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

Developing a genomics strategy for the WA health system

Discussion paper

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Contents

Part 1: A system-wide strategy for genomics

A system-wide strategy (the Strategy) has been developed to set a clear, shared vision for the future of the application of genomics in the Western Australian (WA) health system. The Strategy, currently in draft form, outlines the optimal and appropriate use of genomics within the WA health system to benefit the health and wellbeing of all Western Australians. The Strategy provides leadership and direction, focuses on collaborations and partnerships and builds upon current exemplars of genomics in the WA health system. Aspirations for short, medium and long-term horizons have been identified, which will be accompanied by iterative implementation plans to enhance the utility of the Strategy.

1.1 Strategy governance

In the role of System Manager, the Office of Population Health Genomics (OPHG) at the WA Department of Health guided the development of the Strategy and will lead its implementation.

A WA Health Genomics Strategy Advisory Committee was established to provide oversight to development of the Strategy. This group is comprised of representatives from the Department of Health and health service providers (HSPs), and subject matter experts across a range of fields relevant to genomics.

1.2 Strategy need

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.

1.3 Strategy scope

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.

The Strategy is limited to genomics-related activities that occur within, or are facilitated by, the WA health system, which consists of the WA Department of Health, the Child and Adolescent Health Service, four geographically defined HSPs (North Metropolitan Health Service, South Metropolitan Health Service, East Metropolitan Health Service and WA Country Health Service) and three state-wide services (PathWest Laboratory Medicine WA, Health Support Services and the Quadriplegic Centre). However, we acknowledge that the primary and private health sectors and private industry play a significant role in the provision of genomic services within WA. As

such, we will seek to better understand the impact of these services on the WA health system as well as identify potential opportunities for partnerships that benefit WA patients.

The key people and services involved in the delivery of genomics within, or facilitated by the WA health system, include:

- Health consumers, carers and their families;
- Clinical geneticists and genetic counsellors, such as those employed by Genetic Services of WA (GSWA), WA's state-wide publicly funded clinical genetics service;
- Health professionals (including General Practitioners (GPs) and medical specialists) other than genetics specialists, who facilitate access to genomic tests, either through referral or direct ordering of tests;
- PathWest Laboratory Medicine WA (PathWest), WA's publicly funded pathology provider, which has multiple departments providing genomic testing including Diagnostic Genomics, Anatomical Pathology, Microbiology, Haematology, Immunology and Biochemistry; and
- Other test providers, such as private laboratories in WA, interstate and overseas. Testing through these laboratories can be accessed via PathWest, directly by health professionals and in some cases also by health consumers (direct to consumer testing).

For more detail on the people and services involved, please refer to Appendix A. For more detail on the types of tests that fall within the remit of the Strategy, please refer to Appendix B.

1.4 Strategy development

Between 2019 and 2020, the OPHG undertook a comprehensive scoping exercise in order to: 1) identify whether a Strategy was considered useful by stakeholders; 2) understand the current state of the application of genomics in the WA health system; and 3) begin to explore the 'future state' of genomics. This scoping exercise included:

- An analysis of national and state genomic strategies or plans from within Australia and internationally and a review of recent investments and initiatives in clinical genomics and genomic research within Australia and internationally;
- Literature reviews on potential priority areas and emerging trends in genomics;
- 44 interviews with a range of local stakeholders including healthcare professionals, laboratory staff, medical researchers, consumer representatives and people with lived experience with genomic testing. These were held from September to December 2019;
- Discussion with senior staff in Directorates of the WA Department of Health and clinical service planners within HSPs. These were held from October to December 2019; and
- Consumer engagement activities, which included an online survey and in-depth telephone interviews with consumers (including carers and family members) to better understand the lived experience of those who have had genomic testing in the past in WA.

These initiatives identified that stakeholders do consider a Strategy to be useful, particularly if it addresses the following outcomes:

- Promotes genomics as a priority for the WA health system;
- Demonstrates that investment in genomics is required;
- Provides for the equitable integration of genomics in the WA health system;
- Brings visibility to what the different stakeholders contribute to genomics;
- Identifies key local strengths, needs and opportunities for improvement; and
- Leverages what has been learned nationally and internationally.

Stakeholder perspectives of the current and future state of genomics in the WA health system have been summarised into key themes and included in Parts 3 and 4 of this discussion paper.

The evidence and insights gained from this scoping phase were used by the OPHG to write the initial draft of the Strategy with input and oversight from the Advisory Committee. Horizons of implementation and priority action areas for the first two years were also developed, informed by a process mapping workshop undertaken with the Advisory Committee and informal consultation with key stakeholders as required.

The initial draft of the Strategy and high priority action areas for the first two years of Strategy implementation is currently open for consultation via citizen space. The feedback received will inform the completion of the final Strategy document and development of an accompanying implementation plan for the first two years, which will then be endorsed by the WA Health Genomics Strategy Advisory Committee. The Strategy and implementation plan will then be tabled for approval by the Health Executive Committee (consisting of the WA Department of Health's Director General and four Assistant Director Generals, and all Chief Executives of WA HSPs).

Part 2: The policy context

The current and future potential benefits of genomics are being increasingly recognised. There are a growing number of countries (e.g. England, France, Germany) formulating policy and research programmes to support the development of genomic medicine and the adoption of more personalised approaches to healthcare. In Australia, genomics is also on the national agenda, as evidenced by the allocation of $500 million over 10 years to the Commonwealth's Genomics Health Future Mission and the release of the *National Health Genomics Policy Framework 2018-2021* (the National Framework).

2.1 National Health Genomics Policy Framework

In 2017, the first health genomics policy framework for Australia was released. The National Framework was endorsed by the Council of Australian Governments Health Council and acknowledges the increasing potential of genomics to help people live longer and better through appropriate access to genomic knowledge and technology to prevent, diagnose, treat and monitor disease. The National Framework provides a shared direction and commitment between all governments in Australia to consistently and strategically integrate genomics in the Australian health system.

2.1.1 National strategic priorities

The five strategic priority areas identified in the National Framework are:

1. Person-centred approach – delivering high quality care for people through a person-centred approach to genomics.
2. Workforce – building a skilled workforce that is literate in genomics.
3. Financing – ensuring sustainable and strategic investment in cost-effective genomics.
4. Services – maximising quality, safety and clinical utility of genomics in healthcare.
5. Data – responsible collection, storage, use and management of genomic data.

Oversight for the implementation of the National Framework is provided by the Project Reference Group on Health Genomics (PRG). The PRG includes representation from each state and territory. It is a forum for open dialogue and aims to promote collaboration across jurisdictions on major issues, providing opportunities to align efforts and minimise duplication. The PRG currently oversees several initiatives related to the implementation of the National Framework, including those investigating a sustainable genomic workforce, a national approach to genomic information management, nationally consistent templates and guidance for informed consent to genomic testing, and the current Commonwealth, state and territory legislative and regulatory landscape in the context of current and emerging ethical, legal and social issues related to the collection and use of health genomics information.

The WA Government has committed to supporting the National Framework and its associated implementation plan. The system-wide Strategy for genomics in the WA health system builds on and aligns with the National Framework's five interdependent and interlinked strategic priorities, while taking into consideration local context and needs.

2.2 National Microbial Genomics Framework

In April 2019, the *National Microbial Genomics Framework 2019-22* was endorsed by the Australian Health Protection Principal Committee. The purpose of the framework is to provide a consistent, national, and strategic view for integrating microbial genomics into the Australian health system and for identifying microbial genomics policy issues and challenges that need to be addressed.

The framework aims to support better coordination and consistency of action between Australian laboratories and public health units to ensure the potential benefits of microbial genomics are harnessed in an efficient, effective, ethical and equitable way.

The framework's vision is to protect the health of all Australians from communicable disease and biological agent threats (including foodborne and environmental) through access to microbial genomics information and technology; and, through responsible data sharing, to promote its use for routine surveillance.

