# Supplementary Information

## to the National Health Genomics Policy Framework 2018–2021

**Australian Health Ministers’ Advisory Council**

Supplementary Information to the National Health Genomics Policy Framework

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## Acronyms

|  |  |
| --- | --- |
| AGHA | Australian Genomics Health Alliance |
| AHMAC | Australian Health Ministers’ Advisory Council |
| BRCA1, BRCA2 | Genes that are associated with a predisposition to breast and ovarian cancer |
| CVS | Chorionic villus sampling |
| DNA | Deoxyribonucleic Acid |
| FDA | Food and Drug Administration (United States) |
| Framework | National Health Genomics Policy Framework |
| FSC | Financial Services Council |
| HER-2 | A gene that can play a role in the development of breast cancer |
| HGSA | Human Genetics Society of Australasia |
| HTA | Health technology assessment |
| IVD | In vitro diagnostic |
| IVF | In vitro fertilisation |
| MBS | Medicare Benefits Schedule |
| MPS | Massively Parallel Sequencing |
| MSAC | Medical Services Advisory Committee |
| NATA | National Association of Testing Authorities |
| NHMRC | National Health and Medical Research Council |
| NIPT | Non-invasive pre-natal testing |
| NPAAC | National Pathology Accreditation Advisory Council |
| PGD | Preimplantation genetic diagnosis |
| RCPA | Royal College of Pathologists of Australasia |
| TGA | Therapeutic Goods Administration |
| UM | Uveal melanoma |

This document provides additional information about why the strategic areas for action identified in the National Health Genomics Policy Framework (the Framework) are considered to be a priority.

This document also provides background information for those readers who are less familiar with genomics.

### A 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 to 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.

## 1. An Introduction to Genomics

The information in our DNA is stored as a code made up of four chemicals (bases): A, G, C and T.  Human DNA contains about 3 billion bases. DNA is organised into approximately 20,000 genes. Genes are packaged into chromosomes; a person usually has 23 pairs of chromosomes, or a total of 46 chromosomes, with half inherited from each parent. This entire collection of DNA, genes and chromosomes make up the human genome. Genomics is the study of all the genes in the genome, how they interact with each other and the environment. We inherit our characteristics through our genes.

#### What is a genetic variation?

Every person has two copies of each gene, one inherited from each parent. Most genes are the same in all people, but a small number of genes (less than on per cent) are slightly different between people. These differences contribute to the uniqueness of a person, for example, height or hair colour.

While most variations in a person’s DNA do not influence health, sometimes variations or ‘spelling mistakes’ can mean that the gene (where that variation occurred) is no longer able to work correctly.

#### What is a genetic disorder?

A condition caused by variations in one or more genes that disrupts normal development or causes a medical condition is called a genetic disorder. Genetic variation is essential to evolution. Genetic variants can be present because they were transmitted from a parent (germline variations) or they can also arise in a small subset of cells during the life of an individual (somatic variation). These types of variations are described in more detail below.

* **Germline variations—heritable**  
  Germline variations exist in an individual’s germ cells (eggs and sperm), which can be passed on to future generations. An individual who inherits a germline variation will have that variation in all the cells of his or her body (since the variation was present at conception). Germline variations may be silent, may cause disease, or may generate beneficial genetic diversity. Rare diseases and cancer are often caused by genetic variations; some of these appear randomly, while others are hereditary. Some types of cancer have been linked to predisposition through heritable (germline) variations.
* **Somatic variations—not heritable**  
  Variations of somatic cells can spontaneously arise in any cell in the body, except germ cells, at any time during an individual’s life. This type of variation is limited to the copies of the original cell that developed the mutation—it is not present in other cells in the person’s body and is not passable from parent to child. A somatic variation can be present in the cells that form a cancer tumour but they can be silent, or simply contribute to genetic diversity, which is part of evolution. Usually, many mutations drive transformation of a healthy cell into a cancer cell.

#### Fact

About 80% of all rare diseases have a known genetic association.

#### What is genetic testing?

In genetic testing, the DNA sequence of a single gene is checked for changes that cause, or increase risk of developing a disease. If a particular condition is linked to one or a couple of genes, genetic testing is a way to confirm or rule out the role of these gene(s) as the cause of the condition.

#### What is genomic testing?

Genomic testing investigates many genes at the one time. This is particularly useful when there are a number of genes that can cause a condition, or it is unknown which gene may be the cause. There are a range of different tests that can be used to investigate the variations in multiple genes at the one time:

* Panels can investigate a defined number of genes, for example a panel may investigate 10 genes, or it may contain 400
* Whole Exome Sequencing can investigate the areas of DNA that code for protein (exonic DNA)
* Whole Genome Sequencing can investigate our entire genomic DNA, that is the coding (exonic) and the non-coding (intronic) regions of our DNA.

Until recently, the clinical focus of the application of genomics has been on the investigation and diagnosis of rare single gene disorders (also known as Mendelian diseases). However, with advances in technology there has been a gradual shift from genetics (the study of single genes) to genomics (the study of multiple genes, exome and/or the whole genome). The volume of genetic material being analysed can be considered as a continuum from a single region of a single gene through to the whole genome.

### 1.1 What is clinical genomics?

Clinical genomics is the use of genome sequencing to inform patient diagnosis and care. Clinical genomics is a relatively new and rapidly changing field, which has potential to transform clinical medicine. Knowledge of the human genome is far from complete, but there are already uses for genetic and genomic information in the clinic. Advances in clinical genomics have been accelerated since the introduction of massively parallel sequencing (MPS), or next generation sequencing, which is a method to sequence genes at high speed at an affordable cost. This technique can be applied to any volume of genetic material to identify numerous genetic variants, including in any single cancer. Already, this information is influencing treatment decisions.

Genome sequencing may lead to better prevention, diagnosis, treatment and monitoring. The current focus for clinical genomics is:

* characterising and diagnosing rare and inherited disease (with the potential in the longer term to apply to common diseases)
* classifying cancer tumours to guide treatment
* providing information about an individual’s risk of developing disease or their likely response to different treatments.

The practice of genomics is transformative when sequence information is used to understand human disease. In brief, this begins with a comparison of the DNA sequence obtained from an individual with catalogues of many sequences obtained from other people. There is no single ‘reference’ genome because the extent of DNA sequence variation between people is enormous. A principal task for genomics is to catalogue those DNA variations that cause disease and distinguish them from variants that have no detrimental effects, but this process is in the early stages. This is a complex task as diseases may be caused by a combination of genetic variations in hundreds or thousands of genes. Furthermore, even if the link between genetic variations and disease is determined, this may not lead to new treatment approaches.

Genetic tests usually correlate variations in one gene with a condition or disease. Genomic tests typically involve the analysis of hundreds or even thousands of genes at the same time using sophisticated computer-based analyses (Figure 1 describes the continuum of tests available). While genetic tests are suitable for diagnosing conditions that are based on heritable changes to single genes (for example, Huntington’s disease), they are rarely appropriate for studying complex multifactorial diseases (such as type II diabetes), which result from interactions between many genes, lifestyle and environmental factors.

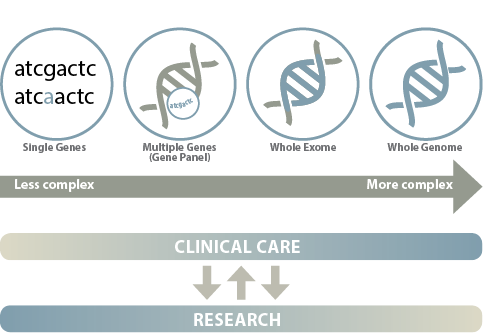


Figure 1: Gene testing continuum

Currently, the main types of gene testing in clinical care use single gene and multiple genes (gene panel) testing methodologies. Recent advances in genomic sequencing has led to the increasing use of whole exome and whole genome sequencing methodologies in research and clinical practice, challenging the traditional research–clinical practice dichotomy.

### 1.2 Why is genomics in health care important?

In recent decades, the Australian health system has been transforming from providing episodic care for those with chronic and complex conditions, to one that better addresses the longer-term health needs of the population. Integrating genomics into health care is one of the key strategies for achieving this transformation.

The potential of genomic medicine for Australia’s health system is considerable. It already means that some diseases can be diagnosed and gene variants detected far more precisely. In some circumstances, treatments can be quickly tailored to reflect a person’s wider genomic make-up, and better identify those at high risk of inherited disease and a range of common chronic conditions.

With rapid advances in technology, such as MPS, it is now possible to analyse panels of sequenced genes, whole exomes or whole genomes quickly and more cost-effectively. While the technology is available to sequence large numbers of genes, understanding the functions of any variations detected is complex. Some of the key milestones in genomics are outlined in Figure 2.

Genomics research is developing rapidly, while at the same time the cost of genomic sequencing is declining. This has led to an increase in the amount of genotypic and phenotypic (the observable characteristics of an individual) data available globally. The cost of genome sequencing has now decreased to a price range similar to many other complex medical tests, increasing the possibilities for its clinical application. While a key challenge is to translate genomic information into better health outcomes, it is likely that in the future more genomic information will be available to guide key health-related decisions.

Most ethical and social considerations raised by genomics already apply generally to health care, including the four commonly accepted principles of respect for autonomy, non-maleficence (do no harm), beneficence and justice. However, the application of genomic knowledge has its own particular challenges, including how the predictive nature of its application may impact on other family members, future generations and communities over time. In addition, progress in understanding how to interpret genomic variation in a single individual requires access to genomics reference data from many individuals. These ethical issues are expanded upon in Section 2.1.

