Government of **Western Australia**
Department of **Health**

# WA Health Genomics Strategy 2021

## Draft

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# Strategy Snapshot

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Maximise the transformative potential of health genomics knowledge and services to benefit all Western Australians.

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# Introduction

## Genomics in health care

Deoxyribonucleic acid (DNA) is a chemical polymer that provides the information needed to build and maintain living organisms. In humans, the complete set of the DNA sequence is called the human genome. Genes, the functional components of the DNA sequence that contain the operational instructions for all cells, are part of the genome.

Variation occurs across human genomes. Some of a person's collection of genomic variation contributes to their individual characteristics such as hair and eye colour. Other variations, depending on their location in the genome, can influence a person's health, such as causing genetic disease through interrupting the body's normal development or other processes. A change as small as a single DNA letter (nucleotide) can cause a severe disease.

Every person inherits their own version of the human genome from their biological parents, inheriting roughly half the variants in each of their parents. However, each of us also has a small number of variants (50-100), so called *de novo* variants, not present in either of our parents.

Variations may only be in an individual's ovaries or testes ("germline" cells) such that the disease-causing variant can be passed to their children, but they don't have the disease themselves. If disease-associated variations are somatic (in cells that aren't part of the germline), they may lead to the individual having genetic disease in only one part of their body – for example in one arm or one leg or only in one cell, which may turn cancerous.

The study of human genomes encompasses the inheritance of genes, their structure and function as well as how the genes contained within the genome interact with each other and with the environment. Knowledge gained from the study of human genomes can be used to enhance the provision of medicine and health care at an individual and population level (i.e., public health). Microbial genomes also influence human health; therefore, knowledge gained from studying microbial genomes can also be used to support health care provision.

Use of human and microbial genomic knowledge to inform health care delivery is referred to as "health genomics". In this document, health genomics also refers to the analysis of single human genes (genetics) and chromosomes. Health genomics is enabled by genetic testing technologies, which can be used to detect variation in the DNA sequence. Genetic testing is not new to health care; however, it has undergone rapid and significant transformation, enabling testing of multiple genes simultaneously, or even sequencing of entire genomes (known as genomic sequencing).

Advances in medical science and data analytics as well as our growing understanding of the genomic causes of diseases have hastened the pace that genomic knowledge is discovered and applied to health care. Big reductions in the cost of genomic sequencing and increased knowledge of clinical implications have also increased the demand for genomics, creating both challenges and opportunities for healthcare systems.

**Established uses of health genomics**

- Accurate molecular diagnostics of people presenting with diseases
- Informed pre-symptomatic predictions of an individual's likelihood of developing a disease enabling proactive surveillance, management and/or treatment
- Better-informed family planning options
- Evidence-based matching of treatments (e.g. pharmacogenomics) or clinical trials for inherited and acquired genetic conditions

Health genomics has already been shown to be able to transform and improve health service delivery and health outcomes for individuals and populations. The ability to apply health genomics across the human lifespan (Appendix 2), creates immense opportunity for further transformation. Health genomics is improving health and wellbeing by providing earlier and more accurate diagnoses, prognostic information, targeted treatments, precision clinical trials, and better-informed disease prevention. Additionally, health genomics can prevent inappropriate or unnecessary treatments and procedures, creating economic savings for the healthcare system in addition to the patient benefits.

Notable applications of health genomics are in the detection of common genetic disorders, cancers and rare diseases.

**Evidence of cost-effectiveness**

Genomic sequencing is perceived to be costly; however, there is emerging evidence of cost-effective applications of genomic sequencing in health care. For example, for paediatric patients in clinical settings, genomic sequencing has been found to increase the diagnostic rate by 16-79% and decrease the cost by 11-64% compared to standard care (1).

Further evidence of cost-effectiveness is needed as potential applications for genomic sequencing continue to grow. This evidence will be integral to the successful integration of genomic sequencing in health care.

## Why is a strategy needed?

All Western Australians should be able to appropriately access and benefit from genomic services. However, the health system presently lacks a state-wide, strategic approach to the delivery of health genomic services. Without this approach, health genomic services have developed in an ad hoc manner resulting in clinical variation and fragmentation across the health system. In the absence of a cohesive approach, health genomic services are also not being delivered as efficiently and effectively as possible.

These issues have been compounded by the rapid advancement of health genomics, which continues to provide new tests and therapies to enhance health care and improve health outcomes. Strategic oversight, new thinking and new approaches are needed to enable genomic tests and technologies to be efficiently integrated into the health system when there is robust evidence of clinical utility, safety and cost-effectiveness. This integration should be focussed on improving outcomes for all Western Australians.

There is an imperative for a system-wide strategy to lead and coordinate the efficient, effective, ethical and equitable translation of genomics knowledge into the health system.

## About the WA Health Genomics Strategy 2021

The Strategy presents a shared vision for all stakeholders, key strategic priority areas and recommended initiatives to lead and coordinate the efficient, effective, ethical and equitable translation of genomics knowledge into the health system. It aims to achieve this by safeguarding current progress, building on local strengths (Appendix 3) and proposing the way forward for enhanced health service delivery to ultimately achieve the best possible outcomes for all Western Australians through health genomics

### Scope

The Strategy has been developed primarily for use by the WA health system. As such its focus is on health genomics activities that occur within or are facilitated by the WA Department of Health and health service providers. It is recognised, however, that there are many existing and potential partners important to supporting the development and optimal application of health genomics for the benefit of the WA population. Collaboration and engagement with these partners will therefore be an ongoing objective.

