

A large, stylized graphic of chromosomes in various colors (blue, green, yellow, orange) arranged in a vertical column, spanning the left side of the page.

## SERVICE PLAN 2025–2030

Genetic Health WA is a statewide service delivered  
by North Metropolitan Health Service



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# List of abbreviations

<b>ABF</b>	Activity-based funding
<b>CAHS</b>	Child and Adolescent Health Service
<b>CaLD</b>	Culturally and linguistically diverse
<b>DNA</b>	Deoxyribonucleic acid
<b>EMHS</b>	East Metropolitan Health Service
<b>FCP</b>	Familial Cancer Program; one of GHWA's subspecialties
<b>FSH</b>	Fiona Stanley Hospital
<b>FTE</b>	Full-time equivalent
<b>Geneticist</b>	Clinical geneticist
<b>GHWA</b>	Genetic Health WA
<b>GP</b>	General practitioner
<b>GPS</b>	Genetic Paediatric Services; one of GHWA's subspecialties
<b>HCC</b>	Health Consumers' Council Western Australia
<b>HGSA</b>	Human Genetics Society of Australasia
<b>HSPs</b>	Health Service Providers
<b>ICT</b>	Information and communications technology
<b>KEMH</b>	King Edward Memorial Hospital
<b>MBS</b>	Medicare Benefits Schedule
<b>MDTs</b>	Multidisciplinary teams
<b>NMHS</b>	North Metropolitan Health Service
<b>OGGS</b>	Obstetrics and General Genetic Services; one of GHWA's subspecialties
<b>OPHG</b>	Office of Population Health Genomics
<b>PathWest</b>	PathWest Laboratory Medicine WA
<b>PCH</b>	Perth Children's Hospital
<b>RPH</b>	Royal Perth Hospital
<b>SCGH</b>	Sir Charles Gairdner Hospital
<b>SMHS</b>	South Metropolitan Health Service
<b>The Act</b>	<i>The Health Services Act 2016</i>
<b>The Plan</b>	<i>Genetic Health WA Service Plan 2025-2030</i>
<b>VUS</b>	Variant of uncertain significance
<b>WA</b>	Western Australia
<b>WA Genomics Strategy</b>	<i>WA Genomics Strategy 2022-2032: Towards precision medicine and precision public health</i>
<b>WACHS</b>	WA Country Health Service
<b>WNHS</b>	Women and Newborn Health Service

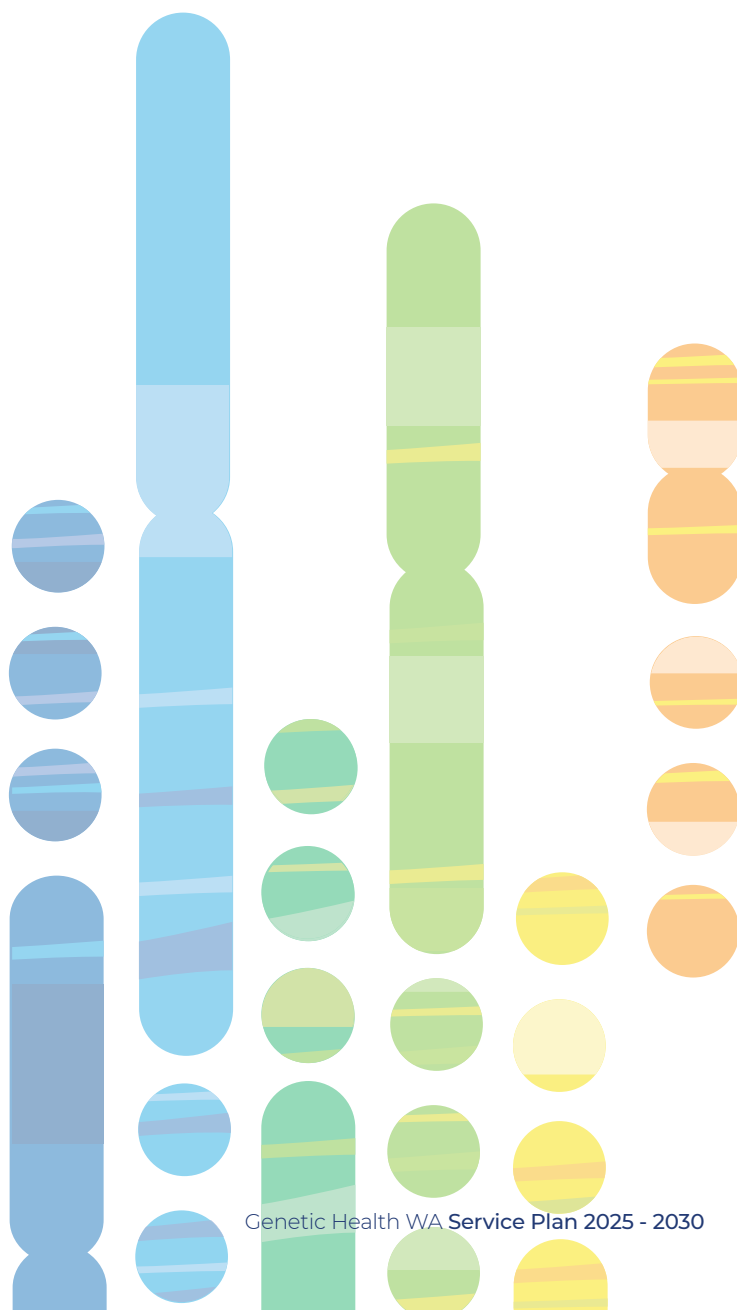


# Glossary

<b>Cascade testing</b>	Testing for changes to an individual's DNA sequence previously identified in a biological relative. Includes predictive, parental, and carrier testing
<b>Consumer</b>	A user of health services, e.g. patient or carer
<b>Clinical genomic specialty services</b>	Diagnosis, genetic counselling, and care and management of individuals and families with, suspected of, or at risk of a genetic condition provided by genomic specialists
<b>Cytogenetic analyses</b>	Study of the structure and properties of chromosomes, encompassing karyotyping, fluorescent in situ hybridisation, microarray, and chromosomal instability studies
<b>Deoxyribonucleic acid</b>	DNA; the molecule that carries the genetic information necessary for the development and function of an organism, e.g. a human
<b>Diagnostic testing</b>	Testing to identify genetic changes causing symptoms in an individual. Results can confirm a suspected diagnosis or exclude a differential diagnosis
<b>DNA sequencing</b>	Testing to identify changes in an individual's DNA sequence
<b>Exome</b>	The approximately 1.5-2% of an individual's DNA sequence that codes for proteins, which in turn perform various functions in cells
<b>Gene</b>	The functional components of the DNA sequence that contain the operational instructions for all cells
<b>Genetic condition</b>	A disease arising from change(s) to an individual's DNA sequence. These can be germline (present in all cells at birth) or somatic (arising in individual cells throughout one's life)
<b>Genetics</b>	The study of the functions and composition of single genes
<b>Genome</b>	All the DNA sequence within an individual
<b>Genomics</b>	The study of genes and how they interact with each other and the environment
<b>Genomic health care/ medicine</b>	Health care informed by genomic knowledge. Includes clinical genomic specialty services and other precision medicine approaches delivered by non-genomic specialists
<b>Genomic specialists</b>	Health professionals who have received formal genomics specialty training, e.g. clinical geneticist, genetic counsellor
<b>Germline testing</b>	Testing for heritable changes in an individual's genetic information (e.g. those genetic changes they are born with, and which can be passed on to the next generation)
<b>High-value care</b>	Health care that is safe, effective, high-quality, and based on clinical evidence. High-value care addresses system waste by directing resources to where they are most needed
<b>Non-genomic specialist/workforce</b>	Health professionals who have not received formal genomics specialty training, e.g. GP, paediatrician, nurse, social worker



<b>Predictive testing</b>	Genetic testing of a currently unaffected individual to predict if they are at risk of developing a particular genetic condition
<b>Primary care representative</b>	GPs, nurses, and other health professionals that provide health care in a primary care setting
<b>Reproductive carrier testing</b>	Testing to determine if prospective parent(s) possess genetic changes that increase their risk of having a child with particular genetic conditions
<b>Somatic testing</b>	Testing for non-inheritable genetic changes acquired within a confined group of cells or tissues in an individual. These genetic changes cannot be passed on to the next generation Also known as “tumour testing” in cells giving rise to a cancer





## Foreword

**It is my pleasure to present the inaugural Genetic Health WA (GHWa) Service Plan 2025-2030. The Plan sets out an ambitious vision for GHWa's future to enable the delivery of equitable statewide services that meet WA's emerging genomic health care needs.**

Partnering with our communities is integral for shaping the future direction of clinical services. The co-design approach taken to develop the Plan has been invaluable for ensuring it addresses the needs of GHWa's consumers and their families. The Plan was influenced by extensive consultation with stakeholders across the WA health system, and I thank everyone who contributed their experiences, perspectives, and advice.

Outlined within the Plan are the priorities, objectives, and actions that GHWa will work towards achieving over the next five years. Central to this work will be engagement and collaboration with consumers and carers to ensure that service improvements reflect their preferences and facilitate the continued provision of person and family-centred care.

Improving service accessibility and sustainability are key focuses for the future, which will be addressed in part through implementation of innovative models of care enabled by digital technologies. Purposeful partnerships with service providers will be utilised to raise awareness and accessibility of GHWa services and ensure that all those who could benefit from its services receive the right care, at the right time, and in the right place.

The Plan acknowledges the vital role played by GHWa staff in delivering high-quality services, and outlines ways in which we can continue to grow this specialist workforce and support their career development. It also recognises the importance of embracing research and innovation to help translate genomic discoveries into tangible health benefits for the WA population.

Whilst these exciting advancements can help to improve health outcomes, it is essential that genomic privacy is safeguarded and consumers are empowered to make informed decisions about genomic testing. GHWa's expertise will be invaluable for informing the safe adoption of genomics into the WA health system and supporting non-genomic specialists to deliver appropriate and effective person and family-centred genomic health care.

It is acknowledged that a genetic diagnosis can have a considerable impact on the physical and mental wellbeing of individuals and their families, and GHWa's consumers may require interaction with many other medical specialties and health and community services. As such, it will be important for GHWa to continue to work together with consumers and other stakeholders to realise the Plan's objectives.

I am excited by the opportunities presented by the Plan and I look forward to seeing its vision of timely, effective, and equitable person and family-centred genomic health care for all Western Australians come to life.

**Jeffrey Williams**

Executive Director

Women and Newborn Health Service



# Executive summary

**The sole public provider of clinical genomic specialty services in Western Australia (WA) is Genetic Health WA (GHWa). To meet the rising demand for genomic health care, GHWa will need to engage in collaborative and coordinated approaches to help grow genomic capacity across the system and implement innovative service models which adapt to the diverse needs of its stakeholders.**

The Genetic Health WA Service Plan 2025-2030 (the Plan) has been developed to guide the strategic direction for the delivery of clinical genomic specialty services by GHWa over the next 5 years. To embed the perspectives of stakeholders in the future delivery of GHWa's services, the Plan was co-designed with people with lived experience of GHWa (i.e., consumers and carers) and was informed by a range of stakeholder engagement activities as well as key Western Australian health system and genomic strategic policies and frameworks.

## GHWa at a glance

As of January 2024, GHWa employs 58 staff to provide clinical genomic specialty services to people throughout WA. This includes the provision of both outpatient and inpatient services delivered from GHWa's central offices at King Edward Memorial Hospital and Perth Children's Hospital, as well as via 2 metropolitan and 6 regional outreach clinics throughout the state. GHWa also delivers 15 collaborative clinics in partnership with other Health Service Providers and provides advice to non-genomic specialists to support the delivery of person- and family-centred genomic health care.

Between 1 January 2018 and 31 December 2022, GHWa received 25,120 referrals and delivered 15,513 appointments to a wide range of consumers across WA. Compared to the 2018 calendar year, GHWa received 14% more referrals and delivered 30% more appointments during 2022. This rising demand for GHWa's services is expected to continue due to population growth, rapid advancements in genomic technologies, and increasing complexity and clinical utility of genomic testing. These drivers, along with issues such as genomic specialist workforce shortages, a limited awareness of GHWa amongst health professionals and consumers, and difficulties with physical and cultural accessibility, will impact GHWa's ability to deliver timely and equitable clinical genomic specialty services. It will therefore be necessary for GHWa to consider and address these key service challenges going forward.

## What the future looks like for GHWa

Outlined within the Plan is a proposed future model for GHWa's service delivery, which considers its key position as the sole public clinical genomic specialty service in WA and its emerging role in supporting the implementation of genomic medicine into clinical practice. In accordance with this vision, GHWa will work to improve service efficiency, enhance its capacity, and explore innovative workforce and service models to meet the increasing demand and ensure consumers, carers, and families continue to receive timely and equitable person- and family-centred services. GHWa's responsibilities will evolve to include surveillance for consumers with inherited multisystem cancer predisposition syndromes, and potentially move towards medical care coordination for these individuals as well as consumers with undiagnosed or rare genetic disorders without a clear managing specialty.

GHWa will also work with its partners and stakeholders to help embed genomic testing for consumers with genetic conditions into non-genomic specialties by contributing to the education, training, and support of the non-genomic health workforce. In keeping with the co-design approach used to develop the Plan, it is vital the voice of people with lived experience of GHWa continues to be at the core of the design, delivery, and evaluation of GHWa's services. It is anticipated this will help to deliver high-value genomic health care that is timely, equitable and safe and benefits all Western Australians, no matter where they live.





### How GHWA will achieve this

To enable GHWA to work towards its future service model, four overarching service directions (outlined in Figure 1) have been identified as priority areas for GHWA over the next 5 years. Central to these service directions is the need to ensure GHWA's services continue to be person- and family-centred so the needs and preferences of all consumers, carers, and families are respected and prioritised. Fulfilling the proposed actions listed under each service direction will be integral to achieving the objectives of the Plan and realising the future role of GHWA within the Western Australian health system.

The Plan will be implemented over a 5-year period with oversight from the North Metropolitan Health

Service. Implementation will require key enablers (highlighted in Figure 1) including sustainable investment, support from senior leadership, strong accountability and clear responsibilities, meaningful partnerships and collaboration, a capable workforce, robust information and communications technology systems, and suitable infrastructure.

The Plan will be reviewed at the completion of the implementation period to determine the success of the Plan and inform the development of future service plans for GHWA. It is anticipated sustained support will be necessary in the years following the Plan's implementation to fully realise its ambitious vision.







