This Framework provides a consistent, national and strategic view for integrating genomics into the Australian health system, and identifies key priorities areas for action to address a range of genomics policy issues and challenges.

Please Note:

Questions 1 to 6 for the National Health Genomics Policy Framework are to be answered through the online submission process.
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Glossary of key terms

For the purposes of the Framework, key terms are defined as follows:

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<tr>
<th>Key Term</th>
<th>Definition</th>
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</thead>
<tbody>
<tr>
<td>Analytical validity</td>
<td>refers to how well the test predicts the presence or absence of a particular DNA sequence, which could be a gene or a genetic change.</td>
</tr>
<tr>
<td>Bioinformatician</td>
<td>person who uses data algorithms and specialised software to analyse biological data, such as DNA or RNA sequences.</td>
</tr>
<tr>
<td>Bioinformatics</td>
<td>the use of algorithms and software to analyse biological data.</td>
</tr>
<tr>
<td>Clinical geneticist</td>
<td>physicians who have undergone speciality training in genetics after general professional training (such as paediatrics and oncology) and see referred patients for diagnosis, management, genetic testing and genetic counselling.</td>
</tr>
<tr>
<td>Clinical genetics</td>
<td>the medical specialty which provides a diagnostic service and &quot;genetic counselling&quot; for individuals or families with, or at risk of, conditions which may have a genetic basis.</td>
</tr>
<tr>
<td>Clinical utility</td>
<td>the usefulness of the genomic test to the patient (i.e. to what extent does it influence the effectiveness of the proposed intervention or clinical decision making).</td>
</tr>
<tr>
<td>Clinical validity</td>
<td>how well the genetic variant being analysed is related to the presence, absence, or risk of a specific disease.</td>
</tr>
<tr>
<td>Big Data</td>
<td>the use of large data sets that may be analysed to reveal patterns, trends, and associations, especially relating to complex interactions.</td>
</tr>
<tr>
<td>DNA</td>
<td>deoxyribonucleic acid, a self-replicating material which is present in nearly all living organisms as the main constituent of chromosomes. It is the carrier of genetic information.</td>
</tr>
<tr>
<td>Efficiency</td>
<td>a measure of whether healthcare resources are being used to get the best value for money. Includes technical, productive and allocative efficiency.</td>
</tr>
<tr>
<td>Embryo</td>
<td>an unborn or unhatched offspring in the process of development, in particular a human offspring during the period from approximately the second to the eighth week after fertilisation (after which it is usually termed a foetus).</td>
</tr>
<tr>
<td>Exome</td>
<td>part of the genome formed by exons, the sequences which, when transcribed remain within the mature RNA after introns are removed by RNA splicing.</td>
</tr>
<tr>
<td>Key Term</td>
<td>Definition</td>
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</tr>
<tr>
<td>Gene</td>
<td>the basic physical and functional unit of heredity. Genes, which are made up of DNA, act as instructions to make molecules called proteins.</td>
</tr>
<tr>
<td>Genetics</td>
<td>the study of genes, genetic variation, and heredity in living organisms.</td>
</tr>
<tr>
<td>Genetic counsellor</td>
<td>healthcare professionals who have undergone speciality training to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions.</td>
</tr>
<tr>
<td>Genetic pathologist</td>
<td>pathologists who have undergone speciality training in genetics and genomics who provide specialist input to genomic tests including test selection, results and interpretation to aid in the diagnosis, management and treatment of patients with a genetic basis for their disease.</td>
</tr>
<tr>
<td>Genetic test</td>
<td>type of medical test that identifies changes in chromosomes, genes, or proteins.</td>
</tr>
<tr>
<td>Gene therapy (or editing) of germline cells</td>
<td>is when DNA is manipulated (inserted or deleted) from reproductive cells, eggs or sperm, in the body. Changes can then passed be down from generation to generation (e.g. preventing the inheritance of a disease trait). Germline therapy is prohibited in Australia under legislation.</td>
</tr>
<tr>
<td>Gene therapy (or editing) of somatic cells</td>
<td>is when the DNA of any human cells, except germline cells, are manipulated (inserted or deleted) to treat disease. These changes are not passed down from generation to generation.</td>
</tr>
<tr>
<td>Genome</td>
<td>the complete set of genetic information in an organism.</td>
</tr>
<tr>
<td>Genomics</td>
<td>the application of genome-based knowledge through the study of genes and other genetic information, their functions and inter-relationships for the benefit of human health.</td>
</tr>
<tr>
<td>Genomic data</td>
<td>refers to data produced from DNA sequencing of a genome. It can be compared with a reference genome.</td>
</tr>
<tr>
<td>Genomic knowledge</td>
<td>includes information about the interpretation of genomic data and the implications of these findings, as well as relevant non-genomic clinical information.</td>
</tr>
<tr>
<td>Genomic medicine</td>
<td>is an emerging medical discipline that involves using genomic information about an individual as part of their clinical care (e.g., for diagnostic or therapeutic decision-making) and the health outcomes and policy implications of that clinical use (also used interchangeably with precision medicine, personalised medicine, stratified medicine).</td>
</tr>
<tr>
<td>Genomic services</td>
<td>whole genome sequencing and analysis available for research, screening, and diagnostic purposes.</td>
</tr>
<tr>
<td>Key Term</td>
<td>Definition</td>
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<tr>
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<tr>
<td>Genomic testing</td>
<td>involves the analysis of hundreds or even thousands of genes from a cell or tissue simultaneously using sophisticated computer-based algorithms.</td>
</tr>
<tr>
<td>Germline cell</td>
<td>the reproductive cells in multicellular organisms.</td>
</tr>
<tr>
<td>Governance</td>
<td>the structures and processes by which the health system is regulated, directed and controlled. It includes the obligations of stewardship ensuring that the system is well sustained for the future as well as serving the needs of the present.</td>
</tr>
<tr>
<td>Health Technology Assessment</td>
<td>a range of processes and mechanisms that use scientific evidence to assess the quality, safety, efficacy, clinical effectiveness and cost effectiveness of health services.</td>
</tr>
<tr>
<td>Introns</td>
<td>sections of the DNA sequence of genes that are not translated into protein. The sections of DNA (or RNA) that code for proteins are called exons.</td>
</tr>
<tr>
<td>Metadata</td>
<td>a set of data that describes and gives information about other data.</td>
</tr>
<tr>
<td>‘omics</td>
<td>suffix that refers to the analysis of all the molecules of one type in a cell or tissue. For example, genomics (investigation of all the DNA molecules in a cell), transcriptomics (all RNA molecules), proteomics (all proteins).</td>
</tr>
<tr>
<td>Personalised medicine</td>
<td>stratifying cohorts of patients by subclass of disease or the likelihood of responding to a particular therapy, intervention, or disease management strategy. The term ‘stratified medicine’ reflects the realistic effects of medicines at population level, while the term ‘personalised medicine’ reflects the possibly overambitious promise of individualised unique drug targeting and development. (also see stratified medicine, precision medicine, genomic medicine)</td>
</tr>
<tr>
<td>Person centred care</td>
<td>an approach involves health care practitioners working in partnership with a patient and their family/carer to manage their health. In the context of genomics and the familial nature of the information generated, privacy and ethical considerations of the individual are paramount but need to be balanced with the right of the family to know clinically relevant information.</td>
</tr>
<tr>
<td>Pharmacogenetics</td>
<td>the study of how the actions of, and reactions to, medicines vary with the patient's genes.</td>
</tr>
<tr>
<td>Precision Medicine</td>
<td>stratifying cohorts of patients by subclass of disease or the likelihood of responding to a particular therapy, intervention, or disease management strategy. The term ‘stratified medicine’ reflects the realistic effects of medicines at population level, while the term ‘personalised medicine’ reflects the possibly overambitious promise of individualised unique drug targeting and development. (also see personalised medicine, genomic medicine, stratified medicine)</td>
</tr>
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<td>Key Term</td>
<td>Definition</td>
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</tr>
<tr>
<td>RNA</td>
<td>ribonucleic acid, a nucleic acid present in all living cells. Its principal role is to act as a messenger carrying instructions from DNA to initiate and control the synthesis of proteins, although in some viruses RNA rather than DNA carries the genetic information.</td>
</tr>
<tr>
<td>Somatic cell</td>
<td>derived from the Greek word soma, meaning “body”. Hence, all body cells of an organism – apart from the sperm and egg cells, the cells from which they arise (gametocytes) and undifferentiated stem cells – are somatic cells. Examples of somatic cells are cells of internal organs, skin, bones, blood and connective tissues. In comparison, the somatic cells contain a full set of chromosomes whereas the reproductive cells contain only half.</td>
</tr>
<tr>
<td>Splicing</td>
<td>process by which the DNA of an organism is cut and a gene, perhaps from another organism, is inserted.</td>
</tr>
<tr>
<td>Stratified medicine</td>
<td>stratifying cohorts of patients by subclass of disease or the likelihood of responding to a particular therapy, intervention, or disease management strategy. The term ‘stratified medicine’ reflects the realistic effects of medicines at population level, while the term ‘personalised medicine’ reflects the possibly overambitious promise of individualised unique drug targeting and development. (also see precision medicine, personalised medicine, genomic medicine)</td>
</tr>
<tr>
<td>Transcription</td>
<td>the first step of gene expression, in which a particular segment of DNA is copied into RNA by the enzyme RNA polymerase.</td>
</tr>
<tr>
<td>Transcriptome</td>
<td>the sum total of all the messenger RNA molecules expressed from the genes of an organism.</td>
</tr>
<tr>
<td>Transcriptomics</td>
<td>the study of transcriptomes and their functions.</td>
</tr>
<tr>
<td>Whole exome sequencing</td>
<td>a laboratory technique for sequencing all the known protein-coding regions of DNA in an organism’s genome (known as the exome).</td>
</tr>
<tr>
<td>Whole genome sequencing</td>
<td>a laboratory process to determine the complete DNA sequence of an organism’s genome.</td>
</tr>
</tbody>
</table>