The scope of the Strategy is the application of genomic knowledge and services to advance medicine and health care, as well as the associated health outcomes and policy implications. All genomic technology for medicine and healthcare purposes are within the remit of the Strategy. This includes genetic and genomic tests (i.e. tests that focus on a single gene and those that focus on multiple genes at the same time), and interventions or therapies utilising genomic knowledge.

### Strategy development process

The Strategy's development occurred with the guidance of the WA Health Genomics Strategy Advisory Committee, which reported to the Executive Sponsor Dr Andrew Robertson, Assistant Director General of the Public and Aboriginal Health Division, WA Department of Health.

Considerable consultation was undertaken to ensure stakeholder engagement and contribution, including more than 50 face-to-face interviews and two online surveys. These were conducted to understand the current application of genomics in the health system, and the current strengths, opportunities and issues for health genomics in WA.

Stakeholders engaged included health consumers and advocates, clinical geneticists, genetic counsellors, medical specialists primarily trained in areas other than genetics, primary healthcare specialists, laboratory scientists, medical researchers, policy makers, and clinical service planners.

Additional advice from the range of experts comprising the WA Health Genomics Strategy Advisory Committee informed development of the draft Strategy document.

## The policy context

A growing number of countries are formulating policies to support the development of health genomics and the adoption of more personalised approaches to health care. In Australia, health genomics has received national recognition at the highest levels, evidenced by the release of the Australian *National Health Genomics Policy Framework 2018-2021* (2) (National Policy Framework) endorsed by the Council of Australian Governments Health Council, and the allocation of $500 million over 10 years to the Commonwealth's Medical Research Future Fund Genomics Health Futures Mission (3).

The National Policy Framework provides a shared direction and commitment between all governments in Australia to consistently and strategically integrate genomics in the Australian health system. The WA Government has committed to supporting implementation of the National Policy Framework. The Strategy aims to build on and align with the National Policy Framework's five interdependent and interlinked strategic priorities (person-centred approach, workforce, financing, services and data), while taking into consideration local context and needs.

In WA, several policies have been developed that embrace the promise of genomics and/or precision health. These include the *Sustainable Health Review Final Report*, which under Strategy 8 prioritises an "enhanced reputation as a world leader in the emerging field of precision medicine and public health that includes new data/digital, informatics, genomics, phenomics and geo-spatial technologies, and their application to health" (4). Priority 2 of the *WA Cancer Plan 2020-2025* also singles out genomics in its strategy to "develop and implement statewide genomic sequencing capability to inform treatment" (5).

Other important strategies that align with and/or support the Strategy are the *WA Health Digital Strategy 2020-2030* (6), the *State Public Health Plan for Western Australia 2019-2024* (7), the *National Microbial Genomics Framework 2019-22* (8), the *National Strategic Action Plan for Rare Diseases* (9) and the *WA Rare Diseases Strategic Framework 2015-2018* (10). Elements of the Strategy will work synergistically with these WA and national policies.

# The Vision for Genomics in WA

## Vision

Maximise the transformative potential of health genomics knowledge and services to benefit all Western Australians.

## Underlying principles

To achieve the vision of the Strategy, implementation of the strategic initiatives will need to be underpinned by strong and clear principles, which are outlined below.

**Empowerment**

Individuals and families are the central focus of health genomics and are empowered to make informed decisions about the use of genomic knowledge in their health care.

**Equity**

Genomics is a key precision health tool that should be available to all Western Australians and their health teams, based on clinical, personal and familial need.

**Diversity and inclusion**

The ethnic, cultural and socio-economic diversity of the Western Australian community is respected. Health genomics services, including genomics data management, is culturally secure, accessible and responsive for Aboriginal people and other people of linguistic and culturally diverse backgrounds.

**Flexibility**

The rapid advancement of genomic technology and knowledge requires an agile and adaptive strategic approach.

**Utility**

The application of genomic knowledge to health care is supported and informed by evidence, person- and family-centred clinical need, and research.

**Trust**

Health genomics is delivered in a way that builds community confidence and trust and minimises potential harm.

**Value**

Investment in health genomics, including genomic technology and infrastructure, is focussed on delivering high value and benefit, primarily for consumers (individuals and their families) but also for services and the health system.

## Enablers of success

To guide the successful implementation of the Strategy, three enablers have been identified as critical building blocks:

- Governance, accountability and roles and responsibilities;
- Senior leadership and support; and
- Meaningful partnership and collaboration.

### Governance, accountability, roles and responsibilities

Robust governance is crucial for driving the visibility, planning, implementing, evaluating and reporting of health genomics services. Governance arrangements should be agile and able to respond quickly to the rapidly developing field of genomics. This supports the health system to remain accountable for the delivery of high-quality services that are fit for purpose and provide value to all Western Australians.

Clear roles and responsibilities for all stakeholders involved in each of the strategic priority areas are paramount to ensuring accountability is upheld and outcomes are achieved in unity.