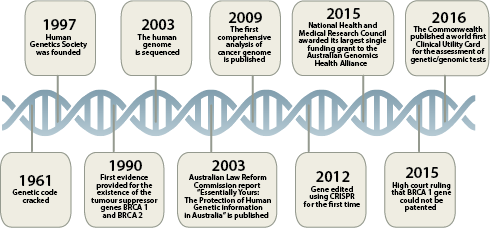


Figure 2: Significant events in genomics

Since the discovery of the DNA helical structure in 1953, there has been an exponential increase in our understanding of our genetic make-up, particularly in relation to improving human health.

### 1.3 Genomics in Australia

Australia is now well placed from a technological perspective to undertake large-scale sequencing and genotyping studies with every state/territory having access to accredited laboratories (refer Section 2.4 for further detail on accreditation) with the capacity to perform MPS. There are still opportunities for Australia to better prepare the community and the health workforce for the potential of genomics, as well as ensuring that genomics knowledge is appropriately embedded in the health system.

Since the human genome was first sequenced in 2003, genomic research in Australia has expanded to the extent that it is now recognised as being highly competitive with research done anywhere in the world. A number of research organisations are participating in global research efforts, such as the International Genome Cancer Consortium.

Australian clinicians, bioinformaticians and researchers now participate as leading members of international genomic alliances—influencing and contributing to global knowledge. Australia has advantages not found elsewhere in its close links between research and clinical practice, as well as access to unique population cohorts. It is important that Australia continues to take advantage of this situation and support innovation through appropriate policies and relevant research, while also ensuring that ethical, legal and social issues are appropriately addressed.

#### Current applications of genomic knowledge to health care

The following table describes the range of genetic and genomic tests currently used in Australia to prevent, diagnose, treat and monitor genetic conditions. There are also a range of different genetic testing techniques that can be used—the most appropriate cost-effective tests are usually determined by the multidisciplinary health care team in consultation with the pathology laboratory.

Figure 3: Range of genetic and genomic tests used in Australia

| **Type of test** | **Description** | **Example of use (Somatic or germline)** |
| --- | --- | --- |
| Diagnostic  testing | Diagnosis of an individual with symptoms of a genetic condition or disease. In relation to rare diseases, genomics is being used to identify gene variations that are responsible for each rare disease and, in some cases, to establish disease severity and determine if there are suitable drug options available. | A young child with physical and developmental delays has had Magnetic Resonance Imaging, X-rays, ultrasounds and blood tests without a diagnosis. The child has their DNA sequenced as a participant in a research study and is diagnosed with a rare syndrome. This information helps the parents make future reproductive decisions and to connect with other families whose children have the same syndrome.  (Germline/Somatic) |
| Prognostic  testing | Prognostic tests can help understand the biology of a tumour or rare disease by looking at the level of expression in certain genes. The goal is to better predict disease outcome (based on risk factors), determine signs of tumour recurrence and optimise treatment strategy and/or palliative care for cancer patients. | An adult male was diagnosed, and treated for uveal melanoma (UM), a type of cancer of the eye two years ago. He decides to undergo tests to determine the risk of secondary disease of the liver (the tumour metastasising). Based on the clinical, histo-pathological and genetic data from the primary tumour, as well as age and gender, he is considered to be at high risk and decides,  in consultation with his oncologist, to have regular liver screening to detect any secondary tumours early.  (Somatic) |
| Pharmacogenomic testing | Better targeting of existing drug therapy avoids wasteful, risky, and sometimes unnecessary, treatments. It can provide savings for the health care system and the economy at large.  In relation to cancer, genomic information can not only enable heritable risks to be identified (germline testing) but also allow the most suitable treatment to be determined (through somatic testing of the tumour). It can also be used to predict how, or if, an individual will respond to a drug and to determine dosage. | A woman has been diagnosed with early stage breast cancer. She undergoes genetic testing of the tumour and it is found that she is HER2-positive, because of a specific gene variation. This type of tumour is known to respond to the drug trastuzumab and she begins a course of treatment. (Somatic) |
| Genetic predisposition (predictive) testing | In some instances, germline testing of individuals who do not have any symptoms can assess predisposition or risk to disease, potentially prompting more focused clinical monitoring and lifestyle changes. | A woman with a strong family history of breast cancer chooses to have genetic testing for mutations of BRCA1 and BRCA2, which have been associated with breast and ovarian cancer. She tests positive for BRCA1 and must decide what to do, including whether to share the information with other family members.  (Germline) |
| Prenatal  genetic testing | Prenatal testing includes testing for chromosome disorders such as Down syndrome. Non-invasive pre-natal testing (NIPT), a maternal blood test,  is available in Australia.  Other current standard tests available include chorionic villus sampling (CVS) and amniocentesis and are performed in pregnancy to confirm chromosomal abnormalities such as Down syndrome or genetic conditions such as cystic fibrosis. | A pregnant woman undergoes prenatal screening uses NIPT and the results detect a possibility of a chromosome disorder. To confirm the results, she subsequently undergoes an invasive technique (CVS). The test result allows her and her partner to better prepare for their new child’s birth or to inform a decision to continue with the pregnancy. (Germline) |
| Preimplantation genetic diagnosis (PGD)—through Assisted Reproductive Technology | A procedure used before implantation to help identify genetic disorders within embryos created through in vitro fertilisation (IVF) to prevent certain diseases or disorders from being passed on to the child. | A child is diagnosed with Wilms’ Tumour (or nephroblastoma) and is successfully treated with chemotherapy and surgery to remove one of his kidneys. As an adult, he and his partner decide to start a family. Their first son is born healthy and has remained unaffected by Wilms’ Tumour but when their second child is three years old, she is diagnosed with Wilms’ Tumour. When considering a third child the couple are informed that an IVF process may help them avoid the genetic risk of another child developing Wilms’ Tumour. With the help of a specifically designed PGD test, embryos are screened for Wilms’ Tumour before transfer to ensure that only a healthy, unaffected embryo is transferred.  (Germline) |
| Carrier screening/testing | Carrier screening/testing is performed to detect whether a person carries a genetic variation that can cause inherited disorders.  Carriers typically don’t have the condition but can pass the variant to their children. If both parents are carriers, they may have a child with  the condition. | A woman has a history of cystic fibrosis in her family. She and her husband decide to start a family and want to know if their children are at risk of cystic fibrosis. The couple undergo carrier screening and discover only the husband is a carrier and they start their family without any further testing.  (Germline) |

#### Other applications of genomic knowledge in health care

There are other applications of genomic knowledge that can contribute to improved health outcomes, including sequencing of the human microbiome (the bacteria, viruses and fungi that live in and on the human body), and sequencing of infectious microbes (which identifies specific strains of pathogens causing infectious diseases).

Microbial genomics is important to public health care as it informs the surveillance, prevention and treatment of infectious diseases. While this application of non-human genomics is not an immediate focus for the first three years of the Framework, embedding genomics into the health care system will also require consideration of microbial genomics in the future.

## 2. Current Arrangements and National Challenges

The World Health Organization suggests that the key building blocks to health systems include good health services; a well-performing health workforce, a well-functioning health information system; and equitable access to technologies of assured quality, safety, efficacy and cost-effectiveness. Leadership and governance, which includes ensuring strategic policy frameworks exist, is also recognised as a health system domain. In the context of genomics, good health services are only possible through the appropriate translation of research to clinical practice.

Underlying these different aspects of the health system are the various ethical, legal and social issues that may arise in the application of genomic knowledge to public health, the provision of health care services and health research. This section describes the arrangements currently in place in terms of these building blocks, and identifies the challenges in each domain. These challenges highlight that there are opportunities to strengthen Australia’s capacity and capability to prepare for genomics as a disruptive technology that will potentially reshape clinical practice.

### 2.1 Ethical, legal and social implications

#### Ethical implications

Genomics presents a number of scientific, clinical, ethical, legal, and social issues and challenges. As there is limited national guidance on ethical decision-making in applying genomic knowledge in research and clinical care, there is a risk of varying approaches being taken in different jurisdictions.

Some ethical implications of the application of genomics are outlined below.

* **Uncertain predictive and familial implications**  
  Unlike other health interventions, genomics has the potential to predict genomic risks of complex human disease. This enables risk estimates of disease for individuals, which may lead to early intervention and improved diagnostic procedures. However, there are also ethical considerations in using some of these tests, and whether their availability may lead to large numbers of individuals seeking further management of a disease of uncertain probability (also known as the ‘worried well’).

#### Case study

A 75-year old woman with four adult daughters was treated two years ago for medullary thyroid cancer, a disease known to run in families. Following her death, her eldest daughter is also diagnosed with medullary thyroid cancer.

The daughter’s doctor tells her that she likely inherited the genetic disorder and encourages her to warn her siblings and her children. The daughter must decide whether to tell her family.

##### The familial challenge

Genomic-related health conditions can also be a family health issue because of the shared nature of genomes. Respecting an individual’s confidentiality by not disclosing the results of genetic and genomic tests to third parties can therefore conflict with the wellbeing of family members, who could benefit from this knowledge. Finding the right balance between the individual’s privacy and confidentiality of their genomic information, and what is in the best interests of family members, is an ongoing ethical and social challenge.

* **Secondary findings**Secondary findings are gene variants that are identified during testing, but are unrelated to the patient’s clinical presentation and the primary investigation for which the genomic test is performed. With the rapid increase in genomic testing, the prevalence of secondary findings has grown. Secondary findings can be complex and sensitive because of the potential to impact other members of the family.   
  There is currently no national agreement on what constitutes a best practice approach to secondary findings, with arrangements differing between states and territories. There is also no agreed position on whether there is a responsibility for existing genomic information to be reanalysed as new genomic knowledge emerges.

##### The secondary findings challenge

Whilst secondary findings offer an opportunity to proactively engage with medical conditions, they also present policy challenges in relation to community literacy and service delivery.

Engaging with secondary findings will depend largely on how well-informed patients are and how consent processes are structured. From a service delivery perspective, a nationally consistent process on how secondary findings are approached should form part of national guidance on bioethics in the context of public health policy. Guidance is also required to clarify the roles and responsibilities of medical professionals in presenting secondary findings.