**Questions:**

7. Are there other key terms referenced in the Framework which should be added to the glossary? If so, please provide details.

8. Are the definitions easy to understand? Do any definitions require amendment? If yes, please provide details.
## Acronyms

<table>
<thead>
<tr>
<th>Acronym</th>
<th>Expanded Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>AGHA</td>
<td>Australian Genomics Health Alliance</td>
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<tr>
<td>AHMAC</td>
<td>Australian Health Ministers’ Advisory Council</td>
</tr>
<tr>
<td>ARTG</td>
<td>Australian Register of Therapeutic Goods</td>
</tr>
<tr>
<td>DNA</td>
<td>Deoxyribonucleic acid</td>
</tr>
<tr>
<td>ELSI</td>
<td>Ethical, legal and social issues</td>
</tr>
<tr>
<td>GA4GH</td>
<td>Global Alliance for Genomics and Health</td>
</tr>
<tr>
<td>HPC</td>
<td>Hospitals Principal Committee</td>
</tr>
<tr>
<td>IVD</td>
<td>In Vitro Diagnostic</td>
</tr>
<tr>
<td>MBS</td>
<td>Medicare Benefits Schedule</td>
</tr>
<tr>
<td>NATA</td>
<td>National Association of Testing Authorities</td>
</tr>
<tr>
<td>NHMRC</td>
<td>National Health and Medical Research Council</td>
</tr>
<tr>
<td>OECD</td>
<td>Organisation for Economic Co-operation and Development</td>
</tr>
<tr>
<td>PBS</td>
<td>Pharmaceutical Benefits Scheme</td>
</tr>
<tr>
<td>RNA</td>
<td>Ribonucleic acid</td>
</tr>
<tr>
<td>TGA</td>
<td>Therapeutic Goods Administration</td>
</tr>
<tr>
<td>WHO</td>
<td>World Health Organization</td>
</tr>
</tbody>
</table>
**PREAMBLE**

**AHMAC and Jurisdictional Working Group**

The Australian Health Ministers’ Advisory Council (AHMAC) brings together Chief Executive Officers of Commonwealth, state and territory and New Zealand departments that have responsibility for health. In March 2016, AHMAC agreed that a whole-of-governments National Health Genomics Policy Framework was required to capitalise on emerging genomic knowledge by better integrating genomics into the Australian health system. The Framework places a strong focus on those policy issues that will benefit from collaboration across all jurisdictions.

The Framework is being developed through AHMAC’s Hospitals Principal Committee (HPC) with input from a Jurisdictional Working Group. Membership of the Working Group comprises a Commonwealth Chair and members from each jurisdiction.

**Purpose**

This Framework is to provide a consistent, national and strategic view for integrating genomics into the Australian health system, and identify genomics policy issues and challenges that need to be addressed. Developing a whole-of-governments and system-focussed Framework, with a person-centred approach to outcomes, is necessary to ensure consistency across Australia.

The Framework will support better coordination and consistency of action across the health system to ensure the potential benefits of genomics are harnessed in an efficient, effective, ethical and equitable way. The Framework will assist in addressing the potential for genomics to contribute to improved patient care, improved population health and containment of healthcare costs in Australia’s health system.

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

**Scope**

The scope of the Framework is intended to be flexible so that new genomic advances can be included over time. Initial priorities are medical and healthcare applications which are informed by, or based on, human genetic/genomic testing (including single gene tests, panel tests and tests based on sequencing exomes or whole genomes). These applications include those used to:

- test for the purpose of diagnosing and monitoring disease;
- treat diseases, including through understanding the genetic variation between underlying differential responses to medicines and how this can be applied to prevent adverse drug reactions and improve healthcare (i.e. pharmacogenetics and stratified medicine); or
- prevent disease including carrier testing and building predictive models based on genomic information as a tool for primary disease prevention.

Future priorities may include:

- other ‘omics technologies that are time and tissue dependent, e.g. transcriptomics or proteomics; and
- gene therapy.
Beyond the human genome and health

While the immediate focus of this Framework is the application of knowledge about the human genome to advance medicine and healthcare, the potential application of genomics is much wider.

There are potential synergies between this Framework and other national strategies and frameworks. Most notably, the Communicable Disease Control Framework has some cross-over as microbial genomic sequencing informs public health surveillance and investigations of communicable disease. In this context, the Public Health Laboratory Network expert advisory group on whole genome sequencing suggests that: Microbial whole genome sequencing (WGS) has the capacity to revolutionise the characterisation of pathogens in clinical and public health laboratories. There are also linkages with the Newborn Bloodspot Screening Framework, the Australian Population Based Screening Framework and the National Maternity Services Framework which may consider the application of genomic sequencing in the context of specific sub-populations.

Other current areas of research include:

- animal and plant therapy to improve population health outcomes as animal/plant genomics can involve modifying animals to prevent the spread of diseases, resist diseases, and improve the nutritional value of food therefore preventing illness; and
- industrial biotechnology (e.g. developing biofuels).

More broadly, there is also the potential for genomics to be applied to sports performance, forensics, employment, defence, anthropology, and national security.

Genomic applications which currently have more limited relevance to population health will continue to be monitored, including the potential development of related policy frameworks.

Audience

The Framework is directed at decision and policy makers at national, state and health service levels. While primarily a tool to provide guidance for the development and implementation of genomic-related policies, strategies, actions and services, the Framework may also be a useful resource for the non-government sector, stakeholder organisations, industry and communities.

Timeframe

The timeframe of the Framework is 3 years, with a review anticipated in 2019 to inform the next iteration.

Questions:

9. Does the preamble provide a sufficient overview of the Framework? If not, please provide further details.

10. Are there linkages with other key frameworks or strategies that should be explicitly referred to in the Preamble? If yes, please provide details.

11. Is a three year timeframe sufficient for the Framework? Please explain your answer.

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2 The Office of the Gene Technology Regulator administers the gene technology regulatory system and has specific responsibility to protect the health and safety of people, and to protect the environment, by identifying risk posed by or as a result of gene technology, and by managing those risks through regulating certain dealings with genetically modified organisms (GMOs).
EXECUTIVE SUMMARY

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

SCOPE
Genomic knowledge (not limited to the human genome) used to inform, and transform; testing; treatment; and prevention of disease to improve human health.