### Senior leadership and support

Leadership and support at the senior executive level is critical to the success of the Strategy. It is important that leadership at this level is ongoing, engaged, recognises the value of health genomics and encourages the translation of the benefits into the health system across all strategic priority areas. This includes providing support to guide the safe and appropriate application of genomics within their respective areas and influencing the organisational culture to adapt to new ways of working.

### Meaningful partnerships and collaboration

Partnerships and collaborations are necessary for maximising outcomes and reducing duplication of efforts in the translation of health genomic advances to benefit all Western Australians. It is imperative that collaboration exists at a state, national and international level. There are already many valuable partnerships and collaborations that exist involving West Australians, and opportunities exist to expand these across multiple sectors to achieve success. These sectors include: Health consumers;

- Public health service providers;
- Primary health care;
- Aboriginal Community Controlled Health Services;
- Private health care;
- Private industry;
- Research;
- Non-government organisations and patient support organisations; and
- Government organisations and policy makers.

## Priority 1: Person- and Family-centredness

### Goal 1: To embed a person- and family- centred approach into health genomics

A person-centred approach to health care involves a way of thinking and delivering services in which the people using those services are considered equal partners in planning and developing care that meets their needs. People seeking care are placed at the centre of decisions and seen as experts, working together with service providers to get the best outcome. They are seen holistically, as a whole person with physical, psychological, spiritual, cultural and emotional aspects that contextualise their experiences of both health and illness.

A family-centred approach to health care extends the partnership philosophy beyond the individual seeking care, to include their family members. This recognises that families can play an integral role in providing care for family members who are ill, particularly when health conditions are serious and life-threatening. Families can provide instrumental levels of emotional and physical support that impact on individual's experiences of health care. Hence, family context can be significant for people accessing health services, and this includes services related to the delivery of health genomics.

A family-centred approach is particularly important for health genomics, due to the familial nature of much genomic information. When a person undergoes genomic testing, the resulting information obtained about them may have implications for family members in terms of their genomic risk of disease. This creates the need to consider intra-familial communication of genetic risk information. Family members may also need to be involved and undergo testing to clarify the likely consequence of findings and to aid interpretation of inconclusive test results (e.g. newly discovered variants). Communicating with families about genomic information can be a difficult matter, as the process is grounded in the broader context of existing family relationships, functioning, communication patterns, cultural and individual beliefs. Individuals and families need to be supported to understand and cope with genomic information.

A person- and family-centred approach should encompass all aspects of health genomics, including pathology services, medical services, the management of genomic data, education and learning and genomic-related research. Being person- and family-centred will involve fostering a mutually beneficial partnership between people engaging with health genomics, their families (where appropriate) and health genomics providers. The focus must be on listening to consumers, enabling information exchange, facilitating choices and sharing decision-making, so that individual and family needs, goals and preferences are met as part of a collaborative approach. In doing so, health genomics providers must respect the ethnic, cultural and socio-economic diversity of individuals and families.

Providing person-centred, equitable and seamless care is one of the enduring strategies of the *Sustainable Health Review Final Report*. This report recognises the importance of providing care with people, not to people, and the need to partner with consumers to design and improve service delivery to meet their needs, including conversations between clinicians and consumers about what is really needed to improve the patient journey and satisfaction; provide consumers with choices; improve communication, relationships and coordination between health services; provide timely, inclusive access to services; empower people through health literacy; and develop meaningful ways of measuring experiences and outcomes.

## Initiatives

1.1. Build strong leadership and commitment to a person- and family-centred culture for the implementation of health genomics.

1.2. Understand and optimise the consumer experience of health genomics, including individual and family outcomes related to the use of genomic testing.

1.3. Ensure the perspectives of consumers are considered in decision-making around health genomics, including in individual care and in the development, implementation and evaluation of policies, programs, services and research.

1.4. Champion equity of access to value-based health genomics services, including for priority populations, such as Aboriginal peoples and people from culturally and linguistically diverse backgrounds.

1.5. Support and improve consumer knowledge, health literacy and capability for informed decision-making about genomics in health care.

1.6. Offer all those considering genomic testing a standardised, culturally appropriate, voluntary and informed consent process, with effective support and counselling around intra-familial communication pre- and post-test.

1.7. Raise public awareness and understanding about the use of genomic knowledge for individual health care and in protecting and improving public health.

## Priority 2: Sustainable Investment

**Goal 2: To attain sustainable and strategic investment in high value health genomics services**

Sustainable investment is crucial to create and maintain an appropriately sized and skilled health genomics workforce. To underpin the services this workforce provides, the necessary infrastructure and data management capability is required, and a thriving translational research environment will enable the delivery of optimal health genomics services that can grow and adapt to meet system-wide demand. With limited health resources, sustainable investment requires transparent and robust decision making. The pursuit of innovative, flexible funding models and partnership opportunities is also desirable.

Current investment and funding models have not kept pace with the rising demands, costs, expectations and opportunities afforded by genomic testing in the healthcare setting. The majority of genomics health services in WA are delivered through the public health system: clinical genetic services (Genetic Services of WA; GSWA) and specialist laboratory services (through PathWest).

