National Health Genomics Policy Framework



Australian Health Ministers' Advisory Council

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Foreword by Health Ministers

The National Health Genomics Policy Framework, endorsed by the Council of Australian Governments Health Council, presents a shared commitment to leveraging the benefits of genomics in the health system for all Australians.

Our vision for genomics in Australia is clear. We aim to help Australians live longer and better by integrating genomics into the health system through taking coordinated action across agreed strategic priority areas.

With completion of the Human Genome Project, and substantial advances in genome sequencing technology, we are beginning to realise the tremendous potential for genomics to contribute to early diagnosis, better targeted treatments and disease prevention. While it is easy to get carried away by the promise of genomics and what can be realistically achieved in the short to medium term, there are also risks to the health system and the community. Therefore the use of genomic knowledge must be underpinned by a coordinated effort that recognises the ethical, legal and social issues. We need to make sure that we have the right infrastructure in place so that genomics can be integrated into the health system in a way that maximises benefits and minimises risks.

This Framework provides an overarching structure for states and territories to take forward specific jurisdictional priorities. It recognises the considerable work already being progressed through governments, publicly funded translational research and the private sector. Success requires a clear understanding of the delivery mechanisms across the health system, clearly identifying the models of care and services required in all cases and maximising the opportunities to use any levers and incentives to drive integration.

This Framework recognises the importance of collaborative partnerships: between clinicians, researchers, private industry and individuals; between clinicians and researchers in Australia and internationally; and between Commonwealth and state/territory governments. The Commonwealth will partner with each state and territory to support the Framework's implementation. This will include specifying actions to address the most important issues, as well as where collaborative action can make the greatest gains.

The health workforce is a critical part of building new clinical pathways that embed genomics and other new approaches for delivering health care in our communities, and within interdisciplinary models of health care. It will be a key force in optimising the application of genomic knowledge within our health care system. Strategic partnerships and clinical leadership are critical for establishing new approaches and ways of operation.

Ultimately all health care is about the individual and this cannot be delivered without their consent, understanding and active participation. We need to support individuals and families to be actively engaged as partners with their health care team. They need to be able to make informed decisions about genomic testing and how their genomic information is shared to contribute to the national and international research effort.

This framework will ensure that our health care system not only meets the challenges of today, but that it is sufficiently flexible to embrace the future age of genomics.



NOTE ON TERMINOLOGY

The term 'genomics' is used throughout this document to refer to both the study of single genes (genetics) and the study of an individual's entire genetic makeup (genome) and how it interacts with environmental or non-genetic factors. While genetic testing for clinical purposes is already embedded in the health system, the term genomics is used for brevity and to acknowledge the cross-over of issues between genetics and genomics, other than where it is necessary to differentiate between genetics and genomics.

The terms genomics and/or 'genomic knowledge' are used in this document and refer to the data, information and learnings derived through genomic research. It also refers to the technologies used for testing, analysing and furthering the discovery of genomic knowledge.

SUPPLEMENTARY INFORMATION

Additional contextual information can be found in the document titled 'Supplementary Information to the National Health Genomics Policy Framework', available at the Commonwealth Department of Health website.

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Introduction: Integrating Genomics into the National Health System

Integrating genomics appropriately into Australia's health system will require new thinking, new approaches and strengthened national collaboration and leadership.

The vision for the future is to ensure that advances in genomics are helping people live longer and better lives. To do this, the health benefits of genomic knowledge and technology need to be harnessed and incorporated within the Australian health system in the most effective possible way.

Genomics has the potential to reshape clinical practice and to fundamentally change the way we prevent, diagnose, treat and monitor illness, providing the opportunity to have more precise and tailored treatments. The ability to respond to this change is dependent on further developing Australia's capacity, capability and infrastructure needed to support integration of genomic technology into the national health system (particularly with regards to clinical utility, workforce, education, data security and sharing, quality and accreditation, cost-effectiveness and research).

It will be important to be realistic about the short-term impact of genomics on health care, so that the issues that present can be appropriately dealt with. The challenge for Australia is to convert genomic information and knowledge into effective and financially sustainable clinical practice to improve individual and population health outcomes. The value of genomic testing needs to be compared to the range of alternative options in terms of its ability to deliver health benefits, its capacity for harm and the costs of testing.

In the future, genomics may have significant impacts on social norms and values, including how society perceives illness, health and wellbeing in relation to individuals. The integration of genomics into the health system will be dependent on community acceptance and confidence.