**Service Direction 1:**  
**Building capacity, capability  
and sustainability to deliver  
high-value care**

1. Review and streamline internal operating models and pathways to improve efficiency and consistency of service delivery
2. Explore and adopt innovations in service delivery (including digital tools where appropriate) to optimise processes and increase productivity
3. Develop a sustainable genomic specialist workforce with the capacity and capability to deliver best-practice, safe, and effective clinical genomic specialty services across WA
4. Engage in evidence-based service planning, monitoring, and evaluation in collaboration with stakeholders to ensure GHWA continues to provide services which meet the needs of the WA population



**Service Direction 2:**  
**Increasing awareness,  
responsiveness and accessibility**

1. Improve the quality, availability, and dissemination of information designed to increase awareness amongst stakeholders of GHWA and the services it provides throughout WA and ensure that consumers with the greatest need can access GHWA's services
2. Increase engagement with stakeholders including consumers, carers, community groups, and health and social care providers to develop strategies to enhance access, care, and support for populations that experience heightened barriers to accessing clinical genomic specialty services
3. Review, develop, and implement strategies, pathways, and innovative models of care in partnership with stakeholders to continue being responsive to the diverse needs of consumers, carers, and family members

## Person- and family-centred care



**Service Direction 3:**  
**Enhancing integration and  
care co-ordination**

1. Refine and develop strategies to improve continuity of care for consumers and carers in partnership with stakeholders, including on-going management, disease surveillance and care coordination
2. In partnership with Health Service Providers and the Department of Health, review, maintain and expand clinical pathways and arrangements that embed access to specialised genomic services and advice into non-genomic clinical services to ensure all Western Australians have timely access to appropriate and coordinated genomic health care



**Service Direction 4:**  
**Fostering excellence in genomic  
healthcare across WA**

1. Collaborate with partners and stakeholders to share best clinical practice, develop clinical and policy guidance, and set direction for future genomic healthcare services
2. Continue to work with stakeholders to support the delivery of genomics in non-genomic clinical services by expanding the provision of advice to non-genomic specialists and providing input on genomics education and training programs for health professionals
3. Participate in and lead research and translation projects to advance the adoption of new genomic technologies and knowledge into clinical practice

### Enablers of success

Skilled, agile and diverse workforce

Robust information and communications technology

Appropriate infrastructure

### Sustainable funding

Senior leadership and support

Accountability, roles and responsibility

Meaningful partnerships and collaboration

**Figure 1:** A summary of the service directions and underlying objectives of the Plan, and the enablers that will help GHWA to realise this vision.



# Introduction

Genetic Health WA (GHWA) was established in 1988 as the Genetic Services of Western Australia. Due to the historic association of clinical genomic specialty services with pre- and post-natal care, the service is part of the Women and Newborn Health Service (WNHS), which was incorporated into the North Metropolitan Health Service (NMHS) in 2006. GHWA is currently located at King Edward Memorial Hospital (KEMH) and began formally operating as GHWA in January 2024.

GHWA provides a clinical genomic specialty service throughout Western Australia (WA) that encompasses the diagnosis, management, and support of individuals with, suspected of, or at an increased risk of themselves or a child having a germline genetic condition. Given the increasing utility of genomic technology and knowledge in health care, GHWA's role as a central source of genomics expertise is invaluable for helping to implement genomics within the Western Australian health system. The rapid growth in genomic medicine over recent years has led to an increase in demand for the clinical genomic specialty services provided by GHWA from both health professionals and consumers across WA.

Since its establishment, GHWA's consumer base has evolved from being primarily comprised of obstetric and neonatal patients to include adults at

risk of familial cancers and children and adults with rare genetic conditions, adding to the complexity of the services GHWA provide. Purposeful and adaptive service planning is needed to enable GHWA to manage future demand in a timely manner and consistently provide equitable, person- and family-centred clinical genomic speciality services. GHWA's inaugural service plan, the "Genetic Health WA Service Plan 2025-2030" (the Plan), has been developed to assist GHWA in achieving this goal. The Plan aims to facilitate the provision of an equitable service able to appropriately support the emerging genomic healthcare needs of consumers throughout WA. As such, it informs the future service profile of GHWA and outlines what actions, enablers and partnerships are required to ensure services are delivered efficiently and equitably and address the diverse needs of its stakeholders.

## Planning context

The Plan was informed and guided by several key state and national policies and strategic frameworks (Figure 2).

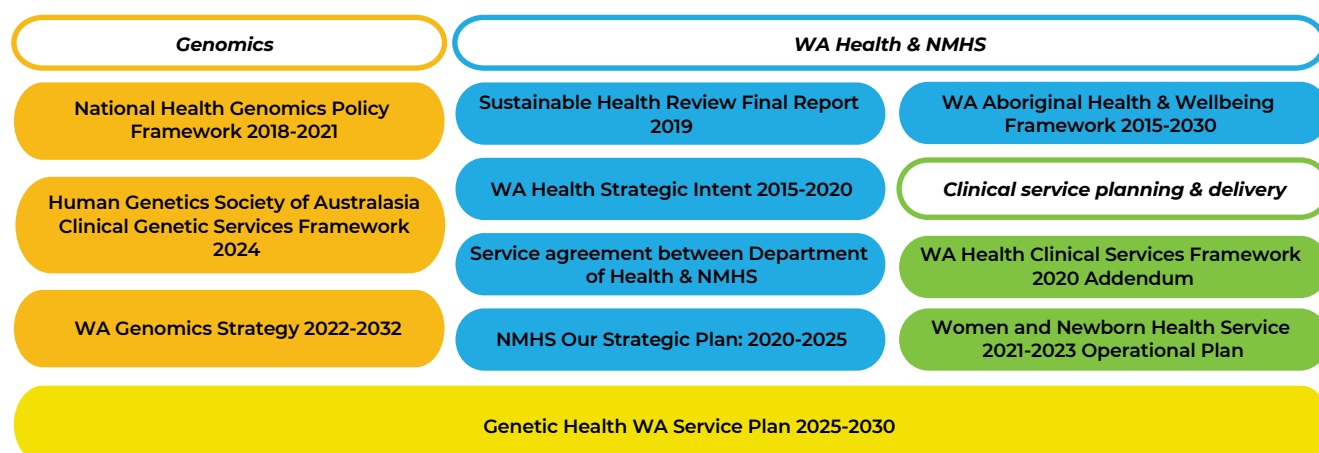


Figure 2: Planning context for the Plan, including relevant state and national documents.



As GHWA is a service governed by the NMHS, yet delivers services throughout WA, the planning context is influenced by the strategic priorities of both the NMHS and the Western Australian Department of Health (Department of Health), including the:

- *North Metropolitan Health Service Strategic Plan: Unleashing our potential 2024-2027*
- *WA Health Strategic Intent 2015 – 2020*
- *WA Health Clinical Services Framework 2020 Addendum*
- *WA Health Digital Strategy 2020-2030*
- *WA Aboriginal Health and Wellbeing Framework 2015-2030*
- *Sustainable Health Review: Final Report to the Western Australian Government*
- *Independent Review of WA Health System Governance Report*

The *National Health Genomics Policy Framework 2018-2021* (currently under review) and the Human Genetics Society of Australasia (HGSA) Clinical Genetic Services Framework 2024 are key national genomics policy and clinical frameworks that have also been considered in the development of the Plan.

The *WA Genomics Strategy 2022-2032: Towards precision medicine and precision public health* (the WA Genomics Strategy) outlines a coordinated and strategic approach to guide the future delivery of health genomics in WA and the development of this Plan is one of many key actions in the first implementation plan for it. The WA Genomics Strategy describes five strategic priority areas and goals to guide the efficient, effective, ethical and equitable translation of genomics into the Western Australian health system and valuably contribute towards the delivery of precision medicine and precision public health. It also outlines four principles to underpin the Strategy's implementation, including the planning and delivery of genomic health care by services such as GHWA (see Box 1).

The WA Genomics Strategy is designed to facilitate the application of genomic knowledge and services to enable precision medicine and precision public health approaches, as well as the associated health outcomes and policy implications. The WA Genomics Strategy has a broad remit, capturing all uses of genomics for precision medicines and precision public health purposes. This includes genetic and genomic tests and therapies or interventions utilising genomic information which have increasing

#### **Principle 1:**

The application of genomic knowledge to health care is equitable and informed by scientific evidence and the diverse needs of consumers, carers, families and the community.

#### **Principle 2:**

Investment in genomic services and applications is strategically prioritised based on their value to consumers, services, and the WA health system.

#### **Principle 3:**

Individuals, families and the broader population are empowered to make decisions about the application of genomics in their health care.

#### **Principle 4:**

Trust in the use of genomics in health care is promoted through adherence to quality and safety standards and compliance with privacy and confidentiality policies.

**Box 1:** Principles for genomic health care in WA as outlined in the WA Genomics Strategy.

applicability to a range of medical disciplines and as such are delivered by a range of health service providers across the Western Australian health system at an individual or population level (see Appendix 1). The clinical genomics specialty services provided by GHWA are one part of this broader application of genomics in WA.

Responsibility for the implementation of the WA Genomics Strategy is shared amongst stakeholders across the Western Australian health system, with system-wide stewardship and leadership provided by the Department of Health and oversight provided by the WA Genomics Strategy Implementation Committee. However, as the only clinical genomics speciality service in the Western Australian health system, GHWA is one of the key implementation partners (see Appendix 2). The Plan aims to guide GHWA in taking the first steps of implementation, with further action and sustained support necessary to continue progress towards achieving the WA Genomics Strategy's ambitious aims in subsequent years.



## Developing the Plan

The development of the Plan was a collaborative undertaking between GHWA<sup>1</sup> and the Office of Population Health Genomics (OPHG) at the Department of Health, and as such was co-sponsored by the NMHS and the Department of Health.

Person- and family-centred care is a central priority in the implementation of the WA Genomics Strategy (Appendix 2). In line with this, the development of the Plan was undertaken using a co-design approach to best address the needs of Western Australians.

As part of this process, over 300 stakeholders shared their insights to inform the Plan (Figure 3), including:

- GHWA consumers and carers
- Staff from GHWA
- Representatives from patient organisations

- Non-genomic specialists (health professionals who have not received formal genomics specialty training, including general practitioners (GPs) and other primary care providers)
- Representatives nominated by the Chief Executives of most Health Service Providers (HSPs) including the WA Country Health Service (WACHS), East Metropolitan Health Service (EMHS), South Metropolitan Health Service (SMHS), Child and Adolescent Health Service (CAHS), and PathWest Laboratory Medicine WA (PathWest)
- Members of the WA Genomics Strategy Implementation Committee

For more details of the co-design approach and stakeholder engagement activities conducted, please see Appendix 3.

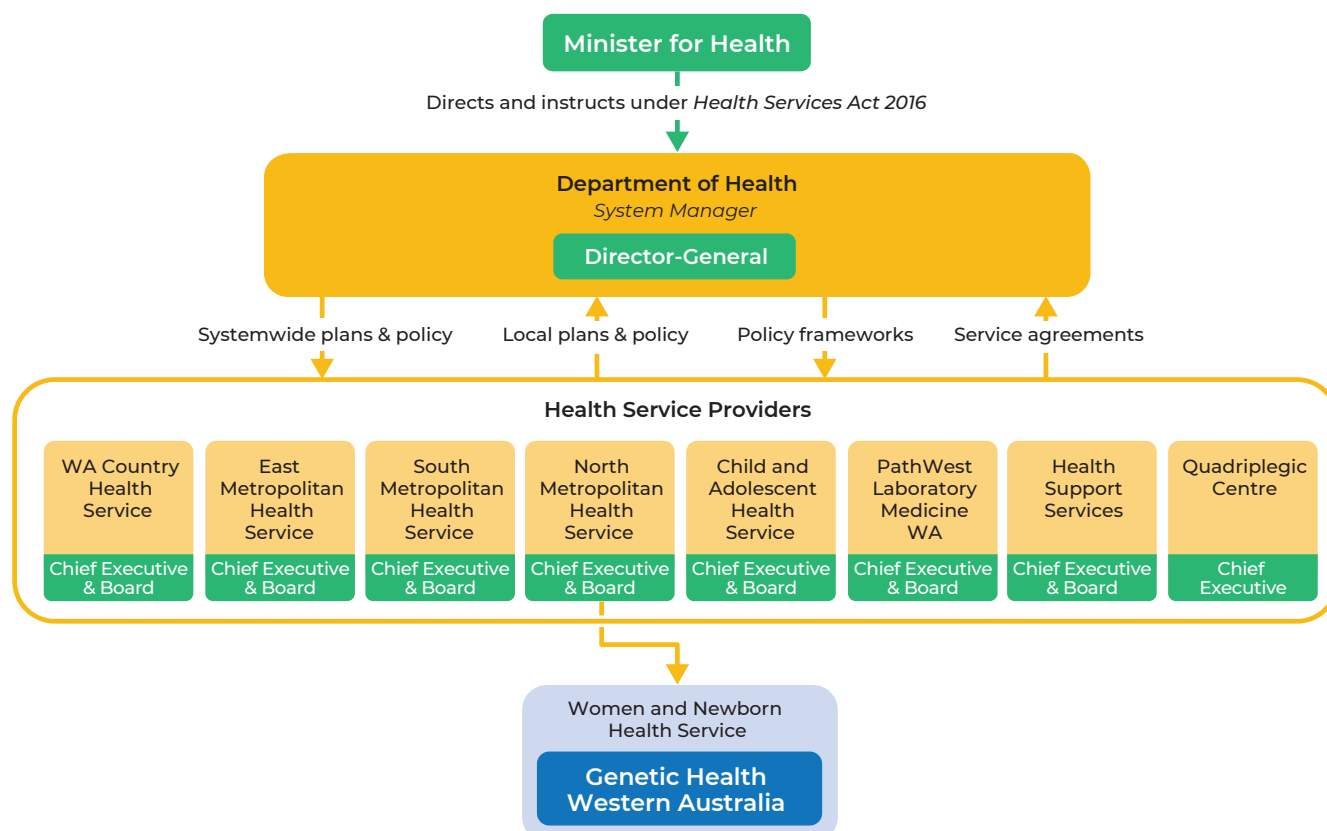
<sup>1</sup> All planning and consultation for the development of the Plan occurred whilst the service was operating as the Genetic Services of Western Australia; however, this document refers to the service as Genetic Health Western Australia or GHWA to reflect the change in name effective 08/01/2024.