PathWest has experienced a significant increase in the volume and scope of requests for genomic tests received from clinicians, and similarly there has been a large rise in demand for genetic counselling and clinical genetics services supplied by GSWA. However, current funding mechanisms (such as activity-based funding, ABF; and block funding) have been unable to support this increased demand, resulting in limited capacity and compromises being made by both GSWA and PathWest in terms of the services delivered. Limitations of the current funding approaches that have contributed to the challenges being experienced by WA's genomic health services include:

- ABF not adequately covering the cost of the clinical genomics services;
- Preclusion of public health service providers from claiming Medicare Benefit Schedule (MBS) rebates for tests provided to public patients (for the albeit small number of genomic tests on the MBS) despite other jurisdictions utilising this funding mechanism;
- Lack of upfront investment in ICT and laboratory infrastructure to support local capacity building and the expansion of genomics services;
- Lack of dedicated funding to enable new tests with recognised clinical utility to be established and validated within the health system;
- Lack of funding for dedicated trainee positions in clinical genetics; and
- Limitations on services that GSWA can provide on a state-wide basis due to placement of the service within a geographically-bound health service provider.

A system-wide approach for investment and funding is needed to address these limitations and realise the opportunities provided by genomics, including the emerging evidence of cost-savings for the health system if genomics is integrated in a strategic way. This can be achieved by establishing systematic processes, which support transparency and robust decision making – the cornerstones of sustainable investment in health genomics. Such processes would help to guarantee that coordinated investment is of high quality and fiscally, ethically and socially responsible, as well as equitable across the state. For example, a rigorous process for assessing the evidence for individual tests such as their utility; safety and quality; and economic impact is needed to inform system-wide policy and decisions on investment (or disinvestment).

The *Sustainable Health Review Final Report* describes a need to introduce methods that shift away from health services being funded based on volume of services provided historically to a more flexible approach based on population health needs and outcomes. This is essential for the desired evolution of genomics health services in WA, to address a clear need for the implementation of contemporary funding methods that enable innovative models of care responsive to patient and family needs. A process for secure, sustainable and transparent funding for translational research and implementation of health genomics will also help to ensure the health system is well positioned to meet the goals of the *Sustainable Health Review Final Report*. Such a process will also capitalise on the opportunities provided to improve efficiencies, maximise health outcomes and minimise waste.

## Initiatives

2.1. Raise the profile of health genomics among health system decision-makers to ensure that health genomics is recognised as a priority for investment.

2.2. Contribute towards establishing evidence that supports the implementation of efficient, cost-effective and value-based health genomic services.

2.3. Secure appropriate funding to enable best practice health genomics services, including funding for genomics infrastructure, workforce and data management that is informed by priorities.

2.4. Establish mechanisms for the timely and evidence-based assessment of health genomic tests, technologies, models of service delivery and therapies for prioritising their translation into the health system.

2.5. Encourage strategic philanthropic contributions and public/private partnerships with a diverse range of organisations including pharmaceutical, medical technology and digital health to support infrastructure, service provision, research and innovation.

2.6. Develop local, national and international collaborations to maximise outcomes and benefits achieved through investment in health genomics.

## Priority 3: Services

**Goal 3: To achieve the optimal integration of genomics knowledge in the health system to deliver high value care that is equitable and safe.**

The full promise of health genomics relies on the widespread, appropriate integration of genomics across the continuum of care. Given the rapid rate of growth in this field, there is a pressing need for a collaborative and coordinated approach to health service planning to support the optimal integration of genomics in the health system. These services should be appropriate, innovative and of high value to meet the needs of individuals, families and the population in a safe, timely and ethical way.

The health system is well positioned to achieve this goal because it has single, public state-wide service providers for both clinical genetics (including genetic counselling) and pathology. This arrangement presents the ideal environment for optimising genomics as it provides the opportunity to centralise referrals for clinical services, genomic testing, data collection and reporting, and in turn maximise the efficiency, equity and cost-effectiveness of health services.

In the absence of system-wide policy guidance and sustainable funding, the integration of genomics in the health system has developed in an ad hoc manner. Some clinical specialties have excelled at integrating genomics as part of routine care by building on their own networks across medicine, diagnostic laboratories and research to aide in the diagnosis and management of genetic disorders. Other specialties have benefited from clinical geneticists and genetic counsellors contributing to their clinics. Both contemporary service delivery models have resulted in more efficient and cost-effective health services than other service delivery models.

Whilst the current approaches to integrating genomics in the health system have occurred in response to the rising clinical utility and demand in health genomics, there has not been a systematic approach aimed at addressing the population's health needs. Despite successes made to date, over the long term, the absence of a systematic approach has significant potential to undermine the equity and quality of genomics services provided by the health system. As an example, limitations in the capacity and capability of the two single, state-wide service providers are already hindering the ability to integrate new advances in genomics whilst balancing an ever-increasing workload.

Optimising the integration of genomics in the health system will become even more important as the applications of genomics continues to expand to mainstream care as well as increase in the private sector. It is crucial that inequity of access to genomics services and the benefits they provide are mitigated. Therefore, the success factors of existing models should be explored so the learnings can be more widely implemented throughout the health system, and new contemporary service delivery models should also be considered.

The goal of this strategic priority aligns with the overall aim of Strategy 5 - Drive safety, quality and value through transparency, funding and planning of the *Sustainable Health Review Final Report*.

## Initiatives

3.1. Increase the capacity and capability of the health system to deliver timely, equitable, accessible and value-based health genomics services in clinical care and public health.

3.2. Support the implementation of innovative and adaptive service delivery models, such as multidisciplinary clinics, which enable the integration of health genomics into mainstream care.