WHY IS A NATIONAL FRAMEWORK FOR GENOMICS NEEDED?

A collaborative and coordinated approach at all levels of government and across stakeholders is necessary to achieve and maximise health outcomes for all Australians. It will help leverage the potential for genomics to contribute to improved care for individuals and families, improved population health and containment of costs in Australia's health system.

The health system is complex and often fragmented, with responsibility for genomics policy issues and funding split between the Commonwealth, and state and territory governments. Numerous national, jurisdictional, professional and consumer-led committees, advisory and community/support groups organise and direct genomic-related activities. At the same time, the private sector has a growing interest in genomic research that could translate into commercial opportunities, as well as laboratories providing private genomic testing.

A more coordinated, strategic approach is becoming increasingly important to embed genomics in the health system in an efficient, effective, ethical and equitable way. While the application of genomic knowledge to health care has the potential to transform the health system, it also presents risks to individuals and society, particularly those related to privacy, security and storage of data, and the potential for genomic discrimination. Other significant issues include different laboratory testing procedures, incompatible data systems, a lack of data standardisation, inconsistent funding decisions, as well as variable legislation that currently limit data sharing, influencing the translation of research to clinical practice. These, and other issues, need to be consistently managed across Australia.

The potential costs to health and the economy are considerable and will increase unless national action is taken.

The Framework recognises the potential for genomics to improve health care, and provides a consistent national and strategic view for integrating genomics into the Australian health system. It aims to address the lack of coordination of activities across Australia and is designed to drive national effort on agreed priorities, and address policy issues and challenges. Developing a whole-of-governments and system-focussed Framework, with a person-centred approach to outcomes, is necessary to ensure consistency of action across Australia.

HOW WAS THE FRAMEWORK DEVELOPED?

The Australian Health Ministers' Advisory Council's Hospitals Principal Committee developed the Framework, with input from a jurisdictional advisory group. This advisory group comprised a Commonwealth Chair and members from each jurisdiction. The draft Framework was open to public consultation through a series of stakeholder forums across Australia and a call for written submissions from December 2016 to March 2017. Extensive consultation with stakeholders confirmed overwhelming support for greater national coordination of the application of genomic knowledge in health care. Further details about the consultation process are in the 'Supplementary Information to the National Health Genomics Policy Framework' document.

THE NATIONAL HEALTH GENOMICS POLICY FRAMEWORK IS STRUCTURED AROUND FIVE KEY PRIORITIES

Following extensive public consultation, five strategic priorities were developed to support the integration of genomics into health care for Australians:

Person- centred approach	Delivering high-quality care for people through a person-centred approach to genomics.
Workforce	Building a skilled workforce that is literate in genomics.
Financing	Ensuring sustainable and strategic investment in cost-effective genomics.
Services	Maximising quality, safety and clinical utility of genomics in health care.
Data	Responsible collection, storage, use and management of genomic data.





The strategic priorities are intrinsically linked to each other; there are interdependencies and linkages within and between priorities that will need to be further considered in implementing the Framework.

The Framework prioritises particular issues for initial consideration and indicates where further work is needed, while also recognising that stakeholders have a role in addressing issues independently. It is not intended to address all issues related to genomics and health. Subsequent reviews of the Framework will identify other emerging issues.

The Framework is the first step towards action. The next step will involve an Implementation Plan that will map current activity, and identify gaps and opportunities. This will allow governments to address current and emerging priorities as resources permit. Leadership across all relevant sectors of the health system is critical to drive effective implementation.

WHO IS THE AUDIENCE FOR THIS DOCUMENT?

The Framework is directed at decision and policy makers at the national, state and health service levels. It is primarily a tool to provide guidance for developing and implementing genomic-related policies, strategies, actions and services.

The Framework will also be a useful resource for the non-government sector, stakeholder organisations, industry and communities.

WHAT IS THE FRAMEWORK'S TIMEFRAME?

The timeframe of the Framework is three years, with a review anticipated in 2020 to inform the next iteration.

NEXT STEPS—IMPLEMENTING AND MONITORING THE FRAMEWORK

The Framework outlines an agreed high-level national approach to policy, regulatory and investment decision-making for genomics. To take the Framework forward, it will be necessary to develop an Implementation Plan.

The Implementation Plan is expected to focus on priority actions for the first three years. This will also allow governments to address current and emerging priorities as resources permit. The Implementation Plan will be a key tool for measuring the progress and success of the Framework. Three-year plans will provide a staged approach to achieving the necessary reforms identified within the Framework.

