

GENETIC HEALTH SERVICES IN WESTERN AUSTRALIA

A strategic plan 2005-2010

Developed by the Genomics Directorate in consultation with the Western
Australian Genetics Council

Executive Summary

A strategic plan for Genetic Health Services was developed following gap evaluation, capability mapping and needs assessment. Consultation occurred with stakeholders under the auspices of the WA Genetics Council (Appendix 1).

Efficiencies have been sought within existing service delivery structures and consideration given to improving cohesion across the networks. To ensure that health delivery keeps pace with the rapid developments in genetics, the requirement for increased investment in this field is unavoidable. There are five areas where improvements are required, which has informed the development of the following strategic initiatives:

Service delivery

Increase capacity of medical genetic services to meet current and projected demand.

Monitoring and Evaluation.

Develop policies from evidence-based evaluation and public engagement that enable the advances in genetic technology to improve health care delivery.

Education

Allocate resources to coordinate existing networks and increase cohesion to provide comprehensive genetics education and training at all levels.

Health consultation

Engage consumers and health professionals in collaborative planning of new genetic health services.

Research and Development

Implement research initiatives designed to monitor and integrate genetic advances into clinical practice.

This strategic plan aligns with the aims of the Health Reform Implementation Taskforce (HRIT) to effect change within the WA health system by "Delivering a Healthy WA".

Vision for Genetic Services in Western Australia

Genetic health services in WA will have high professional and ethical standards, reflecting the needs and cultural values of our diverse community. The component parts will be organised in a coordinated manner so as to provide best use of the available public funds.

Introduction

Genetic conditions, once considered to be rare paediatric or obstetric disorders, are having greater impact on the practice of medicine as more and more diseases are found to have a genetic basis. For example;

- genetic disorders account for up to 40% of the work of hospital-based paediatric practice¹;
- genetic predisposition to disease accounts for 30% of premature deaths².
- 10,000 single gene disorders occur in approximately 5% of the population, many resulting in premature death or requiring long term treatment;
- by the age of 60, six out of ten people will develop a disease that has a genetic component;
- evidence that predisposition to adult chronic disease is determined during fetal development³ promises that a genetic component of disease in every individual can be identified or modified in the future.

Genetics has more recently been determined to be a factor in the development of many common chronic disorders, such as cancer, asthma, diabetes and cardiovascular disease (Figure 1). The benefits of genetics for these conditions are that the pre-symptomatic identification of risk provides opportunities for prevention or early intervention, reduced morbidity and more efficient targeting of therapies. For example, patients who are identified as carrying a gene mutation associated with familial forms of bowel cancer have a 90% risk of developing bowel cancer before the age of 60. Those patients who undergo high level surveillance can expect 39-55 cancer free years.

Genetic testing in WA has expanded rapidly over the last 5 years (Figure 2) and will continue to do so in the short to medium term as more diseases are found to have a genetic basis. Increases in demand will also be fuelled by the discovery of familial subsets in common chronic diseases and as public awareness improves about the role genes play in health. A review of genetic services in 2000 established a short-term plan to improve the existing clinical and laboratory

¹ Scriver CR. Review: assessing genetic risks: implications for health and social policy. *Am J Hum Genetics*. 1995;56:814-816.

² *Health Affairs* 2002;21(2)78-9342

³ Dennison E. Fall C. Cooper C. Barker D. Prenatal factors influencing long-term outcome. *Hormone Research*. 1997;48 Suppl 1:25-29.

services in WA. However, with demand on genetic services increasing by approximately 60% in the last 5 years (Figure 3), new service delivery strategies are required that account for this increased demand and advances in genetic testing capabilities. Furthermore, we need to ensure that advances in genetics are implemented with regard to community values.