3.3. Explore innovative ways to deliver genetic counselling to help meet growing demand.

3.4. Facilitate partnerships and networks to promote and support sharing of health genomics knowledge and reduce duplication of effort across the health system.

3.5. Develop clear pathways, standards and policies to aid the optimal, safe and equitable delivery of health genomic services.

## Priority 4: Workforce

**Goal 4: To have an adequately sized, appropriately skilled, sustainable and agile health genomics workforce**

In recent years there has been a significant increase in the utility of health genomic tests and the demand for genomic testing and clinical genetics services. These services necessitate a health genomics workforce, comprised of clinical geneticists, genetic counsellors, pathologists, bioinformaticians and medical scientists, working collaboratively and as part of broader multidisciplinary teams. However, the capacity of the health genomics workforce, and the education pathways needed to train and upskill both the health genomics workforce and the broader health workforce to deliver genomic health care, are not commensurate with increasing demand for services. Extensive on the job training is also required for medical scientists' post-qualification for these professionals to have the necessary expertise to provide genomics services. Consequently, the needs of the individuals and families to access these services are not being adequately met, often demonstrated by long wait times to see a clinical geneticist and/or genetic counsellor and to receive the results of a genomic test.

Having a skilled workforce with the competence to appropriately deliver genomics in the health system is essential to the provision of high-quality services and improved health outcomes for consumers. Current workforce (and skill) shortages are a major concern for stakeholders, with the need for education and training considered critical. As per Recommendation 27 of *the Sustainable Health Review Final Report*, flexibility is needed within the health system to enable health service providers to manage their workforces within budget. Creating a competent and agile workforce that meets the needs of the WA population will require investment in both the current and future workforce. Present remuneration arrangements and professional development opportunities for key professions, such as genetic counsellors and medical scientists also require consideration, as these affect the retention rate and ongoing upskilling and training of staff in a complex and rapidly changing field.

Making genomics an integral part of mainstream health care will require the appropriate up-skilling and ongoing education of the broader health workforce (e.g., medical specialists, general practitioners, nurses etc.). It is imperative that members of this workforce understand and can provide and/or connect consumers to high quality health genomic services when appropriate. Mainstreaming can be further strengthened by establishing clinical champions who can advocate for and lead the integration of genomics within their respective medical specialty.

Achieving a sustainable health genomics workforce will also necessitate collaboration and partnerships between WA stakeholders, with other jurisdictions, and with education institutions in keeping with Recommendation 26 of the *Sustainable Health Review Final Report*. *The WA Health Strategic Intent 2015-2020* recognises that strategies for greater attraction, induction and retention of staff, as well as more opportunities for professional development, are key enablers for the provision of safe, high quality and accessible health services to all Western Australians.

## Initiatives

4.1. Establish the health genomics workforce capacity needed to meet the growing demand for health genomics, particularly in areas where there are critical workforce shortages.

4.2. Employ mechanisms to grow the health genomics workforce and attract and retain health genomics professionals in WA.

4.3. Foster a sustainable, inter-jurisdictional approach to meet WA's health genomics education and training needs.

4.4. Improve the genomic literacy and capability of the health workforce.

4.5. Increase the competency of the health genomics workforce in the provision of culturally safe and secure care.

4.6. Ensure the health workforce has the competency required to deliver person- and family-centred health genomics services and facilitate informed consumer decision-making about genomics and health care.

## Priority 5: Digital Health and Data

**Goal 5: To accomplish effective, efficient, ethical and timely collection, analysis and use of genomic data**

The successful integration of genomic technologies into the WA health system is dependent on our ability to address several unique challenges relating to data management and digital and technological infrastructure capability. Genomic sequencing involves the generation of large amounts of raw data that needs to be appropriately used, stored and managed.

A key issue with health genomic data is the extent they should be treated as exceptional and accorded special protection. This is due to the complex ethical, legal and social issues related to their predictive capability, familial implications, longevity and uniqueness. For example, in Australia the insurance industry self-regulates with respect to its use of genomic data in life insurance underwriting, which has implications for public trust and willingness to participate in genomic testing. The vast amount of genomic data collected also increases the risk of privacy breaches and data misuse and increases data storage needs. Maintaining data security and confidentiality is a significant challenge.

Harnessing the value of genomic data to improve health care is reliant on national and global cooperation and data sharing. This is particularly the case for rare genetic conditions and cancers, where sharing of patient data can play a major part in achieving a diagnosis or conducting research. Similarly, analysis of variations in microbial genomes can assist with detecting the source of infections and tracking infectious outbreaks.

To optimise the use of genomic data in supporting effective clinical decision making, there is a need to ensure greater consistency in terms of how genomic data is captured, shared, stored and protected across all parts of the WA health system. Health genomic data protocols will need to take into consideration community expectations as well as the legal and regulatory requirements, to ensure public trust and confidence.

Advances in data analytics and bioinformatics, as well as wider digital health technologies (such as electronic medical records and speciality decision making tools) are driving a revolution that can help deliver the promise of health genomics. Considered planning and appropriate and timely investment will be critical to leverage on existing resources and to develop and support the data and digital infrastructure required to capitalise on technological developments. High quality digital health systems and infrastructure can not only help deliver high value care for individuals but has the potential to attract specialised workforce and foster clinical innovation.