# Service profile

## Position within the Western Australian Health System

The *Health Services Act 2016* (the Act) outlines the structure for the public health system in WA. GHWA is part of the NMHS, one of the eight HSPs formed under the Act (Figure 3). The KEMH, the maternity and gynaecology units at Osborne Park Hospital and other specialist services form the WNHS within NMHS. GHWA is situated at KEMH, and therefore the clinical governance for GHWA is provided by the WNHS and the NMHS. Each HSP is assisted by the Department

of Health through a range of systemwide plans and policies and service agreements to deliver healthcare services to a catchment area defined within the Act. Despite being part of one HSP, GHWA provides clinical genomic specialty services on a statewide basis, as set out in clause 7(i) of the *Health Services (Health Service Providers) Order*<sup>2</sup>.



**Figure 3:** Organisational structure of the Western Australian health system as set out by the *Health Services Act 2016*.

<sup>2</sup> *Health Services (Health Service Providers) Order 2016* (WA).

[https://www.legislation.wa.gov.au/legislation/statutes.nsf/law\\_s48154.html&view=consolidated](https://www.legislation.wa.gov.au/legislation/statutes.nsf/law_s48154.html&view=consolidated)





## Service model

GHWA receives referrals for individuals and families with, suspected of, or at an increased risk of themselves or a family member having a genetic condition from health professionals, as well as self-referrals, across WA. GHWA offers comprehensive clinical assessments, genetic counselling, and facilitates access to informed consent and genomic testing services for these individuals.

GHWA's consumer support extends beyond individual consumers to encompass at-risk family members, carers, and their extended healthcare team. This includes the provision of information resources, assistance with sharing genomic test results with family members and, in some cases, direction to other services for further information and support. However, this does not currently extend to broad psychosocial care or ongoing follow up for all consumers.

Like clinical genomic specialty services in other states in Australia at this time, GHWA provides limited follow-up for consumers, with most cases deactivated once the individuals are directed back to their referrers and provided with specialised advice for ongoing care following receipt of genomic testing results. However, consumers are advised they are welcome to re-contact GHWA about questions or concerns in the future, and some consumers are seen at GHWA when needed at a later life stage.

GHWA's central hub is located at KEMH. Outpatient clinics and inpatient ward consultations are provided at both KEMH and Perth Children's Hospital (PCH). GHWA also deliver outpatient services at selected metropolitan and regional outreach clinics (noting that there are some health regions within WA that do not host a dedicated outreach clinic). The locations and scheduling of GHWA's outreach clinics are depicted in Figure 4. Clinics that GHWA collaboratively deliver with health professionals in other HSPs are described under 'Collaborative work'.

Clinics are provided in multi-modal format with telehealth and phone appointments offered where clinically appropriate. Along with outreach clinics, telehealth and phone appointments help to ensure that services are delivered as close to home as possible.

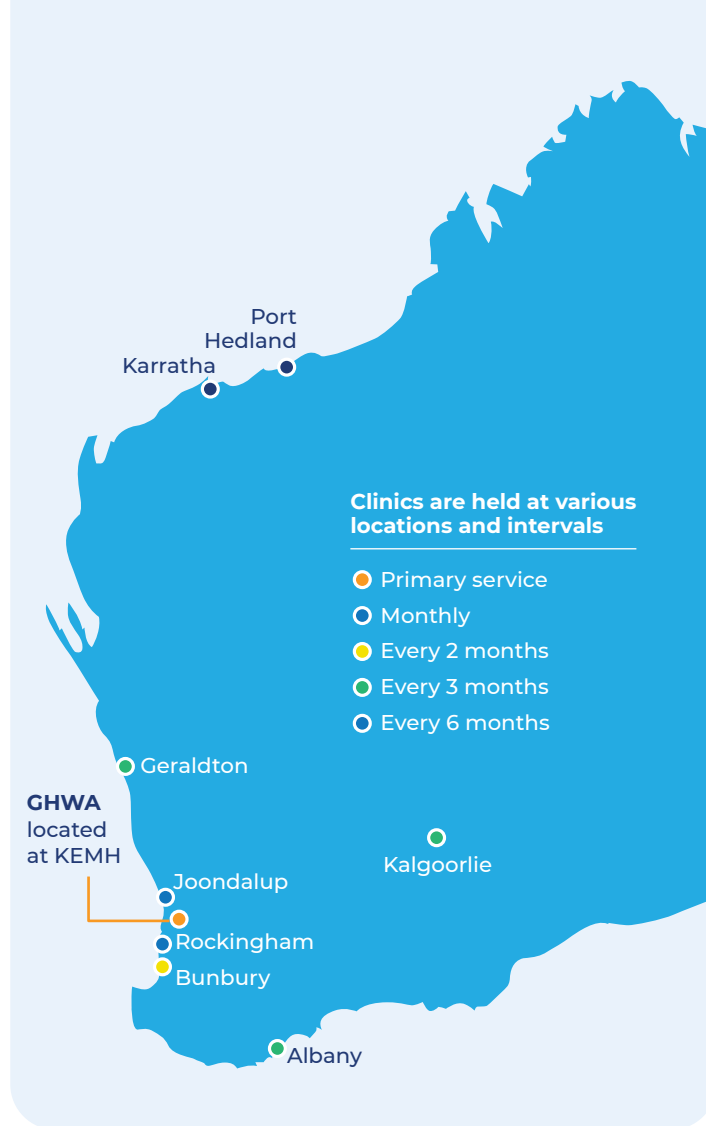


Figure 4: GHWA clinic locations across WA.

GHWA provides services for adults and children across 3 service streams: Obstetrics and General Genetic Services (OGGS), Genetic Paediatric Services (GPS), and the Familial Cancer Program (FCP). Whilst these service streams were historically established to meet demand for clinical genomic specialty services at that time, it is possible this 3-stream model may not remain fit for purpose as genomics evolves in the future.

The OGGS service stream offers services to adults at risk of or affected by a genetic condition, as well as adults prior to or during pregnancy if they are at increased risk of having a child with a genetic condition. This includes adults impacted by cardiac and renal genetic conditions, although there has been some work over recent years to develop these areas into a separate service stream.

The GPS service stream manages children identified with or suspected of having a genetic condition, including those who have congenital abnormalities, are experiencing developmental delay, or have unexplained differences in growth or clinical features. This also includes babies who are identified as likely to have certain genetic conditions through WA's Newborn Bloodspot Screening Program.



The FCP accepts referrals for patients who have or are suspected of having an increased risk of familial cancer. GHWA ensures these individuals are informed of the current cancer screening guidelines and recommendations and have access to regular monitoring procedures.

These functions are aided by the Western Australian Familial Cancer Registry, which is coordinated by and housed at GHWA.

Figure 5 shows a diagrammatic representation of many aspects of the services GHWA provide.



**Figure 5:** A summary of GHWA's current services.





## Eligibility criteria and indications for referrals

Referrals to GHWA are triaged based on the criteria outlined below in Table 1. GHWA does not accept referrals for all genetic conditions. Some genetic conditions or cases not managed by GHWA include familial hypercholesterolaemia, changes in the *MTHFR* gene (a gene involved with folate metabolism), or individuals who are carriers of an autosomal recessive genetic condition such as cystic fibrosis (noting that couples where both individuals are carriers and are planning pregnancy are eligible for referral).

**Table 1:** GHWA referral criteria.

Service Stream	Referral criteria
Obstetrics and General Genetic Services	<ul style="list-style-type: none"><li>• Adults who have or are suspected of having a genetic condition.</li><li>• Adults who are referred due to a sudden cardiac death in the family and are receiving genetic test results (on behalf of the family member who died) and related genetic counselling.</li><li>• Adults who have a family history of a genetic condition and are considering pregnancy.</li><li>• Pregnant individuals whose unborn or future child has an increased risk of a genetic condition.</li></ul>
Familial Cancer Program	<ul style="list-style-type: none"><li>• Adults with, suspected of being at risk, or known to be at risk of an inherited cancer syndrome.</li></ul>
Genetic Paediatric Services	<ul style="list-style-type: none"><li>• Children identified with or suspected of having a genetic condition, including inherited cancer syndromes.</li></ul>





## Triage of referrals

Appointments with GHWA are allocated according to urgency (see criteria in Table 2). In the GPS and FCP subspecialties, the initial contact following receipt of a referral commonly takes place via a scheduled phone call during a work-up clinic. This initial phone discussion provides individuals with an opportunity to ask questions and allows GHWA staff to collect family history regarding the reason for referral. Within the OGGS subspecialty, a non-formalised phone call may be made prior to the initial appointment to gather or clarify family or personal history. This information is important to determine the appropriateness of the referral, and to help triage and prepare for those patients who require an appointment. .

**Table 2:** GHWA referral urgency.

Urgency	Timeframe for appointment	Criteria
1: Urgent	≤ 30 days <i>(Initial contact within 48 hours)</i>	<ul style="list-style-type: none"><li>• A pregnant individual whereby management or treatment decisions may be influenced by genetic testing.</li><li>• An individual whereby potentially life-saving management or treatment options may be influenced by genetic testing.</li><li>• A newborn requiring immediate clinical assessment and possibly genetic testing that may influence lifesaving treatment or management options.</li><li>• An individual who is receiving palliative care.</li></ul>
2: Semi urgent	≤ 90 days	<ul style="list-style-type: none"><li>• An undiagnosed individual who requires a clinical assessment for treatment and management.</li><li>• Known carrier couples considering pregnancy.</li></ul>
3: Routine	≤ 365 days	<ul style="list-style-type: none"><li>• An undiagnosed individual who requires a clinical assessment for a genetic diagnosis.</li><li>• An individual whereby a clinical diagnosis has been identified, however a genetic diagnosis may provide more succinct management or surveillance options.</li><li>• Familial clustering of specific cancers (such as breast or bowel) whereby testing cannot be offered to an affected family member due to death or geographical location.</li><li>• Pregnancy planning for a couple who are biologically related.</li><li>• Cascade testing in an unaffected individual.</li></ul>



## Workforce

GHWA's services are delivered by clinical geneticists (geneticists) and genetic counsellors, supported by clinical officers, administrative and data management staff. A geneticist is a medical doctor specialising in diagnosing and clinically assessing individuals and families affected by, suspected of, or at risk of a genetic condition. Geneticists will assess the consumer's physical attributes, medical and family history to provide individuals and families with information on recurrence and condition management. A genetic counsellor is an allied healthcare professional who helps consumers understand and adapt to the medical, psychological, individual, and familial implications associated with genetic conditions. A genetic counsellor will undertake and interpret personal and family histories, conduct risk assessments, and support individuals and families to make informed and autonomous decisions about their health care. Both geneticists and genetic counsellors educate and provide information on inheritance patterns, testing options, current management or surveillance guidelines, and research opportunities.

Geneticists and genetic counsellors collaboratively deliver clinics, which are all overseen by a geneticist. GHWA's workforce also includes 2 intake support staff known as clinical officers. Clinical officers assist in managing consumer records and gathering comprehensive family history information. Additional clinical staff, administrative support staff, and data or registry manager positions are documented separately in Table 3, which outlines GHWA's current workforce as of January 2024. All GHWA staff are based at KEMH, yet some regularly travel to other locations to deliver certain off-site clinics in the metropolitan area, whilst some share rotational responsibilities for attending outreach clinics.

GHWA are also in the process of recruiting a psychologist (who will provide counselling supervision for genetic counsellors and will also provide counselling to some consumers) and an Aboriginal Health Worker, both of which will be employed part-time at 0.6 full-time equivalent (FTE) hours. These positions are not included in Table 3.

**Table 3:** GHWA's current workforce as of January 2024.

Role	Staff headcount	Total FTE
Genetic counsellors	25	20.4
Clinical geneticists	9	6.6
Trainee clinical geneticists	3	3.0
Clinical officers	2	2.0
Clinical nurse specialists	2	1.2
Data managers	2	2.0
Familial Cancer Registry managers	2	1.6
Administration staff	13	12.1
<b>Total</b>	<b>58</b>	<b>48.9</b>



## Collaborative work

GHWA frequently collaborates with a range of other specialties to provide joint models of care at Fiona Stanley Hospital (FSH), Royal Perth Hospital (RPH), Sir Charles Gairdner Hospital (SCGH), PCH or KEMH. These collaborative clinics are delivered by health professionals from multiple specialties to ensure the care provided to individuals and their family members is timely, holistic, integrated and person-and-family-centred. By involving multiple health professionals with different expertise, these clinics enable mentoring, coaching and knowledge transfer between genomic and non-genomic specialists and thus can help to progress the safe and appropriate integration of genomics into clinical care across a range of medical specialties.

GHWA also supports non-genomic specialists to deliver genomic services by discussing cases with a potential genetic cause and providing advice on the appropriate genomic tests to order, the interpretation of test results, and referral of patients and/or their family members to GHWA. This support is provided through regular attendance at multidisciplinary team (MDT) meetings (at which patients are not present),

or through the provision of *ad hoc* advice (including through an on-call service where a geneticist and/or genetic counsellor answer health professionals' queries) and education. It is likely the number of non-genomic specialists ordering diagnostic testing for their patients will continue to grow as genomic technology becomes increasingly integrated into non-genomic specialties, resulting in rising demand for GHWA's expertise and advice.

Table 4 includes a list of the collaborative clinics and MDTs that GHWA either leads or regularly participates in across the WA health system. In addition to these formalised, regular arrangements, GHWA collaborates with other service providers on a case-by-case basis. For example, GHWA staff occasionally attend joint appointments with the NMHS Neurosciences Unit to support predictive testing for neurodegenerative conditions such as Huntington's disease. Similarly, non-genomic medical specialists such as paediatric oncologists or maternal foetal medicine fellows may attend appointments for some consumers during GHWA's regular clinics when GHWA staff request their specialist input.