A further complex policy consideration is around data retention and the responsibility or expectation that those patients that have consented will have their records retested against new genomic knowledge to uncover health conditions. This may mean that in the future it will be necessary to develop an agreed national position on those conditions for which there is sufficient evidence that existing genomic data should be reanalysed.

* **Managing genomic data**  
  Another ethical issue is the management of genomic data, including collection, security, quality, sharing, privacy and custodianship. Genomic information is personal. Every person has a unique genome, even though individual genes themselves may not be unique. Arguably, it is impossible to de-identify genomic information, particularly if the person has a rare genetic condition. Research suggests the trust that the community has in sharing their genomic information is largely dependent on data security and governance arrangements, the source of funding (for example, government or the private sector), and potential implications for insurance and employment.

##### The data challenge

To effectively harness the value of the genomic data generated in clinical care and research, issues of consent, oversight, data access and data quality need to be addressed. The management of genomic data and associated challenges from an ethical perspective are covered in more detail under Section 2.6—Information systems for genomics.

#### Legal implications

There are a number of regulatory measures in Australia that are relevant to genomics, including legislation and guidelines, as outlined below.

* **Legislation**  
  A range of legislation impacts the application of genomic knowledge to health care, including testing, research, privacy, insurance, discrimination, intellectual property and health records. One of the most comprehensive reports into the legislative arrangements was the Australian Law Reform Commission’s report in 2003 ‘Essentially Yours: The Protection of Human Genetic Information in Australia’, which looked at protection of privacy, protection against unfair discrimination and ensuring ethical standards. Important examples of current Commonwealth legislation relevant to genomics are listed in Appendix B.
* **Guidance**   
  The National Health and Medical Research Council (NHMRC) is a Commonwealth statutory body established to promote the development and maintenance of public and individual health standards. These include guidelines that are sets of non-mandatory rules, principles or recommendations for procedures or practices in a particular field. They only become mandatory if governments turn them into legislation, professional bodies incorporate them into codes of conduct for their members or funding bodies insist on compliance with them.

The NHMRC[[1]](#footnote-1) produces a range of resources on genomics and human health and related issues, including guidelines for, or guidance on:

* assisted reproductive technology, including preimplantation genetic diagnosis
* the use of human embryos
* ethics and framework for genomic research
* direct to consumer tests
* principles for the translation of ‘omics’-based tests from discovery to health care.

Many states/territories also have their own guidelines, for example Victoria has issued donor conception guidelines under their Assisted Reproductive Treatment Act 2008.

##### The legal challenge

It can be challenging for legislation to keep pace with advances in technology and community expectations, particularly in the context of discrimination, privacy and coverage of direct to consumer tests.

At the time of development of this Framework, a Joint Parliamentary Committee is looking into the issue of genomic testing and life insurance as part of a broader ‘Inquiry into the life insurance industry’ (report due to Parliament October 2017).

Any legislative barriers to embedding genomics in health care need to be identified and addressed.

#### Social implications

Genomics has the potential to present social dilemmas and destabilise social norms. For example, technological advances may provide for preventive action to be taken, but in the process, may challenge existing norms around personal responsibility for health.

##### The social challenge

Future advances in genomics need to reflect prevailing social norms.

**The need for action**

Ethical, legal and social issues associated with genomics need to be addressed as part of a national public policy agenda, informed by broad stakeholder discussion, addressed through:

**Framework principles:**

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.

### 2.2 Delivery of genomic services

Person-centred health care means delivering better care through services that are tailored to the individual, including their preferences, needs and values. The Australian Commission on Safety and Quality in Health Care acknowledges the importance of person-centred care as a dimension of high-quality health care. Other national initiatives including the Australian Charter of Healthcare Rights, the National Primary Health Care Strategy, the National Chronic Disease Strategy and the Fifth National Mental Health and Suicide Prevention Plan, all state that a patient-centred approach to health care is needed to improve the quality of health care in Australia. Current Aboriginal and Torres Strait Islander policies also reflect patient-centred principles and focus on family and community.

Engaging patients in health care decisions through shared decision-making is central to the concept of person-centred care. Existing research suggests that members of the general public have some familiarity with genetic and genomic terms but have gaps in understanding of underlying concepts. A considerable body of research has reported associations between low health literacy and less appropriate access to health care services, lower likelihood of self-managing health conditions well, and poorer health outcomes.

How people respond to information about the genetic risk for familial disorders and genomic susceptibility to diseases may also be affected by their level of health literacy. Different audiences need different information on genomic risks.

While consumers are being encouraged to take more responsibility for their health, there can also be risks in directly accessing genomic tests from overseas without the involvement of a health care professional (referred to as direct-to-consumer tests). Consumers can have a range of motives for accessing genomic testing directly, including out of curiosity or a sense of fun. However, such tests can also provide important health information on future disease risk. Due to the shared nature of genomics information, such information may have serious implications for individuals and their families. The information may also be potentially used for a range of commercial purposes that is not clear as part of the consent process and may also need to be declared as part of any application for life insurance.

The analytical and clinical value of direct-to-consumer genomic testing is yet to be established. There is a risk that different direct-to-consumer providers can deliver different results (including false positives and false negatives) from the same sample due to conflicting analytical approaches and methods of interpretation. Genetic risk is also just one factor that contributes to the likelihood of a person developing a particular disease. Although consumers can access direct-to-consumer genetic testing via overseas websites, there are currently restrictions on the supply of such services within Australia.

##### The challenge in delivery of services

Advances in genomic research and their potential application to common diseases that affect large segments of the general population, suggest that individuals with varying levels of health literacy need to be engaged in discussions about genomics. Low genomics literacy for both consumers and health care providers can limit the capacity to access, understand, evaluate and apply information that will facilitate the appropriate application of genomics for the benefit of individuals and their communities.

**The need for action**

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

**Strategic Priority 1:**

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

### 2.3 Funding cost-effective genomic services

Health care for Australians is funded through many different arrangements. Health costs have been rising over time, due to a range of factors including population growth, population ageing, development of new technologies and changing expectations of health care. The Commonwealth and state/territory governments help Australians access necessary health services and technologies by subsidising (in part or in full) the costs of health-related goods and services.

Health technology assessment (HTA) is a key tool for governments to achieve the overall objective of delivering a safe, effective and efficient health system that is fiscally sustainable in the longer term. Australia’s approach to assessing genomics through a HTA system is recognised as an advantage over other countries that have stand-alone genomic assessment processes.

HTA processes and mechanisms provide a means by which new genomic services are assessed against existing health interventions. The key HTA considerations are safety, effectiveness and cost-effectiveness. There is no nationally consistent approach to HTA (or coordinated approach to disinvestment), with decisions made at the Commonwealth, state/territory and, in some cases, the local hospital level.

Funding of genetic and genomic tests in Australia is affected by a number of specific factors including the nature of the disorder, the pathways through which tests are requested, potential participation in research projects and where the individual lives. Private health insurance generally only subsidises genetic tests required as part of treatment when people are privately admitted into hospital, and only if the Medicare Benefits Schedule (MBS) also provides a rebate for the test. The listing of new tests on the MBS is subject to Medical Services Advisory Committee (MSAC) consideration and approval processes[[2]](#footnote-2).

In practice, the majority of genetic and genomic tests (in particular more complex expensive tests) are provided by specialist laboratories and funded on a state by state (or territory) basis. The 2011 Report of the Royal College of Pathologists of Australasia (RCPA) Genetic Testing Survey estimated that up to 75 per cent of genetic tests were funded by states and territories, and/or patients. For this reason, some states are developing their own strategies including processes to better inform the ordering of clinically appropriate genomic tests, which will also guide clinical practice.

Medicare currently funds fewer than 30 genetic and genomic tests in Australia; two examples are tests to detect hereditary hemochromatosis and Fragile X syndrome. While the range of tests funded by Medicare is relatively small, a number of these tests tend to be ordered in high volume. In 2016, over 260,000 genetic tests were reimbursed through the MBS, which is an increase of about 24 per cent since 2012, at a cost of almost $43.5 million[[3]](#footnote-3).

Consequently, funding for genomics is largely embedded in state-funded health systems with increasing clinical activity in the acute care/public health setting and in the private sector.

For example, in 2017 the Victorian Government announced additional funding of $8.3 million over four years for public access to genomic testing for children and adults with rare disease and undiagnosed conditions.

The cost of massively parallel sequencing includes three components: the pre-analytics and assay; bioinformatics; and professional interpretation and services. While the cost of the assay component is declining, there is more uncertainty about the cost trends for the other components.

#### Clinical Utility Card

MSAC is implementing arrangements to assess the utility of germline genetic testing for predisposition to broad disease areas such as cancer, cardiovascular or mental illness.

##### The funding challenge

It is important that the economic evaluation of genomics supports the best use of scarce health system resources. This type of research is critical for resolving uncertainty in health care, but also for the ongoing safety and quality improvement of genomic applications and their cost-effective uptake by the health care system.

Australia’s current HTA process makes recommendations for funding based on the principles of evidence-based health care together with cost-effectiveness analyses. However, the application of genomics to health care presents particular challenges to the current HTA process, including:

* the amount of data generated that is not directly relevant to the clinical investigation, which create demand for health care services with limited clinical utility
* potential predictive nature of the risk of disease
* the familial implications
* clinical trial structures (noting that participation can be limited where the trial involves a rare genetic disorder)
* the links between testing for genetic variations, which indicate the effectiveness of specific pharmaceuticals (co-dependent technology).

New models of care may require new funding models, in particular for patients requiring ongoing genomics related advice and services.