PRIORITY AREAS
1. Strong leadership and governance
   - Support the appropriate use of genomics by applying knowledge of the ethical, social and legal challenges and opportunities into sound national policies and practices.

2. Skilled and genomic literate health workforce
   - Skilled and genomic literate health workforce

3. Appropriate application of genomic knowledge is evidence based, high quality and safe

4. Integration of genomic knowledge into person-centred health care, supported by equity of access to services

5. Sustainable investment in health genomics

6. Effective and appropriate collection, management and utilisation of genomic data

THE FUTURE
1. National oversight supports consistency, coordinated implementation of agreed priority activities, allowing for monitoring review and evaluation.

2. An adequately resourced, well-trained and certified/accredited genomics workforce that delivers best practice clinical genomic services to individuals and families.

3. Genomic knowledge is evidence-based, high quality, safe and is applied responsibly.

4. Individuals are genomic literate and empowered to make informed decisions about their health care, with equity of access in choice.

5. Genomic research and clinical service delivery is cost-effective, ensuring individuals can access the right genomic tests and treatments at the right time.

6. Genomic data and knowledge is protected and data is accessible between jurisdictions and sharing is safe and efficient.
STRAIGHT CONTEXT

In recent decades, the Australian health system has been transforming from one that is responsible for providing episodic care for those with chronic and complex conditions to a more proactive system that addresses the health needs of the population. One of the key strategies for achieving this transformation is through leveraging the opportunities available to the health system by integrating genomics into health care. Critical to this will be “big data” with whole-genome sequencing and communication technologies driving our health system to the cusp of an information-age health system. Although there is huge potential for genomics and big data to improve the health of all Australians, there are also a wide range of challenges, including policy, regulatory, funding and ethical issues that will need to be addressed.

The application of genomic knowledge has the potential to have a major impact on health care in Australia. The Commonwealth and state and territory governments face a major policy challenge in how to respond quickly to integrate genomics appropriately into Australia’s health system, particularly given the public demand already demonstrated. This will likely require new thinking, new approaches and strengthened national cooperation and leadership.

The potential of genomic medicine for Australia’s health system is considerable; it already means that we are able to diagnose diseases and detect variants far more precisely and to quickly tailor treatments to reflect a person’s wider genetic make-up and better identify those at high risk genetically of inherited disease and a range of common chronic conditions.3

Genomics offers considerable potential, but it is important to evaluate the advances of a genomics revolution with pragmatism. While technical advances in sequencing genomes facilitate research, ‘they do not in themselves change patient outcomes’.4 Personalised health care requires the ‘judicious integration of genetic data within existing models of patient care that incorporate individual factors such as age and intercurrent illness’.5 In 2011, the US National Human Genome Research Institute stated that although ‘genomics has already begun to improve diagnostics and treatments in a few circumstances, profound improvements in the effectiveness of healthcare cannot realistically be expected for many years’.6 The challenge for Australia is to convert genetic information and knowledge into effective and sustainable clinical practice to improve individual and population health outcomes.

The Framework is designed to drive national effort on agreed priorities to address the lack of national coordination of genomic activities across Australia. The Framework provides an opportunity to act on divergent approaches to implementing genomics before they form impediments to successful integration of genomics into healthcare nationally. A key enabler for consistency will be the establishment of collaborative governance arrangements that can facilitate actions to address the changing nature of genomics. The development of support structures will allow for coordination across jurisdictional and private sector priorities. Currently, the 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. As a Federation we need to acknowledge the respective responsibilities of all jurisdictions while working together to address gaps in activity which will further drive national value. Integrating genomics into Australia’s health system is a key strategy to ensure that the genomics advances improve health outcomes for all Australians.

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INTERNATIONAL DEVELOPMENTS

- Internationally, there is a considerable range of genomics-related activities being undertaken by national agencies, non-government organisations and international governmental organisations. These activities provide opportunities for partnerships, developing common best practices and clinical guidelines, and leveraging learnings, however, they also potentially represent competitors to Australian industry.
  - In May 2004, the World Health Assembly resolution on Genomics and World Health urged Member States:
    - to frame national genomic policies and strategies, and to set up mechanisms for assessing relevant technologies, cost-effectiveness, ethical review structures, legal, social and economic implications, regulatory systems particularly with regard to safety, and the need for public awareness; and
    - to strengthen existing, or establish new, centres and institutions engaged in genomics research with a view to strengthening national capacity and accelerating the ethical application of the advances in genomics relevant to countries’ health problems.
  - The Global Alliance for Genomics and Health (GA4GH) comprises 427 organisational members from 41 countries who are working together to create a common framework of harmonised approaches to enable the responsible, voluntary, and secure sharing of genomic and clinical data.
  - The Global Genomic Medicine Collaborative (G2MC) seeks to improve global health by fostering the implementation of genomic tools and knowledge into health care delivery worldwide.
  - The UK Government’s 100,000 Genome Project is a key element of, and represents a strategic approach in, the UK Government’s efforts to integrate genomics into the National Health System (NHS).
  - The US Precision Medicine Initiative (launched by President Obama in 2015) aims to accelerate biomedical discoveries and provide clinicians with new tools, knowledge, and therapies to select which treatments will work best for which patients.
  - The US National Cancer Institute’s, Cancer Moonshot Initiative. Including the related Genomic Data Commons and Cancer Genomics Cloud programs.
  - The Organisation for Economic Co-operation and Development (OECD) has a number of guidelines developed guidelines that are relevant to genetics/genomics, including for Human Biobanks and Genetic Research Data Bases, Quality Assurance in Molecular Genetic Testing, and Licensing of Health Care Genetics.

- Countries who have or are developing National Genomics Frameworks include:
  - Canada
  - France
  - United Kingdom

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7 Genomemcanada.ca. (2016). Genome Canada ]. [online] Available at: http://www.genomemcanada.ca/
NATIONAL DEVELOPMENTS

- Jurisdictions are heavily investing in genomics clinical practice and research, including:
  - a $25 million commitment by the Commonwealth Government through the National Health and Medical Research Council’s Targeted Call for Research into Preparing Australia for the Genomics Revolution in Health Care;
  - a $25 million commitment by the Queensland Government, reflected in the recent establishment of Queensland Genomics Health Alliance;
  - a $25 million commitment by the Victorian Government, reflected in the Melbourne Genomics Health Alliance established in 2015;
  - a $24 million commitment by the New South Wales Government in 2014; and
  - a $7 million commitment by the Australian Capital Territory Government.

- Some jurisdictions are developing their own genomics related policy frameworks (New South Wales, Victoria and Queensland).

- South Australia had the first accredited facility for use of massively parallel sequencing in a clinical genetic service and Western Australia has a cohesive genomics pipeline implemented under clinical genetic services, the Rare and Undiagnosed Diseases Diagnostic Pipeline.

- Stakeholder groups, such as the Australian Genomics Health Alliance (AGHA), Melbourne and Queensland Genomics Health Alliances, are important collaborators in building the evidence to:
  - demonstrate the maximised potential of patient benefits through the application of genomic data;
  - inform the analysis on the cost effectiveness of genomic based applications; and
  - build Australia’s research and translation capacity in genomics and healthcare.

Questions:

12. Are the most critical international and national activities referenced? If no, please provide details of what may be included and why it is important.

13. Does the Strategic Context provide a clear case for improved national consistency in genomics policy? Please explain your answer.

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AN AUSTRALIAN PERSPECTIVE – the issues

Governments have a responsibility to ensure genomic technologies are used appropriately and to best effect. The move to person-centred care will mean patients are empowered to ask for, and use, access to information about themselves. This has significant implications for electronic health records, biobanking, 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 risks and benefits of direct to consumer testing.

Four key issues are identified below that are being, and will continue to be, addressed as genomic science evolves. These issues can be interdependent; for example, it is difficult to determine the capability and capacity needs of a health system when the impact of genomics on delivery of healthcare and medicine and individual and population health outcomes is not fully understood.