2.3 Other national genomics initiatives

WA health and policy professionals and researchers are important partners in several innovative, national genomics programs including the Australian Genomics Health Alliance, Mackenzie's Mission, the Australian Genomic Cancer Medicine Program and the Better Indigenous Genetic Health Services project. At the international level, WA is part of multi-national collaborations involving genomics, such as those aimed at unravelling the molecular diagnosis of rare genetic diseases.

2.4 Genomics strategies in other Australian jurisdictions

Several Australian jurisdictions (e.g. New South Wales, Queensland, Victoria and South Australia) are developing or have developed health genomics strategies or plans. These strategies, while taking different approaches, essentially cover similar priority areas to the *National Health Genomics Policy Framework 2018-2021*. Some key issues identified include:

- Community awareness and engagement the community needs a level of genomic competency that enables informed choices about genomics and healthcare; and must be consulted, particularly regarding the ethical, legal and social issues associated with genomics; public trust and confidence in genomics needs to be maintained.
- The patient journey: must be person-centred and provide equitable access to genomics for all; informed choices must be supported; there must be a clear pathway through the health system for patients who require genomics in healthcare.
- Clinical genetics services: must be safe, efficient, cost-effective, have equity of access for health consumers, and be appropriately funded; which may necessitate new service delivery models.
- Laboratory services: applications of genomics must be safe and in line with relevant regulatory guidelines and policies; development of genomic tests must be based on evidence about outcomes such as clinical validity and utility; there is a need for increased capacity and capability to ensure genomics is safely and fairly included in routine healthcare; a need for clear guidelines around when genome sequencing is government funded.
- Workforce: training and education to further build a skilled and genomic-literate workforce is required, with increased capacity and capability in areas such as bioinformatics; a formal workforce needs assessment is required.
- Data and information and communications technology (ICT): The collection, storage, use and management of genomic data must be responsible, effective and appropriate; must have the data/ICT infrastructure and digital connectivity required to enable genomics in healthcare.
- Research: need to grow the knowledge that will enable the use of genomics in healthcare; a focus should be translational research.
- Ethical, legal and social issues (ELSI): need for community engagement, awareness-raising and research on the ELSI associated with genomics.

Multiple states have invested in coordinated programmes or alliances focused on accelerating the building of evidence to support the adoption of genomic medicine into standard clinical care. An example is Queensland Genomics, where a $25 million investment by the Queensland Government is enabling progress in these domains:

- A workforce that incorporates genomics into healthcare;
- An evidence base for clinical genomics;
- Timely and cost-effective diagnostic workflows;
- Use of genomic sequence results to benefit patients;
- Public awareness and understanding;
- A system for managing clinical genomic data; and
- Accelerated research translation.

2.5 Sustainable Health Review

The Sustainable Health Review (SHR) was undertaken to guide the future direction of the WA health system. In 2019, the *Sustainable Health Review: Final Report to the Western Australian Government* (the SHR Report) was released. The SHR Report outlines eight Enduring Strategies to progress the sustainability of the WA health system, which are:

- Strategy 1 – Commit and collaborate to address major public health issues.
- Strategy 2 – Improve mental health outcomes.
- Strategy 3 – Great beginnings and a dignified end of life.
- Strategy 4 – Person-centred, equitable, seamless access.
- Strategy 5 – Drive safety, quality and value through transparency, funding and planning.
- Strategy 6 – Invest in digital healthcare and use data wisely.
- Strategy 7 – Culture and workforce to support new models of care.
- Strategy 8 – Innovate for sustainability.

Under Strategy 8, one of the priorities in implementation for Recommendation 28 is: "*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".*

The Strategy for genomics in the WA health system embraces the opportunities that the SHR Report identifies across the Enduring Strategies.

2.6 WA Health Digital Strategy 2020-2030

The WA Health Digital Strategy 2020-2030 (the Digital Strategy) was developed to take advantage of the innovations transforming healthcare to drive better health outcomes in WA. With person-centeredness at its core, the Digital Strategy has six strategic themes:

1. Empowered consumers
2. Informed clinicians
3. Optimised performance
4. Supported workforce
5. Enhanced public health
6. Embedded innovation and research

Genomics is of particular relevance to strategic themes 3, 5 and 6, especially as the field of precision health (and its related technologies) is anticipated to grow over the life of the Digital Strategy.

2.7 Precision health initiatives

Precision health is an emerging opportunity for the WA Government. In addition to the focus on precision health in the SHR, in April 2019 the WA Minister for Health, the Hon. Roger Cook MLA, announced the establishment of the WA Minister for Health's Council on Precision Health (the Ministerial Council). This Ministerial Council will advise the WA Department of Health on opportunities to further develop and support precision health advances in the WA health system, including identifying enablers for and possible barriers to collaborations that will deliver more effective, efficient, equitable and sustainable healthcare, including in the field of genomics.

Part 3: Proposed key priority areas for the Strategy

The following section outlines key priority areas for the Strategy, which align with the priority areas in the National Framework, with the addition of research and innovation. These are:

- A person- and family-centred approach;
- A workforce that is genomics literate;
- Sustainable and strategic investment in genomics;
- High quality of genomics services;
- Appropriate data considerations and infrastructure; and
- Research and innovation.

Within each priority area, information is provided on a range of needs or issues identified by stakeholders in relation to the current state of the application of genomics in the WA health system. The Strategy promotes a shared vision that will address the issues raised.

3.1 A person- and family-centred approach

Making sure that health consumers are involved in, and central to, their own care is a key component of delivering high quality healthcare. This includes healthcare that is related to the application of genomics. It is important that the experiences, needs, preferences and priorities of people who use genomics inform health system decision-making, including the design of services.

3.1.1 Inequity of access

Consumer access to genomic testing in the WA health system is dependent on who their health professional is. GPs and medical specialists are the primary source of referrals for genomic testing. Currently some health professionals are more likely than others to recommend genomic testing for their patients. Health professionals have varied levels of understanding about genomics and its associated health applications, which can influence their confidence in ordering tests, interpreting results and providing genetic counselling. Further to this, there is a perception that certain hospitals or departments are more engaged than others in promoting access to genomic testing. These factors contribute to an inequity of access for health consumers.

However, a notable exception is somatic mutation testing for cancer patients. Anatomical Pathology within PathWest provides a centralised, state-wide service for pathological diagnosis and prognosis of cancers for all public patients in WA including genomic testing. Decisions regarding which tests should be routinely performed for each cancer type are made by experts in the field consisting of a multi-disciplinary team of molecular pathologists, pathologists and oncologists. As such, which testing occurs, and in which order, is not reliant on a patient's clinical referral.

Some population groups are more likely than others to have inequitable access to genomics in the WA health system. Those that face greater challenges in accessing genomic tests include Aboriginal people, people from culturally and linguistically diverse backgrounds, and those who cannot afford to pay out-of-pocket expenses for genomic testing. People living in rural and remote areas are also likely to be experiencing a disparity in their exposure to health genomics as most medical specialists (particularly those with expertise in genomics) are based in the Perth metropolitan area.

Telehealth is one potential solution to reduce the inequity of access for those living outside of the metropolitan area, but it can be resource intensive and funding of this service delivery model needs improvement. Finally, some tests are not publicly funded (e.g. non-invasive prenatal

testing) and must be paid out of pocket by the consumer, which is not be economically feasible for some people.

3.1.2 Need for clear pathways

Having a clear pathway to genomic testing is important for managing the quality of healthcare provided to consumers and improving health outcomes. To cover the entire span of a person's journey in the health system, including primary and specialist care, and ensure continuity of care, suitable pathways are needed to define aspects such as:

- How should genomic testing be accessed?
- What referrals should be required?
- Who should be able to order different genomic tests?
- Who can be offered certain genomic tests?
- In which situations should testing be offered?
- Who is responsible for pre- and post-test genetic counselling?
- Where will different genomic tests be performed?
- What are the optimal turnaround times for various genomic tests?
- Who provides the test results and post-test management?
- What happens to the genomic data post-test?