All jurisdictions and stakeholders will be able to monitor progress against activities and milestones outlined in the Implementation Plan. A set of performance indicators developed as part of the Implementation Plan will provide another opportunity to evaluate the Framework, and monitor progress and outcomes.

A considerable body of work is already being progressed across Australia through a range of national and jurisdictional research alliances, and leveraging this work will be instrumental to implementing the Framework. A stocktake of clinical genetic and genomic testing is also being conducted in 2017–18 to provide further information on gaps and immediate priorities for implementation.

Overview: National Health Genomics Policy Framework

WHAT IS THE SCOPE OF THE FRAMEWORK?

While the immediate focus of this Framework is the application of knowledge about the human genome to advance medicine and health care, the Framework's scope is also intended to be flexible. This is because the potential application of genomics is much broader. The Framework needs to take into account that genomic knowledge is also advancing at a rapid pace.

The initial priorities covered under the Framework include health care applications that are informed by, or based on, human genetic or genomic testing. These applications include those used to:

- test for the purpose of diagnosing and monitoring disease;
- treat diseases, including through understanding the genetic variation between underlying differential responses to medicines and how this can be applied to prevent adverse drug reactions and improve health care; and
- prevent and predict disease, including carrier testing and predisposition testing.

Genomic applications that currently have more limited relevance to population health (such as sports performance, forensic science and national security) will continue to be monitored, including the potential to develop related policy frameworks.

The major elements of the Framework are described below and a visual summary is provided overleaf.

VISION

Helping people live longer and better through appropriate access to genomic knowledge and technology to prevent, diagnose, treat and monitor disease.

MISSION

To harness the health benefits of genomic knowledge and technology into the Australian health system in an efficient, effective, ethical and equitable way to improve individual and population health.

PRINCIPLES UNDERPINNING THE FRAMEWORK

The following three principles underpin the strategic priorities of the Framework:

- 1. The application of genomic knowledge is ethically, legally and socially responsible and community trust is promoted.
- 2. Access and equity are promoted for vulnerable populations.
- 3. The application of genomic knowledge to health care is supported and informed by evidence and research.

ENABLERS OF SUCCESS

To help guide decision and policy makers in successfully implementing the Framework, three key enablers have been identified:

- collaborative governance and leadership
- stakeholder engagement
- national and international partnerships.

These enablers will help establish a supportive environment for genomics work across Australia and internationally.



A Summary: National Health Genomics Policy Framework

Vision

Helping people live longer and better through appropriate access to genomic knowledge and technology to prevent, diagnose, treat and monitor disease.

Mission

To harness the health benefits of genomic knowledge and technology into the Australian health system in an efficient, effective, ethical and equitable way to improve individual and population health.

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Collaborative governance and leadership

Stakeholder engagement

National and international partnerships

Strategic priority areas

Person-centred approach	Workforce	Financing	Services	Data
Delivering high- quality care for people through a person-centred approach to integrating genomics into health care	Building a skilled workforce that is literate in genomics	Ensuring sustainable and strategic investment in cost-effective genomics	Maximising quality, safety and clinical utility of genomics in health care	Responsible collection, storage, use and management of genomic data
Principles				
The application of genomic knowledge is ethically, legally and socially responsible and community trust is promoted.				
Access and equity are promoted for vulnerable populations.				

The application of genomic knowledge to health care is supported and informed by evidence and research.

Strategic Priorities

There are five strategic priorities outlined in the Framework. Under each of these, a number of priority areas for action have been identified, which are designed to support the integration of genomics into the Australian health system. Each of the strategic priorities is of equal importance and value, and there are interdependencies and linkages within, and between, priorities that will need to be further considered in implementing the Framework.

Strategic priorities	Rationale
Delivering high-quality care for people through a person-centred approach to integrating genomics into health care	Making sure that people are involved in, and central to, their care is a key component of developing high-quality health care, including health care that is informed by genomics.
Building a skilled workforce that is literate in genomics	Upskilling the workforce through increasing capacity and capability in genomics and bioinformatics is necessary to effectively and efficiently support improved health outcomes for the individual and population.
Ensuring sustainable and strategic investment in cost-effective genomics	Australia's investment in genomic research and testing needs to deliver actionable results that lead to people living longer and better lives.
Maximising quality, safety and clinical utility of genomics in health care	The use of genomics in health care should be based on the best available knowledge, evidence and research and the outcomes of treatment should be used to help improve care.
Responsible collection, storage, use and management of genomic data	The collection and analysis of genomic data is essential to driving improvements in health outcomes for all Australians and providing a pathway to truly personalised health care.