**Table 4:** GHWA's clinical collaborations as of 2024.

Collaboration	Location	Led by
<b>Clinics</b>		
Adult inherited cardiac <sup>^</sup>	FSH	FSH
Vascular connective tissue disease <sup>^</sup>	FSH	FSH
Nephrology <sup>^</sup>	FSH/SCGH	GHWA
Adult cancer surveillance	KEMH	GHWA
Inherited gastrointestinal cancer	KEMH	GHWA
Dermatology	PCH	GHWA
Ophthalmology	PCH	GHWA
Paediatric inherited cardiac <sup>*</sup>	PCH	GHWA
Undiagnosed Disease Program	PCH	GHWA
22q11 deletion disorder & CHARGE syndrome	PCH	PCH
Achondroplasia	PCH	PCH
Endocrine bone	PCH	PCH
Paediatric cancer surveillance	PCH	PCH
RASopathy <sup>#</sup>	PCH	PCH
Fabry disease	RPH	RPH
<b>MDTs</b>		
Vascular connective tissue disease <sup>^</sup>	FSH	FSH
Adult inherited cardiac <sup>^</sup>	FSH	FSH
Breast cancer	FSH	FSH
Nephrology <sup>^</sup>	FSH/SCGH/PCH/RPH	GHWA
Maternal Foetal Medicine	KEMH	KEMH
Gynaecology-Oncology	KEMH	KEMH
Ophthalmology	Harry Perkins Institute	Lions Eye Institute
Molecular Tumour Board	PathWest	PathWest
Paediatric oncology	PCH	PCH
Transplant and Cellular Therapy/ Primary Immunodeficiency/Haematology	PCH	PCH
Adult inherited cardiac	RPH	RPH
Breast cancer	RPH	RPH
Breast cancer	SCGH	SCGH

<sup>^</sup> Clinics and MDTs are linked

<sup>\*</sup> Established in early 2024

<sup>#</sup> GHWA's involvement commenced in early 2024



## Genomic testing

Genomic laboratory testing is a core enabling tool of clinical genomic specialty services (see Box 2). GHWA predominantly orders genetic and genomic testing (e.g. deoxyribonucleic acid (DNA) sequencing, cytogenetic analyses) from PathWest, with whom GHWA work collaboratively to maximise the alignment between genomic testing and clinical genomics. This includes helping to assess and classify genetic variants detected through genomic testing.

Consequently, the scope of genomic testing services requested by GHWA is influenced by the testing repertoire and capacity of PathWest. However, testing is sometimes sought from external organisations when required, such as when a particular test is not offered by PathWest or when a specific turnaround time for the result is necessary.

Historically, analyses of genes using a technique called DNA sequencing was limited to analysis of the DNA sequence of only a relatively small section of a single gene. This approach was called “genetic testing”, and it was applied to certain genes known to be causative of specific diseases if they contained particular changes in their DNA sequence. As multiple genes became identified as causing the same or very similar clinical features and diseases, it became difficult for healthcare professionals to accurately select the most appropriate single gene or part of a gene for DNA sequencing, to thereby provide individuals with a genetic cause or diagnosis. If a genetic cause was not established from the initial DNA sequencing of a region/s of a gene, a subsequent gene test might have been selected in an attempt to yield a genetic answer, adding cost and time.

Due to the significant advances in DNA sequencing technology and data analysis, genetic testing has substantially evolved towards genomic testing. Genomic testing enables with a single test the analysis of multiple genes, all genes (an “exome”,

approximately 1.5%-2% of a genome), or even the entire genome (all of a human’s DNA sequence, including all known genes and other regions of DNA that can influence genes). Types of genomic testing include diagnostic, predictive, prognostic, cascade, and reproductive carrier testing. Testing for cancers can be germline or somatic. The knowledge gained from these different types of genomic testing facilitates a range of personal, familial and health system benefits, with the appropriateness of using each type of test dependent on the health scenario.

Whilst the cost of genomic testing has rapidly decreased over time, currently the price can be higher than many single gene tests. However, the timeliness of genomic testing and its greater likelihood of providing information with clinical utility makes this approach desirable in an increasing number of treatment and management circumstances, and indeed can provide economic efficiencies to healthcare systems. As such, genomic testing is now routinely offered to a growing number of Western Australian consumers.

**Box 2:** Genomic testing by DNA sequencing.



# Population serviced

**GHWA is responsible for planning and delivering clinical genomic specialty services to the entire Western Australian population across all age groups and health regions.**

Between 2017 and 2023 the estimated Western Australian population grew by approximately 11.3% to 2,878,563 people. In the year ending 30 June 2023, WA had the fastest population growth rate out of all Australian jurisdictions at 3.1%<sup>3</sup>. In the 2021-22 financial year, the Greater Perth region increased by 1.5% (32,200 people) whilst the rest of the state increased by 1.0% (5,800 people).

Based on the endorsed *WA Tomorrow Population Report No. 12* (see Box 3), the Western Australian population is estimated to grow by 15% from the 2022 population to 3,199,300 by 2029 and 24% to 3,567,000 by 2036<sup>4</sup>. However, it is expected referrals to GHWA will increase at a faster rate as local awareness of genetic conditions grows and more widespread adoption of genomic technologies in routine clinical practice of non-genomics specialties occurs. In addition, more genetic conditions are increasingly being identified as new technologies emerge and methodologies evolve. As such, confirming a genetic diagnosis is becoming more relevant as new interventions and treatments targeted towards the genetic cause of disease are developed. These developments are likely to further increase demand for GHWA's services.

The incidence of disease in the community also affects the population eligible for referral to GHWA and in turn, service demand.<sup>5</sup> For example, the incidence of cancer is expected to continue increasing with the growing size of the population, an ageing population, and advancements in imaging and diagnostic capabilities that can detect earlier or more indolent forms of cancer. In 2019, 13,925 cases of cancer were diagnosed in WA, equivalent to an incidence rate of 463.9 cases per 100,000 persons. Hereditary cancers are estimated to account for 5-10% of cancers, though they are often underdiagnosed.

The size of Australia's population is captured every 5 years in the national Census. In between Census dates, estimates of the resident population are created by several sources, including the Australian Bureau of Statistics and government agencies. The Department of Health uses population projections created by the Department of Planning, Lands and Heritage known as *WA Tomorrow*. These projections were created as 5 bands (A to E), with band C, representing the median forecast agreed by the Department of Treasury, underpinning the Department of Health's modelling.

## Box 3: Population estimate source for the Plan.

It is expected that the number of referrals to GHWA's Familial Cancer Program will grow with increasing cancer incidence, knowledge about cancer genomics, and use of genomic testing to identify affected people and/or guide cancer treatment.

People living with a rare disease will remain a core population group serviced by GHWA given the complex nature of their conditions. Rare diseases can affect multiple systems of the body, which leads to various challenges and unmet needs when clinical specialties predominantly focus on single or localised organ systems. Hence, these consumers normally benefit from multidisciplinary care, which can be coordinated by GHWA. Up to 6% of the Australian population is estimated to be affected by a rare disease, 72% of which are genetic in nature<sup>6</sup>.

<sup>3</sup> Australian Bureau of Statistics (2023). National, state and territory population. ABS [Internet].

Available from: <https://www.abs.gov.au/statistics/people/population/national-state-and-territory-population/latest-release#data-download>

<sup>4</sup> Department of Planning, Lands and Heritage (2024). Western Australia Tomorrow Population Report No. 12. [Internet]. Available from:

<https://www.wa.gov.au/organisation/department-of-planning-lands-and-heritage/western-australia-tomorrow-population-forecasts#wa-tomorrow-population-report-no-12>

<sup>5</sup> Australian Institute of Health and Welfare (2023). Cancer data in Australia. AIHW [Internet].

Available from: <https://www.aihw.gov.au/reports/cancer/cancer-data-in-australia/contents/state-and-territory>

<sup>6</sup> Nguengang Wakap S, et al (2020). Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database.

*European Journal of Human Genetics*, 28(2),165-173





There are more than 6,000 known rare diseases and the majority (70%) affect children<sup>5</sup>.

Based on WA's estimated population of 2,878,563 in the year ending June 2023<sup>7</sup>, approximately 230,285 Western Australians are living with a rare disease, including 48,798 children (aged 16 years and under). Currently, GHWA only services a small fraction of the estimated population affected by a rare disease as these conditions are normally difficult for health professionals to identify due to lack of familiarity, rarity of the disease, and highly complex symptoms. Limited awareness of GHWA and the services it offers may also play a role in the observed referral numbers being lower than expected. However, as genomic technologies become more precise, rare diseases are being discovered more regularly. This, in conjunction with increased awareness of GHWA's services and enhanced data collection on rare diseases, is likely to increase the size of the population eligible for referral to GHWA and demand on the service.

Other priority population groups serviced by GHWA include Aboriginal people and people from culturally and linguistically diverse (CaLD) backgrounds. In 2021, Aboriginal people made up 4.4% of the Western Australian population, a 19.4% increase from the previous 2016 Census<sup>8</sup> with continued population growth forecasted through to 2031<sup>9</sup>. The most populous areas in WA for Aboriginal people include Geraldton, Kalgoorlie, and Rockingham<sup>10</sup> where GHWA has already established outreach clinics. These clinics present the opportunity to have an increasingly important role in delivering care closer to home and improving engagement with consumers, particularly as the Aboriginal population grows.

Additionally, WA is home to the highest proportion of people born overseas (32.2%), compared to other Australian jurisdictions<sup>11</sup>. Whilst the top 10 birthplaces remained the same in both the 2016 and 2021 Censuses, the proportion of each has changed. The birthplaces with the highest growth were India, the Philippines, China, South Africa, Malaysia, and Bhutan. Migration patterns can affect the prevalence and distribution of genetic conditions in the community as ethnic diversity and disease risk profiles change. This has the potential to influence the number of and reasons for referral to GHWA over time.

Some consumers who identify as CaLD may be at higher risk of certain genetic conditions due to an increased prevalence of particular genetic changes or consanguinity (marriage between biological relatives) in their ethnic group. Moreover, the proportion of the Western Australian population born in a non-main English-speaking country is gradually becoming larger (17.5%) than the proportion born in a main English-speaking country (14.7%). This emphasises the growing need to address accessibility barriers affecting CaLD consumers.

<sup>7</sup> Australian Bureau of Statistics (2023). National, state and territory population, ABS [Internet].

Available from: <https://www.abs.gov.au/statistics/people/population/national-state-and-territory-population/latest-release#data-download>

<sup>8</sup> Australian Bureau of Statistics (2023). Estimates of Aboriginal and Torres Strait Islander Australians. ABS [Internet]. Available from: <https://www.abs.gov.au/statistics/people/aboriginal-and-torres-strait-islander-peoples/estimates-aboriginal-and-torres-strait-islander-australians/latest-release>

<sup>9</sup> Australian Bureau of Statistics (2019). Estimates and Projections, Aboriginal and Torres Strait Islander Australians. ABS [Internet]. Available from: <https://www.abs.gov.au/statistics/people/aboriginal-and-torres-strait-islander-peoples/estimates-and-projections-aboriginal-and-torres-strait-islander-australians/latest-release>

<sup>10</sup> Australian Bureau of Statistics (2022). Census of Population and Housing - Counts of Aboriginal and Torres Strait Islander Australians. ABS [Internet]. Available from: <https://www.abs.gov.au/statistics/people/aboriginal-and-torres-strait-islander-peoples/census-population-and-housing-counts-aboriginal-and-torres-strait-islander-australians/latest-release>

<sup>11</sup> Office of Multicultural Interests (2024). Census 2021 Highlights: Western Australia's Changing Population and Cultural Diversity. OMI [Internet]. Available from: [https://www.omi.wa.gov.au/docs/librariesprovider2/statistics/022434omi-census-highlight-report-feb23---web-ready-2-\(1\).pdf?sfvrsn=3913e4d8\\_0](https://www.omi.wa.gov.au/docs/librariesprovider2/statistics/022434omi-census-highlight-report-feb23---web-ready-2-(1).pdf?sfvrsn=3913e4d8_0)

## Service activity overview

Figure 6: A summary of GHWA's service activity from 2018 to 2022



### Referrals

GHWA received **25,120** referrals between 2018 and 2022:

**46%** referrals to the Familial Cancer Program (FCP) **32%** referrals to Obstetrics and General Genetics Services (OGGS)

**21%** referrals to Genetic Paediatric Services (GPS) **1%** referrals to other/unknown



The number of referrals received in 2022 (5,168) was **14% higher** than the number of referrals received in 2018 (4,542).

The most referred age groups were:

**0-4 years** (13% of all referrals)

**30-34 years** (12% of all referrals)

**35-39 years** (11% of all referrals)

This is reflective of GHWA's referral criteria and current service model whereby dedicated service streams are established to meet the needs of paediatric consumers, and adults seeking clinical genetics expertise for pregnancy and/or family planning reasons among others.

## Consumer Geographical Distribution

Of the consumers who attended at least one appointment between 2018 – 2022:



**29%**

resided in the North Metropolitan Region



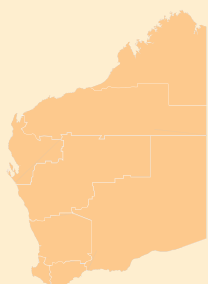
**27%**

resided in the East Metropolitan Region



**24%**

resided in the South Metropolitan Region



**6%**

resided in the South West

**2%**

resided in the Wheatbelt

**2%**

resided in the Great Southern

**2%**

resided in the Midwest

**2%**

resided in the Goldfields

**2%**

resided in the Pilbara

**1%**

resided in the Kimberley

**3%**

resided in other/unknown

This distribution aligns closely with (within 1% of) the geographical distribution of the entire WA resident population over the same time period<sup>12</sup>.



## Appointments and Clinical Service Events

**17,504** appointments were scheduled with **15,513** delivered<sup>13</sup> between 2018 and 2022:

**31%** appointments delivered by FCP clinics

**25%** appointments delivered by OGGS clinics

**24%** appointments delivered by GPS clinics

**14%** appointments delivered by Outreach clinics

**3%** appointments delivered by Cardiac and Renal clinics

**2%** appointments delivered by Collaborative clinics<sup>14</sup>

**1%** appointments delivered by other/unknown

### Of the appointments delivered by Outreach clinics:

**55%** were delivered in Metropolitan Perth

**19%** were delivered in the South West

**10%** were delivered in the Midwest

**6%** were delivered in the Great Southern

**6%** were delivered in the Goldfields

**4%** were delivered in the Pilbara



The number of appointments delivered in 2022 (3,520) was **30% higher** than the number of appointments delivered in 2018 (2,702).



The median Did Not Attend rate was **10%**.



**35,760** other service events<sup>15</sup> were delivered.

*Note: The activity data presented above spans a 5-year period from 1 January 2018 to 31 December 2022. The data were sourced from Kin Trak, GHWA's internal database for recording consumer information. When considering the data, it is worth bearing in mind the profound impact the COVID-19 pandemic had on the Western Australian health system and its consumers during a large proportion of this period. The COVID-19 pandemic disrupted the way public outpatient services were delivered due to hospitals needing to shift priorities and the allocation of resources at the time. Staff shortages due to COVID-19 infection also affected service capacity. Additionally, many consumers were affected by various public health and social measures (e.g. community-wide lockdowns, social distancing, self-isolation), which may have altered normal patterns of health seeking behaviour. Any trends that emerged may therefore not reflect the trends that would have been observed had the circumstances remained ordinary across the entire 5-year period.*

<sup>12</sup> Australian Bureau of Statistics (2023). Australian Bureau of Statistics, Australian Government. [Internet]. Available from: <https://www.abs.gov.au/>

<sup>13</sup> This figure signifies the number of distinct appointments delivered by GHWA and therefore does not encompass all claimable events generated in the delivery of family-based appointments.