Ideally, a clear pathway should exist and be communicated to the consumer at the start of their journey. However, at present, genomic medicine pathways are not very clear, to either health consumers or healthcare providers. The exception to this is in clinical areas that have long-established local capabilities in genomics, such as haematology and neuromuscular diseases. Yet these capabilities may rely on person-specific expertise, which is not sustainable in the long-term.

3.1.3 Need for information and support

Health consumers must be enabled to make informed decisions about the use of genomics in their healthcare. This can occur through the provision of the right amount of information and support from the health professionals about genomic tests, including provision of information to take away and refer to after an appointment. There are some existing sources of genomic-related information and education for the public (e.g. The Centre for Genetics Education in New South Wales), but a pressing issue is how to connect people (including patients, families and health professionals), to these resources at various stages of their journey. Health professionals need access to up-to-date and easily accessible resources to assist health consumers in making informed decisions. Within appointments, clinical geneticists and genetic counsellors would like to be able to access online resources on portable devices such as tablets to help with providing information to health consumers. This option is presently limited for public genetics services in WA, due to a lack of wireless internet access.

3.2 A genomics-literate workforce

Having a skilled workforce with the competencies to appropriately deliver genomics in the WA health system is critical to high-quality services and improving health outcomes for health consumers. Current workforce (and skill) shortages are a major concern for stakeholders, with the need for education and training considered critical.

3.2.1 Increasing demand amid a workforce shortage

In recent years, the demand for genomic testing and services has increased significantly, for example approximately 10% per annum for GSWA and Diagnostic Genomics, and 25% per annum for Molecular Anatomical Pathology. This demand is likely to increase further, with both the increased utility of genomic testing and the introduction of a Medicare items, such as an item for whole exome sequencing of children with suspected monogenic diseases. This item requires consultation with a clinical geneticist, effectively increasing the clinical geneticist (i.e. GSWA) workload. There has also been an increase in complexity of testing and associated counselling. However, the workforce to deliver these services has not increased at a rate consistent with this increased demand, placing a strain on existing staff as they attempt to keep up with demand.

There are long wait times to see a clinical geneticist and/or genetic counsellor and result turnaround times for many tests are also considered too long by health consumers, health professionals and laboratory staff. This will be exacerbated by the expected future increase in demand for testing and/or genetic counselling, which is being driven by factors such as population growth, increasing clinical and personal utility of testing for certain diseases, the increasing role of genomics in the standards of care, patient demand, and direct-to-consumer genetic testing. This will further compromise the timely, sustainable and equitable provision of services.

3.2.2 More pathways for education and upskilling

The broader healthcare workforce needs to be literate in genomics, including medical students, general practitioners, medical specialists, nurses and allied health staff, particularly as the application of genomics becomes more mainstream. Some health professionals interviewed said that they have undertaken some degree of training in genomics, but it was thought that the amount of genomics education most receive is insufficient. Currently, there are limited education and training pathways in WA to support workforce capacity building in genomics and continuing professional development. Funding limitations and geographic isolation are some of the barriers in relation to accessing further learning opportunities; however, virtual attendance is one possible solution to this problem. Another solution is “on-the-go” education built into hospital digital health platforms, such as electronic test ordering. Formalised networks of knowledge transfer may also be helpful. Adequate and effective avenues to enable upskilling need to be increased as the landscape of genomics is changing rapidly. Keeping up to date with the latest evidence and tests is important for delivering the best possible care.

A key contributing factor to the skills shortage in the laboratory setting is the absence of a formalised training pathway (or upskilling and promotion of existing scientists) in data analysis, interpretation, report writing and bioinformatics, despite the field being recognised as a critical component of genomic testing. Training for certain genomics positions within laboratories relies on direct supervision from a suitably- (often medically-) trained person with the appropriate skills and experience (e.g. Fellowships in Genetics from the Human Genetics Society of Australasia and the Royal College of Pathologists of Australia (RCPA); Fellowship in Genetic Pathology, RCPA).

In clinical genetics, there are only two trainee clinical geneticist positions offered in WA each year and these are dependent on non-recurrent funding. Lack of sustainable funding has contributed to the risk of shortages in the clinical geneticist workforce. Further to this, there are no courses in genetic counselling offered in WA, which creates a dependence on other states to produce qualified genetic counsellors. Even then, the number of new graduates is low, at approximately 40 per year.

Other factors contributing to the shortage in genetic counsellors include the current limited number of positions available at GSWA, competition with the research sector for new graduates, and lack of recognition of genetic counselling as a specialist health profession (and consequently inadequate remuneration for the work they do). Training of nursing and allied health staff in particular aspects of genetic counselling (e.g. micro-credentialing), may be one way to complement the current and future genetic counselling workforce for certain aspects of healthcare involving genomics.

3.2.3 Expanding the role of genetic counsellors

The involvement of genetic counsellors is required across primary care, private practice and different medical specialties, including roles in educating and advising clinicians, counselling health consumers, and participating in multidisciplinary teams. Genetic counsellors are seen to have greater capacity than medical specialists to explain “basic genetics” and “basic inheritance patterns” to health consumers, improving health literacy and ensuring the implications of genetic conditions are understood for a person and their family. Therefore, it has been suggested that there could be an expanded role for genetic counsellors within hospitals as well as more direct access for medical specialists to genetic counsellors within GSWA. This would support medical specialists and facilitate their direct access to genomic testing, rather than always needing to refer to GSWA.

3.2.4 Crucial need for a bioinformatics workforce

Genomic testing is creating new challenges for the health workforce, requiring an investment in staff training not only at a technical level in the wet laboratory, but also the training of staff to be able to interrogate large datasets and appropriately integrate complex results in a clinical setting. DNA sequencing using next generation sequencing technologies (also known as massively parallel sequencing) can generate huge amounts of data, which require interpretation. Consequently, there has been a rise in the need for computational and bioinformatics capabilities in laboratories. PathWest has experienced challenges in recruiting staff with the appropriate skills in genomics, particularly in bioinformatics. Globally, there is a shortage of people with the necessary disease-specific knowledge and experience in next generation sequencing methods and the associated data analysis and interpretation. There is an opportunity to address the need for bioinformatics expertise by creating clearer pathways to obtain bioinformatics skills. Such pathways are currently limited in Australia, contributing to the bioinformatics skills shortage.

3.2.5 Opportunities to build on existing relationships and networks

Strong working relationships have developed organically between GSWA, PathWest and some medical specialists and these are perceived as a strength of the current state of genomics in the WA health system. These relationships have enabled collaboration and information sharing between these parties, particularly when it comes to developing new tests, interpreting test results and communicating the implications of genomic testing to health consumers. Not all health professionals have yet developed such relationships with GSWA and PathWest, and this has contributed to a fragmented genomics network in the WA health system. Those who perceive that they currently do not have strong links with GSWA and PathWest would welcome the opportunity to develop closer ties, and this is particularly true for those clinicians who are

new to WA or new to genomics. This will help ensure equitable access to genomics for all medical specialties in the health system, and thus for all health consumers who could benefit from genomic testing.

3.3 Making sustainable and strategic investments in genomics

Financing has been identified as a key enabler to the delivery of high-quality services, to ensure the genomics workforce is appropriately sized and skilled and has access to the required data, ICT and other infrastructure. Key themes arising in relation to sustainable and strategic investments in the application of genomics in the WA health system are:

- Genomics should be a priority for investment;
- Dedicated, sustainable funding is required for genomic tests;
- Investment is needed in clinical genetics services, laboratory infrastructure, bioinformatics and translational research;
- Multi-sector partnerships should be explored; and
- Costs associated with genomic tests and therapies must be considered.