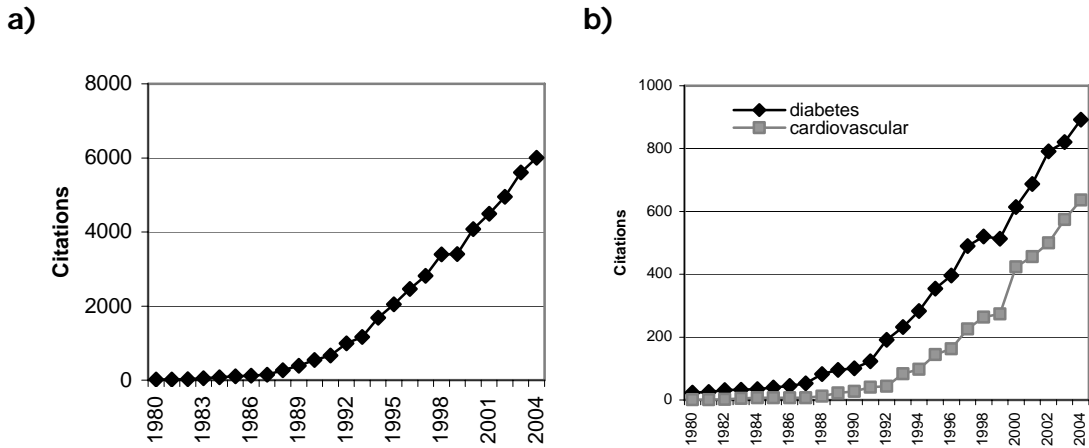


Figure 1. Increase in the number of Ovid citations for human genes associated with a) cancer and b) diabetes and cardiovascular disease between 1980-2004.

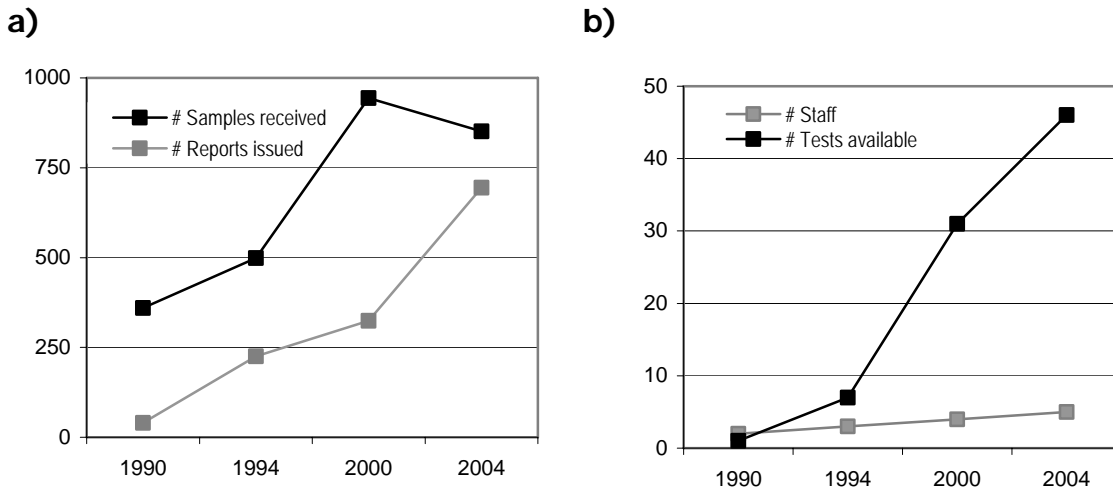


Figure 2. Increase in genetic testing for Neurogenetic conditions 1990-2004. Demand on testing for neurogenetic conditions has increased significantly over time (2a) as has the number of conditions that can now be tested for while staffing levels have remained static (2b). This has increased pressure on staff and increased turnaround times for issuing of reports. (Source: Neurogenetics Laboratory RPH). The situation is comparable for other laboratories offering genetic tests in WA.