<sup>14</sup> GHWA participates in a range of collaborative clinics with other (non-clinical genomics) medical specialties across the Western Australian health system. These encompass clinics hosted by GHWA as well as external clinics hosted by other Western Australian public hospitals.

<sup>15</sup> Other service events comprise of activities such as taking or updating medical and family health history, discussing reason for referral and purpose of an appointment (or why an appointment is not being offered), providing anticipatory guidance about testing and clinical matters to consider after test results are received, notifying of test results, and providing psychosocial support (counselling) after an appointment, results or procedure.



# Service challenges

There are several issues that impact upon GHWA's ability to deliver timely and equitable clinical genomic speciality services. Some of these are system pressures resulting from the rapid advancement in genomic technologies and the consequent increasing clinical utilisation of genomic testing, which adds to the demand on GHWA's services. Whilst many of these challenges are outside of GHWA's control, it is necessary for these to be considered in planning for future service delivery. Additionally, there are some issues relating to the efficiency, accessibility, and sustainability of GHWA's services that would be beneficial for GHWA to address going forwards. These key service challenges are outlined below.

## Increasing complexity of genomic testing

- The complexity of genomic testing has expanded the work GHWA must undertake in pre-appointment preparation to ensure the most appropriate testing options are offered, the amount and type of information needed to be conveyed to consumers to enable informed consent for testing, and in the interpretation and explanation of (sometimes ambiguous) test results.
- As more disease-causing genes and gene variants are discovered, this will further increase demand for GHWA's expertise.
- There are limited criteria and guidelines available to support clinicians to select the most appropriate genomic test for patients, with complex disease presentations frequently requiring multiple approaches to confirm a genetic diagnosis.

## Extended turnaround times for genomic testing

- The capacity and capability of pathology services to meet the increasing demand for genomic testing influences the testing turnaround times and subsequently GHWA's timeliness of communicating results to consumers and referrers.
- Currently the wait time for genomic testing through WA's public pathology provider PathWest is variable depending on the test complexity and clinical urgency and there are no processes or systems to support the monitoring and communication of genomic test status with referrers.

## Limitations of funding for clinical genomic speciality services

- GHWA is primarily funded by activity-based funding (ABF), a national funding system used in Australian public hospitals so that hospitals receive funding based on the number and types of services they provide. GHWA claims ABF for outpatient services it delivers or leads (in the case of collaborative clinics).
- ABF should encompass all the costs associated with delivering a service, and is calculated on an average price for that service. However, the ABF price allocated is based on retrospective costing data from across Australia and is not sufficiently responsive to the fast-paced advancements in utility and complexity of genomic tests that are often associated with relatively significant costs.
- Without dedicated extra funding (except some funding through research programs), GHWA and PathWest have been hindered in facilitating consumer access to new genomic tests, including tests added to the Medicare Benefits Schedule.<sup>16</sup> This has the potential to lead to inequity of access to these tests for GHWA consumers.
- GHWA does not receive funding for certain activities it performs, such as inpatient consults, multidisciplinary or collaborative clinics where it is not the lead, and when it provides advice and training to other health professionals (e.g. on-call geneticist or genetic counsellor, MDT meetings, or case discussions with PathWest).

<sup>16</sup> Under MBS rules, Medicare rebates are only available to private patients. As such, public hospitals cannot claim Medicare rebates for pathology tests performed for public consumers as part of a hospital admission or outpatient clinic.



### Increasing support required by non-genomic specialists

- As the clinical utility of genomics increases, a growing number of non-genomic specialists are seeking expertise and support from GHWA around ordering and interpreting genomic tests, which has contributed to a rise in GHWA's workload for advising, counselling, and variant interpretation, which is often not adequately funded by ABF.
- No state-wide formal education program aimed at improving the genomic-related capabilities of non-genomic specialists currently exists.

### Private clinical genomic specialty services

- Like other clinical healthcare specialties, private services play a role in the delivery of clinical genomic specialty services, although accessing private services can be a financial burden for some consumers and creates inequity in access, including to genomic testing.
- Additionally, although the scale of private clinical genomic specialty services in WA is growing, it remains relatively modest in size in relation to public services when compared to other healthcare specialties. Due to the small and highly specialised nature of the specialist genomic workforce, GHWA must take into consideration how a significant growth in private clinical genomic specialty services may impact GHWA's workforce.
- Furthermore, consumers attending a private genomics clinic may have at-risk family members requiring cascade counselling or testing who, for a number of reasons, may choose to go through the public health system. In these situations, they will be referred to GHWA, thus further increasing wait times for public clinical genomic specialty services (including genomic testing).

### Specialised genomic workforce shortages

- Training opportunities for both geneticists and genetic counsellors are constrained by the number of institutes offering certified programmes. The Royal Australasian College of Physicians oversees all Australian geneticist training, while only 2 universities nationwide offer a Master of Genetic Counselling.

- Following training and certification, limited positions, opportunities in other states, and competition with the private and research sectors influence GHWA's ability to recruit additional staff and retain them.
- This has resulted in a gap between GHWA's current workforce numbers and recommended national benchmarks for geneticist and genetic counsellor positions<sup>17</sup>.
- GHWA staff, in particular genetic counsellors, have limited workforce development opportunities (e.g. a lack of protected time and funding for participating in research and training activities) due to constraints within the current professional award. This limits genetic counsellors' ability to participate in professional development required for professional registration.

### Lack of formal agreements to work across Health Service Providers

- GHWA currently develops collaborations with specialists on an *ad hoc* basis as capacity allows, which potentially results in inequity in the delivery of clinical genomic specialty services across HSPs.
- Furthermore, there is a lack of formal agreement between GHWA and the HSPs as to how they deliver and fund collaborative clinics.

### Limited awareness of the service

- Some medical practitioners and staff working in primary and community care in WA are not aware of GHWA, the scope of service, the criteria for referral, or the ability to seek *ad hoc* advice from the service. This can prevent some consumers who would benefit from GHWA's services being referred.
- It also appears some consumers and carers do not fully understand the reason for their referral to GHWA and may not be informed of the services available to them upon acceptance of this referral, leading to reduced accessibility for these individuals.

<sup>17</sup> Human Genetics Society of Australasia (February 2024). Clinical Genetic Services Framework (2024GL01). HGSA [Internet]. Available from: <https://hgsa.org.au/Web/Consumer-resources/Policies-Position-Statements.aspx>



### Difficulties with accessibility

- Some stakeholders feel waiting times for appointments are unclear at the time of referral and can be too long. This can be challenging for consumers and carers, some of whom experience significant anxiety or uncertainty during this time, as well as referring clinicians, who may not receive communication about their patient until the completion of their appointment.
- The location of GHWA's central hub at KEMH, a hospital with ageing infrastructure and limited space for expansion, and limitations on clinic spaces at other hospitals, impacts upon GHWA's ability to increase capacity to meet the growing demand for services.
- KEMH's clinic spaces are not appropriate to conduct all clinical examinations required and are not always suitable for adults who do not have a maternity-related health concern or consumers with accessibility requirements, such as those with physical or sensory impairments due to their genetic condition.
- The physical location of GHWA's services also poses a geographical barrier for consumers living in rural, regional, and remote areas for whom an in-person appointment is clinically required or preferred. This is particularly difficult for those living in regions not serviced by GHWA's outreach clinics.

- People who identify as Aboriginal or CaLD experience an inequity of referrals and access to clinical genomic specialty services. These inequities are the result of several cultural and language barriers such as a lack of information and resources available in their first language, difficulties in awareness and understanding of how to access the service, stigmatisations of certain health issues, or insufficient provision of culturally informed care.

### Outdated clinical genomics database

- GHWA's current data entry and management system (Kin Trak) is a legacy software no longer supported by contemporary information and communications technology (ICT), resulting in barriers to integration with other clinical data systems including the hospital patient database, webPAS.
- This hinders the ability to collect the required ABF data, leading to inefficiencies due to the need to double enter patient data into the hospital patient database webPAS and inaccuracies in data entry and reporting.
- It also inhibits GHWA's ability to accurately monitor its service activity, which is critical for measuring true service demand and informing future service planning.<sup>16</sup>





## Future service model

This section outlines the optimal service model for GHWA in the future (see Figure 7 for a conceptual diagram of this model). It is acknowledged this ideal state will likely not be fully realised within the 5-year period covered by this Plan, and that achieving this ambitious vision will require ongoing commitment and support in the years following its implementation.

### Ensuring equitable, responsive, person- and family-centred services

As emphasised in the WA Genomics Strategy, person- and family-centredness is a central priority for all genomic services and will remain at the core of any future service developments for GHWA. Going forwards, GHWA will continue to provide a person- and family-centred service throughout WA for the diagnosis, management, and support of individuals with or suspected with genetic conditions, as well as their families and carers. Furthermore, GHWA will work with its partners to promote and support the delivery of person- and family-centred genomic health care in a manner that aligns with the underlying principles of the WA Genomics Strategy.

GHWA's current operational model, encompassing centralised services with networked outreach clinics, will be enhanced to facilitate Western Australians to have equitable access to services closer to where they live. This will require improved service efficiency, enhanced capacity, and the exploration of innovative workforce and service models to meet increasing demand, particularly in rural and remote areas. A mixed mode of service delivery, including face-to-face consultation and virtual or digital health approaches to care, will ensure GHWA is responsive to consumer preferences and accessibility requirements in line with their clinical needs and will provide further opportunities to minimise the burden of travel for both consumers and GHWA staff. In partnership with stakeholders, GHWA will also develop innovative approaches to service delivery that consider how the diverse needs of consumers across various ethnic, social, and cultural groups can be met to help address existing barriers to access. GHWA will also partner with providers within hospital, primary, social, and community healthcare services to ensure consumers receive appropriate follow up and support for their genetic condition across the care continuum.

### Working at the highest scope of practice

GHWA will continue to offer clinical genomic specialty services to all eligible Western Australians. However, with the demand for genomic services likely to increase over time, GHWA will need to focus its resources on optimising care for consumers, carers, and families with complicated symptoms and/or who require a greater level of support. GHWA will therefore prioritise the diagnosis and management of individuals and families with complex, multisystem, and rare genetic conditions, including inherited cancers. This will involve collaborating with stakeholders to develop clearly defined referral pathways to ensure consumers with complex needs are directed to GHWA and/or other appropriate clinical services. These pathways will facilitate timely referrals to GHWA for consumers who most need GHWA's specialty services, as well as family members who are eligible for cascade testing and counselling.

In the long term and as capacity allows, GHWA will explore opportunities to coordinate the ongoing medical care and management of consumers with rare genetic disorders or undiagnosed conditions where clinical expertise and management is not available through other services. This will include facilitating access to clinical trials and emerging therapies for these consumers. In addition, GHWA will drive care coordination and health surveillance for complex, multisystem genetic disorders, for which a priority will be placed on adults with a genetic predisposition to cancer (with management and surveillance of children with inherited cancer predisposition syndromes to be led by PCH).





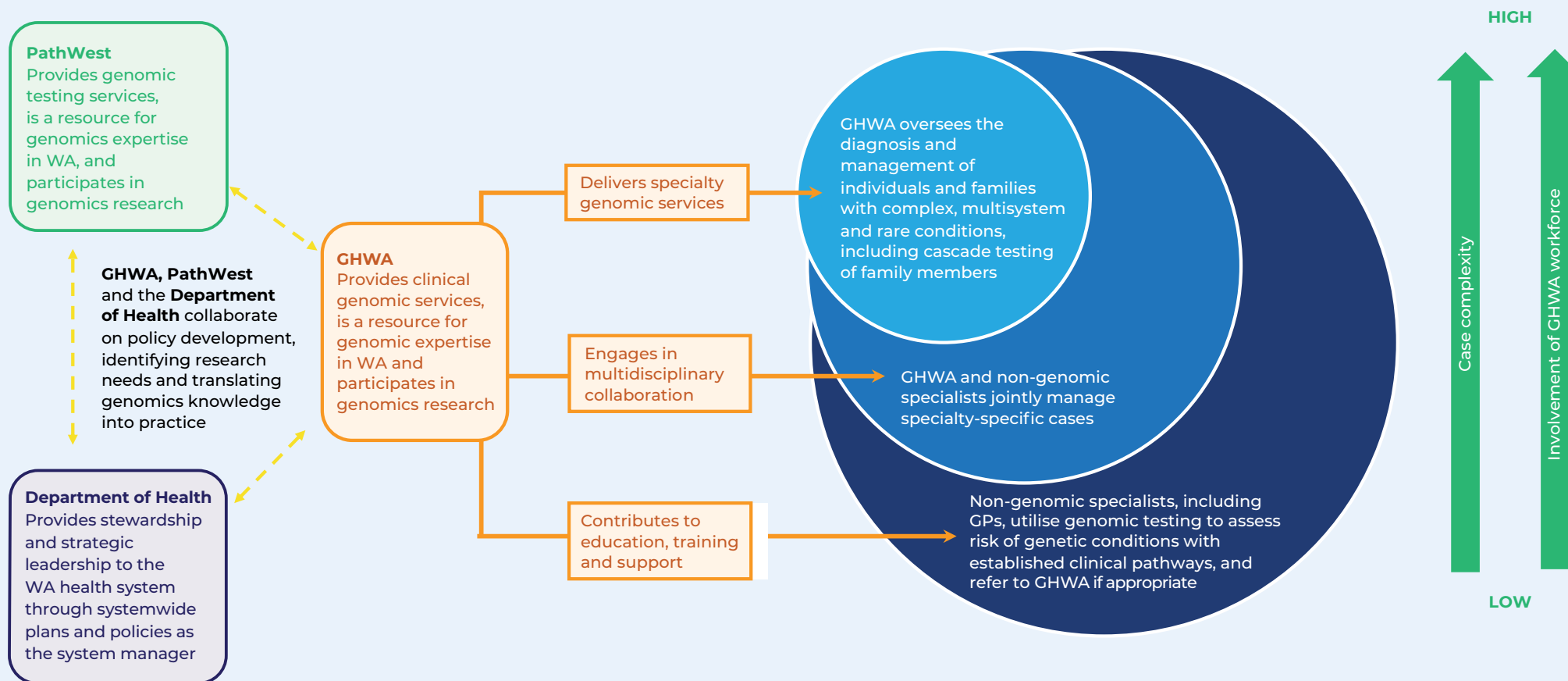
## Supporting the implementation of genomics across WA

The rapid advancement in genomic knowledge and technology has contributed to the broader implementation of genomics across the Western Australian health system as clinicians in other specialties become more aware of the advantages of utilising genomics to inform precision medicine approaches. Whilst this can help to relieve pressure on GHWA and provide consumers with timelier access to genomic testing by a specialist they are already familiar with, there are recognised challenges to the delivery of genomic medicine by non-genomic specialists. There is a pertinent need for the Western Australian health system to provide education, training, and development opportunities along with appropriate policies and clinical pathways to ensure genomic health care is delivered by non-genomic specialists in a safe, person- and family-centred manner. This includes upholding consumer autonomy and informed consent and minimising potential harms such as over-diagnosis, data misuse, discrimination, and stigmatisation.