3.3.1 Genomics should be a priority for investment

The WA Minister for Health, the Department of Health and HSPs should support investment in genomics in the WA health system. Genomics has the potential to reshape clinical practice by providing the opportunity for more precise and tailored healthcare. The ethos at the top needs to be that genomics is a priority. Decision-makers need to be aware of the effect in the future if investment is not made now to increase capability, capacity and equity of access in genomics. To increase the visibility and priority given to genomics, a local evidence base is needed that demonstrates the value proposition of genomics to the WA health system. Published studies internationally confirm, and local anecdotal evidence suggests that genomic testing can have clear benefits for health consumers, reduce duplication and waste, and can be cost-saving and cost-effective.

3.3.2 Dedicated, sustainable funding of genomic tests

Clear policies at multiple levels and dedicated, sustainable funding is needed to facilitate GSWA, hospital departments and general practitioners ordering appropriate genomic tests. Within the WA health system, there has been a lack of systematic and strategic investment in the funding of genomic tests. This is compounded by only a small proportion of the genomic tests being reimbursable through Medicare and that under the National Health Reform Agreement public hospitals cannot claim Medicare rebates for pathology tests performed for public patients as part of a hospital admission or outpatient clinic, meaning that most genomic tests are paid for by the WA public health system.

Historically, the costs of genomic tests have either been absorbed by PathWest, or paid for by GSWA, or by the health service in which the medical specialist requested the test. The current approach is problematic as designated funding for genomic testing either does not exist or is insufficient. An appreciation is needed among funders that these tests can often be crucial in providing the best standard of care for health consumers and are likely to produce cost-savings and cost-effectiveness for the health system.

The inability to pay for tests is highlighted as the biggest issue for many health professionals and impacts quality of care, in that health professionals are not requesting all the genomic tests that they consider could be beneficial for their health consumers due to cost constraints. This problem will be exacerbated as the demand for genomic testing increases. When clinicians within the WA health system do not have the means to fund certain genomic tests, health consumers are sometimes electing to order these tests through private companies and paying

themselves if they can afford it. Thus, health consumers are not getting equity of access to genomic testing.

3.3.2 Investment in clinical genetics services

A more appropriate funding model is required for the delivery of clinical genetics services in WA. As with many other hospital-based services in WA, GSWA is predominantly funded through Activity Based Funding (ABF). However, the ABF model under-represents the complexity of providing genetic services and the high cost of emerging and new genomic tests and as such GSWA faces ongoing budget pressures. A clinical geneticist can see significantly fewer health consumers per week than other specialties, due to the current and increasing complexity of cases; and ABF doesn't sufficiently consider the amount of work that geneticists and genetic counsellors need to do outside of clinic appointments. In recognition of this, the WA Department of Health has historically provided GSWA with annual block funding, but this has not grown in line with the consumer price index, or in line with the dramatic increased demand for GSWA services. The gap between what GSWA needs financially to operate its services and what it is funded to do is growing year by year.

GSWA is a state-wide service, yet it is based within a geographically defined HSP, namely North Metropolitan Health Service. This creates challenges in relation to the provision of genomics services to other HSPs, including clinical geneticists and genetic counsellors participating in multidisciplinary clinics at hospitals in other HSPs. However, there is presently uncertainty as to where the funding for these clinics should be coming from, including which HSP can claim the activity for ABF. Other clinics across the health system find themselves facing similar challenges. For example, the neurogenetics clinic at Royal Perth Hospital, which provides a state-wide service from within the East Metropolitan Health Service. Currently there is no WA Department of Health policy and guidance relating to the governance and operation of state-wide services being delivered by HSPs.

3.3.3 Investment in laboratory infrastructure and bioinformatics capability

Funding is required to enable WA laboratories to perform all types of genomic testing deemed appropriate for the WA health system, and to match demand in terms of test quantities and timeliness. PathWest is currently funded through a mixture of fee-for-service activities (including consumer charges and service level agreements with each HSP), external revenue and research funding, and block funding provided by the WA Department of Health via a service level agreement. This service level agreement is high level and does not include detailed information about the individual services purchased from PathWest, and it does not specifically mention genomic testing.

The budgets provided internally to each department within PathWest are largely historical and senior departmental executives reported funding constraints that have hindered their ability to meet the increased clinical demand for genomic services. In particular, the ability to access funding for equipment and to employ new staff were recognised as significant problems that limited the growth and diversification of services. To help overcome these funding constraints, external revenue sources have been sought, including research funds and provision of testing for health organisations in other jurisdictions or pharmaceutical companies.

Strategic investment in laboratory equipment, including DNA sequencing instruments and robotics, is required to increase the capacity of PathWest to deliver genomic sequencing, particularly higher-throughput massively parallel sequencing including whole exome and whole genome sequencing. A centralised sequencing service for the WA health system with all samples for genomic testing being directed to PathWest for processing, or subsequent referral to designated external laboratories, is considered to be advantageous. This is for multiple

reasons, including clear referral pathways, equity, data storage and consistent data interpretation, and obtaining trusted results.

Some interviewees suggested that rather than purchasing new DNA sequencing equipment to meet growing demand for genomic testing, PathWest could develop partnerships with research institutes to utilise existing sequencing infrastructure that is currently underutilised. However, for this to occur, there are several challenges, including differences in regulatory standards for genomic testing in the routine clinical service environment versus in research settings, and accreditation requirements of the National Association of Testing Authorities and National Pathology Accreditation Advisory Council. These challenges would need to be overcome to allow test results obtained using research equipment in research laboratories to be used in a clinical setting. Also, experience in other states supports that this model could be more expensive, prevent capacity building, have significant sustainability challenges and pose the risk of sudden loss of genomic testing services, as compared to provision of appropriate genomics infrastructure within the WA health system.

There is also a need to invest in further capacity in high-performance computing, bioinformatics platforms and analytical pipelines compatible with the clinical use of genomics. As such, there may also be merit in considering a centralised core bioinformatic service in WA, e.g. at PathWest, which is secure and has appropriately addressed the ethical, social and legal issues related to genomic analysis, including reporting of incidental findings and data re-analysis.

3.3.4 Investment in innovation and translational research

There is a need for further investment in innovation and translational research. Many health professionals expressed that it is challenging to conduct and translate research without appropriate funding. It is acknowledged that the success rate for grants through the National Health and Medical Research Council (NHMRC) is low in WA and thus novel funding mechanisms, such as the WA Government's Future Health Research and Innovation Fund, should be considered to maximise the potential of translational genomics research within the WA health system.

3.3.5 Multi-sector partnerships should be explored

The involvement of philanthropic funding in genomics in the WA health system was very welcome and giving incentives to philanthropists to invest could be useful. Philanthropic contribution made most sense to stakeholders in the areas of innovation and research, particularly translation of research into clinical practice, clinical trials, drug repurposing and the development of gene therapies. Funding could be for researchers both inside the public health system (e.g. for clinicians to do research) and outside (e.g. to researchers in medical research institutes).

For more routine testing, there could also be a role for philanthropy, for example such funding could be used to build capacity and capability of clinical services. This could include investment in infrastructure and technology such as sequencing equipment, robotics, data storage, artificial intelligence, new models of genomic education, and digital health, or in funding staff positions within GSWA or PathWest. Models of funding include public-private partnerships, for example, government funding to match donations made by philanthropists.

It was suggested that the health system could partner with a range of private organisations including pharmaceutical, medical technology, digital health, bioinformatics companies. For example, partnerships might facilitate access to equipment and set up pipelines for genome sequencing. These partnerships could be used for demonstration projects, such as collecting evidence that genome sequencing delivers better diagnosis and provides cost-savings to the health system.

However, in general there was less support for the involvement of private industry compared to philanthropy. Issues raised included access to data/genomic information and privacy if private companies were involved.

3.4 Maximising the quality of genomics services

The delivery of genomics-related services is evolving, and more and more health professionals are wanting to access genomic testing for their patients. There are, however, perceived barriers to this, including: lack of access to local genomic testing; lack of clear pathways for the setup of new genomic tests; clinicians not being up-to-date on what tests are possible and available, and when to perform certain tests; lack of access to genetic counsellors; and not having an established relationship with GSWA and/or PathWest between that clinician/their department. These issues are addressed in the key themes that arose in relation to genomics services, which are:

- The rise of contemporary models of service delivery;
- Greater local, equitable access to genomic tests;
- More guidance on how and when to order genomic tests;
- Expanding the role for genetic counsellors; and
- Building on existing relationships.