(i) Genomics challenges social norms and clinical practices for health and wellbeing, illness and injury, medicine and healthcare.

Genomics is recognised as a disruptive technology that will reshape clinical practice. It has the potential to change healthcare delivery particularly through improved diagnosis and therapy. Genomics will fundamentally change the way we improve health and prevent, diagnose and treat illness, requiring a redesign of the Australian health system. This will include transforming the health workforce to provide high quality and safe genomic services. Genomics may have significant impacts for social norms and values, including how society perceives illness, health and well-being in relation to human individual, as well as enhancement versus medicine. 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. The introduction of genomic medicine may have an impact on the financial sustainability of the health system. There will also be broader clinical, ethical, legal, cultural and social implications, including the potential for discrimination based on an individual’s genomic information.

(ii) Genomics could lead to disproportionate outcomes for individuals and populations.

Genomics is expected to have a transformative impact on personal and population health and wellbeing. However, the widening gap between the rapid progress in genome sequencing and the comparatively slow progress in the functional characterisation of specific genetic variants and novel mutations, and elucidation of their clinical utility requires management. In short, genomics is a known unknown, the full potential of which is not yet understood. Furthermore, the inequity of access to genomics testing and services is resulting in varied individual and population outcomes, especially for the remote locations in Australia, and must be addressed.

(iii) Lack of national coordination of genomic activities across Australia.

Activity is already occurring at both the national and state level to enable genomics, but divergent approaches are starting to appear. Some areas of the health system are underprepared for the technology advances and implementation is considered too ad hoc across Australia. There is also an uncoordinated effort in establishing capacity and infrastructure needed to support integration of genomic technology into the national health system (workforce, research, education, data security and sharing, cost-effectiveness and clinical utility).
(iv) Australia’s health system does not currently have the capacity or capability to fully integrate genomics.

Australia has lagged behind other countries in developing national genomic policies, regulations, and capacity building since the genome was first sequenced in 2003. The health system capability to integrate genomics and harness its benefits efficiently and effectively is a major concern for governments.

**System capability and capacity issues:**

Genomics has presented Australia with an urgent need to consider system capability and capacity mechanisms for its integration into healthcare, due to the far-reaching impacts and flow-on effects of the swiftly advancing technology improvements. One of the most pressing underlying issues is the management of genomic data, including collection, security, quality, sharing, privacy and custodianship. A related but crucial challenge is to relate aggregated sequence data to phenotype data collected from medical records and routine diagnostic tests. Other key themes emerging from previous genomic related work/reviews include:

- inequitable access to genomic services due to geographical, cultural and socioeconomic factors;
- supply and demand: unmet need in clinical settings is a significant driver; technology suppliers are also driving testing; consumer demand is impacting on limited resources; and the widening gap between the supply of available workforce and demand on clinical genomics specialists continues to grow;
- poor genomic and genetic literacy of consumers and the health workforce, which hinders informed decision-making;
- uncertainty about specific indications for genomic testing (including consideration of the clinical utility, costs, benefits and risks of genomic sequencing);
- validity of genomic data analysis;
- direct-to-consumer genetic/genomic testing;
- research investment; and
- absence of a comprehensive analysis of our current human clinical genomics activities (baseline) in Australia, which means we cannot evaluate current practice, nor determine the true cost of clinical genomics.

**Question:**

14. Are there additional barriers, issues or challenges to integrating genomics into the health system that should be included in this section? If yes, please provide details.
A NATIONAL HEALTH GENOMICS POLICY FRAMEWORK FOR THE NEXT 3 YEARS

A Whole-of-System based approach

- Future-proofing the health system to ensure we continue to achieve better health outcomes requires a whole-of-system approach with a clear vision and roadmap to achieve this, including how any proposed reforms will complement and build upon each other to achieve our overall vision for the health system.

- The World Health Organization (WHO) has developed a framework for strengthening health systems to improve health outcomes, using a whole-of-system approach. This Framework builds upon the WHO systems approach\(^\text{10}\), which considers how changes in one part of the system (e.g. medicines and technology domain) will impact on the rest of the health system (governance, financing, medicines and technologies, information technology, workforce/human resources).

Principles

The development of the Framework was supported by the following guiding principles:

i. **National** – developed jointly by Commonwealth and state/territory governments (i.e. whole-of-governments) with the Framework facilitating national coordination and government priority setting and decision making.

ii. **High-level, strategic framework** – identification of themes, principles (including bioethical principles) and considerations for embedding consistency and national coordination as enablers for more efficient, equitable and effective utilisation of genomics. It will be overarching and, as such, will align with existing, or to be developed, regulation, guidelines and discussion papers addressing specific genomic issues.

iii. **System-focussed** – an understanding of what the system can deliver for the patient and consideration of how a change within one system domain (i.e. leadership and governance; system financing; human resources (workforce); information systems; medicines and technologies; and service delivery) will impact, interact, and change the other domains and affect the system as a whole.

iv. **Person-centred care** – an approach which involves health care practitioners working in partnership with the patient and their family/carer to manage their health. In the context of genomics and the familial nature of the information generated, privacy and ethical considerations of the individual are paramount but need to be balanced with the right of the family to know clinically relevant information.

v. **Evidence-based clinical policy and evidence-informed public policy** – ensuring that the best available research and information is used to guide decisions at all stages of policy processes.

vi. **Flexible to keep up with scientific advances** – will contain flexibility to enable it to be adapted to reflect the evolving nature of genomics technology and take into account the latest scientific findings and advances and potential shifts in genomics policy issues and challenges.

vii. **Identify priority areas** – gives consideration to prioritising the genomics policy issues and challenges that need to be addressed (including clinical utility, and ethical and social issues) and identifies directions for change/opportunities for action and areas that require further work.

Enablers

- Willing coalition of Commonwealth and state/territory governments
- Partnerships:
  - engaged professional bodies;
  - genomic academics;
  - professional colleges and universities (to upskill future cohorts); and
  - engaged community, including patient representative groups
    - improved genomic literacy
    - public acceptance and valuing of genomics.
- Effective regulatory levers to enable the potential benefit of genomics, whilst minimising the potential risk to individuals and society:
  - NHMRC ethical guidelines and framework for genomic research;
  - NATA accreditation system for diagnostic laboratories;
  - national approach to evaluation of applications for Medicare Benefits Schedule (MBS)/Pharmaceutical Benefits Scheme (PBS) funding of genomic applications;
  - national approach to assessment of quality, safety and efficacy of genomic based medicines and diagnostic tests (as provided for in the Therapeutic Goods Act 1989);
  - review of any legislation which may support or align with the principles; and
  - uniform or consistent clinical pathways.
- Commonwealth and state/territory funded genomic research programs
- State/territory strategies for genomic services
- Clinical genomics capacity – sequencing / functional analysis
- National disease-specific networks / clinical registers

Strategic Intent

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

It is important that the Framework aligns with the longer term vision for an integrated health system that effectively improves health care for individuals and populations.
Strategic Priority Areas

A range of priorities has been identified for future genomics activities which reflect the WHO domains for system change. These priorities are areas where we can make the most difference to maximise the health benefits of genomics for all Australians. The priority areas are not necessarily discrete and there will be inter-relationships and inter-dependencies. For example ethical, social and legal considerations have been identified as an over-arching priority given that these aspects of genomics should be addressed across all areas of action.

These priorities are about:

- embedding ethical, social and legal considerations in all activities;
- establishing governance arrangements to take the lead on matters of national significance;
- building a multi-disciplinary competent genomics health workforce to leverage existing resources and support good clinical and ethical practice;
- growing and applying genomic knowledge that is evidence-based, high quality and safe;
- putting the individual at the centre of health genomics and supporting equity of access;
- making sure that our investment in health genomics is cost-effective and sustainable into the future; and
- managing genomic data in a way which realises the benefits of genomics for all, while protecting individuals.

