opportunities to improve the impact of MRFF funded research.

Related Australian Government entities and committees

Australian Commission on Safety and Quality in Health Care

The Australian Commission on Safety and Quality in Health Care (ACSQHC) works in partnership with patients, consumers, clinicians, managers, policy makers and healthcare organisations to achieve a sustainable, safe and high-quality health system. The ACSQHC is funded jointly by the Australian Government and by state and territory governments. It is accountable to the Australian Parliament and the Australian Government Minister for Health and Aged Care. Its purpose is to lead improvements to the safety and quality of health care so all Australians receive better care, everywhere.

Australian Digital Health Agency

The Australian Digital Health Agency (ADHA) aims to improve health outcomes through delivery of national digital health services and systems. ADHA focuses on putting data and technology safely to work for patients, consumers and the healthcare professionals who look after them. ADHA is responsible for My Health Record and other e-health programs under the National Digital Health Strategy (2023 – 2028).

Cancer Australia

Cancer Australia aims to reduce the impact of cancer, address disparities and improve outcomes for people affected by cancer by leading and coordinating national, evidence-based interventions across the continuum of care. Cancer Australia works collaboratively and liaises with a wide range of groups, including those affected by cancer, key stakeholders and service providers with an interest in cancer control. It also focuses on populations who experience poorer health outcomes, including Aboriginal and Torres Strait Islander peoples, and people living in rural and remote Australia.

Cancer and Population Screening Committee

The Cancer and Population Screening (CAPS) Committee provides strategic policy advice for national population screening and cancer control. The CAPS Committee is established under and reports to HCEF. This includes progressing recommendations from relevant program management committees and groups to HCEF.

Gene Technology Regulator

The Gene Technology Regulator assesses and approves activities with genetically modified organisms (GMOs) to protect the health and safety of people and to protect the environment from any risks posed by gene technology. This oversight includes research involving GMOs, clinical trials with therapeutics that are or contain GMOs and commercial release of these therapeutics.

Medical Research Future Fund (MRFF)

The Medical Research Future Fund (MRFF), established under the *Medical Research Future Fund Act 2015* (MRFF ACT), is a long-term investment supporting Australian health and medical research. The MRFF aims to transform health and medical research and innovation to improve lives, build the economy and contribute to health system sustainability. The Health and Medical Research Office (HMRO) advises the Minister for Health, Disability and Ageing on MRFF policy matters and is responsible for the overall administration of the MRFF.

Medical Services Advisory Committee (MSAC)

The Medical Services Advisory Committee (MSAC) is an independent non-statutory committee appointed by the Australian Government Minister for Health, Disability & Ageing. It advises the Australian Government on applications for public funding of health services and technologies.

National Health and Medical Research Council (NHMRC)

The National Health and Medical Research Council (NHMRC) funds high quality health and medical research to build research capability, support researchers, encourage the translation of research into better health outcomes and promote the highest ethical standards for health and medical research.

Pharmaceutical Benefits Advisory Committee (PBAC)

The Pharmaceutical Benefits Advisory Committee (PBAC) is an independent statutory body appointed by the Australian Government. The PBAC is established under the National Health Act 1953. Its primary role is to recommend new medicines for listing on the Pharmaceutical Benefits Scheme (PBS).

Therapeutic Goods Administration (TGA)

The Therapeutic Goods Administration (TGA) is responsible for evaluating, assessing and monitoring products that are defined as therapeutic goods. The TGA regulates medicines, medical devices and biologicals.

Policy documents at the state and territory level

While this framework primarily focuses on priorities and activities that would benefit from national coordination, embedding genomics into the health system will require activity at the state and territory levels. The use of genomic medicine is the focus of several state and territory policy documents.

State and Territory Strategies and Policy Documents	
ACT	<p>ACT Child and Adolescent Clinical Services Plan 2023-2030</p> <p>The Child and Adolescent Clinical Services Plan 2023-2030 sets out a holistic roadmap for children’s health services funded by the ACT Government. This plan builds on work across the ACT’s health and early childhood systems to improve and strengthen existing services and programs for children, adolescents and their families.</p> <p>ACT Health Services Plan 2022-2030</p> <p>The ACT Health Services Plan 2022 to 2030 provides a roadmap for redesign, investment and redevelopment of health services funded by the ACT Government. It also sets out ACT Government priorities for working with Australian Government funded health services, private providers, primary care and allied health services. The strategies outlined in this plan include a focus on equity and ensuring that services are flexible and inclusive.</p> <p>Better together – A strategic plan for research in the ACT health system 2022-2030</p> <p>Better together outlines the eight-year vision for the ACT Government and its partners to drive research which strengthens the health of their communities through a strategic approach to investment and collaboration. The plan has three strategic objectives:</p> <ul style="list-style-type: none"> • the ACT health system becomes a learning health system • ACT people have capacity to undertake high value research in the health system <p>ACT research infrastructure supports high value research.</p> <p>Digital Health Strategy 2019-2029</p> <p>Digital Health Strategy presents a vision and direction to guide future activities and investments in technology across the Territory. It outlines the direction for the ACT public health system in building the digital capabilities needed to support a sustainable, innovative and world-class health system for the ACT.</p>

