Companion Document to the draft National Health Genomics Policy Framework

Purpose
This document provides context for the draft National Health Genomics Policy Framework (the Framework) by:

- describing how genomics will be used to inform and guide health care;
- clarifying why a Framework is needed;
- outlining the scope of the Framework;
- describing current funding arrangements in Australia for genetic/genomic testing; and
- (when available) summarising the results of consultation in relation to the Framework.

Background
The genome is the complete set of genetic material in a cell or an organism. Consequently, genomics is the study of the entire DNA sequence and how the different elements of the sequence interact with one another¹.

Whereas genetics tests usually correlate mutations 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. While genetic tests are suitable for diagnosing conditions that are based on heritable changes to single genes (e.g. Huntington disease), they are rarely appropriate for studying complex multifactorial diseases (e.g. Type II diabetes), which result from interactions between many genes, lifestyle and environmental factors.

While genetic testing is the focus of current efforts, genomics research is developing rapidly and in time genomic tests will be available to guide health care. The scope of the Framework includes the use of genetics, genomics and other types of “omics”² in human healthcare, especially in relation to testing and emerging techniques to manipulate the genome (such as gene therapy). For simplicity, the Framework uses the term “genomics” to refer broadly to the use of the above technologies in healthcare.

Some of the more common applications of genomics are often referred to as precision or personalised medicine, or stratified medicine. All of these terms form part of an emerging medical

¹ Genomics refers to a person’s entire genetic code whereas genetics considers the impact of each gene individually.
² Other types of “Omics” include transcriptomics (all the RNA molecules in cell or organism), proteomics (all the proteins in a cell or organism) and metabolomics (all the metabolites in a cell or organism).
discipline that involves using an individual’s genomic information to inform their clinical care (e.g. for diagnostic or therapeutic decision-making). This approach is commonly referred to as ‘genomic medicine’.

Genomics is one of the fastest transforming areas of human health. The question is not whether genomics will change healthcare but how quickly it will change it and how the health system can best respond.

**How will genomics inform and guide healthcare?**
The potential of genomic medicine to improve health care is considerable; it already means that in some instances we can diagnose diseases and quickly tailor treatments to reflect a person’s wider genetic make-up and assess their predisposition to disease, potentially prompting more focused clinical monitoring and lifestyle changes.

Two areas where genomics is impacting health care are in cancer prevention and treatment and in diagnosing rare diseases. In relation to cancer, genomic information can enable heritable risks to be identified, it can allow the most suitable treatment to be determined and it can be used to predict how, or if, an individual will respond to a drug and to determine dosage (known as precision medicine). In relation to rare diseases, genomics is being used to identify gene mutations that are responsible for each rare disease and, in some cases, to determine if there are drug options available.

Precision medicine may extend into many areas of clinical practice; for example, in the prescribing of drugs such as antidepressants, analgesics and anticoagulants. Better targeting of existing drug therapy avoids wasteful, risky, and sometimes unnecessary, treatments and can provide savings for the health care system and the economy at large.

Clinical applications of genomics can be classified as being primarily for prediction, diagnosis or treatment as outlined in the following diagram.
Current uses of genomic testing to inform healthcare include\(^3\)

- **Screening** – testing of a group in the population (usually defined by age) to identify the possible presence of an as-yet-undiagnosed disease in individuals without signs or symptoms. Examples include the Newborn Screening program which operates in each state and territory, carrier screening which can identify individuals who do not have any symptoms but carry mutations for conditions or diseases that are inherited recessively (e.g. cystic fibrosis) and screening of individuals with a family history of developing a condition (e.g. mutations to the genes BRCA1 and or BRCA2 can increase susceptibility to developing heritable cancers, such as breast cancer).

- **Preimplantation genetic diagnosis (PGD)** – used in conjunction with in vitro fertilisation (IVF) to select embryos without specific known gene mutations prior to implantation.

\(^3\) Note that the examples given are genetic tests. However for the purposes of the Framework and Companion Document, all are regarded as being within the scope of genomic testing.
- Prenatal testing – can be used during pregnancy to genetically confirm a condition in the fetus that may have been indicated through ultrasound.

- Diagnostic testing – diagnosis of an individual with symptoms of a genetic condition or disease.

- Somatic cell testing – testing of DNA changes in the non-germline cells of an individual (e.g. to test for mutations in cells that have led to cancer). Because the mutations tested for are not inherited or passed onto future generations, somatic cell testing does not have implications for family members.

- Pharmacogenomics – using genomic testing to predict responses to, or side effects from, particular drugs (also called precision medicine). For example, HIV/AIDS patients with a certain gene type known as HLA-b*5701 may react badly to the antiretroviral medication, abacavir.

- Tissue typing – using genetic testing to predict which cells, tissues or organs, including stem cells, may be used in transplants to minimise the risk of rejection.