Questions:

15. Are the key guiding principles appropriate? Please explain your answer.

16. Are there additional enablers that should be included? If yes, please provide details.

17. Is the Strategic Intent of the Framework appropriate? If no, what would you suggest?

18. Are the priority areas appropriate? Please explain why or why not.
OVERARCHING PRIORITY: Ethical, social and legal (regulatory) issues

Ethical, Legal and Social Issues (ELSI) Context

Realising the expected benefits of integrating genomics into the health system will depend, in significant part, on the confidence placed by the public in the information and services available. The Framework is an important step towards building assurance that appropriate safeguards exist to support individual and population level engagement in the system. The Framework needs to be sufficiently flexible to consider emerging societal issues and the broad spectrum of community views, in particular around the use of data.

The Nuffield Council on Bioethics suggests that:

*Decisions and actions informed by the use of data in research and health care may have both beneficial and harmful effects on individuals or on broader groups of people including families, companies, social groups, communities or society in general. Different people may value these potential benefits and harms very differently – what may be profoundly troubling for one person might be a matter of indifference to another.*

Engaging with patient representative groups in developing this Framework, and the actions to implement it, is a key component to successfully integrating genomic knowledge into clinical care within an ethical framework.

Ethical Principles

Most ethical considerations raised by genomics already apply generally to health care. However, the application of genomic knowledge has its own particular complex and unique 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 reference genomics data from many individuals. In considering these challenges, the following ethical principles/concepts are proposed to inform discussion of ethical dilemmas arising in the application of genomics the Australian health care system.

- **Respect for persons**, includes respecting autonomy, which values the right of the individual to self-determination. In the context of genomics, respect for persons, 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. Furthermore, consent must be current and specific and the individual must have the capacity to understand and communicate their consent.
  - **Privacy and confidentiality**: individuals have a right to protection of data and for confidentiality to be maintained.

- **Do no harm (non-maleficence)**: the interests and welfare of the individual should have priority, including respect for human rights.

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• **Act in the best interest of the patient (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:** understands that, in certain circumstances, common interests are served by sharing genetic information for mutual benefit.

• **Protection of vulnerable individuals and communities.**

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.

**Genetic Information**

• Genetic information about an individual is considered to be sensitive information under the *Cth Privacy Act 1988* (Privacy Act).

• The Privacy Act contains thirteen Australian Privacy Principles (APPs) which outline how Australian Government agencies, private sector organisations with an annual turnover of more than $3 million, all private health service providers and some small businesses must handle, use and manage personal information.

**Consent**

• Consent, particularly informed consent, is a multifaceted issue for clinical research and practice. Genomic research can include third party access to a person’s genetic data, and secondary uses for their data.

• Where genetic data is stored and shared, the opportunity for participants to revoke their consent or opt out of particular uses of their data is often difficult.

• Incidental discoveries also have implications for consent and release of results.

• Complex issues associated with consent, proprietary and other rights related to pregnant women, newborn children and foetuses, people with disabilities (including mental disability and dementia) and deceased persons also require resolution.
Incidental findings

- Incidental genetic discoveries, which are found when testing for other things and may be clinically relevant or of uncertain relevance.
- Incidental findings pose challenges to the rights of patients and obligations of health providers, particularly with regard to disclosure to family members of results that could have implications for them.  
- Dual-Use dilemma in research - the results of well-intentioned scientific research can be used for both good and harmful purposes. There has been growing debate about the dual-use nature of life science research, including genomics.

Use of data

- Protecting individuals or populations from discrimination resulting from genetic testing, particularly with regard to employment, health insurance and life insurance.
- Sensitivity - cultural and ethnic identification. Members of ethnic groups may face stigmatisation if the group is found to have a genetic disposition to particular diseases.
- Risk of being able to re-identify individuals, in particular if data is released for small populations/sample sizes.
- The benefits of the use of genomic data should be equitable.

Genetic data

- Ethical, legal and social issues (ELSI) relating to data include:
  - the collection, storage and sharing of genetic data, including the legal and governance requirements for repositories of genomic information;
  - ownership of genetic data;
  - patient access to their data;
  - third party access to an individual’s genetic information and the right ‘not to know’;
  - linking of data;
  - technologies such as cloud computing; and
  - privacy.

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13 Note – Section 95AA of the Privacy Act authorises the Information Commissioner to approve guidelines (the s95AA Guidelines), issued by the NHMRC, which specify the requirements that must be met by health practitioners, in the private sector, if they choose to use or disclose genetic information without patient consent. See National Health and Medical Research Council & Office of the Privacy Commissioner, (2009). Use and disclosure of genetic information to a patient’s genetic relatives under Section 95AA of the Privacy Act 1988 (Cth) - Guidelines for health practitioners in the private sector.

Social issues

- Genomics carries a number of ethical and social issues beyond health care. Issues such as the possibility of genetic determinism, reductionism, essentialism, and exceptionalism have been discussed in relation to public attitudes and in public policy. For example, as individuals increasingly learn critical details about their personal genome and the implications of that information, societal understanding, free-will and individual responsibility may change.¹⁵
- Societal acceptability of genomics may also be influenced by religious and cultural beliefs.
- The genomic literacy of the public will also impact the acceptance of genomics within society.

Research

- Consideration will have to be given to the blurring of lines between research and clinical genomics, particularly with regard to the roles and responsibilities of research ethics committees and clinical ethics committees - issues around scope of consent, ownership of data, feedback to participants on outcomes of research etc.
- NHMRC produces a range of resources on genomics and human health and related issues. For example, assisted reproductive technology including preimplantation genetic diagnosis, biobanks, use of human embryos, clinical ethics and human research, Guidelines under Section 95 of the Privacy Act (s 95 Guidelines) and Guidelines approved under Section 95A of the Privacy Act (s 95A Guidelines).¹⁶
  - The s95 Guidelines and s95A Guidelines provide a framework for Human Research Ethics Committees to approve researchers’ proposals to collect, use or disclose identified information without consent.
- Research ethics - while the Australian Health Ethics Committee (AHEC) is the Australian representative body at the World Health Organization’s global Summits of National Bioethics Advisory Bodies, its accountability is limited to the NHMRC.

Utility

- Utility requires that genomics provides the greatest possible benefit across a population. It is therefore necessary for genomics interventions to be scientifically sound and to have clinical utility.
- The clinical utility of genomics tests can take a number of years to assess and this raises issues in relation to the balance between regulation and innovation.

**Regulation – current**


- Therapeutic Goods Act 1989 – In vitro diagnostic (IVD) medical device framework for diagnostic tests that are either commercially supplied to laboratories or are developed “in-house” (also known as in-house IVD medical devices or laboratory-developed tests) are regulated by the TGA. This includes genomic tests that are associated with TGA approval of a medicine and are used to define suitable patient groups for treatment (often referred to as companion diagnostics). There is limited regulation by TGA if it is part of a clinical trial.

- Laboratories that develop in-house genomic tests are required by the TGA to be accredited by the National Association of Testing Authorities (NATA) and comply with the National Pathology Accreditation Advisory Council (NPAAC) standard, Requirements for the development and use of in-house in vitro diagnostic medical devices, which sets out requirements for validation of these tests and includes the need to establish clinical validity and utility of the test. Laboratories are required to hold this data and notify the TGA of the in-house IVD medical devices being used. Laboratories that develop higher risk IVD medical devices (Class 4 IVDs) to diagnose serious diseases are required to apply to TGA for inclusion in ARTG. Exemptions can be requested for specific situations – Special Access Scheme, Emergency Exemption.

- To access Medicare (Health Insurance Act) - Royal College of Pathologists of Australasia run accreditation scheme. NPAAC– develop standards and provide education. NATA accredit pathology laboratories – requirement that they participate in quality assurance program.

  The Australian Law Reform Commission produced a report in 2004 ‘Essentially Yours: The Protection of Human Genetic Information in Australia’ which considered legislative arrangements and put forward a range of recommendations.

**Direct to consumer tests**

- Available through the internet. Risks to consumers if test intended for health purposes (e.g. indicate predisposition to disease) – analytical validity, clinical validity and clinical utility.

- Currently, there is no mechanism to integrate this information into a patient’s health record (similar issue with doctor-initiated testing).

- Consumers request clinicians (GPs and clinical geneticists) to explain/interpret results of direct to consumer tests, which then impacts on broader access and budgets. Consumers need health genomic literacy and counselling to understand implications of test results.