The potential of data and digital innovation to improve healthcare efficiency and transform health care has been highlighted in the *Sustainable Health Review Final Report* in Strategy 6 – Invest in digital healthcare and use data wisely.

## Initiatives

5.1. Ensure that digital and data solutions within the health system support the appropriate integration of health genomics into health care.

5.2. Develop and implement recommendations for how health genomics data should be effectively and efficiently recorded, shared, stored and protected in line with evolving community expectations, legal and regulatory requirements, and international standards.

5.3. Improve the capability and capacity of the health system to analyse and interpret health genomics data.

5.4. Improve the ability to capture, access and transfer phenotypic information to aid genomic health care, including appropriate ordering of health genomic tests and data analysis.

5.5. Investigate opportunities for utilising health genomics data to enhance cohort identification for designing services, interventions, treatments, clinical trials and research.

5.6. Promote culturally secure and appropriate health genomics and phenotypic data collection, storing and sharing that reflects the ethnic diversity within the Australian population.

5.7. Contribute towards a national health genomics data governance framework and explore a potential national genomic data collection, storage and sharing facility.

## Priority 6: Research and Innovation

**Goal 6: To have a health system that values and supports the translation of genomics research and innovation**

There is an increasing body of evidence demonstrating a range of benefits to clinical practice that arise from the translation of health genomics research and innovation. These include faster and more accurate diagnoses, and the ability to tailor health care for individuals and their family members. WA is home to many successful research groups that continuously pursue a range of health genomics research and innovation opportunities both locally and around the world. Some notable examples are included as vignettes.

Considering the established and growing prominence of health genomics research and innovation in WA, there is significant potential for the resulting benefits to be applied across the health system. There are several factors critical to this, which include:

- Clear pathways and adequate funding for medical scientists and health professionals to collaborate in planning and conducting health genomics research and innovation for enhanced translation into clinical practice;
- Building capacity and equal access to local DNA sequencing and bioinformatics capabilities;
- Appropriate data storage facilities to hold the volume of health genomics data being generated; and
- An efficient ethics and governance process for approving health genomics research and innovation projects.

A coordinated system-wide approach is needed to provide the optimal translation of genomics research and innovation into the health system. Ideally, this approach would recognise the value of translational health genomics research and innovation, and embrace an organisational culture that encourages translation of the resulting benefits for high value care and ultimately see greater investments made in this area. The critical factors for success would also be addressed and by doing so, the benefits of health genomics research and innovation would be dispersed to the WA population.

The following initiatives were developed to achieve the critical factors for success, like those mentioned above. The strategies align to Strategy 8 – Innovate for sustainability of the *Sustainable Health Review Final Report* and the mission of the WA Government's Future Health Research and Innovation Fund.

### Initiatives

6.1. Ensure local, translational health genomics research and innovation projects are informed by clinical utility and the needs of individuals, their families and the WA population.

6.2. Facilitate opportunities for health professionals and medical scientists to be involved in translational health genomics research and innovation.

6.3. Facilitate a culture that encourages a timely, transparent and shared approach to the integration into the health system of health genomics research and innovation advances.

6.4. Foster and develop productive local, national and international partnerships and networks to inform and conduct translational health genomics research and innovation projects that should benefit Western Australians.

6.5. Explore avenues to secure funding for local health genomics research and innovation projects into priority areas for the health system.

## Outcomes

It is anticipated that the Strategy will lead to significant positive outcomes at multiple levels, including for:

- People who utilise genomics services within the health system and their families ('consumers');
- The health system and the genomics services that operate therein; and
- The broader Western Australian community, including the state as a whole and its economy.

A range of anticipated outcomes from the Strategy are highlighted below. The implementation of the Strategy stages will include processes to capture, measure, monitor and evaluate the extent to which these outcomes are achieved.

***Table 1. What success looks like***

| **Person and family** |
|---|
| • Understand the benefits, risks and limitations of health genomics<br>• Experience improved genomic literacy<br>• Are informed and empowered to ask questions and make decisions<br>• Can take greater accountability for and ownership of their health<br>• Have clear pathways to access genomic services<br>• Get the care they need when they need it<br>• Have their needs and preferences met<br>• Receive safe, high quality care<br>• Trust their genetic information is managed and used in accordance with their wishes and their privacy is protected<br>• Experience respect for their ethnic, cultural, and socio-economic diversity and how these factors influence the way they relate to genomic knowledge and use genomic services<br>• Can easily access the support and information they need to make choices about genomic testing and the ways genomic knowledge is used in their health care |
| **Community** |
| • The overall health of Western Australians is improved through optimal access to and use of genomic knowledge in health care<br>• Community members are engaged as partners in determining the safe, fair and ethical use of genomics in health care<br>• Data are shared, stored and protected in line with community expectations (and regulatory requirements)<br>• The community understand the benefits of sharing genomic data to support research<br>• There is community awareness, acceptance, confidence and trust in health genomics |