3.4.1 Contemporary models of service delivery

Stakeholders identified two key emerging models of service delivery for genomics in the WA health system. These were “mainstreaming” genomics and multi-disciplinary clinics.

Mainstreaming genomics is a concept that refers to the integration of genomics as part of routine care, facilitated by the increased use of genomics by all health professionals. Mainstreaming could be a useful way to address the increasing demand for genomic testing, but there should be a level of gatekeeping or regulating as to which tests are made readily accessible to non-clinical geneticists. Less complex tests may be more appropriate to mainstream, but more complex tests (e.g. whole exome sequencing, whole genome sequencing, tests that provide predictive risk of disease) should ideally be ordered by a clinical geneticist or a limited subset of specialists who are well aware of the tests’ benefits, harms, risks and limitations.

Mainstreaming is already happening in some parts of the WA health system, particularly in haematology and neurology. Within these medical specialties, genomic testing and genetic counselling is provided as standard care and has been for many years. This was achieved through forging strong partnerships in the field and the involvement of world-leading clinicians and researchers who applied their expertise locally.

In other parts of the health system, however, ordering of genomic tests by health professionals is patchy and fragmented. This is partly attributed to the personal knowledge and level of interest a practitioner has in genomics, including how well connected they are to staff at PathWest and/or GSWA. Other times, it is because practitioners have appropriate knowledge (e.g. that a gene mutation can be detected by another test, or that the test results would not make any difference to the diagnosis, prognosis or management decisions for a health consumer) that a genomic test is not performed. For health consumers, this means that their access to genomic tests and the appropriateness of the test they are offered, varies depending on which health professional they see.

Multidisciplinary clinics are viewed as the ‘ideal’ service delivery model for genomics, being more efficient and cost-effective than other service delivery models. Some clinics have already been established for a few medical specialties in WA and these most commonly consist of a

clinical geneticist, a genetic counsellor, and at least one health professional from a specific clinical area. The purpose of each clinic includes the review of whether a patient should be offered genomic testing and, if so, either the clinical geneticist or the health professional will request a test through PathWest. When the test results come back, they are discussed at the clinic to interpret the findings with the multiple experts who are present.

There is a belief that health professionals participating in multidisciplinary clinics become the genomics experts in their given discipline, and as such gatekeepers to genomic tests, particularly for health consumers with more complex disease presentations (or phenotypes). There is acknowledgement by those involved in multidisciplinary clinics that for health consumers whose situation is more straightforward, all health professionals should be able to request the test directly, rather than referring on to a multi-disciplinary clinic.

There is a strong sentiment that the genetic counsellor is key to the success of these multidisciplinary clinics. They play a major role in coordinating the meetings and gathering the information that informs the review. However, there have been difficulties associated with the funding and physical location of these clinics.

3.4.2 Local, equitable access to genomic testing for health professionals

Health professionals need to have timely access to appropriate and dependable genomic testing from trusted and reputable laboratories. GSWA and some medical specialists indicated that they do not routinely order certain genomic tests, largely because these tests are not performed locally by PathWest and/or funding is not readily available. There have been difficulties in accessing some genomic tests from external laboratories and medical specialists feel unguided trying to find an appropriate test provider themselves. Therefore, most health professionals want the genomic tests relevant to them and their patients to be available at PathWest. PathWest laboratory staff support this preference.

Having a wider range of in-house massively parallel sequencing tests, such as whole exome and whole genome sequencing, would build WA health system genomics capacity. This would support high quality interpretation and facilitate interaction between the laboratory staff and health professionals to discuss and decipher genomic test results. Additionally, health consumers would be more likely to get the right tests at the right time. However, some health professionals indicated that they are likely to continue accessing external laboratories for these tests, where these tests have faster turnaround times and may be perceived to be cheaper than PathWest. Building capacity in PathWest would ameliorate the concerns underlying these requests.

Health professionals should have equitable access to the genomic tests offered by PathWest. They should have equal opportunity to request and order tests, and to have new tests developed by PathWest that are relevant for their specialty area. There should be clear pathways for how and when new tests can be established. It has been suggested that any current shortcomings in local and equitable access are a resource issue and that more investment should be made into PathWest, particularly in terms of their access to sequencing machinery and workforce to undertake data analysis and interpretation.

3.4.3 Need for guidelines and keeping up to date

Health professionals should have access to guidance and a clear pathway for how and when to order a genomic test. Guidelines that provide this information would be useful and would contribute to equity of access to tests for requesting clinicians. Further, many practitioners have identified genomics as a new area of medicine that creates an ‘information overload’ that makes it difficult to keep up to date. There is a need to have access to a good quality evidence base (e.g. a repository) that provides the latest information on genomics relevant to their profession

or area of specialty. Given the rapidity and scale of change of information in genomics, and to ensure sustainability, consideration should be given to how these are integrated in digital health systems and their interoperability with Laboratory Information Systems.

3.5 Data

A key issue with genomic data is the extent to which they should be treated as exceptional and accorded special protection, due to the complex ethical, legal and social issues related to their predictive capability, familial implications, and uniqueness. The vast amount of genomic data collected also increases the risk of privacy breaches and data misuse. Maintaining data security and confidentiality is a significant challenge. From the stakeholder consultations thus far, three key themes have arisen in relation to data. These are the need for:

- Data-related policies and guidance in clinical settings;
- Electronic access to genomic test results; and
- Data infrastructure.

3.5.1 Need for data-related policies and guidance in clinical settings

There is a requirement for guidance or policies on the collection, use and management of genomic data, particularly in the following areas:

Data privacy and overseas providers: Some health professionals are requesting genomic testing directly from commercial providers overseas. It is unclear whether the data privacy policies of international testing providers are examined or whether this information is appropriately shared with the health consumers undergoing testing in a manner that enables informed consent.

Disclosure of genomic test results to family members: The shared nature of genomes means that genomic-related health conditions can also be a family health issue. Clinicians working in the WA public health system are not covered by the national Privacy Act[1] and therefore, have no provision for disclosing genomic information to family members, without consumer permission. Some clinicians have faced ethical challenges regarding their inability to provide genomic test results to known relatives when the person who has been tested is unwilling to disclose this information to their relatives. The WA Government is proposing to introduce state privacy legislation; however, it is unclear whether this legislation will include guidance on disclosure of genomic information to family members.

The implications of genomic results on insurance: In Australia, protection against discrimination by life insurers based on genomic test results is self-regulated by the life insurance industry. A need for national legislation to prevent the use of predictive genomic tests results by insurance companies has been expressed by some. In response to a Parliamentary Joint Committee inquiry into the Australia life insurance industry, in mid-2019 the Financial Services Council announced an industry-regulated moratorium which allows Australians to get up to $500,000 of life insurance without disclosing genomic test results.

In addition, there is a desire for more guidance and support regarding how to appropriately counsel health consumers regarding the insurance implications of test results, particularly in relation to predictive test results and incidental findings.

[1] The federal Primary Act 1988 (Cwlth) permits the disclosure of genetic information to at-risk family members when there is a serious threat to the family members' life, health or safety.

3.5.2 Electronic access to genomic test results

Electronic access to genomic test results across the WA health system should be prioritised where appropriate. Historically, laboratory test results have been provided to requesting clinicians in paper form. More recently, some results for genetic tests (i.e. single gene tests) ordered directly by health professionals have been uploaded by the laboratory to the electronic iSOFT system. However, all genomic test results are still only provided on paper. GSWA does not have access to iSOFT and thus receives all test reports in paper form. It is planned that during 2020, with the introduction of the new Laboratory Information System within PathWest, genomic reports will be available electronically delivered to the requesting doctors.