- Direct to consumer genetic tests which are intended for health related use are prohibited for supply in Australia but there is no authority to extend this prohibition to tests ordered from overseas. [Potential for lack of regulatory oversight of testing performed in overseas laboratories]. US Food and Drug Administration has taken regulatory action to ensure US based genetic tests for specific markers to inform health care are approved by FDA (23andme).

- NHMRC has prepared a statement on use of direct to consumer tests.\(^{18}\)

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What are opportunities for improvement?

- Identification of priority areas for national positions on specific issues, including the adequacy of current protection for personal genomic information.

- To further examine and develop legislation, regulations that provide for optimal use of genomic knowledge to improve health, while protecting consumers from misuse of genomic information.

- Addressing emerging regulatory challenges:
  
  o Regulating a diagnostic test based on rapid genomic sequencing of large segments of an individual’s genome compared to a commercial test "kit," (for example a panel test which examines genetic markers for a particular disease or condition).

    - 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 as 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 IVD test.

  o There is increasing complexity in the way genetic testing services are being delivered. Components of the service may be outsourced to third parties (e.g., sequence analysis and/or result interpretation services). Potentially, there may be components of a testing service that would not be subject to regulation by the TGA or require NATA accreditation.

  o If a laboratory uses commercially supplied genetic tests (that are included in the ARTG) and is not receiving Medicare reimbursements then there would be no requirement for them to obtain NATA accreditation.

  o Need to review the current regulatory options for direct to consumer tests and consider whether additional controls (both regulatory and non-regulatory) are required (noting that a small number of tests are performed overseas as the technology is unavailable in Australia¹⁹).

  o Appropriately qualified and experienced evaluators needed to manage the pace of technological change.

  o Appropriate processes to facilitate efficient drug approval processes, to enable timely access to new drugs which are developed through specific targets identified through genomics technology, while maintaining safety and efficacy standards. (Important where a new drug may be effective across several indicators).

  o Secure and trusted processes to control the use and access to population-based data regarding outcomes for treatments based on genomics.

What does the future look like?

- Ethical, social and legal issues will be considered for all genomic clinical, research and policy activities; ethical principles are used as a basis for resolving ethical dilemmas.

- Options for creating nationally consistent ethical and legal arrangements for genomic clinical, research and policy activities will be explored.

- Regulation is sufficiently flexible and adaptable to keep pace with advances in genomic technology while also providing adequate oversight and protection.

- Information/data used to improve care and patient experience.

Questions:

19. Is the placement of ethical, social and legal (regulatory) issues as an overarching priority appropriate?

20. Should these issues be considered prior to the following six priority areas, or after?

21. Are there any other broad ethical, legal or social issues that should be addressed under this priority? If yes, please provide details.
PRIORITY AREA 1: Strong leadership and governance

What is the current situation?

- Key challenge lies in our federated system where responsibility of genomics policy issues is split between the Australian Government and state and territory governments.
- Some jurisdictions are moving towards state-wide genomics strategies and plans.
- No national coordination of effort, consistency in approach, and prioritisation of genomics activity.

Why is strong leadership and governance a priority?

- Strong leadership and governance will be critical to achieve improvements in all priority areas of the Framework.
- Joint national leadership will:
  - establish a shared purpose and commitment to achieving the Framework intent;
  - drive future genomics policy directions;
  - ensure a whole-of-system view is taken, rather than the limited view usually determined by the Commonwealth or state and territories discrete roles and responsibilities within the health system; and
  - foster public confidence and trust through transparency, accountability and ethical oversight.
- To succeed, joint national leadership must be underpinned by strong governance arrangements at the state and territory, national and international levels.
  - Articulation of clear roles, responsibilities and performance expectations will enable accountability and transparency of the health system stewards.

What are the opportunities for improvement?

- National governance arrangements should:
  - facilitate a clear understanding and shared commitment to the integration of genomics into Australia’s health system;
  - provide effective strategic advice and guidance to Governments and other stakeholders on emerging genomics policy issues;
  - bring together Government and non-Government stakeholders to jointly;
    - articulate the agreed and complementary roles and responsibilities of Governments and other key partners in implementing actions to support the Framework goals and priorities
      - map impacts on the health system
      - embed continuous evaluations and improvements
    - establish and maintain key partnerships to ensure a coordinated effort at state and territory, national and international levels;
facilitate effective and coordinated collaboration with multilateral organisations, such as the World Health Organization, and other nations to ensure shared learnings and coordination of effort;

- oversee the implementation of the Framework, including the prioritisation and coordination of activity, and the establishment of performance measures and accountability expectations; and

- establish reporting mechanisms to periodically monitor, review and evaluate the Framework.

What does the future look like?

- The Framework is endorsed by, and implemented, across all jurisdictions with actions taken across all priority areas.

- National oversight that:
  - enables consistent and coordinated implementation of agreed priority activities across Australia; and
  - allows for periodic monitoring, review and evaluation; and accountability and transparency to support the Framework implementation.

- Coordination and consistency in genomics activities across Australia enabled by strong and visible leadership that provides nationally unified directions.

- More efficient prioritisation given the limited resources available, and less duplication of effort where unnecessary.

Question:

22. With regard to Priority Area 1 – Strong leadership and governance, is anything missing or what should change, for:

   (a) the current situation;

   (b) why is this important;

   (c) opportunities for improvement; and/or

   (d) what the future looks like?
PRIORITY AREA 2: A skilled and literate genomics workforce

What is the current situation?

- Demand for clinical genetic services exceeding current workforce capacity, has placed enormous pressure on the small and highly specialised clinical genomics workforce.

- AGHA preliminary mapping of the genomics workforce in Australia in 2015 identified the following:
  - very small clinical genetics/genomics workforce comprising approximately 62 qualified clinical geneticists, with a further 20 in training, and 18 qualified genetic pathologists, with a further 2-3 in training.
    - Australasian Society of Genetic Counsellors, a special interest group of the Human Genetics Society of Australasia, has 280 members representing accredited genetic counsellors and trainees, which provides an insight into the small size of the trained genetic counsellor workforce. However, anecdotally, it has been suggested that the number of people training each year exceeds available workforce positions.
    - Smaller jurisdictions such as Northern Territory and Tasmania face significant challenge in access to services and currently contract some of their specialist workforce from interstate.
  - Need for bioinformaticians to support research and translation of genomic information into routine clinical care.
  - Scope of practice – health genomics workforce does not have clear scope of practice and guidelines to support clinical decision making. Also there are no clear clinical pathways.
  - Low level of genetic/genomic literacy in health profession, particularly in the primary health care sector.
  - Limitations of the current genomics workforce are impacting on patient care: the Victorian Government estimates the waiting times for public genetic clinic consultations are currently up to nine months from initial referral.
  - Linkage with clinical guidelines - the absence of nationally agreed approaches to best practice (detailed in Priority Area 3), the rapidly evolving science translating to clinical practice in ad hoc way, and the complex clinical, ethical and social impacts of genomic information presents challenges to the clinical application genomics as well as to the broader health workforce.

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20 Information on a clinical genomics workforce is limited.
22 Defined in Glossary of key terms
Why is a skilled and literate genomic workforce a priority?

- Genomics requires a highly specialised workforce including clinical geneticists, genetic counsellors, genetic pathologists and clinical bioinformaticians, among others, who can understand the benefits and limitations of genomics findings in the context of health and disease.

- Multi-disciplinary teams, with specialities across diverse disciplines, to support the delivery of genomic services to consumers. The Human Genetic Society of Australasia suggests that the use of multidisciplinary teams also improves the efficiency and effectiveness of clinical genomics services.

What are the opportunities for improvement?

- Build on work underway by the AGHA to undertake a needs assessment to provide an evidence base for future policy directions.