| Services and health system |
| --- |
| • Genomic services are safe, high quality, sustainable, contemporary, transparent, adaptive, agile, evidence-based, efficient, ethical and cost-effective<br>• There is equitable access for all Western Australians based on personal and familial need<br>• Services are culturally safe for Aboriginal people and people from culturally and linguistically diverse backgrounds<br>• Services deliver an ideal experience for consumers that achieves their goals and result in improved health for them and their families<br>• There is capacity and capability to meet demand, with timely access to clinical genetics services and the results of genomic testing<br>• A system-wide culture that fosters person- and family centred care and that values integration and innovation of translational research<br>• Genomics tests are conducted with a view to achieving benefits for the individual and/or family members, rather than being driven by technology<br>• Genomic tests, technologies and therapies used in the health system will have clinical utility<br>• Use of these tests, technologies and therapies will lead to higher diagnostic yields and will improve disease management<br>• Seamless consumer and clinical pathways that facilitate flexible and coordinated care<br>• There are high levels of genomic literacy among health professionals, allowing optimal decision-making that is informed by genomic knowledge<br>• Services connect consumers and health professionals to trusted, relevant, accurate and timely information about genomics and health care, which is linguistically and culturally safe and appropriate<br>• Best practice technologies and infrastructure, including digital solutions, support the appropriate and ethical collection, analysis, management and sharing of genomic data<br>• There are high levels of employee satisfaction<br>• There are strong strategic partnerships between consumers, services providers and researchers, resulting in shared knowledge and decision-making<br>• Health genomics is a priority for systemic investment<br>• Improved integration of genomics leads to high value care and lower costs of care<br>• There is reduced pressure on health services, less duplication and waste of resources, less variation in clinical practice and more appropriate use of health services<br>• There is a strong local evidence based, which can be used to demonstrate the value of genomics to the health system |

## Next Steps

The Strategy has been developed to unite key stakeholders involved in the delivery and/or utilisation of health genomics services in WA. It provides a shared vision and six strategic priority areas to streamline investment of resources to achieve the optimal application of genomics across the health system. This is premised on a belief that more outcomes may be achieved to benefit all Western Australians by working collaboratively and coherently, rather than in silos.

It is important to note that a strategy document on its own cannot deliver its vision. Rather, it must be implemented appropriately and effectively to ensure that its well-intended initiatives are translated into continued action. The essential steps to achieving the vision include establishing governance arrangements, developing and carrying out implementation plans, and building on a culture of accountability.

### Governance

A new governance body, the WA Health Genomics Steering Committee (the Committee), will be established to provide ongoing stewardship to the implementation of the Strategy. The Committee will bring together key leaders and stakeholders from across WA including representatives from the health system's health service providers and other subject matter experts. It is anticipated that this Committee would interact with other relevant bodies such as the Minister for Health's Precision Health Council.

The Committee will play a pivotal leadership role, which will include facilitating and supporting partnerships and initiatives to progress the strategies outlined in the previous section. The Committee will also provide ongoing monitoring, review and evaluation of the Strategy. In addition to this, working groups may be formed to guide the implementation of specific projects as required.

### Implementation and evaluation

The Strategy provides a guide to achieving the optimal application of genomics in health care for the next ten years. Given the rapid pace at which new evidence emerges and technology advances in the field of health genomics, an agile approach to implementation (including planning, evaluation and monitoring) will be required.

The Strategy will be implemented through a series of short-, medium- and long-term horizons. However, it is anticipated that some larger projects and initiatives will span two or more phases of implementation. The first Implementation Plan is currently in development and will be finalised in first half of 2021; subsequent implementation plans will be developed with consideration to the evolving health landscape.

The implementation plans are a key tool for measuring the success of the Strategy. An evaluation of each implementation plan will be undertaken to help inform the development of the next implementation plans. The benefits of these are two-fold as they will also serve as mid-point evaluations on the success of the Strategy.

## Appendix 1 – Glossary of terms

**Activity-based funding (ABF)**: A method of funding whereby health services are paid based on the number and mixture of patients they treat, taking into account the complexity of particular service events.

**Autosomal recessive inheritance**: A disease exhibits autosomal recessive inheritance when a disease-causing genetic variant must be present on both chromosomes in a pair of non-sex chromosomes before the disease is expressed. If both parents have one copy of the same disease-causing variant, then there is a one in four chance their children will inherit both copies of this variant and express the disease.

**Bioinformatician**: Someone who uses computer science and mathematics to study genomic information and solve biological (i.e. health) problems at the molecular level.

**Block funding**: A method of funding whereby health services are paid based on the historical cost of delivering services.

**Chromosome**: DNA is packaged into long, thread-like structures called chromosomes. Humans have 46 chromosomes, which can be arranged into 23 pairs, with one chromosome from each pair inherited from either biological parent. Of the chromosome pairs, 22 contain autosomes, which appear the same for males and females. The remaining pair of chromosomes are sex chromosomes, which determine biological sex.

**Consumers**: People who utilise services within the health system and their families.

**Deoxyribonucleic acid (DNA)**: A chemical polymer that provides the information needed to build and maintain living organisms. In humans, the complete set of the DNA sequence is called the human genome.

**Diagnostic rate**: The number of diagnoses achieved over a set period.

**Gene**: The functional components of the DNA sequence that contain the operational instructions for all cells in the human body. Genes code for molecules that influence the expression of a person's characteristics. Changes in the DNA code of a gene can potentially result in a person having a health condition.

**Genetics**: The study of the functions and composition of individual genes.

**Genetic test**: A test whereby DNA material is extracted from cells and analysed in a laboratory for changes to chromosomes, genes, RNA and/or proteins.