- Gene therapy – transfer of genetic material (i.e. DNA or RNA) into the somatic cells of an individual, usually to produce a therapeutic effect.

Over the last 30 years there has been considerable research into mutations in single genes associated with disease, but only a few diseases are linked to single genes. The focus is now on identifying changes in sequences of multiple genes (genomics) that may be associated with complex diseases like obesity, but this makes interpretation, counselling and decision making more difficult and requires large amounts of data and access to significant computing power.

While genomics has tremendous potential in health care, a significant challenge for the healthcare system and society is how to minimising the potential harms, which include data misuse, discrimination and stigmatisation.

Why a Framework – and associated policies – are needed

The World Health Organization (WHO) defines health policy as:

Decisions, plans, and actions that are undertaken to achieve specific health care goals within a society. An explicit health policy can achieve several things: it defines a vision for the future which in turn helps to establish targets and points of reference for the short and medium term.

Australia’s health system is characterised by a complex interaction of public, private and non-government organisations providing services that may be funded by the Commonwealth, the states or territories, and through private payments by individuals. This complexity can lead to health service funding and delivery occurring in an uncoordinated and poorly integrated fashion, creating service
fragmentation and gaps, inequitable access, unnecessary duplication of research effort and potentially less than optimal outcomes for the public. In this context it is critical that coordinated action is taken at the Commonwealth, state/territory and local levels to enable a more harmonised approach to integrating genomics into the health system through national policy.

WHO also notes that:

*Policies, strategies and plans are not ends in themselves. They are part of the larger process that aims to align country priorities with the real health needs of the population, generate buy-in across government, health and development partners, civil society and the private sector, and make better use of all available resources for health – so that all people in all places have access to quality health care and live longer, healthier lives as a result.*

While genomics has the potential to offer significant benefits for the future of medicine and public health, from a policy point of view, there are many things that need to be done to facilitate translating genomics into improvements in human health. Decisions about genomics policy involve many complex issues about ethics, costs, benefits, individual interests (privacy), and societal interests. Areas like financing of genomic research, including patenting and intellectual property issues, equitable access and affordability of genomic information and services, evaluating the utility of genomic testing, raising the level of public awareness on genomics, as well as maintaining ethical standards in genomics also need to be considered carefully by policy makers.

The expanding role of genomics in medicine and health requires not only a national effort but also an international effort to create sound frameworks from which to build and further the research and applications of genomics. Policy makers have a significant role to play in guiding the investment in genomic research and development to target the specific health needs of their communities.

Against this background, the Health Policy Advisory Committee on Technology recommended in 2014\(^4\) the urgent need for a “national coordinated approach to genomic policy development across jurisdictional boundaries”. In early 2016, the Australian Health Ministers’ Advisory Council (AHMAC) agreed the Commonwealth would lead a project, in consultation with states and territories, to develop the Framework.

\(^4\) Massive Parallel Sequencing – A discussion Paper. August 2014. Health Policy Advisory Committee on Technology
Scope of the Framework

The scope of the Framework includes all aspects of human genomic medicine that are legal under Australian law, including genetic/genomic testing, other types of “omic” testing and various recombinant DNA technologies, including gene therapy. While gene therapy has been used in research for several decades with limited success, recent advances in gene editing (e.g. the CRISPR-Cas 9 gene editing) mean that it may one day be possible to cure or prevent genetic diseases such as muscular dystrophy, sickle-cell anemia and certain cancers. For instance, many cancers develop the ability to switch off immune cells that are trying to attack them; with gene editing technology it may be possible to modify a patient’s immune cells to turn them against their cancer.

Given the rate with which new advances in genomics research and development are occurring, the scope of the Framework is likely to change over time. This will require ongoing evaluation of the relevant scientific, ethical, policy environment that applies to genomic medicine.

Funding arrangements for genetic/genomic tests

Funding of genetic/genomic tests in Australia is affected by a number of factors, including the nature of the disorder, the request pathways through which tests are ordered, potential participation in research projects, and where the individual lives. Private health insurance generally only provides subsidies for genetic tests that are required as part of treatment when patients are privately admitted into hospital, and only if the Medicare Benefits Schedule (MBS) also provides a rebate for the test.

Medicare currently funds fewer than 30 genetic tests in Australia; two examples are tests to detect hereditary hemochromatosis and Fragile X syndrome. While the range of test funded by Medicare is relatively small, a number of these tests tend to be ordered in high volume. In fact, it is estimated that around 20 to 30 per cent of all genetic tests are funded through Medicare and the remainder funded by state and territory health departments\(^5\). If medical practitioners wish to have a new test listed in the MBS, they must apply to the Medical Services Advisory Committee (MSAC). MSAC provides advice to the Commonwealth Minister for Health about the strength of evidence relating to the safety, effectiveness and cost effectiveness of new and emerging medical services and technologies and under what circumstances public funding, including listing on the MBS, should be supported.