- There is opportunity to improve the current situation, in collaboration with the relevant Royal Colleges, through:
  - nationally consistent certification and/or accreditation of the clinical genomic workforce;
  - collaborative efforts among health professionals, through partnerships and networks, to share information and implement nationally consistent evidence-based approaches in clinical genomics;
  - ensuring the health workforce understands the impact of genomic knowledge on their practice and effectively communicates this to patients and families;
  - promoting the integration of appropriate genomic education and training (including ethics) into specialist health professional education from undergraduate study through to continuing professional development;
  - ensuring that relevant health workforce strategies consider the capacity of, and access to, clinical genomic health professionals, including the opportunity to facilitate innovative delivery methods such as telemedicine/telehealth;
  - other healthcare providers, especially GPs and obstetricians, are more aware of the range of services available through the specialist genomic workforce and appropriate clinical pathways. Also there is the opportunity to integrate genomics into other medical specialties e.g. oncology, cardiology, etc;
  - workforce strategies and planning support capacity of the workforce and access to services. Essentially, the clinical genomics workforce is well funded and has the capacity to manage the growing demand for clinical genomic services; and
  - health professionals working together in multi-disciplinary teams to deliver safe and quality clinical genomic services to consumers.

- Workforce considerations outside health are also supported to better integrate the benefits genomics offers to healthcare. This should be inclusive of:
  - providers of undergraduate study and continued professional development;
  - health-economists specialising in genomics health; and
  - genomic-health legal expertise.

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What does the future look like?

- A well-trained and certified/accredited genomics workforce with clear roles and responsibilities built around core competencies to effectively support improved patient outcomes.
- A genomics workforce supported at early stages at an education and career pathway level that meets the needs of the increasing use of genomic services in the Australian health system.
- Increased genomic literacy and application of genomic knowledge in the health workforce, particularly in the primary health care sector.

Question:
23. With regard to Priority Area 2 – A skilled and literate genomics workforce, is anything missing or what should change, for:
   (a) the current situation;
   (b) why is this important;
   (c) opportunities for improvement; and/or
   (d) what the future looks like?
PRIORITY AREA 3: Application of genomic knowledge is evidence based, high quality and safe

What is the current situation?

- Widely recognised as a disruptive technology that will significantly reshape clinical practice in the future.
- Applications of genomic knowledge are currently being used in clinical practice in Australia, e.g. new-born screening to identify those at risk of rare, but serious medical conditions. Further, while pharmacogenomics is a relatively new application in Australia and only, certain indications are funded by the Commonwealth Government through the MBS\(^{26}\) and the PBS for the treatment of certain cancers and HIV.\(^{27}\)
- No nationally agreed clinical genomics approaches to ensure the application of genomic knowledge is evidence based, high quality safe and used responsibly (e.g. Genetics Services Standards Framework, developed in 2013 and reviewed in 2015 - as starting point).
- Lack of evidence based guidelines for genomic testing for specific indications, particularly in terms of clinical validity, clinical utility, costs, benefits and risks of testing (e.g. ACCE\(^{28}\) as a helpful process for evaluating genetic tests in respect of analytical validity, clinical validity, clinical utility and ethical, legal and social implications of a test).

Why is ensuring the application of genomic knowledge is evidence based, high quality and safe a priority?

- Risks of potential harm to patients, their families, health professionals and society.
- Effectiveness of genomic information in informing clinical practice must be evidence-based to avoid harm to patients, improve outcomes and ensure efficient use of scarce resources.

What are the opportunities for improvement?

For diagnostic-based genomic applications, evidence is needed in three domains: analytic validity (how well tests perform in the laboratory), clinical validity (how well do tests correlate with clinical endpoints), and clinical utility (whether the use of testing improves health outcomes).

There is also opportunity to:

- develop evidence-based genomic guidelines and standards for ethical, clinical and regulatory purposes;
- actively monitor, identify, and report evidence-based genomic clinical guidance domestically and internationally;
- build on current work to consider appropriate investment in, and distribution of, nationally agreed infrastructure to collect, share and evaluate clinical validity and clinical utility of genomic tests and treatments; and
- leverage research opportunities appropriately to resolve uncertainty in genomics.

\(^{26}\) Australian Government Department of Health (Operating from 01 April 2016) Medicare Benefits Schedule Book Category 6 Group 7 Genetics pg. 47; 91-94

\(^{27}\) Ibid. pg. 93, 94.

What does the future look like?

- The application of genomics knowledge is evidence based, and nationally consistent.
- Genomic knowledge is used safely, effectively and efficiently in clinical practice, and is supported by appropriate regulation.
- Health professionals have current knowledge of appropriate clinical pathways, and able to safely and effectively use genomics knowledge to inform and integrate with patient care, via established clinical pathways.

Question:

24. With regard to Priority Area 3 – Application of genomic knowledge is evidence based, high quality and safe, is anything missing or what should change, for:

(a) the current situation;
(b) why is this important;
(c) opportunities for improvement; and/or
(d) what the future looks like?
PRIORITY AREA 4: Integration of genomic knowledge into person-centred health care, supported by equity of access to services.

**What is the current situation?**

- Person-centred care - health care that is respectful of, and responsive to, the preferences, needs and values of patients and consumers.
- Low genomic literacy and understanding in the community and even lower genomic knowledge awareness.
- Health literacy - how people understand information about health and health care, and how they apply that information to their lives, using it to make decisions and act on it.
- Patient and their family need to be able to make informed decisions about their care and treatment.
- Equity of access to services is multi-dimensional: location, cost, availability, appropriateness (including cultural acceptability). Currently inconsistent across Australia.

**Why is person-centred care a priority?**

- Person-centred care recognises the importance of better understanding the patient experience and their needs within a complex and fragmented health system.
- Shared decision-making between patients and clinicians produces better health outcomes – for example, a person considering their options and fully understanding the potential implications of screening or diagnostic tests.
- Standard 2 of the National Safety and Quality in Healthcare Standards\(^{29}\) recognises the significance of partnering with consumers as a key indicator of safety and quality.
- Australia’s ability to fully integrate genomics into the health system, and maximise the benefits of genomics research and innovation, depends on achieving broad consumer recognition, acceptance, engagement, interaction and support.
  - This requires that there needs to be meaningful engagement between the community and the genomics sector and that there is an opportunity for participation and feedback.
- Equity of access is integral to person centred care – making sure that people have the right access to the right services and care when they need it.

**What are the opportunities for improvement?**

- Clinically appropriate integration of genomics into patient pathways and the patient experience.
- Develop community engagement strategies to better understand the application and impact of genomic advances on the public.

• Informed national discussion to take place, involving:
  o Empowering individuals to take responsibility for their health care and make informed decisions;
  o Engaging the community to build confidence and trust in clinical services provided, including discussions about ethical, social and legal issues;
  o Educating the public to manage consumer demand and expectations about unnecessary testing to avoid waste in the health system; and
  o Educating the public about the gap between clinical testing and treatment options – just because you can test doesn’t mean you should test.
• Investigate how genomics data can be integrated with electronic health records to improve coordination of care and seamless clinical pathways.
• Identify gaps and inconsistencies in equity of access to genomic services and opportunities to address them (e.g. nationally consistent processes to assess clinical and cost-effectiveness).
• Promote choice for genomic services which are culturally appropriate and consistent across Australia.
• Public awareness and understanding of genomics through availability of linguistically and culturally appropriate information resources.

What does the future look like?
• Improved patient health outcomes and satisfaction.
• Improved equity of access to services.
• Improved public genomic literacy leading to increased public acceptance of genomic knowledge and its function in the health system – community trust and confidence.
• Empowerment of individuals to take responsibility, and make informed decisions, for their health care.
• Genomic literacy informs consumer expectation of the benefits, risks and limitations of genomic knowledge.

Question:
25. With regard to Priority Area 4 – Integration of genomic knowledge into person-centred health care, supported by equity of access to services, is anything missing or what should change, for:
   (a) the current situation;
   (b) why is this important;
   (c) opportunities for improvement; and/or
   (d) what the future looks like?
PRIORITY AREA 5: Sustainable investment in health genomics

What is the current situation?

- Funding for tests – combination of:
  - state and territory programs - a mix of block-funding and activity based funding for public genetic clinics, clinical testing and genetic counselling services, and teaching, training and research activities;
  - Commonwealth Government funding - MBS and the PBS;
  - Patients pay full costs for genetic and genomic testing as private services; and
  - Services dispersed geographically - delivered predominantly through public health system.