Health professionals indicated that paper-based reporting and the restriction of genomic test results to one or a limited number of clinicians has significant flow on impacts on clinical decision-making and management. All health professionals interviewed said they would find it useful to access genomic test results electronically, with many recognising that consumer confidentiality must be safeguarded. Sharing of genetic test results ordered by speciality services with the patient's GP (with consent) needs to be prioritised.

3.5.3 Need for data infrastructure

Increased data storage capacity

One of the key enablers for the continued integration of genomic sequencing into the WA health system will be an increase in data storage infrastructure and capabilities that allow the secure storage of large genomic datasets. It is recognised that the most sustainable long-term solution that can provide the elastic scale needed for storing DNA sequences are cloud based solutions. The WA Department of Health is transitioning to a cloud-based ICT system. However, the proposed cost structure is perceived by stakeholders to be prohibitive for the storage of genomic sequencing data.

Data sharing infrastructure

The benefits to be obtained from genomic testing are reliant on data sharing, particularly for the diagnosis of rare genetic conditions. A critical mass of data needs to be available for analysis, to validate the link between variations in the genome and specific health risks, conditions/disorders and treatment responses. Sharing of variant interpretations is needed between clinical laboratories and to genotype-phenotype databases such as ClinVar. PathWest is currently participating in the pilot of the Shariant platform, which enables real-time sharing of expertise and evidence about clinically curated variants between Australian laboratories and clinical services.

There is also the need to share phenotypic data to help guide diagnostic investigations for complex and rare genetic diseases. Clinicians would benefit from a data-sharing platform that allows them to efficiently share a list of standardised phenotypical ontological terms about their consumer. This would increase diagnostic yield and also crucially aid in finding other health consumers globally who have a similar phenotype, and to then learn from the clinical investigations for those health consumers.

Electronic health record to improve clinical genomics

Health professionals should have access to a comprehensive state-wide electronic medical record (EMR) or another way of linking data from various clinical and administrative databases in real time. The WA Health Digital Strategy (2020-2030) has an EMR system as the foundation for many of the digital technologies planned. Benefits of an EMR for genomics include the availability of more comprehensive phenotypic information to assist genomic test analysis for increased diagnostic yield and reduced findings of uncertain significance, as well as the likely prevention of duplication of genomic testing, which has significant potential cost-savings. In the absence of an EMR, and also when an EMR exists, measures to support system interoperability

with new and existing databases, registers (e.g. Western Australian Register of Developmental Anomalies, Western Australian Cancer Registry) and digital health platforms will be critical.

One PathWest department for example, Diagnostic Genomics, currently upload to electronic records genomic test reports (not raw sequencing data) for diagnostic and prenatal tests after six months from when the test results are provided to the requesting clinician. This decision was made in consultation with GSWA, to enable appropriate time for genetic counselling to take place to ensure that results are explained properly and are not open to misinterpretation. The decision to date is that predictive test results will not be uploaded to a patient's electronic records.

More guidance is desired regarding how genomic information could be recorded in a health consumer's medical file, particularly in an electronic form. With regards to the national My Health Record platform, there is no consistent approach across jurisdictions to the inclusion of genomic information, and it has been acknowledged that different strategies may be required depending on the nature of the condition and test result. Consumer preference at an individual level should also be included in these decisions.

GSWA clinical database

A fit-for-purpose and contemporary clinical database is required to support the provision of clinical genetic services in WA. GSWA have a clinical database called Kintrak that is outdated, unsupported and computer- (not web-) based and is not interoperable with other administrative databases such as webPAS. The database has administrative burdens, adds inefficiencies to GSWA's clinical operations and cannot produce the required data for ABF reporting. As GSWA is the state-wide provider of clinical genetics, the GSWA clinical database potentially contains valuable data about the distribution of genetic conditions in the WA population. However, limitations of the current database prevent the use of these data for clinical and research planning purposes.

Collection of standardised phenotypes

The accurate and detailed recording of a consumer's phenotype is essential to appropriately target genomic analysis and accurately interpret genomic variation. Laboratory staff have reported that phenotype data included on genomic test requests are highly variable and that the information provided is often limited, non-specific or irrelevant. As a result, pathologists frequently contact the requesting clinician with the aim of recording a more comprehensive phenotype. Some health professionals feel that specialised skills are required to accurately and comprehensively phenotype health consumers and, in some cases, specialised diagnostic equipment is required. It is recognised that standards and software tools that support and guide appropriate phenotyping are likely to be beneficial.

3.6 Research and innovation

There is significant potential for genomics research to expand in WA. This requires mechanisms to translate innovation and research findings into clinical practice. Local exemplars of this approach (e.g. the development of targeted gene panels for neuromuscular and other disorder groups, and comprehensive genomic profiling of 5 common cancers at PathWest) could be replicated in other medical areas. Also, innovations in clinical service redesign under the Undiagnosed Diseases Program, as identified as the exemplar of Innovate for Sustainability in the SHR, are notable.

At present there appear to be limited local pathways to support the involvement of clinicians in innovation and research translation, with barriers including lack of time, insufficient funding, limited access to DNA sequencing and bioinformatics capability, insufficient data storage, limited uptake of national and international pipelines, and a resource-intensive and lengthy process for ethics approval.

To facilitate increased translation of research findings into clinical practice, clinicians need to be supported to become more involved in genomics research. Therefore, there is a need for hospitals to value genomics research and embed this within a culture that embraces research and innovation. Research that is conducted by health professionals is usually completed in their own time mostly without any compensation.

There are examples of successful research and innovation programs, such as models to allow rapid translation of projects from bench to bed side, that could be considered to enable WA to be competitive in the field of genomics research. Strengthening WA's research and innovation capacity in genomics would also benefit from decisions around which areas research efforts should be focused on, determined by the State's current research strengths and comparative advantages.

Part 4: The future state of genomics

The Strategy identifies opportunities to build on the strengths of the current approach to applying genomics technology in the WA health system. Stakeholders have identified current strengths and what an ideal future state for genomics in the WA health system might look like.

4.1 Building on current strengths

Several strengths already identified related to the current application of genomics in the WA health system include:

- A single, state-wide public pathology provider, PathWest, with a proven track record of delivering high-quality genomic testing across multiple disciplines. This helps avoid duplication of testing and ensures some consistency in the way tests are offered within a public healthcare system;
- PathWest being a world-leading centre of expertise in genomic testing for certain disciplines, such as neuromuscular disorders, adult cancer tumours, donor-organ and recipient matching and undiagnosed rare diseases;
- A single, state-wide clinical genetics service, GSWA, with a proven track record of delivering high quality, appropriate genetic counselling and clinical genetics services;
- Having single providers of clinical genetics and pathology services supports the consolidation of data for both clinical and research purposes;
- A history of strong and productive relationships and collaborations between staff at GSWA, PathWest, medical specialists, laboratory staff and medical researchers;
- State-wide clinics that facilitate genomic testing, such as the neurogenetics clinic and the lipid disorders clinic at Royal Perth Hospital (which provides cascade screening and treatment for familial hypercholesterolaemia).
- An existing data-linkage ecosystem that can partner to clinical and laboratory data sets for unique state-wide, cross-sectional and longitudinal (health economic) impact and health system monitoring studies of the impact of genomic diseases, and methods to improve their diagnosis and care; and
- Having excellent, internationally recognised researchers in WA in the field of medical genomics, for example in the diagnosis of rare diseases.

Opportunities identified to build on these strengths include:

- Ensuring the sustainability of and replicating the success of state-wide genetics clinics, such as the neurogenetics clinic at Royal Perth Hospital;
- Utilising PathWest's genomic testing expertise to attract funding from international pharmaceutical companies as a testing site for clinical trials;
- Building on WA's strengths in cancer genomics and becoming a provider of services for other jurisdictions to attract revenue into the State;
- Building on WA's unique transplant immunogenetics service, which allows HLA typing to be done on the day of transplantation enabling better matching, to be the best kidney transplant centre in Australia; and
- Providing beta testing for new technologies, which WA has a history of doing for international centres.