The disconnected nature of genomics funding has led to inconsistent decision-making, which, in turn, undermines equity of access, as the cost and financing of genomic services is widely reported to have a significant impact on access to services. Equity of access to genomic services, and individual choice about courses of treatment across Australia, will continue to be challenging without any nationally consistent approach to decisions about what genomic services are funded and to which groups within the population. Equity of access can be strengthened through the cooperation of Commonwealth and state/territory governments, noting that equity of access is a broader challenge across the health system.

To realise the broad public benefits from genomics, cost-effective genomic testing needs to be available to all, rather than just those that can afford to pay privately or happen to live in an area where the service is funded through their local health services. If equity of access to genomics testing and services is not actively promoted, there is a risk that the gap in health outcomes for vulnerable populations will become wider.

**The need for action**

Australia’s investment in genomic services needs to support equity of access and deliver actionable results that lead to people living longer and better lives, addressed through:

**Framework Principle 2:**

Access and equity are promoted for vulnerable populations.

**Strategic priorities:**

3. Ensuring sustainable and strategic investment in cost-effective genomics.

4. Maximisingquality, safety and clinical utility of genomics in health care.

### 2.4 Safety and quality

Safety and quality is central to the delivery of health care. In August 2012, Australian Health Ministers agreed to the first set of Australian Safety and Quality Goals for Health including:

* **Partnering with consumers**—that there are effective partnerships between consumers and health care providers and organisations at levels of health care provision, planning and evaluation
* **Safety of care**—that people receive health care without experiencing preventable harm
* **Appropriateness of care**—that people receive appropriate, evidence-based care.

For diagnostic-based genomic applications to be high-quality and safe, evidence in three domains needs to be evaluated in combination with the economic, ethical, legal and social implications of the test. These domains are:

* analytical validity—how well the test performs in the laboratory
* clinical validity—how well the test correlates with clinical condition
* clinical utility—whether use of the test is likely to subsequently improve health outcomes.

Utility requires that genomics provides the greatest possible benefit across a population. This is necessary if genomics interventions are to be scientifically sound and have clinical utility.

#### Medical device regulatory framework

The Therapeutic Goods Administration (TGA) assesses the safety and efficacy of health technologies for market regulation and is responsible for administering the In Vitro Diagnostic (IVD) medical device regulatory framework, which came into full effect on 1 July 2017. Under this framework, all commercially supplied genomic tests must be registered with the TGA and any Australian laboratory developing their own genomic tests (including MPS) on human samples that can affect the diagnosis, treatment or management of a patient must be accredited by the National Association of Testing Authorities (NATA)/RCPA and notify the TGA of the tests being used. All genomic tests, including laboratory developed (i.e. in-house) tests, must be validated, including establishing the clinical validity and utility of the test.

#### Diagnostic pathology laboratories

It is a legislative requirement under the Health Insurance Act 1973 that Medicare benefits for pathology services cannot be provided unless the laboratory is accredited under a recognised scheme. The NATA/RCPA run an accreditation scheme for this purpose with the National Pathology Accreditation Advisory Council (NPAAC) overseeing the standards and providing education to the sector. NATA/RCPA accredited pathology laboratories are required to participate in both internal and external quality assurance programs.

There is no requirement for laboratories that do not seek reimbursement under Medicare, or that do not provide services to public hospitals to be NATA/RCPA accredited. However, under the Therapeutic Goods Act 1989, laboratories that are using in-house genomic tests to provide patient results are required to maintain NATA/RCPA accreditation against the NPAAC standards.

There are different requirements for the safety and quality of genetic and genomic tests that are applied in research compared with the clinical setting.

#### The NPAAC standard

‘The Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies’ came into effect on 1 April 2017.

This standard applies to all genomic applications (single gene, panel of genes, somatic testing, while exome or whole genome sequencing).

##### The safety and quality challenge

A major challenge lies in balancing clinical validity, clinical utility and cost-effectiveness of testing. Some tests have excellent analytical validity, but are not viable from the clinical or economical point of view. On the other hand, some tests have poor analytical validity, but nevertheless impact on patient and family management. Therefore, it is important that the safety and quality of genetic or genomic tests is considered in the clinical context, as well as the laboratory.

There is increasing complexity in the way genomic testing services are being delivered in Australia, with different elements of the test undertaken by different parties. Some genomic tests are undertaken by laboratories overseas either because the test is not available in Australia, or the consumer orders the test from overseas, usually through the internet (known as direct to consumer tests). This creates a regulatory challenge given that Australia has no jurisdiction in other countries.

Another challenge is that unlike traditional diagnostics, which typically detect the presence of one, or several, biomarkers or genes, genomic sequencing can screen for numerous biomarkers and relevant mutations simultaneously. The capabilities of genomic sequencing and the rapid evolution in the field pose regulatory challenges. This is because, once sequence data is obtained, it can facilitate ongoing analysis for any additional or emerging biomarkers/mutations. This form of testing may require recurrent scrutiny of clinical evidence to assess the ongoing safety and effectiveness of a given genomic test, particularly as interpretation of the same data can change quickly.

**The need for action**

Safety and quality of genomics in health care should be supported by regulation that is sufficiently flexible to accommodate emerging technologies and new models of testing, addressed through:

**Framework Principle 1:**

The application of genomic knowledge is ethically, legally and socially responsible and community trust is promoted.

**Strategic Priority 4:**

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

### 2.5 The workforce

The provision of clinical genetics services involves multidisciplinary teams. These typically comprise clinical geneticists, genetic counsellors and specialist social workers. Generally, these services work collaboratively with other medical specialties, pathology laboratories and researchers. Australian genetic services are available in all states and territories.

Preliminary mapping of the genetics and genomics workforce in Australia in 2015 by the Australian Genomics Health Alliance (AGHA) indicated that:

* while genetic counsellors are not a registered profession, there is work progressing to strengthen the self-regulatory arrangements through the National Alliance of Self-Regulating Health Professions
* smaller jurisdictions such as Northern Territory, Tasmania and the Australian Capital Territory face significant challenges in supporting access to specialist genetic and genomic services, and currently contract services from interstate.

#### Genomics is dependent on a cross-cutting health workforce

A new trans-disciplinary workforce model is required that provides for bioinformaticians as a new component and broader clinical involvement.

As genomics evolves so will workforce needs and roles.

##### The workforce challenge

The emergence of genomics is having a significant impact on the genetic and genomic, as well as the broader, health workforce.

There are a number of workforce challenges to integrating genomics into clinical care, including:

* the low level of genetic and genomic literacy in the non-genetic specialist health professions, noting the emerging role for the broader health workforce in the delivery of clinical genomics
* the lack of agreed clinical pathways
* the lack of clarity about who is best placed to coordinate individual care, for example, the general practitioner, the genetic specialist or the referring specialist
* ensuring services are culturally safe and appropriate
* some clinical genomics activity is carried out in the research setting, rather than as part of routine clinical care, changing the composition of multidisciplinary team.

Demand for clinical genetic and genomics services currently exceeds workforce capacity and capability. This has placed pressure on the small and highly specialised clinical genetics and genomics workforce, and increasingly the broader health workforce. For example, in some jurisdictions, waiting times for some public genetic clinic consultations are currently up to 12 months from initial referral. Anecdotally, it has been suggested that the number of people training each year is insufficient to meet the need for workforce positions.

Another significant challenge is that advances in genomics in health care are changing the composition of the workforce that provides clinical genomics services. For example, other medical specialities such as cardiologists, oncologists and obstetricians are having an increasing role in the provision of clinical genomics services, including providing genetic counselling. Currently, there are limited workforce training and education opportunities to ensure the broader health workforce has the genomic literacy and capability to deliver quality and safe clinical genomic services.

Typically, a multidisciplinary team for clinical genomics involves collaborative efforts between the individual, their family, the genetic counsellor, psychologist, nurses, genetic/genomic pathologists and scientists, specialist bioinformaticians, clinical geneticists and other clinical specialists.

For genomic knowledge to be integrated into the health system, it is critical that the broader health workforce develop an understanding of the application of genomics to health care.

#### The specialist genomics workforce:

* 62 qualified clinical geneticists, with a further 20 in training, and 18 qualified genetic pathologists, with a further six in training
* 280 accredited genetic counsellors and trainees (as members of the Australasian Society of Genetic Counsellors, a special interest group of the Human Genetics Society of Australasia HGSA)
* 80+ clinical geneticists and those training in this discipline (as members of the Australasian Association of Clinical Geneticists, another special interest group of the HGSA)

**The need for action**

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 health, addressed through:

**Strategic priority 2:**

Building a skilled workforce that is literate in genomics.

### 2.6 Information systems for genomics

Integrating genomic knowledge into the health system is dependent on Australia’s capacity to store, use and manage the large volume of data generated through genome sequencing. The information generated from sequencing a person’s entire genome (genomic data) involves reading a code made up of around three billion units, generating between 700 megabytes and 180 gigabytes of data depending on the sequencing methodology used.

Genomics data carries ethical, legal and social complexities as it unravels critical biological factors that determine who we are and possibly how we will live our lives. In this context, the pressing underlying issues are the management of genomic data including collection, security, quality, sharing, privacy, use and custodianship. These issues are further complicated because clinical data is subject to different requirements than research data.

A critical mass of data needs to be available for analysis to validate the link between variations in the genome and specific health risks, conditions/disorders or treatment responses. Consequently, the wider the data is shared, the more accurate the analysis and correlation of genotypic and phenotypic data. This is particularly true for building the data to diagnose and treat rare diseases and cancers. Currently there is no agreed national approach to building Australia’s bioinformatic infrastructure, including high-performance computing and data storage.

The vast amount of genomic data recorded also increases the risk of privacy breaches and data misuse. Communication technologies and improved analytics are rapidly driving Australia’s health system to the cusp of an information-age health system, where genomic data may be better integrated into health care. Internationally, work is being undertaken to develop viable electronic medical record systems capable of handling family history and genomic data required to fully utilise genomic information for patient care. This recognises that existing clinical informatics architectures are largely incapable of storing genome sequence data in a way that allows the information to be searched, annotated and shared across health care systems over an individual’s lifespan.