**Genomics**: The examination of genes and how they interact with each other and the environment. Differs from genetics, which is focused on analysing single genes.

**Genomic testing/sequencing**: Testing for variants in many genes, specific regions of the genome, or the entire genome sequence at one time. Genomic testing can detect changes in many genes at once using a gene panel, in the exome (i.e. sections of the genome that contain the instructions for proteins) using whole exome sequencing, or throughout the entire genome using whole genome sequencing.

**Health genomics**: Use of human and microbial genomic knowledge to inform healthcare delivery.

**Nucleotide**: The chemical 'letters' (nucleotides) that make up the DNA sequence. There are four chemical letters that make up the DNA sequence; adenine (A), guanine (G), thymine (T) and cytosine (C).

**Person-centred care**: Focuses on the delivery of quality healthcare while respecting a person's values, needs, wants and goals.

**Pharmacogenomics**: The study of how the human genome affects an individual's response to different drugs.

**Value-based health care**: Delivers the best outcomes for patients and the best value for the health system.

**X-linked recessive inheritance**: A disease exhibits X-linked recessive inheritance when a disease-causing genetic variant must be present on one (in males) or both (in females) copies of the X-chromosome before the disease is expressed. recessive

## Appendix 2 – Current and emerging application of genomic tests across the life cycle

| Type of test | Description |
|---|---|
| **Diagnostic** | Test to investigate the cause of disease. Follows onset of symptoms, a clinical discovery or a positive screening test. |
| **Microorganism** | Testing of microorganisms that impact human health. Used to understand and trace infection, outbreaks and antimicrobial resistance. Emerging applications include investigation of human microbiomes and influence on immunity, drug interactions and disease expression. |
| **Newborn bloodspot screening** | Screening of newborns to identify infants at increased risk of one of many rare and serious genomic conditions to enable early intervention. |
| **Pharmacogenomics** | Screening for genomic variants that influence drug metabolism to inform drug choice and dosage. |
| **Predictive/pre-symptomatic** | A test to establish whether an asymptomatic at-risk individual has predisposition to disease development. |
| **Preimplantation testing** | Screening or diagnosing of embryos to determine those without a specific genomic variant or a chromosomal abnormality for subsequent use in vitro fertilisation (IVF). |
| **Prenatal testing** | Screening or diagnosing of a fetus during pregnancy for specific genomic variants or a chromosomal abnormality to inform reproductive choice, preparedness and early intervention. |
| **Prognostic** | Identifies gene variant/s to predict disease progression, severity and outcomes and guide and monitor therapeutic interventions. |
| **Reproductive carrier screening** | Testing to determine the carrier status of couples planning a pregnancy or in pregnancy to inform reproductive choice, preparedness and/or early intervention. |
| **Posthumous** | Molecular autopsies to identify cause of sudden unexplained death, including *in utero* death. |

Table adapted from: Bilkey et al, Genomic Testing for Human Health and Disease Across the Life Cycle: Applications and Ethical, Legal, and Social Challenges, Frontiers in Public Health 2019 (11)

## Appendix 3 – Local strengths

The presence of single, state-wide providers of public pathology and clinical genetic services in WA place the State in a strong position to build on and grow its health genomics service offerings. PathWest provides high-quality genomic testing services to the health system, facilitated by the centralisation of pathology and medical science expertise. The public pathology provider has developed fit-for-purpose genomic tests across multiple disciplines and is an international centre of expertise in genomic testing for neuromuscular disorders and adult cancers. Similarly, GSWA provides high quality clinical care and genetic counselling services to Western Australians and helps to build genomic literacy through participation in multi-disciplinary clinics across health services providers. Together these state-wide services act as a clear, single point of referral for health genomics in WA and have an assurance function, guiding appropriate genomic testing and a consistent approach to health genomics.

WA has also been a hub of clinical research and innovation in health genomics. Excellent, internationally recognised Western Australian researchers in the field of medical genomics have made strides in rare disease diagnostics, which through strong partnerships between researchers, scientists and clinicians, have been translated into clinical care. There have also been nation-leading improvements in the model of service delivery for ultra-rare, genetic diseases, which would previously have remained undiagnosed. Similarly, WA has led the way in its novel approach to screening and clinical service provision for familial hypercholesterolaemia, a genetic disease that predisposes people to high cholesterol and increased risk of heart attacks at a young age.

WA has also made important advances in donor-recipient matching for bone marrow and organ transplants using genomic testing, has been an important jurisdictional partner in gathering evidence for carrier screening and has been fortunate in having prominent researchers lead the bench-to-bedside development of several promising gene therapies. There will be additional opportunities to build on WA's strengths in health genomics research and innovation with the introduction of the Future Health Research and Innovation (FHRI) Fund, which recognises the need to support new or existing infrastructure (such as 'omics' technologies) to elevate WA as an international leader in research and innovation fields (12).

Prominent WA projects and plans such as the adoption of cloud services, and the intent to progress towards state-wide electronic medical record functionality, provide additional opportunities to strengthen health genomics services in WA. These projects recognise there is an imperative to transition WA towards a uniform, digitally-connected and enabled health system. This will be a critical piece of work for health genomics, given the large volumes of data produced, analysed and stored as part of genomic testing.

## Appendix 4 – References


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