4.2 An ideal future state for genomics

Stakeholders have identified a range of scenarios that would contribute to an ideal state for genomics in the WA health system in 5 years' time. Not surprisingly, these scenarios largely addressed needs and issues identified with the current state. In five years from now, there is a vision for:

- Improved person-centredness of healthcare related to genomics;
- Health professionals having easy, timely and equitable access to genomic testing, diagnoses and therapies for the people for whom they provide healthcare;
- Funding of genomic tests being transparent and increased, from both the WA and Commonwealth Governments;
- Health professionals being aware of the genomic tests suitable for their patients and having the required competency to provide counselling, interpret test results and report back to their patient;
- Multidisciplinary clinics being expanded across the WA health system to equitably cover the breadth of medical specialties across the lifespan;
- Appropriate funding for activity and workforce to deliver high-quality state-wide clinical and diagnostic genomics services;
- All genomic tests being accessible through one local laboratory and available to rural and remote practitioners, as well as those working in the private setting. This includes in-house capability at PathWest laboratories for clinically relevant whole exome sequencing, whole genome sequencing, non-invasive prenatal testing, preconception carrier screening, and newborn bloodspot screening;
- More equitable and culturally-appropriate access to genomic testing and genetic services, including for Aboriginal people and other people of culturally and linguistically diverse backgrounds.
- Precise genomic classification of tumours for every cancer patient in WA and for patients to obtain coverage for biomarker testing that would enable them to receive targeted therapies.
- Access to diagnostic genomic testing for all people living with an undiagnosed rare and/or genetic disease.
- State-of-the art genomic laboratories, with the necessary infrastructure and skilled personnel to deliver equitable services to all who could benefit from genomic testing, including in relation to data handling, bioinformatics and data storage;
- A digital health platform for managing genomic test results, that is compatible with the information systems of laboratories and hospitals;
- Cross-sector collaboration leading to improved research translation; and
- System-wide guidance and policies to facilitate the equitable, ethical, efficient and effective implementation of genomics.

Part 5: Glossary

Activity based funding: 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.

Bioinformatics: A scientific field where computational tools are used to organise, analyse, visualise, interpret and store biological data from prepared biological specimens, including DNA.

Clinical exomes: The exome is made up of sections of the genome that contain instructions for creating proteins. A clinical exome is a genomic test, which tests for genetic variation in specific regions of the exome that are known to be associated with diseases.

Clinical geneticist: A clinical geneticist is a doctor with specialist training in diagnosing and treating genetic conditions.

Cloud-based: Cloud-based refers to data, applications and resources stored and processed on a network of remote servers hosted on the internet, rather than on local servers or personal computers.

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

Cytogenetic testing: Testing of chromosomes or their structure to identify any changes that would contribute to a diagnosis, treatment and monitoring of treatment of a disease or condition.

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.

Elastic scaling: Allows databases to grow and shrink on demand making it easier for cloud applications to manage multiple levels of data.

Electronic medical record (EMR): Contains information that is created and held within a single healthcare organisation. These are digital versions of the paper charts used by clinicians in clinics and hospitals. EMRs enable providers to track data over time, identify patients for preventative visits and screening, monitor patients and improve the quality of care provided.

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 condition: A medical condition, disease or disorder caused by one or more variations in a person's DNA sequence.

Genetic counsellor: Healthcare professionals who help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of genetic health conditions.

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.

Genome: The complete set of genetic information in an organism.

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 data/information: Refers to the data/information produced from DNA sequencing of a genome (either in part or in full).

Genomics literacy: The knowledge to understand and use genomic information to make informed healthcare decisions.

Genomic testing/genome 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*.

Genotype: The genetic makeup of an organism or individual, which determines a specific characteristic (phenotype) of that organism/individual.

Informed consent: A voluntary decision to accept or decline a test, which is underpinned by sufficient information, including understanding of the benefits and risks, to support an appropriately informed decision.

Inheritance patterns: Describe how genetic variants are distributed in families. Understanding these patterns is crucial to predicting disease risk in family members of an affected individual.

Jurisdiction: A geographic area with a set of laws which are different from neighbouring areas. In Australia, the Commonwealth and each state and territory are separate jurisdictions.

Multidisciplinary: Combining or involving a range of academic disciplines, fields of expertise or professional specialisations.

Next generation sequencing: Refers to latest technology used in genomic sequencing, currently Massively Parallel Sequencing, which can sequence multiple genes rapidly and simultaneously.

Pathogen genomics: The use of genomic sequencing for the identification and surveillance of pathogens in hospitals and the broader community

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

Phenotype: The observable characteristics or traits for an individual, which include physiological, morphological, behavioural and biochemical properties/traits. A phenotype results from the interactions between a person's genetic code (genotype) and the influence of environmental factors.

Sanger sequencing: The first technique for determining the nucleotide sequence of DNA, which was developed by Frederick Sanger and his team in 1977. Capable of sequencing short DNA fragments.

Somatic mutation: A genetic mutation (i.e. variation of the DNA sequence) in a somatic cell. Somatic cells are cells of a living organism apart from reproductive cells.

Variant: A genetic variant is an alternation in the DNA sequence, which can be benign or pathogenic.

Appendix A: People and services involved in applying genomics in the WA health system

Health consumers and carers

Health consumers are referred by a health professional for genomic testing within the WA health system. They may already be inpatients at a public hospital, or they may be referred from the community or private hospitals to outpatient services. A person is only referred for genomic testing if there is anticipated clinical and/or personal utility. That is, the result of the test will be relevant and useful to the person taking it, particularly in relation to the diagnosis, prognosis or management of a condition, or reproductive decision-making. The referring clinician usually makes the final decision on whether a test will be useful based on several considerations including the benefits, risks and perceived value to the individual.

Service providers

There are several service providers who contribute activity related to genomics in the WA health system. These include:

- Genetic Services of WA (GSWA), a publicly funded clinical genetics service.
- Health professionals other than clinical genetics specialists, including general practitioners.
- PathWest, a publicly funded pathology laboratory service.
- Other test providers, such as private laboratories.

Genetics Services of WA

GSWA provides a comprehensive, publicly funded, state-wide service in clinical genetics via three subspecialty areas: Genetic Paediatric Services; Familial Cancer Program; and Obstetrics and General Genetic Services. Housed at King Edward Memorial Hospital, GSWA provides their services through a network of metropolitan and regional outpatient clinics. Their primary role is to help health consumers in relation to the diagnosis and management of their genetic condition, including testing for inherited cancer predisposition, prenatal diagnosis and carrier screening for family planning. As such, health consumers are referred to GSWA through multiple avenues, such as health professionals and general practitioners from both public and private sectors, child development centres, and a small number of self-referrals. Referrals are seen based on urgency as determined by GSWA.

GSWA employs clinical geneticists, genetic counsellors and administrative staff. The clinical geneticists will determine whether genomic testing is required. If so, they will order the test from a laboratory, which is usually PathWest. They will coordinate the testing, obtain informed consent from the person prior to testing, interpret the test results and provide the results back to the person, their carers and/or families. The genetic counsellors are qualified to provide specialised pre- and post-test counselling to inform the consumer's decision about the test (e.g. on the risks and benefits of the test, or on the familial implications of the test results). This form of counselling is provided when deemed necessary to ensure that people can provide informed consent to testing.

Health professionals other than clinical genetics specialists

Many public and private health professionals refer their health consumers to GSWA for genomic testing and counselling. This is particularly true for health consumers whose situation is considered more complex and when the health professional doesn't feel confident to provide the required counselling. However, increasingly health professionals are themselves selecting and ordering genomic tests for their consumer, either with or without first consulting a clinical

geneticist. In these cases, the health professionals provide counselling to health consumers, obtain informed consent, interpret the test results and provide the results back to the consumer. At present, there are no WA standards to guide practitioners in undertaking these activities.