At present, there are pockets of activity occurring across Australia. The Australian Digital Health Agency, as a Commonwealth statutory agency, is currently working with the AGHA on the future development of Australia’s digital health foundations (including the My Health Record system) to support the integration of genomic information with other health data. The AGHA and the NHMRC are involved in the Global Alliance for Genomics and Health, which aims to facilitate access to, and exchange of, genomics data across international borders.

##### The data challenge

The collection of genomic data for the purposes of research is critical for the discovery and the application of clinically effective interventions within the health system. The informational value of genomic data to improving health care can only be harnessed by collecting a high volume of data, as well as by being able to share and analyse this information. The types of data that are valuable to share for research, interpretative and diagnostic purposes range from genetic data to associated clinical, genotypic, phenotypic and metadata. Accurate determinations and diagnosis is dependent on two key factors:

* quality and completeness of the underlying knowledge base
* knowledge of the variants in an individual’s genome.

Individuals can support development of personalised health care by agreeing to share their data. There is a need to balance policy to reflect the individual’s right to own their data against the benefits to population health. Public recognition that sharing of genomic information serves the common interest for mutual benefit is critical to advancing genomic knowledge.

Correspondingly the safeguards and protections in place must reflect the needs of the community and ensure that the public is reaping the benefits of advances in genomic knowledge.

Currently in Australia, there is no nationally consistent or coordinated approach to the collection, generation, storage, analysis, translation and utilisation of genomic data, as well as investment in the physical infrastructure required to support storage and analysis. There is inconsistency and fragmentation in approaches to data governance and management across and within jurisdictions. The AGHA is undertaking some critical foundation work in exploring a national approach to data collection, storage and analysis.

Through future governance structures, a collaborative decision will need to be made on how data collection, storage, management and use are managed in Australia. This includes options on system architecture models, quality and standards. Planning for this will require engagement with consumers as well as the organisations within the genomics sector. Approaches to data will need to be flexible to prepare for advances (such as machine learning) that will shape analytics and the workforce required to investigate genomic data.

**The need for action**

Establishing a seamless flow of genomic data from research to clinical application (and vice versa) requires coordinated governance, collaboration, investment in infrastructure, workforce training and community trust, addressed through:

**Strategic priorities:**

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

2. Building a skilled workforce that is literate in genomics.

5. Responsible collection, storage, use and management of genomic data.

### 2.7 Establishing the evidence-base: Translational research

Advances in genomics, machine learning and ‘big data’ is leading to an increased use of next-generation sequencing in health care and a higher integration of research and clinical practices. Jurisdictions are currently heavily investing in genomics translational research over the next two- to four-year period, including:

* $25 million commitment by the Commonwealth Government through the NHMRC’s Targeted Call for Research into Preparing Australia for the Genomics Revolution in Health Care (awarded to AGHA)
* $25 million commitment by the Queensland Government, reflected in the recent establishment of Queensland Genomics Health Alliance
* $25 million commitment by the Victorian Government to the Melbourne Genomics Health Alliance in 2015
* $24 million commitment by New South Wales Government in 2014 for the Sydney Genomics Collaborative and NSW Genomics Collaborative Grants Program
* $7 million commitment by the Australian Capital Territory Government in 2016 for the new Clinical Genomic Service in Canberra.

The research sector currently provides substantial amount of genomic testing in association with clinical services. This leads to a blurred boundary between research and clinical services; increasingly research participants are identified in the clinic, have their genome sequenced in the research setting, which generates research findings used to inform their clinical care in real time. This is often an iterative feedback loop, particularly where there is uncertainty about the causal relationship between the patient’s genomic information and their condition. This blurred boundary challenges the usual paradigm of research and clinical care, which is often represented as a linear pipeline (refer to Figure 4).

##### The translational research challenge

As a result of the blurred boundary, it is often difficult for individuals and regulators to distinguish between research and clinical activities. It is also increasingly common for clinical pathways that incorporate genomic tests carried out in the research setting to be embedded in health care services. Different ethical frameworks can apply to research than clinical care and the levels of evidence required also differ. Different regulatory levers also apply to tests carried out in the research setting, compared with the clinical setting.

There is currently debate within Australia and internationally that conventional clinical trials are no longer the best solution and that even more innovative approaches that focus on individual, not average, responses to interventions, may be required.

**The need for action**

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

**Framework Principle 3:**

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

**Strategic Priorities:**

3. Ensuring sustainable and strategic investment in cost-effective genomics.

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



Figure 4: Genomics is changing the research transition pathway

Genomics research is increasingly being undertaken in the clinic, with research informing clinical practice informing research, leading to a research-clinical care cycle.

## 3. What Can Governments Do?

Governments have a responsibility to ensure genomic technologies are used responsibly and appropriately and to best effect. A person-centred approach to genomics in health care will mean individuals are empowered to ask for, access and use information about themselves. This has significant implications for electronic health records, data storage and sharing. Society needs to consider ethical implications, balancing individual and population needs, data and information privacy and sharing, potential inequalities, regulation of devices and diagnostics, and the implications of direct to consumer testing.

The Commonwealth and state and territory governments face a major policy challenge in integrating genomics into Australia’s health system. This challenge requires new thinking, new approaches and strengthened national cooperation and leadership.

Appropriate leadership and governance by the Commonwealth and states/territories includes ensuring strategic policy frameworks exist. A national policy framework sets out how the Commonwealth and states and territories will work collaboratively to integrate genomics into the Australian health system over time. It provides a shared direction and commitment between governments to address current and emerging genomics priorities in a nationally coordinated and consistent way, as resources permit.

While there is a clear and compelling case for concerted action to be taken in the short to medium term to integrate genomics in the delivery of health care, the longer-term objective must be for genomics to become part of the mainstream health system. The integration of genomics into the health system will require social acceptance, education and valuing of genomic services, particularly genomics literacy among clinicians and the wider community.

### 3.1 Benefits of government action

A concerted and nationally coordinated effort to embed genomics into the health care system, will:

* help people access culturally safe and appropriate services, irrespective of their geographic location
* promote legislation, regulation and financing models that keep pace with technological advances, mitigating risks to the community, in particular vulnerable groups or discrimination (for example, life insurance)
* strengthen the case for governments and individuals to fund genomic tests or services that have clinical utility and lead to improved health outcomes
* build community trust and confidence in the use of genomics
* support research through quality data that can be linked to other national/international data
* maximise the use of resources through avoiding duplication of effort.

Ultimately, the Australian community will reap the benefits of a national health system that avoids the costs associated with fragmented, uncoordinated and inconsistent health care.

## 4. The First Step: A National Health Genomics Policy Framework

The Framework is a shared commitment between governments, endorsed through the national Council of Australian Governments Health Council to enhance national coordination on agreed priorities.

### 4.1 Development of the National Health Genomics Policy Framework

The Australian Health Ministers’ Advisory Council (AHMAC) Hospitals Principal Committee developed this Framework with input from a Jurisdictional Advisory Group. Membership comprised a Commonwealth Chair and a member from each state/territory.

An extensive consultation process was undertaken during the development of the Framework, with feedback gathered through seven public consultation forums, around 80 written submissions, and targeted discussions. The consultation process was integral to shaping the Framework from the perspectives of a range of stakeholders, including consumers, researchers, academics, private industry, clinicians and governments.

### 4.2 Vision

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

### 4.3 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.

### 4.4 Enablers of Success

Key enablers to guiding implementation of this Framework include:

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

#### Collaborative governance and leadership

Effective national governance arrangements will guide the successful implementation of this Framework, and ensure it achieves the ultimate objective of better health outcomes for individuals and the population. Governance arrangements will need to promote:

* openness and transparency
* accountability to the community
* leadership and oversight
* stakeholder participation
* inter-jurisdictional and inter-sectoral collaboration
* ongoing evaluation and review, to enable quick and flexible responses to emerging issues.

National coordination of effort, through a national governance body, will be critical to achieve improvements in all strategic priority areas of the Framework. Collaborative leadership will:

* establish a shared purpose and commitment to achieving the Framework principles
* drive future genomics policy directions
* establish consistent ethical and legal approaches
* ensure a whole-of-system view is taken
* foster public confidence and trust.

#### Stakeholder engagement

This Framework was informed by feedback from a broad range of stakeholders. It is expected that implementation will similarly be guided by a multi-stakeholder approach, including input from states/territories, ethicists, consumers, clinicians, researchers and industry representatives.

Consumers are the ultimate stakeholder in the health care system and must continue to be engaged in developing the actions to implement the Framework. Implementation planning will be informed by the experience of people living with a genetic condition, carers and their families.

#### National and international partnerships

National and international collaboration will be necessary to maximise outcomes and reduce duplication of effort in the translation of genomics research or applications of genomics for population health. In this context, Australia is already making a valuable contribution to international efforts with a considerable range of genomics-related activities being undertaken by state and national agencies, non-government organisations and international governmental organisations. These activities provide opportunities for partnerships, developing common best practices and clinical guidelines. Opportunities for Australia to continue to contribute to international efforts will be maximised through a coordinated approach to creating and sharing genomic knowledge.

### 4.5 Principles underpinning the Framework

The following principles underpin the Framework and implementation:

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

#### Principle 1: The application of genomic knowledge is ethically, legally and socially responsible and community trust is promoted

The Framework is an important step towards building assurance that appropriate safeguards exist to support integration of genomics into the health care of Australians. It needs to be sufficiently flexible to incorporate emerging societal issues and the broad spectrum of community views, in particular around the use and storage of data. The following ethical principles are proposed to inform discussion of ethical dilemmas arising in the application of genomics in health care.