Delivering high-quality care for people through a person-centred approach to integrating genomics into health care

Making sure that people are involved in, and central to, their care is a key component of developing high-quality health care, including health care that is informed by genomics.

Priority areas for action

- **1.1** Improve support for individuals, and their families, to make informed choices about genomic testing, and take responsibility for those choices and related risks.
- **1.2** Encourage appropriate referrals of genomic testing, that put the welfare and needs of the individual first, thereby avoiding unnecessary testing.
 - **1.2.1** Developing and promoting clinical practice guidelines and decision support tools for engaging with individuals on their personal context and health goals.
- **1.3** Engage relevant community/patient advocacy organisations and consumers in discussion of the consumer experience, as well as on the ethical, legal and social issues of genomics.
 - **1.3.1** Developing community engagement strategies to promote an understanding of the application and impact of genomic advances in health care, including the gap between testing and treatment options.
 - **1.3.2** Exploring how the consumer experience can be captured and measured to inform priorities and establish a baseline.
- **1.4** Promote public awareness and understanding of genomics, including through linguistically and culturally safe and appropriate information resources for targeted consumer groups.
- **1.5** Identify barriers to equity of access and develop a national approach to address these, noting that access is multi-dimensional and includes location, cost, availability and appropriateness (including cultural acceptability). This includes, but is not limited to:
 - exploring barriers to the uptake of genomic services including the potential for discrimination (life insurance, employment, lifestyle, access to services); and
 - evaluating the delivery of genomic services in terms of being accessible, appropriate and culturally secure and responsive for Aboriginal and Torres Strait Islander peoples.
- **1.6** Investigate how genomics data can be integrated with electronic health records to improve coordination of care, support better clinician decision-making and facilitate seamless clinical pathways.
- **1.7** Explore the potential to develop integrated person and family-centred care delivered by multidisciplinary teams (where appropriate).
- **1.8** Identify and promote a standard model of consent that is sufficiently flexible to support a person's understanding of the potential implications of having their genome sequenced, familial aspects, and decision-making about any secondary findings, as well as including provision for access by researchers if appropriate.

- Individuals, and their families, are empowered to take greater accountability and ownership for their health.
- ✓ All testing is conducted with a view to achieving benefit for the individual and/or family members, rather than being driven by technology.
- ✓ Community understanding, acceptance and trust is built and maintained.
- Improved public health genomic literacy leading to increased public understanding of the place of genomics in the health system. Increased understanding will inform consumer expectations of the benefits, risks and limitations of genomics.
- Equity of access is supported, and consumers receive timely and appropriate genetic and genomic services.
- Continuity of care is improved with clarity about the consumer journey and to avoid unnecessary testing.
- Individuals and/or families undergo a voluntary and informed consent process that clearly explains associated risks and benefits.



STRATEGIC PRIORITY 2: Workforce

Building a skilled workforce that is literate in genomics

Upskilling the workforce through increasing capacity and capability in genomics and bioinformatics is necessary to effectively and efficiently support improved health outcomes for the individual and population.

Priority areas for action

- **2.1** Improve the genomics literacy and capability of the health workforce through the development, delivery and ongoing maintenance of appropriate genomic education, training and skills.
- 2.2 Build the capacity for, and promote access to, a skilled and literate genomics workforce, through workforce strategies and planning at the national level.
- 2.3 Facilitate partnerships and networks to promote and support sharing of knowledge.

- Competent genomics workforce with clear roles and responsibilities built around core competencies to effectively and efficiently deliver a person-centred approach and improved individual outcomes.
- Improved genomics literacy of the broader health workforce, leading to the appropriate referrals of, and support for, consumers requiring genomics services.
- ✓ Genomic literate and appropriately skilled health professionals working together in multidisciplinary teams to deliver equitable, safe and quality clinical genomic services to consumers.
- People can access health professionals appropriately trained in genetics and genomics when they need to, and when it is appropriate to do so.



STRATEGIC PRIORITY 3: Financing

Ensuring sustainable and strategic investment in cost-effective genomics

Australia's investment in genomic research and testing needs to deliver actionable results that lead to people living longer and better lives.