It is anticipated that GHWA's involvement in this work would be stratified based on genomic complexity, GHWA's expertise, patient numbers and impact to consumers and families and would complement other measures introduced by the Department of Health to guide the safe adoption of genomics into the Western Australian health system as part of the implementation of the WA Genomics Strategy. It will also be necessary to gain input and support from other services such as PathWest and utilise clinical and genomic testing data to identify the clinical areas of need for GHWA's support. Given the value of collaborations for both consumers and clinicians in genomic health care delivery, GHWA's role will include increased participation in joint clinics and MDT meetings. In addition to this, GHWA will collaborate with stakeholders to co-develop clinical guidelines or decision aids which will support a person- and family-centred model of genomic healthcare delivery while reducing the time to access genomic testing and facilitating ongoing patient management.

GHWA will also support the Department of Health, HSPs, and other relevant organisations in training and educating health professionals in other clinical specialties, including primary care, to develop their knowledge and skills to directly request appropriate genomic testing within their scope of practice and understand how results can impact on patient care and their family. It is recognised these efforts will vary depending on the unique needs of different non-genomic specialties and should take into consideration existing genomic knowledge, models of care, and specialist and consumer preferences.

The rapid pace of development in genomic knowledge means research is recognised as a fundamental part of clinical genomic service delivery, to enhance the understanding, prevention, and treatment of genetic conditions. GHWA will play a key role in supporting the timely translation of genomic research into clinical care by participating in local, national, and international research programs, as well as contributing to the development of evidence-based guidelines and innovative service design.



**Figure 7:** A conceptual framework showing GHWA's role in the future state of genomic healthcare delivery in WA.



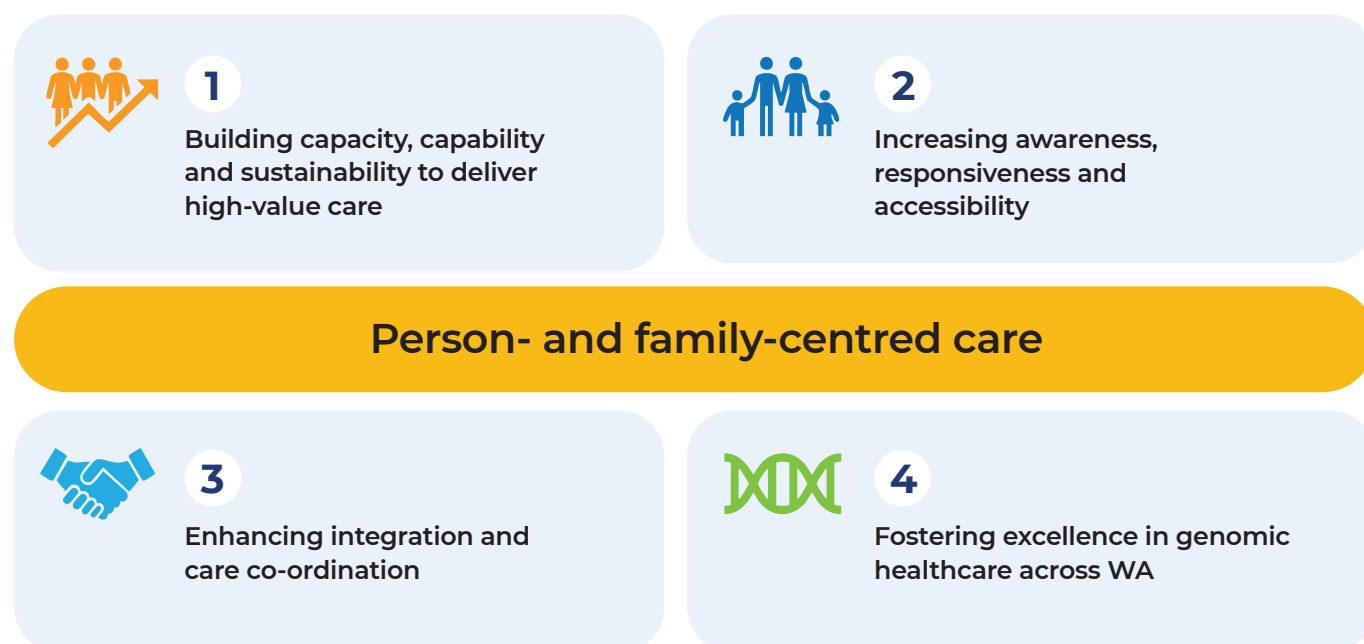
## Service directions

Four service directions have been outlined below to provide an overview of the key objectives and actions to guide the delivery of GHWA's services over the next five years:

1. Building service capacity, capability, and sustainability to deliver high-value care
2. Increasing awareness, responsiveness, and accessibility
3. Enhancing integration and care coordination
4. Fostering excellence in genomic health care across WA

Person- and family-centred care is essential to the delivery of safe, high-quality care, and is a central priority underlying all service directions for GHWA (see Figure 8).

Engagement and partnership with consumers and carers in the planning, delivery, and evaluation of GHWA's services is paramount, and should be prioritised across all service directions to ensure that value-based, person- and family-centred care is continued into the future.



**Figure 8:** GHWA's service directions for the next five years.



## Service Direction



### Building service capacity, capability, and sustainability to deliver high-value care

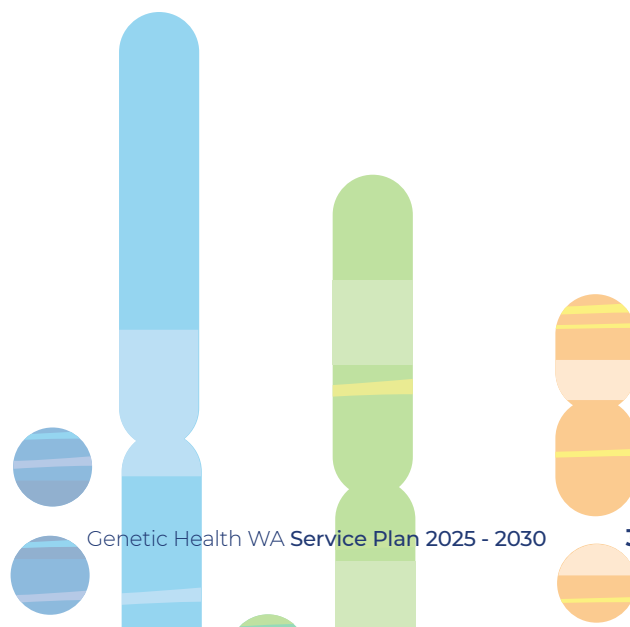
High-value health care aims to reduce system waste, eliminate unwarranted clinical variation, and direct resources to areas of greatest need. This is particularly important to realising an integrated genomic healthcare environment where consumers, carers, and family members receive safe, high-quality, and person- and family-centred care based on the best available evidence.

Resources, workforce, and processes are fundamental building blocks to efficient health service delivery and are crucial elements to ensuring GHWA's provision of sustainable, high-quality, safe, and person- and family-centred clinical genomic specialty services into the future. Dedicated planning, leadership and investment will be required to support the design and implementation of effective, sustainable, and innovative service models.

Expanding service capacity will be necessary and this will be partly addressed by GHWA in the immediate term by optimising operational efficiency (including through streamlining processes, utilising digital tools, and exploring alternative workforce models) and implementing innovative models of service delivery. However, in optimising service processes, the experience of consumers, carers and family members will remain a key priority, and GHWA should continue to ensure that they receive high-quality, person- and family-centred care amidst operational changes.

GHWA will undertake regular service and workforce planning, which will be underpinned by strengthened data collection systems that enable ongoing service monitoring, and evaluation and horizon scanning. Service improvements will be designed and implemented in collaboration with stakeholders to ensure that these are aligned to best practice and meet the needs of consumers and families, the health workforce, and the Western Australian health system. This will include drawing on service-level data to inform the development of new evidence-based models of care and to drive consistency in genomic service delivery across the Western Australian health system.

Achieving the objectives in this service direction will support the ongoing delivery of timely, equitable and effective person- and family-centred clinical genomic specialty services for Western Australians. It will also allow GHWA to operate at the top of their scope of practice, implement key priorities arising from this service plan, and utilise their expertise to explore and undertake new key roles within the Western Australian health system to help optimise the provision of clinical genomic specialty services in WA into the future. Ultimately, new resources will be required to continue expanding the capacity, capability, and sustainability of GHWA to achieve this vision. These additional resources will be sourced through normal budgetary processes, supported by high-quality service data and evidence-based service planning.





## Objectives

- 1.1** Review and streamline internal operating models and pathways to improve efficiency and consistency of service delivery (*Actions 1-5*).
- 1.2** Explore and adopt innovations in service delivery (including digital tools where appropriate) to optimise processes and increase productivity (*Actions 6-7*).
- 1.3** Develop a sustainable genomic specialist workforce with the capacity and capability to deliver best-practice, safe, and effective clinical genomic specialty services across WA (*Actions 8-11*).
- 1.4** Engage in evidence-based service planning, monitoring, and evaluation in collaboration with stakeholders to ensure GHWA continues to provide services which meet the needs of the Western Australian population (*Actions 12-14*).

## Actions

- 1.1** Complete the implementation of a new clinical database and establish consistent processes for data recording to improve service flow, data control, and reporting efficiency and accuracy.
- 1.2** Develop, implement, and maintain standard operating procedures, guidelines, and protocols to reduce service variation.
- 1.3** Review and optimise the consumer pathway through GHWA, including refining communication around referral procedures and pathways.
- 1.4** Develop and regularly review Western Australian health system Referral Access Criteria and supplementary educational materials and explore the potential integration of referral forms with the Smart Referrals WA system to standardise the referral process by improving communication between referrers and GHWA, consistency in referral acceptance and triage practices, and quality of referrals.
- 1.5** Continue to explore the optimal subspecialty model for GHWA to ensure it best reflects the evolution of genomics knowledge and demand, such as in areas of adult cardiac and renal genetics.
- 1.6** Explore and pilot innovative methods to streamline the collection of family history and medical information, such as digital consumer-driven forms.
- 1.7** Implement digital mechanisms to improve communication and information-sharing with referrers, including secure messaging systems and linking of digital records to ensure accessibility of clinical letters.
- 1.8** Utilise service data and clinical frameworks to support workload projections and identify staffing needs.
- 1.9** Explore alternative workforce models to improve service efficiency, capacity, and sustainability (e.g. allied health workers/nurses/psychologists trained in genomics, intake and data entry assistants).
- 1.10** Increase staff access to continuing professional development and networking opportunities to ensure staff knowledge and skills meet the evolving needs of GHWA.
- 1.11** Contribute to the development and implementation of education, training, attraction, and retention programs for the relevant specialised genomics workforce in WA.
- 1.12** Establish key performance indicators for GHWA to facilitate regular service monitoring, evaluation, and quality improvement.
- 1.13** Pilot the prospective collection of service quality data (e.g. patient-reported experience and outcome measures) to understand and optimise service delivery.
- 1.14** Explore and advocate for sustainable activity-based funding models for specialised genomic services.



## Service Direction

# 2

### Increasing awareness, responsiveness, and accessibility

Despite its increasing utilisation in the health system, it is recognised that the awareness and understanding of clinical genomics varies considerably across the community and healthcare professionals. This includes knowledge of genetic risk factors and pathways to accessing clinical genomic specialty services, representing a key barrier to the access to and uptake of genomic health care. GHWA will ensure that information provided to stakeholders is clear, comprehensive, and accessible to help promote and increase awareness of GHWA, the services that it provides, and the referral pathways into the service. Where appropriate, this information will be tailored to and developed in collaboration with relevant stakeholders to ensure that information is fit-for-purpose and disseminated widely and effectively. This will also improve the ability and confidence of stakeholders to identify those who have or are at risk of having a genetic condition and enable them to access clinical genomic specialty services as needed.

Person- and family-centred care involves working with consumers, carers, and their families to undertake shared decision-making and care planning. Quintessential to this is clear, effective communication to facilitate consumer understanding of genomic concepts, the reason/s for their referral, and what will occur during their appointment/s. In addition, person- and family-centred care considers and respects the preferences, culture, and beliefs of consumers, and acknowledges the familial implications of genomics. GHWA will continue

to provide high-quality, person- and family-centred care to consumers, carers, and their family members that is responsive to their needs. Purposeful and responsive stakeholder engagement will be utilised to help understand the needs and preferences of consumers, carers, and family members, and develop strategies and improved models of care to address these, such as through improving communication, exploring person- and family-centred models of care and utilising digital tools to improve consumer access and experiences.

Partnerships with community services are particularly important to understand and address the diverse needs of the wider community, and specifically communities that experience inequity of access. GHWA will focus on refining, developing, and implementing models of care, strategies, and pathways to increase equity of access for populations that experience heightened barriers to receiving genomic health care and which require additional support needs. This will provide GHWA with a strong foundation to explore and expand its strategies to increase access to clinical genomic specialty services for all Western Australians more broadly into the future.

Overall, achieving these objectives will continue to enable consumers, carers, and their family members to be genuine partners in the decision-making and planning of their care, and facilitate the equitable, timely and appropriate provision of clinical genomic specialty services in WA.

## Objectives

- 2.1** Improve the quality, availability, and dissemination of information designed to increase stakeholder awareness of GHWA and the services it provides throughout WA and ensure consumers with the greatest need can access GHWA's services (*Actions 1-3*).
- 2.2** Increase engagement with stakeholders including consumers, carers, community groups, and health and social care providers to develop strategies to enhance access, care, and support for populations that experience heightened barriers to accessing clinical genomic specialty services (*Actions 4-6*).
- 2.3** Review, develop, and implement strategies, pathways, and innovative models of care in partnership with stakeholders to continue being responsive to the diverse needs of consumers, carers, and family members (*Actions 7-8*).