* Respect for persons—includes respecting autonomy, which values the right of the individual to self-determination. In the context of genomics, respect for people, includes:
  + Voluntary informed consent—the right of the individual to make their own decisions without coercion, based on an understanding of benefits, risks and any limitations. Consent must be current and specific, and the individual must have the capacity to understand and communicate their consent. Autonomy should be promoted through a person-centred approach.
  + Privacy and confidentiality—individuals have a right to the protection of their data and for confidentiality to be maintained.
* Do no harm (non-maleficence)—the interests and welfare of the individual should have priority, including in respect of human rights, genetic discrimination and testing-induced harms.
* Act in the best interest of the individual (beneficence)—all clinical genomic interventions must be scientifically sound, clinically indicated and employed after assessing the risks and benefits.
* Equity of access and sharing the benefits of research (justice)—a concept that emphasises fairness among individuals and between communities (to address disparities) and sharing the benefits of research.
* Solidarity—the notion that, in certain circumstances, common interests are served by sharing genetic information for mutual benefit.

In putting forward these principles, it is acknowledged that they will not necessarily be readily translated into the development of specific or unambiguous rules of conduct. In particular, the need to balance maintaining confidentiality and permitting wide access to data is fundamental to building and maintaining public trust and confidence (balancing individual privacy with solidarity through data sharing).

#### Principle 2: Access and equity are promoted for vulnerable populations

Among the issues raised by developments in genomics, some of the most complex and important involve the effects on vulnerable populations. There is a risk that without any coordinated effort, there will be greater inequity of access to genomics testing and services, resulting in variations of individual and population outcomes. Equity in health care means working towards fair access, informed decisions and fair resource distribution to alleviate any disadvantage experienced by at-risk or vulnerable populations.

Access to testing and services is not simply about geographic location. Economic and social factors may also affect an individual’s ability to access services, including availability, cost and appropriateness of services, as well as demand factors such as knowledge, attitudes and skills and self-care practices. Literacy and bias is a specific challenge for disadvantaged populations.

While access and equity must be promoted, consideration must be given to balancing this with the sustainability of the broader health system.

##### Aboriginal and Torres Strait Islander peoples

Genomics can contribute towards achieving health and life expectation equality for Australia’s Aboriginal and Torres Strait Islander peoples (known as Closing the Gap Campaign). However, at present Aboriginal and Torres Strait Islander peoples tend to be underrepresented as consumers of genomic services and in clinical trials.

Aboriginal and Torres Strait Islander concepts of health extend beyond the physical wellbeing of an individual to the social, emotional and cultural wellbeing of the whole community. Culture can influence decisions about when and why Aboriginal and Torres Strait Islander peoples seek health services, acceptance or rejection of treatment, the likelihood of adherence to treatment and follow up, as well as the likely success of prevention and health promotion strategies. Recognising the centrality of culture to health, and respecting Aboriginal and Torres Strait Islander peoples and cultures is necessary to enhance service access, equity and effectiveness.

Aboriginal and Torres Strait Islander patients, families, carers and communities are entitled to have the opportunity and capacity to make well-informed decisions to access culturally safe and appropriate genomic services. Aboriginal and Torres Strait Islander people need to be empowered to be active in health genomic research specific to their cultural and/or population health needs.

##### Children

Children, in particular young children, may lack informed decision-making capabilities and informed consent must be obtained from a legally appropriate health care decision-maker (usually the parents), who are obliged to act in the child’s best interest. However, this is a complex and controversial concept because the child’s interests are often debatable and may change once they become an adult. While potentially useful for a child who is ill, the decision is less straightforward when the test is being performed to predict a heritable condition that might not occur until later in life and there is uncertainty about how the information may be used for other purposes.

##### Culturally and linguistically diverse communities

Cultural factors such as beliefs, values and ethnic backgrounds play an important role in determining genomic risk profiles, genomic risk assessment and testing, and responses to genetic risk information. Linguistic diversity and literacy are important factors to consider in the communication of information, gaining of consent including consideration of risks and provision of results.

##### People living with disability

The application of genomic knowledge must respect the rights and autonomy of individuals, and particular care is needed to ensure that these principles are applied to those living with disabilities. The application of genomic knowledge has the potential to prevent, diagnose, treat and manage genetic conditions resulting in disability. While these advances in genomics are generally welcomed, there is also a risk that society may marginalise people with disability who do not choose medical intervention. Different individuals, communities, cultures and religions have different perceptions of disability and this may raise additional issues.

Some adults living with disability lack capacity to make informed health care decisions. In such cases, a legally appropriate health care decision maker will act on their behalf by upholding the wishes of the person expressed while they had capacity, or in their absence, acting in their best interests.

##### Other vulnerable populations

There are several groups in Australia with poorer health outcomes than the general population due to a range of environmental and socio-economic factors. Priority population groups for health interventions also include people who are socioeconomically disadvantaged and people in rural and remote areas.

#### Principle 3: The application of genomic knowledge to health care is supported and informed by research and evidence

Genomic applications are like other medical services in that their use in health care should be based on evidence and supported by research. While evidential standards for the clinical use of genomic applications are still evolving, ongoing data collection is vital for establishing comparative safety and cost effectiveness. Health and medical professionals are used to dealing with some level of uncertainty. This level is increased for genomics because genome sequencing can reveal information about future disease risk that is secondary to the initial clinical investigation. While some of these findings (refer Section 2.1—secondary findings) will be clinically proven and useful, others will be relevant to research only. This demonstrates how genomics challenges the linear research pathway that is common to many disciplines because questions raised in health care can be fed back to research (refer to Figure 1 and 4).

The translation of genomic knowledge into clinical practice must be supported by the assessment of safety, effectiveness and cost effectiveness through a robust HTA process. Without this, there is a risk of premature adoption and overuse of new tests that are not supported by clinical evidence, and which may impose a cost burden on consumers and our health system in comparison to conventional tests with established clinical utility and validity.

### STRATEGIC PRIORITY 1: Person-Centred Approach

**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 genomic knowledge.*

**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.**

There are different levels of understanding about genomics in the community. New partnerships between the education system, the health care system, the government, community advocacy organisations, consumers and the media are required so that if a person is referred for testing, or offered testing, they can make an informed health decision. The consent process should clearly communicate the potential benefits and risks of testing, including the value of sharing genomic data for research purposes to further advance our knowledge of the role of genes in health and illness.

As the cost of whole exome or whole genome sequencing becomes progressively more affordable, the key challenge is how to equip consumers with the information they need to make an informed decision to undertake a test.

**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.***

Clinical practice guidelines support health professionals in assisting patients with appropriate and informed decision-making about genomic testing, and interpreting results in the context of clinical decision-making. This applies across all relevant areas of the health system.

**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.***

Respectful debate of differing views and active participation by consumers and the broader community is part of collaborative decision-making. Knowing what consumers experience through their health care journey is a key component in ensuring the delivery of high- quality health care. An integral part of improving person-centred care is measuring the patient’s experience and identifying opportunities to prioritise initiatives to optimise that experience.

**1.4 Promote public awareness and understanding of genomics, including through linguistically and culturally safe and appropriate information resources for targeted consumer groups.**

Public understanding of genomic concepts and associated ethical and policy issues enables informed deliberation and decision-making about genomic testing. Health literacy in genomics and an understanding of the expectations of the public will help to shape clinical practice guidelines; consumer resources; and recommendations on the ethical, legal and social implications of genomics.

**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.***

Equity of access is an underlying principle of the Framework and is integral to person-centred care, as well as safe, ethical, effective, evidence-based and holistic health care.

There are a range of barriers to the uptake of clinical genomics. The potential need to disclose genomic information for life insurance or employment purposes has been identified as one of the most significant barriers. Aboriginal and Torres Strait Islander people are under-represented as a population group for the uptake of services, likely influenced by cultural and access issues, as discussed under Principle 2.

**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.**

There is the potential for electronic health records to capture genomic information as part of a patient’s clinical record (see Section 2.6). As opportunities arise to build flexibility into these electronic systems, it will be necessary to consider how they can best support the sharing of genomic information to inform research and clinical care.

**1.7 Explore the potential to develop integrated person and family-centred care delivered by multidisciplinary teams (where appropriate).**

In the context of genomics, sometimes the term ‘family-centred care’ is used, rather than person-centred care. This concept encompasses the concepts of parental participation (where a child is involved), partnership and collaboration between the health care team and parents in decision-making, and care of other family members. Care of other family members can be particularly important given the conditions, or predispositions that are associated with genetic variations are often inherited. In considering ethical issues around consent, health professionals have a responsibility to clarify not only what testing may mean for the health of the individual (particularly if the analysis is likely to guide/improve treatment or interventions), but also what the information may mean for their relatives, and their responsibilities towards those relatives.

**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.**

One of the core attributes of person-centred care, and also a widely recognised cornerstone of the ethical application of genomic knowledge, is informed consent that is voluntarily provided. The goal of informed consent is to ensure that individuals and their families are aware of the risks and potential benefits and make a voluntary decision about having their (or their child’s) genome sequenced.

A flexible approach to consent is also important in the context of the implications if the genomic sequencing reveals a clinical finding that is not related to the condition being tested for. The consent process needs to reflect the potential psycho-social, personal, financial consequences and the risk of harm to the individual as well as the potential benefits.

The consent model should be respectful of, and responsive to individual cultural health beliefs and practices, preferred language and health literacy level. The community decision-making processes of Aboriginal and Torres Strait Islander peoples is to be taken into account in developing a consent model in consultation with Indigenous communities, noting that Aboriginal and Torres Strait Islander societies view genomic knowledge as family knowledge and therefore a community, rather than individual, issue.

### 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.*

The future integration of genomics into health care is highly dependent on the development and expansion of an appropriately literate, skilled and resourced workforce.