- Funding for research.
  - Multiple sources – some intrastate, interstate and national alliances formed.
  - No coordinated national investment in research for genomics.

- Commonwealth and states/territories all make own decisions about what genomic services will be funded - no nationally agreed Health Technology Assessment (HTA).
  - HTA - what is it and why is it important (i.e. Governments cannot financially support every new health technology that comes onto the market, so aims to direct government funding, in the form of subsidies, to health technologies that are both clinically effective and cost effective).
  - Inconsistent processes – disparity in the provision of services can limit choice for patients.

Why is investment in genomics that is sustainable a priority?

- Health care expenditure is continuing to rise in Australia, as in many other countries.
- Consumer expectations of access to increasing sophisticated and more expensive treatments and technology.
- Commonwealth and state/territories governments are responsible for ensuring that level of spending can be sustained.
- One of key levers to support financial sustainability is to ensure that funded health care interventions are clinically effective and cost-effective (i.e. through HTA process).
- Inconsistent HTA processes also creates imbalance equity/choice – affordability (i.e. if funded in some jurisdictions and not others, individuals may experience barriers of access to genomic medicine, and can also lead to cost-shifting) (links also to equity of access - Priority area 4)
- Need to ensure that maximise return on investment in genomics research across Commonwealth and states/territories.
**What are the opportunities for improvement?**

A strategic and nationally consistent and coordinated approach to investment in genomics clinical services and research can be facilitated by the strong leadership and governance arrangements outlined in Priority Area 1. This will provide an opportunity to:

- strengthen collaboration in strategic investment in genomics to achieve shared goals and maximise benefits, now and into the future;
- encourage effective and efficient genomic financing and funding arrangements through consistent HTA processes;
- support timely, clinically appropriate and cost-effective mainstreaming of genomic research outcomes into clinical practice; and
- engage with industry to better understand their role in supporting the development and application of genomics.

**What does the future look like?**

- Genomic services across Australia are safe, clinically effective, efficient and cost-effective.
- Investment by Governments, and other partners, in genomic knowledge is leveraged for maximum health outcome gain and supports Australia’s research, innovation and infrastructure priorities.

**Question:**

26. With regard to Priority Area 5 – Sustainable investment in health genomics, is anything missing or what should change, for:

(a) the current situation;
(b) why is this important;
(c) opportunities for improvement; and/or
(d) what the future looks like?
PRIORITY AREA 6: Effective and appropriate collection, management and utilisation of genomic data

What is the current situation?

- Size of genomic data - code made up of around three billion units generating between 700 megabytes and 180 gigabytes of data depending on the sequencing methodology used.
- No nationally consistent or coordinated approach to the collection, generation, storage, analysis, translation and utilisation of genomic data. There is inconsistency and fragmentation in approaches to data governance and management across and within jurisdictions.
- Work underway - The Australian Genomics Health Alliance (AGHA) is undertaking critical foundation work in establishing a national data repository in collaboration with key partners, including the Kinghorn Centre for Clinical Genomics and the National Computational Infrastructure. Big data strategies being discussed across governments.
- Australia is involved, through government and non-government alliances, in progressing work to help integrate international efforts in genomic data. However, the current effort of partners across Australia is uncoordinated and there is no national strategic direction guiding international engagement.
- International developments to develop viable electronic medical record systems capable of handling family history and genomic data are required to fully utilise genomic information for patient care.
- Genomic data – cross over regarding ethical, legal and social issues. ALRC report, Essentially Yours (2004) made a number of recommendations to further protect human genetic information.
- Currently, the collection, storage and sharing of genetic data and patient access to their data is regulated by the Cth Privacy Act 1988 and the Australian Privacy Principles (APP).

Why is effective collection, management and utilisation of genomic data priority?

- Challenge not in generating data but in analysing and storing genomic data of an agreed standard that allows for sharing and linking - “big data” problem.
- Critical to genomics value (links to clinical utility) - generation of comprehensive catalogues of genomes to enable the study of genetic variation and disease. The generation of, and need to link and share such data sets, requires large-scale efforts to: develop robust data standards; ensure high quality data and broad utility; and develop computational intensity.
- Increased information facilitates more accurate diagnosis, prognosis and treatment, and management of patients with a genetic condition.
- Issues around the:
  - analytical validity of the sequencing methods and quality of the data generated;
  - clinical validity of the analysis and interpretation of the data;
  - clinical utility of the genomic knowledge in clinical practice and consumer decision-making and whether this leads to better individual and population health outcomes;
  - ensuring decisions made on evidence of analytical and clinical utility are still in the best interest of the patient; and

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need for strong data custodianship and stewardship arrangements to address issues around
data ownership and protection.

- Confidence of community in providing broad consent is linked to trust in governance
  arrangements – use of data for greater public good balanced against its potential economic value
  (noting that commercial companies also play a valuable research role).
- Inaction in moving forward in the management of genomic data, at individual health record and
  comprehensive data catalogue level, in Australia, will undermine our ability to harness the
  benefit of genomic data in healthcare to improve individual and population health outcomes.
- Moving forward, consumers, health care providers, insurers and regulators face a difficult
  balancing act to protect the privacy of genetic and other health information while also ensuring
  its availability and use for medical research and clinical decision making.

What are the opportunities for improvement?

- Building capacity so that data can be utilised to assist patient diagnosis through clinical and
  phenotypic data.
- The development of clear guidelines on reasons for data being requested and collected. Any
  such approach should have the confidence of the community.
- Opportunity to build upon the work of the AGHA and other genomic alliances across Australia
  and to work collaboratively with our international partners to enable the full potential of
  genomics in healthcare, by:
  - establishing a national data governance framework aligned with international frameworks.
    This includes developing and administering quality standards for the management, storage
    and use of data from research through clinical usage; \[31\]
  - developing nationally consistent requirements for consent, data sharing, custodianship and
    privacy requirements;
  - developing nationally agreed transparent guidelines for data collection, safe storage,
    analysis and reporting;
  - contributing to international genomic data sharing where appropriate; and
  - supporting data collection and sharing that is reflective of the ethnic diversity within the
    Australia population.

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\[31\] United Kingdom, Department of Health, (2012). *Building on our inheritance: genomic technology in
What does the future look like?

- Australian health professionals and researchers will have shared and timely access to data and relevant clinical information which will increase the effectiveness and efficiency of genomic knowledge and ensure the potential benefits of genomic knowledge apply to all Australians.
- The Australian public will have confidence in the ethical and culturally safe use and protection of genomic data and relevant clinical information.

Question:
27. With regard to Priority Area 6 – Effective and appropriate collection, management and utilisation of genomic data, is anything missing or what should change, for:
   (a) the current situation;
   (b) why is this important;
   (c) opportunities for improvement; and/or
   (d) what the future looks like?
IMPLEMENTING THE FRAMEWORK

The National Health Genomics Policy Framework sets out how the Commonwealth and states and territories will work collaboratively to integrate genomic approaches into healthcare over time.

While the policy Framework outlines an agreed national approach to policy, regulatory and investment decision-making for genomics, it does not identify all of the specific actions needed to take the Framework forward. However, the Framework does provide for a governance body to be established and it is envisaged that this body would be responsible for developing a genomics action plan (the Implementation Plan) which will underpin the Framework by identifying explicit actions, who is responsible and timeframes.

The Implementation Plan is expected to focus on the actions agreed to for the first three (or five) years and will outline their priorities, resourcing and timing. The Implementation Plan will be a key tool in measuring progress and success of the Framework. Three (or five) year action plans could provide a staged approach to achieving the necessary reforms identified within the Framework. They also allow governments to address current and emerging priorities as resources permit.

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

Questions:

28. Is the suggested approach to implementing the Framework reasonable and appropriate? Please explain your answer.

29. Is the structure of the Framework appropriate and easy to follow? Please explain your answer.

30. How could the review and evaluation of the Framework be strengthened?

31. Do you have any other feedback on the Framework?

32. Are there any issues you would like covered at the stakeholder consultation forums in February 2017?