## Actions

- 2.1** Develop a single website to best support stakeholders' information needs and improve awareness of GHWA's services, including by providing:
  - a. Information in different formats (e.g. Easy English, graphics and videos).
  - b. Information translated into languages other than English.
  - c. Additional details of services provided (e.g. clinic locations and types, referral triage principles or categories, conditions not seen and alternative care pathways for these, and indicative waiting times for appointments within each GHWA subspecialty).
- 2.2** Develop and implement a communication and/or engagement plan to promote GHWA to consumers, primary care, and other non-genomic service providers, and increase awareness and understanding of the services provided. This plan should prioritise stakeholders and consumers who historically have not been reached or experience barriers to access.
- 2.3** Contribute to, develop, and promote education material and guidelines for primary care and other non-genomic specialists to enhance and support referrals to GHWA, including materials that build knowledge about how to recognise consumers likely to have a genetic condition and how to gather information necessary for referral to GHWA.
- 2.4** Develop strategies to improve the provision of culturally appropriate care and equity of access for Aboriginal consumers and carers, such as:
  - a. Establishing service models and pathways that integrate Aboriginal values and knowledge and are consistent with Aboriginal sovereignty and self-determination.
  - b. Recruiting Aboriginal staff and providing all staff with cultural awareness training to ensure culturally safe and respectful service provision.
- 2.5** Develop strategies to improve the provision of culturally appropriate care and equity of access for consumers who identify as CaLD, such as:
  - a. Establishing service models and pathways that are specific to individuals from CaLD backgrounds.
  - b. Recruiting staff who are of a CaLD background and providing staff cultural awareness training to ensure culturally safe and respectful service provision.
  - c. Investigating avenues to upskill interpreters in genomic concepts.
- 2.6** Informed by service utilisation data and stakeholder consultations, explore requirements and opportunities to provide additional outreach clinics in rural and regional areas to increase accessibility for underserved populations.
- 2.7** Explore and pilot person- and family-centred models for service delivery to enable consumers to be active partners in decision-making around their care, such as:
  - a. Mechanisms to keep consumers informed of the status of their genomic testing.
  - b. Taking into consideration consumers' preferred times and format (e.g. telehealth or face-to-face) where possible when scheduling appointments and delivering genomic testing results.
  - c. Providing accessible educational resources (e.g. fact sheets or videos on genomic testing and genetic conditions in multiple languages) and digital consenting for genomic testing to increase consumer genomic literacy and allow for more informed consent.
  - d. Developing visual, Easy English, and/or translated summaries for consumers following their appointments to facilitate understanding of appointment outcomes.
- 2.8** Work with WNHS to improve and redesign GHWA infrastructure to better meet rising demand for services and consumer needs such as cultural and physical accessibility





## Service Direction

### 3

#### Enhancing integration and care coordination

Integrated care involves the provision of seamless, effective, and efficient health care that reflects the whole of a person's health needs in partnership with consumers, carers, and family members and other health service providers. This is particularly important to consider in clinical genomics, which is a complex clinical area that spans nearly every medical discipline and has an extensive range of care and management implications (both short- and long-term for the patient, as well as potential implications for family members). Genomic specialists have expertise in providing diagnosis, counselling, and management advice to consumers, carers, and family members at risk of or affected by an inherited condition. However, the ongoing management and support of the patient's health and social needs can involve many other services and health professionals. Providing care that minimises duplication and achieves shared outcomes will be reliant on the development of purposeful partnerships that improve coordination, communication, and connectivity between GHWA and service providers across community health, social and primary care, and hospital services. This will ensure consumers receive the right care, at the right time, and in the right place.

GHWA will extend the emotional and psychosocial support currently provided to consumers and develop strategies to enhance continuity of timely and coordinated services across the care continuum. This includes empowering consumers to access support services, ensuring that clear, comprehensive, and accessible information is provided to referring clinicians about ongoing management, referring patients to other medical and psychological services when required, and developing strong partnerships with professional and community organisations. GHWA will also explore opportunities to promote coordinated care for consumers, carers, and family members who are currently not being adequately served in the Western Australian health system (e.g. those with rare diseases) through potential care coordination roles and joint clinics with other health professionals, ensuring these additional collaborations are formally established based on evidence and need, and are sustainably implemented into the future. It is anticipated these collaborative service models will also enhance the knowledge and confidence of non-genomic specialists to provide person- and family-centred genomic health care and allow GHWA to focus its clinical genomic specialty services on consumers with complex needs.



## Objectives

- 3.1** Refine and develop strategies to improve continuity of care for consumers and carers in partnership with stakeholders, including ongoing management, disease surveillance and care coordination (*Actions 1-6*).
- 3.2** In partnership with HSPs and the Department of Health, review, maintain and expand clinical pathways and arrangements that embed access to specialised genomics services and advice into non-genomic clinical services to ensure all Western Australians have timely access to appropriate and coordinated genomic health care (*Actions 7-9*).

## Actions

- 3.1** Expand GHWA's involvement in evidence-based screening for and management of Tier 1 genomic applications with recognised public health benefits,<sup>18</sup> including:
- a. Enhance and maintain the Familial Cancer Registry and formally embed an adult familial cancer surveillance clinic within GHWA to improve the identification, surveillance, and treatment of individuals and families with familial cancer syndromes for which surveillance is currently not available.
  - b. Explore avenues for GHWA to support the implementation of pathways to improve the identification, counselling, and management of individuals with familial hypercholesterolaemia and the provision of cascade screening and counselling for their family members.
- 3.2** Contribute to efforts to increase accessibility of relevant consumer genomic information (e.g. clinical summary letters) for consumers and clinicians to support ongoing patient management and continuity of care.
- 3.3** Develop and implement standardised processes for consumers to be directed to support services, including peer support groups and psychological support
- 3.4** Work with PathWest to investigate options for re-analysis of inconclusive genomic testing results.
- 3.5** Explore innovative models of care (e.g. co-consultation with primary care providers) to improve continuity of care for consumers, carers, and their family members.
- 3.6** Scope the feasibility of an expanded role for GHWA in coordinating medical care, managing ongoing care and complex psychosocial support needs, and providing disease surveillance for consumers with unmet needs, including those:
- a. With an undiagnosed condition.
  - b. With rare, complex multisystem conditions (e.g. familial cancer syndromes and rare diseases without a clear "home" medical specialty).
  - c. Transitioning from paediatric to adult care.
- 3.7** Evaluate the clinical, economic, and social impact of existing joint clinics and use the outcomes to establish decision-making criteria to guide the sustainable and equitable expansion of GHWA's involvement in collaborative service delivery models.
- 3.8** Formalise arrangements for services provided by GHWA to other HSPs.
- 3.9** Identify the clinical areas requiring additional support from GHWA (e.g. via multidisciplinary work, the provision of education or advice to non-genomic specialists, or assisting with referrals to GHWA) by examining current clinical service and testing data.

<sup>18</sup> US Centres for Disease Control and Prevention (March 2014). Tier 1 Genomics Applications and their Importance to Public Health. CDC [Internet]. Available from: <https://archive.cdc.gov/#/details?url=https://www.cdc.gov/genomics/implementation/toolkit/tier1.htm>



## Service Direction



### Fostering excellence in genomic health care across WA

As the sole provider of public clinical genomic specialty services, GHWA is uniquely positioned to support innovative, high-value patient- and family-centred genomic health care by sharing their specialised genomics expertise across the WA health system. GHWA will partner with the Department of Health, PathWest, and other service providers to develop a strategic and stepwise approach to improve the integration of genomics into clinical practice of a range of medical specialities. A flexible approach will be required with different pathways needed for clinical specialities dependent on the existing clinical pathways, the conditions their patients have, workforce capability, and specialists' awareness and knowledge of genomics.

This will include expanding GHWA's involvement in joint clinics and multidisciplinary team meetings and exploring digital tools and other innovative ways to increase the accessibility and provision of advice to non-genomic specialists to support appropriate genomic testing requests, referrals to clinical genomic specialty services, and communication to consumers about the implications of genomic information. GHWA will also work with the Department of Health, PathWest, and other HSPs in the design and delivery of education and training programs, best practice guidelines and other resources aimed at improving the genomic-related capabilities of the multi-professional healthcare

workforce across all service levels. This may require additional funding to support the procurement and development of educational resources and dedicated staff time for training activities.

Research and innovation are fundamental components in evidence-based service delivery. This is particularly important to consider in the dynamic environment of clinical genomics, with newer, faster diagnostic technologies and emerging range of gene therapies meaning that Western Australians can gain greater access to timely genetic diagnoses, more personalised care, and better outcomes. GHWA will continue to be involved in, utilise, and explore research, innovation, and digital technologies, and together with consumers, clinicians, laboratory scientists, academics, and policy makers, translate genomic research and innovation into clinical practice. This may require investment in research infrastructure, resources, and staff time to better enable GHWA to lead and participate in clinical research activities.

Achieving these objectives will help to ensure the optimal integration of genomic knowledge into the Western Australian health system and facilitate the delivery of high-value genomic health care which benefits all Western Australians.



## Objectives

- 4.1** Collaborate with partners and stakeholders to share best clinical practice, develop clinical and policy guidance, and set direction for future genomic healthcare services (*Actions 1-3*).
- 4.2** Continue to work with stakeholders to support the delivery of genomics in non-genomic clinical services by expanding the provision of advice to non-genomic specialists and providing input on genomics education and training programs for health professionals (*Actions 4-6*).
- 4.3** Participate in and lead research and translation projects to advance the adoption of new genomic technologies and knowledge into clinical practice (*Actions 7-10*).

## Actions

- 4.1** Continue to explore the most effective and suitable pathways for genomic testing which will ensure appropriate and timely access to clinical genomic specialty services for consumers.
- 4.2** Undertake horizon scanning across medical specialties for genomic applications that may have clinical utility in the short- and medium-term.
- 4.3** Continue to contribute advice and expertise to the Department of Health to inform health genomics policy and guidelines.
- 4.4** Investigate options for sustainable dedicated funding to support GHWA's participation in the development and delivery of genomics education and training activities for consumers and health professionals.
- 4.5** Explore digital avenues to provide advice to non-genomic specialists, including enhancing access or linkage to specialty-specific clinical geneticists or genetic counsellors.
- 4.6** Provide input into and support the delivery of education strategies and training models to upskill non-genomic specialists and enable delivery of safe person- and family-centred genomic healthcare services in WA.
- 4.7** Explore opportunities to invest in and grow research capabilities within GHWA to enable greater involvement in clinical research.
- 4.8** Increase participation in the development of evidence-based models of care.
- 4.9** Lead and participate in local, national, and international research translation and quality improvement projects that are informed by the needs of consumers and the health system, and which can be translated and adopted in a timely manner. and participate in local, national, and international research translation and quality improvement projects that are informed by the needs of consumers and the health system, and which can be translated and adopted in a timely manner.
- 4.10** Explore opportunities to increase consumer access to clinical trials and innovative treatments, including those for novel genetic therapies and other precision medicines.



# Future considerations

## Implementation

The Plan will be implemented in a phased approach, with as many actions implemented as possible over the 5-year period depending on the timeframe for each project, the availability of resources, and the strategic priorities of the NMHS and the Western Australian health system more broadly. To achieve this, it will be necessary for GHWA to develop an implementation approach to prioritise the actions for commencement and define measures to assess progress towards realising the objectives.

Building on the co-design approach employed in the development of the Plan, implementation of the actions will be led by GHWA and undertaken in consultation with key partners, including (but not limited to) consumers, other healthcare providers, community support organisations, the Department of Health, and the WA Genomics Strategy Implementation Committee. Additional implementation partners may be identified throughout the implementation period. This will ensure the approach best reflects the needs of all stakeholders and aligns with the implementation of the WA Genomics Strategy across the Western Australian health system.

## Enablers of success

There are several key enablers that will be necessary to successfully implement this service plan. This includes the enablers outlined in the WA Genomic Strategy such as sustainable funding of genomic services; senior leadership and support; accountability, roles and responsibilities; and meaningful partnership and collaboration.

The WA Genomics Strategy highlights the vital role played by senior leadership, and the need for clear roles and responsibilities for all stakeholders to ensure accountability for genomic health care. As such, GHWA will need to identify individuals who can guide the adoption of innovative service models or care pathways and lead behaviour change across the system, as well as work closely with senior leadership at the executive level across HSPs to ensure effective implementation and monitoring of the Plan.

As touched on above, meaningful partnerships and collaboration with all stakeholders at the local, national, and international level will enable GHWA to achieve

the objectives of this plan. While additional resources are likely to be required to establish and build meaningful partnerships, this will ultimately help to expand service capacity and capability through leveraging efforts, identifying synergies, and reducing duplication over the duration of the Plan's implementation and beyond. As a priority, GHWA will explore the ideal mechanisms to embed consumer and carer voices in line with the Department of Health's *Working with Consumers and Carers Toolkit*<sup>19</sup> and the *NMHS Partnership Model*.<sup>20</sup>

As identified in the WA Genomics Strategy, the key areas of genomic workforce, infrastructure and data management would benefit greatly from upfront investment. Such focused investment for GHWA will provide greater opportunities for improvement, growth and expansion, all necessary to realise the ideal approach to future clinical genomic service delivery in WA.

GHWA will need a skilled, agile, and diverse workforce that is committed to delivering high-quality clinical genomic specialty services and able to embrace new ways of working. GHWA will also benefit from contemporary and fit-for-purpose ICT which can facilitate seamless collection and sharing of relevant information to enhance continuity of care. Finally, appropriate infrastructure that enables GHWA to meet the increasing demand for services and the physical, cultural, and psychosocial needs of staff and consumers will be key to ensuring high quality service delivery into the future.

## Review and evaluation

It is anticipated some actions within this plan may require modification over time in response to the rapidly changing genomics environment in WA. Furthermore, it is noted that the Plan includes an ambitious range of actions to implement in the timeframe allocated. As such, any actions unable to be completed within the 5-year timeframe will be carried over and pursued as part of the review and evaluation process. Regular review of progress against the objectives will be undertaken by GHWA in conjunction with NMHS Clinical Planning and reported via the usual clinical governance pathway. The Plan will be evaluated by NMHS Clinical Planning following implementation to assess the impact of the plan on service delivery and inform the development of future GHWA service plans.