Priority areas for action

- **3.1** Consider genomics in the context of any broader review of health technology assessment to support national consistency.
- **3.2** Develop partnerships, funding and data sharing approaches for genomics that promote access to safe, efficient and cost-effective services.
- **3.3** Develop a national research agenda for genomics and identify opportunities to link to Commonwealth and state/territory research priorities.
- **3.4** Better understand the role of the private industry, and the opportunities for partnerships to support the development and sustainable application of genomic knowledge.
- **3.5** Collaborate across governments and stakeholders to maximise investments and reduce duplication of resources and effort.

- All Australians, who have a clinical need, have equitable access to genomic services, regardless of barriers such as their geographic location.
- ✓ The partnerships, funding and data sharing approaches for genomics promote access to safe, efficient and cost-effective services.
- ✓ A nationally consistent approach to genomic investment and disinvestment is in place.
- Innovation and healthy competition is encouraged through integrated research translation.

STRATEGIC PRIORITY 4: Services

Maximising quality, safety and clinical utility of genomics in health care

The use of genomics in health care should be based on the best available knowledge, evidence and research and the outcomes of treatment should be used to help improve care.

Priority areas for action

- 4.1 Review and build on guidelines, regulations and standards to ensure genomic applications:
 - are evidence-based;
 - nationally consistent (where appropriate);
 - demonstrate clinical utility; and
 - align with agreed national ethical approaches.
- **4.2** Strengthen processes to identify, promote, monitor and report best practice in clinical genomics, including sharing of data and information.
- **4.3** Maximise genomics research opportunities that aim to resolve clinical uncertainty and improve quality and safety.

- Genomic applications in health care are evidence based and are underpinned by agreed national approaches to ethical considerations (such as secondary findings).
- Comprehensive national guidelines, regulations and standards support the quality and safe use of genomics in health care.
- ✓ Uncertainty in relation to the use of genomics in health care is addressed through research.
- An increase in the number of participants in clinical genomic research studies that are diagnosed through the application of genomics.

STRATEGIC PRIORITY 5: Data

Responsible collection, storage, use and management of genomic data

The collection and analysis of genomic data is essential to driving improvements in health outcomes for all Australians and providing a pathway to truly personalised health care.

Priority areas for action

- 5.1 Establish a national genomic data governance framework that aligns with international frameworks.
 - 5.1.1 Explore infrastructure options for national genomic data collection, storage and sharing.
 - **5.1.2** Strengthen public trust of data systems and mechanisms so that people are empowered to engage with genomic interventions in the health system.
- **5.2** Promote culturally safe and appropriate genomic and phenotypic data collection and sharing that reflects the ethnic diversity within the Australian population, including for Aboriginal and Torres Strait Islander peoples.
- **5.3** Develop nationally agreed standards for data collection, safe storage, data sharing, custodianship, analysis, reporting and privacy requirements.
- **5.4** Promote public awareness of the contribution of all research activities, including those funded through private industry, to advancing the application of genomic knowledge to health care.
- **5.5** Support sector engagement with international genomic alliances to promote shared access to data for research and global harmonisation of data where appropriate.

- Genomic data and knowledge are used in an effective, efficient, ethical and timely manner.
- The public is confident that genomic data and other clinical information is protected and culturally safe.
- The public understands the societal value of agreeing to share genomic data to support genomic research, including those funded through private industry.
- International data sharing is facilitated to increase knowledge and application in Australia.
- ✓ National approaches are developed that facilitate the interoperable sharing of genomic data.
- Benefits of genomics are available to all Australians, particularly to vulnerable population groups.
- Genomic and phenotypic databases reflect the ethnic diversity within Australia.



Appendix A: Commonly Used Terms

A number of glossaries of commonly used terms in genomics are available on the internet (see below). While some definitions may vary slightly, this is not critical for the purposes of the Framework.

New South Wales (NSW) Centre for Genetics Education The website also includes a glossary for families and professionals who may not be familiar with terminology

http://www.genetics.edu.au/Publications-and-Resources/glossary-1

The Australian Genomics Health Alliance Genomics Glossary https://www.australiangenomics.org.au/for-participants/genomics-glossary/

The National Human Genome Research Institute (NGHRI) talking glossary https://www.genome.gov/glossary/

Genome Quebec Glossary

http://www.genomequebec.com/glossary.html

Genomics England—Understanding genomics resources https://www.genomicsengland.co.uk/the-100000-genomes-project/understanding-genomics/



www.health.gov.au

All information in this publication is current as at August 2017