**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.**

There is a pressing need to address the low level of genetic and genomic literacy and capability of health professions, who are having an increasingly expanded and central role to the provision of clinical genomic services. The workforce needs to have the skills to respond to and adopt new research, technology and innovation, to ensure that the potential benefit of genomics to people are realised. Nationally consistent action is required to develop and maintain the genomic literacy and capability of the broader health workforce.

An important approach to improving genomic literacy across the health workforce is to target the emerging health workforce by embedding genomics education modules in undergraduate and postgraduate health professional programs. Improvements in genomics literacy and capability can also be achieved and maintained across the existing and emerging health workforce, through the availability of appropriate genomic education and training in continuing professional development programs.

The development of genomics education and training programs must apply a person-centred approach, and include competencies for delivering culturally safe and appropriate care to Aboriginal and Torres Strait Islander people, and people from culturally and linguistically diverse backgrounds. Further, the genomics training and education programs must be responsive to advances in genomics and be available to the broader health workforce, including clinicians and allied health professionals.

Specialised knowledge and skills are required to accurately analyse, validly interpret and appropriately use genomic information in health care. Genomics education and training programs should be supported by a nationally consistent certification and/or accreditation to build consumer confidence in the health workforce and clinical genomics services.

**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.**

The integration of genomics into health care will lead to an increased demand for access to clinical genomic services provided by a skilled and literate workforce. Noting that current demand for clinical genetic services is placing pressure on the clinical genetic workforce, there is a need for workforce strategies and planning at the national level. This is important to ensure that people can access health professionals appropriately trained in genetics and genomics when they need to and when is appropriate to do so.

The Commonwealth and state and territory governments need to work together to develop health workforce strategies to support the integration of genomics into health care, and promote equity of access. This may involve exploring innovative service delivery methods, such as through telemedicine/telehealth platforms to address workforce shortages.

Workforce planning and strategies should also reflect diversity within Australia and be drawn from a variety of backgrounds, including Aboriginal and Torres Strait Islander peoples. Consideration must also be given the capacity of the health system to manage the flow-on impacts of genomic testing and treatment, including the impact on the broader health workforce.

**2.3 Facilitate partnerships and networks to promote and support sharing of knowledge.**

Genomics in human health requires a multidisciplinary team approach, involving a range of specialised skills and knowledge across the clinical and allied health workforce. Integral to such an approach is: establishing clear roles and responsibilities of all providers involved in delivering genomic services; sharing relevant clinical information within the team; and effectively communicating the impact of genomic knowledge on individuals and their families.

There is also a need to develop agreed consistent, evidence-based and informed best practice approaches to guide genomics multidisciplinary teams in supporting and empowering individuals to make informed autonomous decisions. Genomics presents complex and challenging ethical considerations that must be worked through by the multidisciplinary teams, in collaboration with individuals and their families. Sharing information is an important aspect of promoting best practice approaches and building the evidence-base.

### 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.*

**3.1 Consider genomics in the context of any broader review of health technology assessment to support national consistency.**

It is important that integrated and nationally consistent approaches to HTA are adopted to ensure that genomics is not siloed from the rest of medicine, and decisions are made on the basis of the best comparative health outcome and cost. HTA processes across Australia should be established and aligned to ensure that the best available evidence informs robust decisions about the subsidised use of technologies. Any broader reviews of HTA may consider the ability to best serve the clinical journey of a consumer with a suspected or diagnosed genomic-related condition.

**3.2 Develop partnerships, funding and data sharing approaches for genomics that promote access to safe, efficient and cost-effective services.**

Increased investment in genomics is occurring both nationally and internationally, as it is recognised as a pathway to improving public health, managing demand on health systems and driving economic development. Australia has the opportunity to maximise existing linkages (both nationally and internationally) to develop partnerships, funding and data sharing approaches that promote access to safe, effective and cost-effective services. New models of funding that best reflect community needs should also be developed.

**3.3 Develop a national research agenda for genomics and identify opportunities to link to Commonwealth and state/territory research priorities.**

One of the foremost issues in genomic research policy is allocation of funds and research prioritisation. While there are currently multiple sources of funding for genomics research, including the Commonwealth, states/territories and the private sector, there is no coordination and alignment of national investment in genomics (beyond research alliances that have evolved nationally and at a jurisdictional level) and few public/private partnerships.

The development of a national research agenda for genomics would ensure investment is complementary, and opportunities for cross collaboration are maximised.

**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.**

Many genomic advances are occurring outside of the public sector, given that the for-profit private industry has a strong commercial incentive to develop new drug targets, new tests and new technologies. Governments must improve their understanding of the role the private sector fulfils, with collaboration and engagement being ongoing objectives. This will support the development and application of genomics through maximising funding and encouraging innovation and competition.

**3.5 Collaborate across governments and stakeholders to maximise investments and reduce duplication of resources and efforts.**

As a Federation, Australia needs to acknowledge the respective responsibilities of all jurisdictions. It also needs to work together to drive national value through financial sustainability of the applications of genomic knowledge, while also supporting affordability for vulnerable populations. The importance of non-government stakeholders must also be recognised, with collaboration and engagement being an ongoing objective.

### 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.*

The principles for safety and quality in genomics are generally the same as those that apply to other areas of health care, namely that care is driven by information, organised for safety and used responsibly. However, genomics can reveal information that can be predictive of future health, which also may have implications for family members. It is important this Framework identifies priority areas for action where a nationally coordinated approach to improvement can lead to high-quality and safe genomics in health care for all Australians.

**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.***

Acknowledging the range of national, state and professional guidelines, regulations and standards that exist to promote safe and quality genetic and genomic applications, the Framework presents a timely opportunity to review and build on previous efforts. It is important that guidelines, regulations and standards are driven by information and informed by:

* up-to-date knowledge and evidence to guide decisions about care
* safety and quality data, which is regularly collected, analysed and fed back to ensure continuous quality improvement
* the lived experience of patients, carers, family members and others
* agreed national ethical approaches.

It is important to undertake a comprehensive review of existing legislation, regulatory and other safety and quality levers. This review should assess the effectiveness of current levers and identify opportunities to further promote safety and quality, through identifying gaps and future needs particularly given the rapidly evolving advances in genomics in health care.

**4.2 Strengthen processes to identify, promote, monitor and report best practice in clinical genomics, including sharing of data and information.**

The rapid advances in genomics in health care is challenging Australia’s capacity to establish best-practice approaches that promote safe and high-quality genomics testing and services for Australians. It is becoming increasingly important to share information and data, and generate evidence to identify, promote, monitor and report best practice in clinical genomics. There is an opportunity to strengthen processes nationally to support best-practice approaches that can be translated into guidelines, regulations and standards   
as appropriate.

**4.3 Maximise genomics research opportunities that aim to resolve clinical uncertainty and improve quality and safety.**

While our understanding of the human genome has increased substantially in recent years, translating these findings into the clinical practice to truly transform patient care remains difficult. Time and further research is required to effectively embed genomics into clinical practice. It is also important to understand that other determinants of health, such as socioeconomic, environmental and lifestyle factors also have a role in the development of disease.

Dealing with uncertainty is a key component of genomics, as it is with other areas of health care. However, the ease with which data can be generated in genomics can increase uncertainty because correlations that appear significant may not have a basis that is biologically plausible.

While randomised clinical trials remain the gold standard for evaluating evidence in genomics, there is an emerging trend for evidence to be gathered through other study designs. This is due to the small size of target populations, which makes recruitment for clinical trials difficult and assessment of results more problematic. As conventional research translation pathways may no longer apply, there is a need to consider new methods for assessing and promoting safety, quality and efficacy.

Furthermore, given the substantial yet fragmented, investment in genomics research across Australia, it is timely to consider the future priorities for genomics investment.

### 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.*

**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.***

It is critical that activities to support genomic data are guided centrally to ensure national coordination, and that the available resources are used efficiently to give precedence to actions that provide the most value for the Australian public. Appropriate governance also offers the opportunity to leverage international efforts, and align and strengthen Australia’s access to genomic knowledge.

* + - Governance arrangements will also help formally explore options for developing infrastructure critical for delivering and supporting national consistency for data collection, storage and sharing.
    - Governance should also play a primary role in building and maintaining public trust through the development of best practice standards and reflecting ethical, legal and social standards.

**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.**

It is imperative that the benefits genomics may offer be available to all Australians regardless of ethnic diversity or geography. The value of genomic data to health care requires collection and sharing of standardised genotypic and phenotypic data. This will offer clinicians the ability to compare variants, and researchers the opportunity to investigate areas of new discovery that could benefit patients in the short term and consumers of health care more broadly.

**5.3 Develop nationally agreed standards for data collection, safe storage, data sharing, custodianship, analysis, reporting and privacy requirements.**

The development of nationally agreed standards for the collection, storage, sharing, custodianship, analysis, and reporting of genomic data will provide lasting impacts for clinicians and researchers. The benefits will also better support consumers in the delivery of high-quality, safe and appropriate health care, and build trust with the community.

**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.**

Private industry plays a valuable role in furthering research into genomic knowledge. Industry’s capability to serve this function is dependent on accessing genomic data for research purposes. The broad availability of data underlying all research studies (public and private) optimises data sharing and the potential for translation to improved health care. Public and private sectors often work in partnership to deliver on advances in genomics in 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.**

For genomics to realise its potential, sector engagement both nationally and internationally will be crucial as it will fuel research into genomic knowledge that can benefit health care through scientific and technological advancements. Investments in secure and interoperable computational infrastructure will offer access to robust data and lead to an increase in genomic knowledge and its application.

## 

## 5. Adopting a National Framework

Improving the Australian health system is an iterative process that involves identifying the problem, formulating policy options and setting a direction, implementation of the policy direction and ongoing review and evaluation (see Figure 5), in consultation with stakeholders.