Sometimes, a health professional may consult a clinical geneticist after the test results are received for help in interpreting the test results or refer their consumer to GSWA following a positive test result, so that counselling and management of the consumer (and possibly their family) is provided in collaboration with clinical geneticists or genetic counsellors. In the primary care sector, general practitioners are also becoming increasingly involved in genomic testing. In many cases they refer health consumers suspected of having a genetic disease to health professionals or GSWA. However, there are occasions where they will order tests directly, for example, NIPT and Fragile X testing.

PathWest Laboratory Medicine WA

PathWest is the only public provider of pathology laboratory services in WA. Multiple departments within PathWest offer genomic testing, which includes a range of assays such as cytogenetics, microarrays, Sanger sequencing of targeted gene variants or sections of genes, and massively parallel sequencing of large collections of genes (e.g. gene panels, clinical exomes). Decisions regarding which tests each department will provide are made by senior departmental staff in collaboration with specialist clinicians and scientists. Decisions are based on multiple factors including clinical demand, evidence of clinical utility, available resourcing and funding avenues. Examples of human genomic tests provided by each department are outlined below.

Diagnostic Genomics offers a comprehensive range of testing pathways in prenatal diagnosis and reproductive genetics, familial cancer, developmental disorders, neurogenetics, cardiac genetics, haematology and oncology genetics and rare diseases. Working closely with local researchers, Diagnostic Genomics has developed some internationally renowned bespoke gene panels in areas such as neuromuscular and cardiac disorders.

In July 2020, Diagnostic Genomics introduced Whole Exome Sequencing (WES) technology to replace several of its targeted gene panels. While the sequencing data resulting from WES can be filtered using bioinformatics to analyse only a select number of genes relevant to specific conditions or broader medical specialties, this technology provides far greater scope to investigate other genes if further diagnostic investigation is required. Patient data is stored and can be re-analysed in the future as more information becomes available relevant to a genetic condition.

Anatomical Pathology provide state-wide public pathology somatic molecular testing as part of a standardised and equitable reflex protocol for cancer diagnosis, prognostication and biomarker testing. Currently testing is performed as sequential single and/or multiple gene tests on a wide range of sample types using complex and broad test panels and platforms. The laboratory is one of the very few laboratories in Australia to obtain accreditation from the National Association of Testing Authorities for somatic massively parallel sequencing using the Illumina TruSight Tumour 26 panel that analyses twenty-six genes at once, and this has been in practice for more than three years, thus replacing multiple single gene tests. The laboratory has a state-wide and well-established complex and integrated workflow for cancer testing by experts in these fields and performs comprehensive somatic genomic profiling using the Illumina TruSight170 panel for selected cancer types refractory to chemotherapy. The laboratory is transitioning to a larger targeted panel of 523 genes (Illumina TruSight Oncology 500 panel), capable of analysing microsatellite instability and tumour mutation burden.

Haematology offers a wide range of genetic and genomic tests, many of which have been provided as standard of care (in both malignant and non-malignant haematology) for years. These tests include:

1. Haematological malignancy
    a. Karyotyping and FISH analysis for leukaemia cases (including adult and paediatric, acute and chronic, myeloid and lymphoid leukaemia's), multiple myeloma, myelodysplastic syndromes and myeloproliferative/myelodysplastic disorders.
    b. Targeted sequencing of multiple genes for diagnosis of myeloproliferative neoplasms.
    c. Single gene sequencing of specific mutations relevant to leukaemia treatment where rapid turn-around is required.
    d. A myeloid targeted NGS gene panel (with support of diagnostic genomics and utility of their sequencing equipment).
    e. Quantitative PCR for minimal residual disease monitoring in acute and chronic leukaemia.
    f. Testing for treatment resistance in leukaemia (e.g. BCR-ABL1 kinase domain mutations).
2. Thrombosis and Haemostasis
    a. Targeted sequencing for the Factor V Leiden and Prothrombin gene mutations
    b. Specialised sequencing for haemophilia and other bleeding disorders is referred interstate and overseas via PathWest Haematology.
3. Transfusion Medicine
    a. Red cell and platelet antigen genotyping.
4. Haemoglobinopathy and thalassaemia diagnosis
    a. HPLC in haematology. Multiplex gap PCR, MLPA and globin gene sequencing performed in collaboration with diagnostic genomics with reporting through haematology.

Immunology performs specialised testing for immunogenetic-related assays such as full human leukocyte antigen (HLA) typing and matching for solid organ and bone marrow transplantation.

Microbiology provides routine and specialist diagnostic testing for the management and treatment of infectious diseases (which includes some genomic sequencing), supporting public health surveillance and prevention programs.

Other test providers

Clinicians within the WA health system are most likely to request their genomic tests through PathWest. Although clinical exomes are performed, whole exome sequencing or whole genome sequencing is not presently available at PathWest, due to lack of infrastructure and workforce capacity. Instead, if these tests are deemed appropriate for the consumer, DNA samples will be sent away to an accredited laboratory interstate or overseas for processing. Other genomic testing that PathWest does not currently perform yet coordinates is non-invasive prenatal testing.

Genomic testing of paediatric cancers is currently performed at laboratories interstate and overseas, and this is organised by paediatric oncologists. Similarly, there are some practitioners in other medical fields who do not request their tests through PathWest, but rather order tests direct from laboratories interstate or overseas. This is for reasons such as that these tumours are relatively rare and testing is conducted as part of international or national clinical trials.

Appendix B: How genomics is currently applied in the WA health system

The WA health system

The WA health system consists of the Department of Health, Child and Adolescent Health Service, four geographically defined HSPs (North Metropolitan Health Service, South Metropolitan Health Service, East Metropolitan Health Service and WA Country Health Service) and three state-wide services: PathWest Laboratory Medicine WA (PathWest); Health Support Services; and the Quadriplegic Centre.

The Department of Health, through the Director General as the system manager, provides leadership and management of the WA health system to ensure the delivery of high quality, safe and timely health services. The Director General enters service level agreements with each HSP, who are then responsible for the delivery of services that aim to optimise outcomes relative to the costs of providing the services.

Genomic testing

In the context of healthcare, genomic tests are medical tests that can identify changes in the structure or function of a person's DNA. Genomic tests require a person to provide a sample of their DNA from a biological source such as blood, cheek cells or tumour biopsies obtained by various modalities. Their biological sample is then sent to a laboratory, where DNA is extracted and subsequently analysed to determine changes or "variants" that could be causative of a genetic condition in them and/or their offspring.

People who utilise the WA health system may be referred (by a health professional) to have a genomic test for multiple reasons. The types of genomic tests that are currently conducted within or facilitated through the WA health system include:

- Diagnostic testing in symptomatic persons to determine the presence of a genomic variant/s associated with the observed phenotype.
- Predictive testing in asymptomatic persons to identify whether they have a genomic predisposition to a condition that may arise later in life.
- Somatic mutation testing in persons diagnosed with cancer to look for genomic variants in cancer cells to provide valuable information about diagnosis, prognosis or targeted treatment options.
- Pharmacogenomic testing to screen for genomic variants that alter drug response with the aim to inform drug dosages and regimes to improve drug efficacy and consumer compliance, whilst reducing side effects and avoiding life-threatening reactions.
- Cascade screening to find and test family members of a person affected by a genetic condition, to see whether the family members also have the same genetic variant that causes the condition.
- Carrier screening in asymptomatic persons (or couples) to find out whether they carry any genetic variants linked to conditions that can be passed down to future children.
- Prenatal screening and testing, where an unborn child's DNA is analysed to determine if a genetic condition is likely to be present or is present respectively. An example is non-invasive prenatal testing (NIPT) that tests fetal DNA present in the mother's blood.
- Preimplantation diagnosis, where genetic material from embryos are tested for the presence of one or more genetic abnormalities that indicative of disease, prior to embryo implantation as part of an in vitro fertilisation procedure.

This document can be made available in alternative formats on request for a person with disability.