Most genomic tests are provided by specialist state-based laboratories and funded on a state by state (or territory) basis. For that reason some states are developing their own strategies, including processes to better inform the ordering of clinically appropriate genomic tests, which will also guide

the development and planning of genomic-related services. This can result in inequities in access and different outcomes for individuals, in particular if they have a rare disease⁶. The community bears the inherent costs associated with fragmentation, lack of coordination and inconsistent outcomes.

What is the Framework
The Framework 2017-2020 is a whole-of-governments policy aiming to identify priority areas for national action to ensure that the benefits of genomics are harnessed to optimise health outcomes in a cost effective way. It represents the highest level of strategic planning undertaken by Commonwealth, state and territory departments of health. All other jurisdictional specific genomic plans should reflect, and support, the implementation of the National Health Genomics Policy Framework. Examples of the range of key genomic initiatives currently being funded by the Commonwealth and states/territories are listed in Appendix A.

The National Health Genomics Policy Framework 2017-2020:
- identifies the main priorities areas for action and opportunities to work towards a vision of the future
- provides a direction for collective action by Governments and guidance for the non-public sector, and
- promotes leadership across the Commonwealth and all state/territory Governments agencies to institute a 'whole-of-government' approach to policy and planning, research, service delivery and evaluation.

It is important to note that while AHMAC is the custodian of the Framework, it will not be solely responsible for delivering all of the potential activities involved in implementing the Framework. The Framework identifies the pressing need to establish overarching governance arrangements to guide the development of concrete plans of action to align with the Framework.

Other partners such as research institutions, non-government organisations and consumer groups are also likely to have an important role in contributing to implementing the Framework. Health service organisations and healthcare leaders have a role in planning how they will incorporate genomics into healthcare delivery, where clinical utility⁷ and cost effectiveness have been demonstrated.

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⁶ It is estimated that 80% of rare diseases are genetic in origin (https://globalgenes.org/rare-diseases-facts-statistics/)
⁷ Clinical utility refers to the ability of the test to enable a diagnosis to be made (or the best course of treatment to be determined).
Australian Genomics Health Alliance
The National Health and Medical Research Council (NHMRC) announced in 2015 a $25 million commitment to support integration of genomic medicine into healthcare across Australia. The Targeted Call for Research into Preparing Australia for the Genomics Revolution in Health Care grant—the second largest in NHMRC history—will fund the Australian Genomics Health Alliance (AGHA), a national network of more than 50 partner organisations including research institutes, hospitals and universities.

ARC Centre of Excellence in Bioinformatics (ACB)
ACB is an interdisciplinary centre that brings together researchers in genomic and post-genomic bioinformatics, cell and structural biology, mathematics, statistics, computer science, information technology, and systems analysis. Headquartered at the Institute for Molecular Bioscience at the University of Queensland, partner institutions are:

- Macquarie University (Sydney)
- The University of Newcastle (Newcastle)
- The University of Queensland (Brisbane)

Canberra Clinical Genomics (CCG)
The Australian Capital Territory (ACT) Government has provided $7.3 million to establish Canberra Clinical Genomics (CCG). CCG is a partnership the ANU and ACT Health which aims to realise the diagnostic potential of genomics by identifying patients with complex diseases, sequencing their genomes and finding treatments that are personalised to their condition.

Melbourne Genomics Health Alliance
The Victorian Government provided $25 million funding over 4 years from 2015 to enable greater access to genomic medicine for Victoria’s hospital patients. The funding for the Melbourne Genomics Health Alliance brings together clinical, research and teaching strengths among Victoria’s leading hospitals and research organisations to allow children and adults to receive early diagnoses of their conditions, enabling them to receive treatment earlier and improve their health outcomes.

Queensland Research Consortium
A research consortium, coordinated by The University of Queensland and consisting of Berghofer QIMR, the CSIRO, the Queensland University of Technology and Queensland Health, is to receive $25 million from 2015 to 2020 to increase engagement between diagnostic services and genomics research centres and create a clinically driven and patient-focused program of research to ensure Queensland is part of a national diagnostic network. The Queensland Centre for Medical Genomics (QCMG) has been established with funding from the NHMRC, the Queensland State Government and the University of Queensland.

Sydney Genomics Collaborative
The Sydney Genomics Collaborative is a $24 million, four-year investment by the New South Wales (NSW) State Government in using genetic technologies to improve patient outcomes. The Collaborative Program comprises: Medical Genome Reference Bank; NSW Genomics Collaborative Grants; and Genomic Cancer Medicine Program.

\[8\] Note that there will be an opportunity to add to and finalise this list following public consultation.