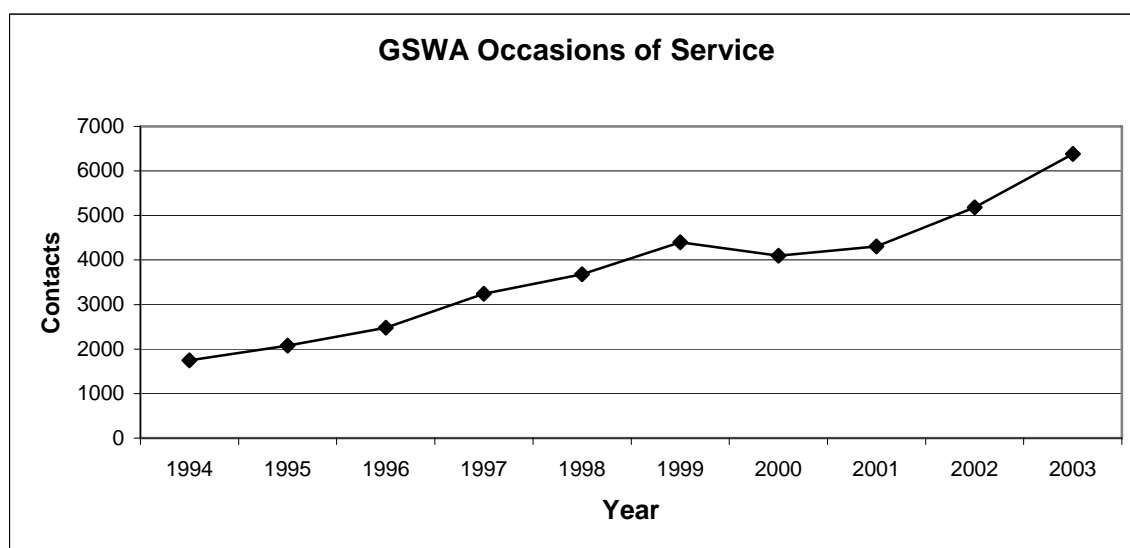


Figure 3. Increased demand for clinical genetics services (Source: Genetic Services of WA)

Genetics is an exciting and rapidly progressing field of medicine, but the health benefits attributable to genetic healthcare will become evident only if a cohesive vision is developed in this early phase of expansion. The key initiatives set out in this strategic plan aim to define this vision and are based on five identified areas of need:

- staffing and coordination of clinical services,
- improving genetics knowledge of health professionals and the community,
- development of sound, evidence-based policy,
- engaging professionals and consumers in genetics health policy;
- improved methods to monitor the effectiveness of genetic programs and services.

The specific goals within each key initiatives, intended to align with the HRIT agenda, are broken down into specific strategies described on the following pages with measurable outputs that can be used to map progress towards each goal. Allocating timeframes for the implementation of the strategies into short (1year), medium (2-3 years) or long (3-5 years) will provide focus for those involved in allocating resources and implementing these strategies. A discussion document which describes current services and provides more detail about the key initiatives is available for interested parties and can be found at http://www.population.health.wa.gov.au/Genomics/resources_genomics.cfm.

Enquiries relating to this strategic plan or the discussion document should be directed to the Genomics Directorate (Katy.Roberts@health.wa.gov.au). An implementation plan, prioritising the goals set out in the following pages and identifying resource needs will developed as the next phase of the project.

Service delivery: Increase capacity of medical genetic services to meet current and projected demand

Principle: Clinical, counselling and laboratory genetic services will be organised in the most effective way possible to ensure quality, efficiency and ongoing development.

Goal: Improve coordination and efficiency of genetic service delivery

(Healthy Hospitals, Healthy resources, Healthy communities)

Strategies

- Increase clinical and counselling services in adult hospitals **(S)**
- Ensure appropriate and cost-effective development of laboratory services **(S)**
- Ensure that genetic laboratory testing and supporting technologies are validated and have clinical utility **(S)**
- Ensure prioritisation of future genetic test development according to clinical need and available funds **(S)**
- Participate in national strategies to rationalise genetic testing for rare disorders **(M)**.
- Improve availability of genetic services outside the metropolitan area **(M)**.

Goal: Build infrastructure to address current and emerging demands arising from integration of genetics into mainstream healthcare.

(Healthy workforce, Healthy resources)

Strategies

- Increase investment in existing clinical and laboratory networks in medical genetics to ensure future sustainability. **(S)**
- Provide postgraduate education to assist in the integration of genetics across all branches of medicine. **(S)**
- Investigate workforce development strategies to address future needs **(M)**
- Develop appropriate benchmarks to ensure that state genetic services compare favorably against national and international standards and operate in a manner consistent with national, state and community priorities **(M)**

(S) – short term, **(M)** – medium term, **(L)** – Long term

Monitoring and Evaluation: Develop policies from evidence-based evaluation and public engagement that enable the application of genetic technology developments to improve health care delivery.

***Principle:** Genetic health policy should incorporate new discoveries in an ethical and economically sustainable manner to ensure the community reaps the benefits of genetic medicine. Integral to this process is the systematic evaluation of genetic tests and services to ensure the greatest health benefits are delivered to those in need.*

Goal: Evaluate new gene-based technologies that can be incorporated into health services, within an ethical, legal and social framework

(Healthy Communities, Healthy Leadership)

Strategies

- Monitor advances in genomics science and their proposed or demonstrated application to public health **(M)**
- Establish processes to evaluate current and emerging technologies with regard to their ethical, legal and social implications **(M)**
- Participate in the development of national policies for genetics in healthcare **(M -L)**
- Ensure public policy takes account of diverse cultural and social factors **(L)**

(S) – short term, **(M)** – medium term, **(L)** – Long term

Education: Allocate resources to coordinate existing networks and increase cohesion to provide comprehensive genetics education and training at all levels

***Principle:** Public awareness and understanding of genetics are prerequisites to the acceptance of genetic technologies in the community. An increased awareness of genetics in mainstream medical services such as cancer and heart disease will improve the management of familial subsets of these diseases.*

Goal: Provide and promote relevant public and professional education and training programs

(Healthy workforce)

Strategies

- Collaborate with genetic services providers, public and professional education groups to develop a comprehensive educational program **(S)**.
- Develop and implement a coordinated plan to improve genetics education at secondary school, undergraduate and postgraduate training **(M)**.