<sup>19</sup> WA Department of Health (2023). *Working with Consumers and Carers Toolkit*. Health Networks [Internet]. Available from: <https://www.health.wa.gov.au/-/media/Corp/Documents/Health-for/Health-Networks/WWCCT/Working-with-Consumers-and-Carers-Toolkit.pdf>

<sup>20</sup> North Metropolitan Health Service (2023). *Partnership Model*. NMHS [Internet]. Available from: <https://www.nmhs.health.wa.gov.au/-/media/HSPs/NMHS/Documents/NMHS-Partnerships-Model.pdf>

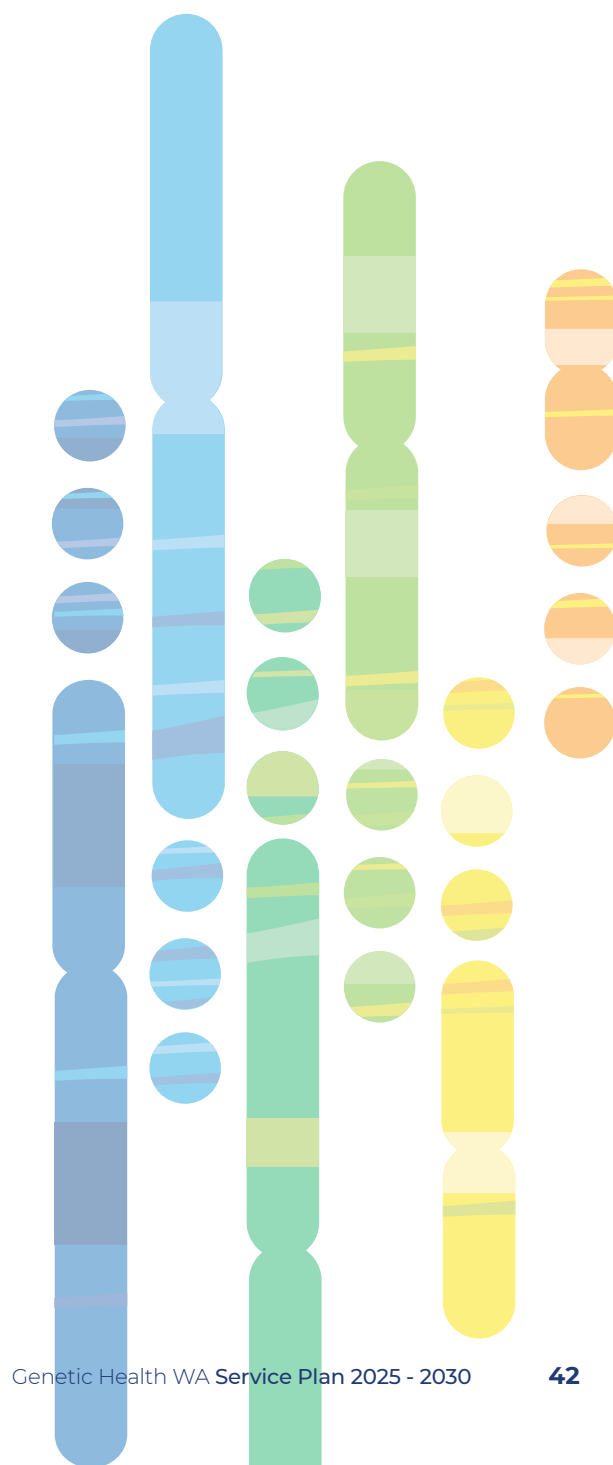


## Appendix 1

### Other genomic healthcare providers in WA

Whilst GHWA is the sole public provider of clinical genomics speciality services in WA, there are other services within the state that deliver genomic health care to the Western Australian public. As WA's only public provider of pathology services, PathWest provides genomic testing services across the state. These services include diagnostics, predictive testing, tumour profiling, carrier screening and prenatal testing for high-risk pregnancies, which PathWest provides to GHWA as well as other services within public hospitals, GPs, and certain other health professionals working in the private sector in WA. PathWest can also facilitate DNA samples being sent to an accredited laboratory interstate or overseas for a specific test to be conducted. Alternatively, some health professionals order tests direct from laboratories interstate or overseas. This is facilitated by their own networks or through research projects.

Additionally, other medical specialties have integrated genomics as part of routine health care without the involvement of GHWA. This includes the application of genomics to inform diagnosis and treatment, administer novel therapies, and monitor emerging public health threats. In these circumstances, the treating health professional has full oversight of the genomic healthcare pathway of the health consumer. For example, GPs are becoming increasingly involved in genomic testing. In many cases they refer health consumers suspected of having a genetic disease to the relevant medical specialist or GHWA. However, there are occasions where they will order tests directly. These are mostly non-invasive prenatal tests or single gene tests available through the Medicare Benefits Schedule.







## Appendix 2

# GHWA's role in the implementation of the WA Genomics Strategy

The WA Genomics Strategy outlines a framework for future genomic health care in WA, guided by its five priority areas. A coordinated systemwide approach will be required to ensure this vision is realised, with collaborative efforts from all stakeholders and oversight from the Department of Health. As the sole public provider of clinical genomic specialty services in WA, GHWA will play a crucial role in supporting the future genomics ecosystem as outlined in the WA Genomics Strategy.

All five priority areas of the WA Genomics Strategy (see Box 4) are relevant to GHWA's current operational model and future service planning. In recognition of the familial nature of genomic information, the WA Genomics Strategy emphasises that person- and family-centredness should be embedded at the core of all genomic services delivered within WA. This means a person and family-centred approach will be taken in all aspects of genomic healthcare delivery, the management of genomic data, and genomic-related research. The focus will 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, while also supporting safer and more effective care. In doing so, genomic providers will respect the ethnic, cultural and socio-economic diversity of consumers and families.

The document also highlights the crucial role of genomic healthcare services such as GHWA in supporting the equitable and person- and family-centred integration of new genomic advancements into the Western Australian health system in a timely manner. This will require a coordinated approach between PathWest, GHWA and other providers of health services to grow genomic capacity across the Western Australian health system. New and innovative service models will be needed to ensure genomic advancements can be incorporated into the Western Australian health system in a safe and equitable manner to bring the greatest benefit to Western Australians close to where they live.

It is advised in the WA Genomics Strategy that the efficient and sustainable implementation of genomic advances will require a coordinated effort and investment to develop, attract and retain the required

specialised genomic workforce (which includes GHWA's staff), as well as increase the genomic literacy across the broader healthcare workforce. It also notes the need for digital solutions that enable the appropriate access and use of genomics in health care, which encompasses the efficient, appropriate and secure collection, storage and sharing of genomic and phenotypic data across the Western Australian health system. Finally, the WA Genomics Strategy recognises the importance of encouraging health professionals (including those employed by GHWA) to collaborate in research and innovation projects to support the translation of genomic knowledge into healthcare services.

### **Priority 1: Person- and Family-centredness**

**Goal 1:** To ensure consumers, carers, families and communities are at the heart of how genomic healthcare is designed, delivered and evaluated.

### **Priority 2: Genomic Healthcare Services**

**Goal 2:** To achieve the optimal integration of genomic knowledge in the WA health system to deliver high value care that is timely, equitable and safe.

### **Priority 3: Workforce, Education and Training**

**Goal 3:** To develop a sustainable health workforce with the appropriate capacity and necessary education and training to deliver genomic healthcare services.

### **Priority 4: Digital Health and Data**

**Goal 4:** To establish the digital health and data solutions, protocols and standards needed to optimise the delivery of responsible genomic health care.

### **Priority 5: Research and Innovation**

**Goal 5:** To have a health system that values and supports the creation and translation of genomics research and innovation.

**Box 4:** The WA Genomics Strategy priorities and goals.



## Appendix 3

### Co-design approach

#### What is co-design?

Co-design is a deliberate and collaborative approach to designing policies, services and research done in partnership with consumers and the community to ensure that results effectively address the needs of end-users<sup>21</sup>. Co-design places equal value in the input from consumers as other stakeholders, seeking to actively learn from their experiences to identify issues, define problems and design a solution. Rather than other engagement approaches where feedback from end-users is sought after the development of projects, co-design involves them at the outset of a project. It is widely recognised that co-designed services are more likely to meet the needs of end-users, improve services and generate more innovative ideas.<sup>22 23</sup>

#### The co-design approach for the Plan

The approach to co-design adopted for the development of the Plan brought together people with lived experience of GHWA, representatives from community organisations, clinicians, and other health staff from the beginning to shape and provide input on the project planning, design and delivery of the stakeholder engagement activities, interpretation of findings from the stakeholder engagement activities and the drafting of the Plan. Two project oversight groups were established with representatives from the range of GHWA's stakeholders, including a GHWA consumer and GHWA carer representative, and both were co-chaired by a lived experience consultant. This enabled a discrete co-design team to be formed, with a balanced representation of people with lived experience of GHWA and people with learned experience of GHWA or treating people with genetic conditions (e.g., GHWA staff or clinicians) to advise on the co-design of the Plan.

#### Project oversight

##### Co-Design Team

The GHWA Service plan Co-Design Team (Co-Design Team) was established to bring together people with lived and learned experience of GHWA and involve them in all stages of development of the Plan.

- 2 consumer representatives (one patient and one carer)
- 2 representatives from community organisations
- 1 representative from GHWA
- 1 lived experience consultant (co-chair)
- 1 Department of Health Project Manager representative (co-chair)
- 1 Department of Health Project Team member

The Co-Design Team met 8 times from March 2023 to July 2024 to collaborate throughout the project, including on the development of the stakeholder engagement plan. As GHWA is a service operating throughout WA with a wide range of stakeholders, the Co-Design Team prioritised engaging with as many different stakeholders as possible by using a range of engagement methods (including surveys, focus groups, interviews, and workshops) and recruitment strategies.

The Co-Design Team was guided by the *Working with Consumers and Carers Toolkit* (Department of Health) guiding principles:

1. Accessible and inclusive
2. Respectful and safe
3. Authentic
4. Clear communication and transparency
5. Purposeful
6. Committed and accountable

##### Working Group

The role of the GHWA Service Plan Working Group was to provide strategic guidance and expertise on clinical planning and the provision of genomic health care. The Working Group included all the members of the Co-Design Team in addition to members representing:

- Primary care referrer to GHWA
- Primary care liaison
- Non-genomic medical specialist referring to GHWA
- Non-genomic medical specialist not currently referring to GHWA

<sup>21</sup> Blomkamp, E (2018). The Promise of Co-Design for Public Policy. *Australian Journal of Public Administration*, 77(4), 729-743.

<sup>22</sup> McKercher (2020). *Beyond sticky notes: Co-design for real: Mindsets, methods and movements*. Sydney, Australia: Beyond Sticky Notes.

<sup>23</sup> Steen, M, Manschot, M, & De Konig, N (2011). Benefits of Co-design in Service Design Projects. *International Journal of Design*, 5(2), 53-60.



- Aboriginal Community Controlled Health Service
- Genomic testing provision
- Clinical planning
- WACHS

The Working Group met in April 2023, December 2023, and July 2024 to discuss the stakeholder engagement plan, outcomes from the stakeholder engagement activities, and the incorporation of stakeholder feedback into the Plan.

### Stakeholder engagement activities

Eight distinct stakeholder engagement activities were conducted by the Health Consumers' Council of WA (HCC) and the OPHG to inform the Plan (Figure 9). The aim of these activities was to understand the experience and perspectives of stakeholders on the current and future service delivery of GHWA. Each activity was tailored to the stakeholder group targeted and covered a broad range of topics.



**Figure 9:** The Stakeholder engagement activities undertaken for the development of the Plan.

#### Community survey

A community survey was hosted from July to August 2023 by the HCC. The survey was completed by 252 consumers and carers from GHWA, with representation from the 3 subspecialties currently offered by GHWA. This included consumers who self-identified as:

- Aboriginal and/or Torres Strait Islander (3%)
- CaLD (10%)
- Currently living in a rural, regional or remote area (15%)
- Living with and/or caring for someone with a disability (26%)

#### Lived experience focus groups and interviews

Three focus groups and 9 interviews were conducted by HCC with 17 consumers and carers from August and September 2023, including:

- 9 consumers who identified as CaLD
- 2 consumers who identified as Aboriginal and/or Torres Strait Islander
- 5 consumers who identified as living in a rural, regional or remote area

These consumers were evenly split across the subspecialties at GHWA.



### Clinician and Support Service survey

The Clinician and Support Service survey was hosted from June to August 2023 by the OPHG for primary care specialists, other medical specialists, and support service staff. This survey was completed by 26 clinicians, including 7 GPs and 19 non-genomic medical specialists. No support service staff completed the survey.

### GHWA staff survey

A survey for GHWA staff was internally hosted by the OPHG in August 2023. This survey was completed by 18 staff members in various roles including clinic coordination, data management, clinical geneticists, and genetic counsellors.

### GHWA staff focus groups

Three focus groups with GHWA staff were held in August 2023, with 25 staff members participating. This included staff across a range of roles, including genetic counsellors, clinical geneticists, administration staff, data management and clinic coordination staff.

### Primary care focus group

One focus group for primary care providers run by an external facilitator was held in August 2023. It was attended by 2 GPs practicing in metropolitan Perth.

### Clinician interviews

Eleven interviews were held with 13 healthcare professionals throughout August and November 2023. Interviewees included 4 non-genomic specialists participating in multidisciplinary clinics with GHWA, 6 non-genomic specialists who refer to GHWA, one GP who refers to GHWA and 2 staff members from a community health service. Specialists interviewed were from a range of clinical disciplines and were employed at various HSPs within the Western Australian health system.

### Multistakeholder workshop

A multistakeholder workshop was held on 12 October 2023, with 40 participants attending including:

- 15 consumers and carers who have attended an appointment with GHWA
- 4 GPs
- 10 other medical specialists
- 8 GHWA staff members
- 3 Western Australian health system representatives

As part of the workshop, guiding statements for GHWA's future service delivery were proposed and tested with participants, and these were used to inform the drafting of this service plan.

### Health Service Provider representative discussions

Discussions were held with representatives nominated by the executive leadership of EMHS, SMHS, CAHS, WACHS and PathWest in November 2023. Representatives held varying roles within their HSP, including consultant physician, Head of Department or Executive Director.

### Public consultation on the draft Plan

The draft Plan was open for public consultation from 6 to 31 May 2024 in the form of an online survey hosted on the Department of Health's consultation platform, Citizen Space. There were 23 responses to the survey, as well as 3 submissions sent directly to the OPHG via email. Respondents included health consumers, health professionals, representatives from patient support organisations, and policymakers.

### Outcomes of stakeholder engagement activities

The learnings from all stakeholder engagement activities were discussed with the Co-Design Team and Working Group and were used to inform the actions, objectives, and service directions within the Plan, alongside findings from desktop reviews of relevant strategies, guidelines, plans and literature.

Suggestions for future service delivery put forward by stakeholders that were deemed too specific to be included within the Plan itself were noted for consideration during implementation of the Plan.