NSW	<p>Future Health: Guiding the next decade of health care in NSW 2022-2032</p> <p>Future Health provides the strategic framework and priorities for the whole NSW Health system over the next decade. The strategic framework outlines six strategic outcomes, each with key objectives. The strategic outcomes are:</p> <ul style="list-style-type: none"> • patients and carers have positive experiences and outcomes that matter • safe care is delivered across all settings; people are healthy and well • staff are engaged and well supported • research and innovation, and digital advances inform service delivery • the health system is managed sustainably. <p>NSW Health Genomics Strategy (2017) and Implementation Plans</p> <p>The NSW Health Genomics Strategy articulates a shared vision for genomics in NSW. It describes pathways to implementation and makes recommendations as to how NSW can remain responsive to this transformation in healthcare and position itself as a recognised leader in this field.</p> <p>The related NSW Health Genomics Strategy Implementation Plan 2021-2025 was developed with consumers, clinicians, researchers and health managers using a co-design approach. It builds on the achievements in delivering foundational initiatives described in the NSW Health Genomics Strategy Implementation Plan 2018-20. The 2021-2025 Implementation Plan focuses on implementing Phase 2 of the Health Genomics Strategy to enhance disease management and prevention.</p>
NT	<p>Strengthening our Health System Strategy (2020-2025)</p> <p>Northern Territory Health, Aboriginal Medical Services Alliance NT and Northern Territory Primary Health Network have formed a collaborative partnership to drive and coordinate the opportunities emerging through digital health technologies and new ways of working across the health system. The Strengthening our Health System Strategy (2020-2025) sets out the intent and commitment to strengthening the NT health system by pursuing opportunities to better connect communities, workforce, systems, and approaches enabled by digital health capabilities and technologies.</p>

QLD	<p>Genomics and Precision Health – A strategic 5- year roadmap 2021- 2026</p> <p>Genomics and Precision Health – A Strategic 5-Year Roadmap sets out Queensland Health’s vision for embedding genomics into the health system through coordinated leadership, equitable access, and system-wide integration. It prioritises ethical, legal, and social considerations, supports the mainstreaming of genomic services in healthcare delivery, and fosters a collaborative ecosystem that enables commercial partnerships with research and industry. The roadmap is structured around seven key priority areas:</p> <ol style="list-style-type: none"> 1. Person and family-centred care 2. Services 3. Resources 4. Workforce 5. Data and biobanking 6. Research 7. Governance <p>Digital Health 2031 – A digital vision for Queensland’s health system</p> <p>This digital strategy outlines the vision for a digitally enabled, world-class healthcare system in Queensland. Its primary goal is to improve health outcomes for all Queenslanders by positioning digital as a key enabler of safe, quality, and sustainable care. It defines the future state of digital healthcare, provides a strategic framework for implementation, and sets out the roadmap and success factors for delivering and sustaining digitally enabled services.</p>
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SA	<p>Precision Medicine Blueprint</p> <p>The Commission on Excellence and Innovation in Health’s Precision Medicine Blueprint outlines the first steps in establishing an agency agenda to guide the co-development of a system-wide framework for precision medicine in South Australia. The system priorities of the blueprint are:</p> <ul style="list-style-type: none"> • supporting infrastructure and service models • workforce, education and training • integrated big data and analytics • research, innovation and translation • community education and health literacy. <p>The South Australian Health and Wellbeing Strategy 2020-2025</p> <p>The Health and Wellbeing Strategy 2020 - 2025 informs the work, priorities and direction for the public health system in South Australia from 2020 to 2025. This direction emphasises the importance of keeping people healthy and refocuses on prevention, promotion and early intervention initiatives, as well as expanding service capacity in community settings to support people to avoid unnecessary interactions with the hospital sector.</p>
TAS	<p>Digital Health Transformation – Improving Patient Outcomes (2022-2032)</p> <p>The vision for the Tasmanian Digital Health Transformation Strategy is to empower consumers and healthcare workers to deliver better patient outcomes through system-wide, digitally enabled technologies. The four focus areas of the strategy are to improve community care, engage patients in their care, optimise clinical and operational workflows, and foster statewide collaboration.</p> <p>Tasmanian Genomics Framework 2024-2029</p> <p>The Tasmanian Genomics Framework 2024-2029 offers guidance to administrators, system planners and health professionals to recognise the system-transformative changes genomics can bring. Its principles and priorities also provide guidance for the broader health sector in Tasmania.</p>

VIC	<p>Victorian Department of Health - Our Strategic Plan 2023-2027 (2024 update)</p> <p>The vision for the Victorian Department of Health Strategic Plan 2023-27 (2024 update) is that Victorians are the healthiest people in the world. Victoria will partner with the community to support every individual to lead a healthy life. To achieve this vision, the Plan identifies seven strategic directions: keeping people healthy and safe in the community, providing care closer to home, keep innovating and improving care, improving Aboriginal health and wellbeing, moving from competition to collaboration, a stronger and more sustainable workforce, and a safe and sustainable health, wellbeing and care system.</p> <p>Genetic and genomic healthcare for Victoria 2021</p> <p>Genetic and genomic healthcare for Victoria 2021 provides a framework outlining what needs to be done to further embed the appropriate use of genomics to benefit the health and wellbeing of Victorians. It outlines four areas in which additional work is needed to include genomic information into routine healthcare: strengthening the healthcare system; building trust; raising awareness; and growing knowledge.</p>
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WA	<p>WA Genomics Strategy 2022-2032: Towards precision medicine and precision public health</p> <p>The WA Genomics Strategy outlines an ambitious vision to benefit all Western Australian through timely and appropriate translation of genomics. To achieve this vision, the WA Genomics Strategy identifies five strategic priority areas::</p> <ul style="list-style-type: none"> • person and family-centredness • genomic healthcare services • workforce, education and training • digital health and data • research and innovation. <p>WA Health Digital Strategy 2020-2030</p> <p>The WA Health Digital Strategy aims to take advantage of the innovations transforming healthcare to drive better health outcomes for all Western Australians. Initiatives, investment priorities and resources for the WA Health Digital Strategy will be focused on six strategic themes:</p> <ul style="list-style-type: none"> • empowered consumers • informed clinicians • optimised performance • supported workforce • enhanced public health • embedded innovation and research.
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