Goal: Promote integration of genetics into healthcare practice

(Healthy Leadership)

Strategies

- Increase health care provider awareness of genetic services and resources **(S)**
- Strengthen primary care provider capacity to provide primary level genetic services **(S)**
- Increase health care provider understanding of genetic concepts and how they relate to health and health care and raise awareness in all clinical groups of the potential and relevance of genetics in their field of practice **(M)**

(S) – short term, **(M)** – medium term, **(L)** – Long term

Health consultation: Engage consumers and health professionals in collaborative planning of new genetic health services

***Principle:** Collaboration between consumers and health professionals in establishing new genetic services and programs will ensure that these programs have high clinical standards and remain consistent with current societal values.*

Goal: Increase public awareness about the role of genetics in health and disease.

(Healthy Communities)

Strategies

- Promote family history as a tool to increase awareness of the importance of genetics in health **(M)**
- Increase public understanding of basic genetic principles and health issues **(L)**

Goal: Increase genetic support group and community consultation in the planning and design of health programs

(Healthy partnerships)

Strategies

- Increase public awareness of genetic services and resources in WA **(M)**
- Decrease cultural barriers to accessing genetic services and programs **(M)**
- Facilitate wider community and stakeholder involvement in health service planning **(M)**
- Maintain individual and community confidence in genetic technologies by ensuring genetic health privacy and informed consent are upheld in clinical programs and genetics-related research **(M)**

(S) – short term, **(M)** – medium term, **(L)** – Long term

Research and Development: Implement research initiatives designed to monitor and integrate genetic advances into clinical practice.

Principle: Systematic monitoring of genetic advances is needed to guarantee their translation into better patient care. Establishing partnerships between clinical services and the research community will ensure that future developments in genetics can be identified and planned for.

Goal: Implement processes to collect and analyse data about inherited conditions and genetic services for health resource planning and improvement

(Healthy Partnerships, Healthy Leadership, Healthy Communities)

- Strategies**
- Establish accurate, up-to-date information about incidence and health outcomes for selected genetic conditions and birth defects **(S)**
 - Increase availability of information about utilisation, access and quality of genetic services **(M)**
 - Monitor efficacy of services **(M)**

Goal: Strengthen links with academic institutions involved in genetics-related research

(Healthy Partnerships)

- Strategies**
- Increase interactions between genetic service providers and scientists conducting genetics research **(M)**
 - Foster research initiatives designed to monitor and promote the integration of genetic advances into clinical practice. **(M)**

(S) – short term, **(M)** – medium term, **(L)** – Long term

Appendix 1

Membership of the Western Australian Genetics Council 2005

Mr Michael Jackson (Chair 2002-2005)	Executive Director Population Health, DOH
Dr Simon Towler (Chair)	Executive Director Health Policy and Clinical Reform, DOH
Clinical Professor Jack Goldblatt	Genetic Services of WA
Dr Philip Montgomery	Clinical Services, RPH
Professor Carol Bower	Birth Defects Registry of WA
Dr Moira Sim	General Practice Divisions of WA
Dr Peter O'Leary	Genomics Directorate, DOH
Justice Robert French	Federal Court of Australia
Dr Barry Lewis	Newborn Screening Program, PMH
Clinical Professor Jan Dickinson	School of Women's and Children's Health, UWA
Dr Ian Walpole	Genetic Services of WA
Dr Ted Edkins	Molecular Genetics Laboratory, PMH
Dr Ashleigh Murch	Cytogenetics Laboratory KEMH
Professor Nigel Laing	Neurogenetics Laboratory, RPH
Mr Mitch Messer	Cystic Fibrosis WA
Mr Terry Slevin	Cancer Council WA
Professor Peter Leedman	Laboratory for Cancer Medicine, WAIMR
Professor David Ravine	UWA Chair of Medical Genetics
Mr Terry Keating (resigned)	Genetic Support Council of WA Inc.
Ms Sharon Van der Laan	Genetic Support Council of WA Inc.
Professor Lyle Palmer	UWA Chair of Genetic Epidemiology
Professor Jon Emery	UWA Chair of General Practice