### 5.1 Implementation

Consistent with the process outlined above, this Framework sets out the policy direction and identifies priority areas for action. However, implementation planning will be a separate process that will require further consideration by governments around priorities and funding. An Implementation Plan will be developed specifying activities and responsibilities.

As a first step to support implementation, AHMAC is commissioning a national genetic and genomic testing and activity stocktake during 2017–18. This will provide a baseline for future ongoing monitoring and reporting of progress against the Framework priorities. This, together with a comprehensive gap analysis, needs assessment and stakeholder consultation will inform further advice to governments.

### 5.2 Monitoring, review and evaluation

The Implementation Plan will be a key tool in monitoring progress and measuring the success. Ongoing monitoring and review must be undertaken to support a flexible and agile approach to implementation given the rapid advances in genomics.

An evaluation strategy will also need to be developed to systematically assess the appropriateness, effectiveness and efficiency of the Framework and implementation, and increase public accountability through reporting on key performance indicators.

### 5.3 Governance

Strong leadership and governance at all levels has been identified as a key enabler of success for achieving improvements in all priority areas of the Framework, particularly given the challenge of Australia’s federated health system. The Framework sets out how the Commonwealth and states and territories will work collaboratively with key partners and stakeholders to integrate genomics into health care over time.

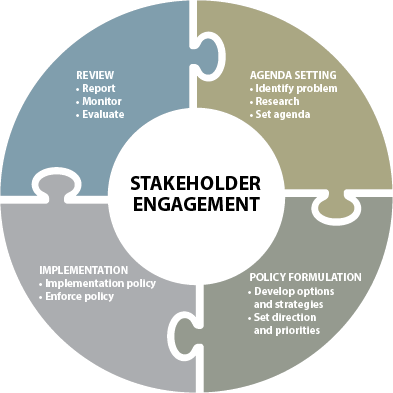


Figure 5: The policy cycle

[adapted from the WHO Health in all policies training manual, 2015]

Implementation planning will be overseen by AHMAC governance arrangements in 2017-18 and will be informed by appropriate stakeholder consultation. However, there may also be scope for a formal mechanism to engage and seek advice from genomic clinical specialists/researchers, genetic pathologists, bioethicists and consumer group representatives.

Strong leadership and governance arrangements will be required in the longer term for ongoing monitoring, review and evaluation of the Framework, including accountability and transparency of implementation activities.

### 5.4 What success looks like

Embedding this Framework over time is expected to lead to the mainstreaming of genomics in the Australian health system, with the health benefits of genomic knowledge being harnessed in an efficient, effective, ethical and equitable way to improve individual and population health. Achieving this will have different impacts on different stakeholders as outlined in the table below.

|  |  |
| --- | --- |
| **Stakeholder** | **Success is when …** |
| Consumer | individuals with a suspected genetic disorder are diagnosed and treated in a timely and appropriate way and their experience in navigating the health system is appropriately coordinated and supported. Consumers can make informed decisions about their health care and have confidence in the management of their personal information and data. |
| Provider (public or private) | there is confidence in the provider’s skills and genomics literacy when providing appropriate genomics services to consumers with an identified need. Providers have access to resources to make clinical recommendations and request referrals based on the latest evidence and support consumers and their families in voluntary informed consent process. Consumers have a clear clinical pathway, where providers work as a multidisciplinary team to achieve the best outcomes for individuals and their families based on individual needs and circumstances. |
| Researcher | there is a strategic approach to investment in genomic research that promotes collaboration, and leverages national and international initiatives including data standardisation and interoperability of genomics and phenotypic data platforms. Consumers are provided with a choice to share their genomic information to contribute to medical research and translation of genomics into clinical practice. |
| Private industry | innovation in genomics is fostered leading to long-term economic productivity and growth. |
| Governments | genomics is part of routine clinical care, where appropriate, and services are efficient, cost effective, ethical and equitable. Individual and population health is improved through building on person-centred care, financial sustainability, safety and quality, workforce planning and data management. |

## APPENDIX A: Source List

If you are interested in further reading about genomics, some suggested sources include:

1. Australian Commission on Safety and Quality in Health Care (2016). Patient and Consumer Centred Care | Safety and Quality. Available at: <https://www.safetyandquality.gov.au/our-work/patient-and%20-consumer-centred%20care/>

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5. Department of Health & Human Services (2016). Genomic health Care for Victoria—A Discussion Paper.

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10. Health Policy Advisory Committee on Technology q1 (August 2014). Massively Parallel Sequencing—A discussion paper.   
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16. National Human Genome Research Institute (NHGRI) (2016). ELSI Research Priorities. Available at: <https://www.genome.gov/27543732/elsi-research-priorities/>

- National Human Genome Research Institute (NHGRI) (2016). Regulation of Genetic Tests. Available at: <https://www.genome.gov/10002335/regulation-of-genetic-tests/>

17. National Pathology Accreditation Advisory Council (NPAAC) (2017). Guidelines and standards. Available at: <http://www.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-publication.htm>

18. Newson AJ, Schonstein L. (2016). Genomic testing in the paediatric population: ethical considerations in light of recent policy statements. Molecular Diagnosis and Therapy, 20 (5):407–14.

19. Newsroom | Inserm (2016). Presentation of the French Plan for Genomic Medicine 2025. Available at: <http://presse.inserm.fr/en/presentation-of-the-french-plan-for-genomic-medicine-2025/24328/>

20. Northern Territory Department of Health. Northern Territory Health Aboriginal Cultural Security Framework 2016–2026. Available at: <http://digitallibrary.health.nt.gov.au/prodjspui/handle/10137/730>

21. NSW Centre for Genetics Education. Available at: <http://www.genetics.edu.au/publications-and-resources/facts-sheets>

22. Nuffield Council on Bioethics (2015). The collection, linking and use of data in biomedical research and health care: Ethical issues. London.

23. Office of the Australian Information Commissioner (2014). Australian Privacy Principles guidelines Privacy Act 1988.

24. PHG Foundation (2007). The evaluation of clinical validity and clinical utility of genetic tests: Summary of an expert workshop 26 and 27 June 2006. National Genetics Reference Laboratory Manchester.

- Principles for the translation of ‘omics’-based tests from discovery to health care.

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## APPENDIX B: Examples of Commonwealth Legislation Relevant to Genomics in Health Care

| **Topic** | **Legislation** | **Relevance** |
| --- | --- | --- |
| Gene editing/cloning | *Prohibition of Human Cloning for Reproduction Act 2002*—administered by the National Health and Medical Research Council  *Research Involving Human Embryos Act 2002*—administered by the National Health and Medical Research Council | This legislation prohibits or limits the application of genomic knowledge to the cloning of humans and research involving human embryos. |
| Regulation of genetic/genomic tests for therapeutic use\* | *Therapeutic Goods Act 1989*—administered by the Therapeutic Goods Administration (TGA) | Genetic/genomic tests that are used for a clinical purpose need to be approved by the TGA for supply in Australia, based on evidence of their safety, quality and performance. TGA does not assess cost-effectiveness. |
| Health Records | *My Health Records Act 2012* —administered by the Commonwealth Department  of Health  Health Records Act—state and territory health records legislation | The privacy of health information, including genomic information, handled by private and public sectors is regulated under relevant state-based legislation.  The My Health Record system is the Australian government’s digital health record system. It contains My Health Records that are online summaries of an individual’s health information, which may include (or link to) genomic information in the future. The My Health Records Act limits when and how health information included in a record can be collected, used and disclosed. Unauthorised collection, use or disclosure of this information is both a breach of the Act and an interference with privacy. |
| Privacy | Privacy Act 1988—administered  by the Commonwealth  Attorney-General’s Department.  Some states and territories regulate privacy through legislation, others use administration regimes. | Sharing of genomic information must comply with privacy regimes. |

\*Defined as preventing, diagnosing, curing or alleviating a disease, ailment, defect or injury/influencing inhibiting or modifying a physiological process/testing the susceptibility of persons to a disease or ailment/influencing, controlling or preventing conception/testing for pregnancy, including goods that are used to replace or modify a part of the human body.

|  |  |  |
| --- | --- | --- |
| Insurance | *Health Insurance Act 1973* —administered by the Commonwealth Department  of Health  Life Insurance Act 1945 | Health insurers are prohibited from charging a premium based on a person’s state of health or history of claiming.  Members of the Financial Services Council (FSC) (the industry association) are required to comply with Standard No 11 (Genetic Testing Policy) which covers genetic testing and life, disability and trauma insurance. However, membership of FSC is voluntary. |
| Discrimination | Australia has anti-discrimination legislation at the federal level as well as legislation in all states and territories  (Eg, Cth Disability Discrimination Act 1992) | Discrimination concerns may act as disincentives to testing and research participation and have negative consequences for individual and public health outcomes. |
| Intellectual Property | *Patents Act 1990*—administered by IP Australia | A patent is a right that is granted for any device, substance, method or process that is new, inventive and useful. Australian patent rights are legally enforceable and give the owner exclusive rights to commercially exploit the invention for a period of up to 20 years. The way in which the Patents Act is interpreted determines what constitutes an invention and whether it is patentable. While naturally occurring genes are not patentable, patents in relation to biotechnology are important to promote innovation. |

1. For a list of the most up-to-date NHMRC guidelines see [www.nhmrc.gov.au/guidelines](http://www.nhmrc.gov.au/guidelines). [↑](#footnote-ref-1)
2. MSAC provides advice to the Commonwealth Minister for Health about the strength of evidence relating to the comparative safety, effectiveness and cost effectiveness of new and emerging medical services and technologies, as well as under what circumstances public funding, including listing on the MBS, should be supported. [↑](#footnote-ref-2)
3. Department of Human Services, <http://medicarestatistics.humanservices.gov.au/statistics/mbs_item.jsp> (data provided on 25 May 2017). [↑](#footnote-ref-3)