

THE CHANGING LANDSCAPE OF THE GENETIC COUNSELLING WORKFORCE FINAL REPORT

14 FEBRUARY 2017
PREPARED FOR NSW MINISTRY OF HEALTH

URBIS

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Project Code	SPP12016
Report Number	Final Report



Urbis's Public Policy team has received ISO 20252 Certification for the provision of public policy research and evaluation, social planning, community consultation, market research and communications research

Template version 2015.5.1

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The purpose of this document is to outline the methodology, approach and themes raised by allied health stakeholders as per the consultation phase of the NSW Ministry of Health’s workforce planning methodology. It should be noted that the views expressed in the report are not necessarily those of the NSW Ministry of Health.

EXECUTIVE SUMMARY

INTRODUCTION

Background

The NSW Ministry of Health (the Ministry) is committed to work that supports small but critical workforces in NSW Health, including the genetic counselling workforce. As a first step in investigating this workforce, the Ministry commissioned an evidence check through the SAX Institute to inform understanding of the current genetic counselling workforce, and to provide an evidence-based resource for planning the future composition and strategic development of the workforce so that it best serves the future needs of the NSW population. The results of this evidence check suggest that the landscape of the genetic counselling workforce is changing (Barlow-Stewart, Dunlop, Shalhoub, & Williams, 2015). Within this context, the Ministry commissioned Urbis to undertake a stakeholder consultation process to gather evidence on the changing landscape of the NSW genetic counselling workforce.

Methodology

The research design incorporated collection and analysis of primary and secondary data. Urbis conducted a literature review to inform the research project, with a focus on issues in relation to the provision of genetic counselling, and the role of genetic counsellors in Australia and internationally. Subsequently, 60 stakeholders took part in in-depth interviews or focus groups, including:

- representatives of Local Health Districts (LHDs) and Specialty Networks
- staff working within NSW Health cancer clinics
- private sector providers
- primary healthcare professionals
- representatives of peak bodies/patient organisations
- staff of the University of Sydney and University of Melbourne.

This research was especially focussed on seeking perspectives from professionals with an interest in genetic counselling other than genetic counsellors (including clinical geneticists and other specialists), to identify alternative service and workforce models for the provision of genetic counselling.

SUMMARY OF KEY FINDINGS

Taken together the results of this consultation suggest that genetic counsellors are an adaptable workforce that is providing a valued and important services across the NSW health system. The specific tasks performed by genetic counsellors, however, appear to rarely fall outside of the domains of genetics (including ethics and informed consent), counselling, and multidisciplinary team care, and most public sector counsellors seem to work across all three domains. These tasks include:

- initial contact with individuals who are referred, and the provision of information
- medical history taking, and the gathering of evidence (former test results, other clinical records)
- taking informed consent
- taking samples
- ordering (under a clinical geneticist) tests within certain limits (which may differ according to the service setting)
- providing test results to patients and families
- providing counselling and psycho-social support to patients and families
- liaison with other clinicians
- conducting follow-up or cascade testing
- administrative and record keeping tasks.

While it is not possible to quantify with the available data, most genetic counsellors employed in the public system appear to be currently working in a delegated clinical role under the supervision of a clinical geneticist. The genetic counsellor can, under this model of care, be the first point of contact for a patient or family, taking a detailed medical history, providing initial information regarding the pertinent medical issue and outlining the risks and implications of any proposed or potential tests, and even ordering tests in select instances. The genetic counsellor and the geneticist will sometimes meet with a patient or family, either before or after a test, and provide co-counselling to the patient; alternatively, the geneticist may provide the results to a patient and the genetic counsellor then provides the subsequent counselling and assistance to the patient in understanding the results and their implications.

Based on the evidence collected for this review, genetic counsellors tend to work more independently of clinical geneticists when they are: employed in the private sector; employed by medical specialties other than clinical genetics; work in a regional or remote location (including hospital and community based clinics). The evidence further suggests that this service delivery model (i.e. independent practice) only negatively impacts patient care very occasionally (e.g. when a newly qualified genetic counsellor works outside of their defined scope), and can help to reduce wait times for genetic services.

The literature review identified several emerging challenges currently affecting the delivery of genetic care in NSW, many which were also identified by stakeholders. Most of these challenges relate to the growing field of genomic testing, increased demand for genetic services and the related workforce issues. The common factors that stakeholders expect will influence the future delivery of genetic counselling services typically fell under three interrelated categories:

- the transition from genetic counsellor to ‘genomic counsellor’
- the current (and anticipated) mismatch between demand for and supply of genetic counselling services
- challenges associated with ensuring consistent service provision and adequate governance of an evolving workforce.

Workforce planning and service delivery re-design will assist in ensuring that these challenges are overcome, and genetic counsellors can continue to safely service NSW patients and consumers. Elements of planning and re-design could focus on:

- the *role* of genetic counsellor – increasing the types of settings in which genetic counsellors may work, increasing the delegated scope of the role, creating a career structure along the lines of nursing through which one may progress to more senior roles (e.g. from genetic counsellor to senior to consultant)
- the *activity* of genetic counselling – increasing the training available to other health professionals so that genetic counselling may be provided by a range of clinicians.

Workforce implications of limited formal training opportunities

The University of Sydney and the University of Melbourne are the only two universities in Australia that have offered a postgraduate course for genetic counselling (Master’s of Genetic Counselling) and, in 2016 the University of Melbourne decided not to offer the course in 2017. The Universities of Sydney and Melbourne have each taken a maximum of 12-14 students annually into the Master’s of Genetic Counselling, and the courses have, according to education stakeholders and program data, frequently been oversubscribed. For instance, in 2014, the University of Sydney received a total of 85 applications for the 2015 intake (Barlow-Stewart et al., 2015). After completion of the Master’s course, postgraduate students can seek employment as genetic counsellors, and enrol in the Human Genetic Society of Australian certification program.

The decision of the University of Melbourne to withdraw the genetic counselling training program for 2017 has highlighted the limited opportunities in Australia for formal training as a genetic counsellor. The requirements for entry to the certification program, as well as the long period of training, also limit the potential of the genetic counsellor workforce to expand to meet expected future demand.

The literature is not expansive with regard to best practice and workforce models for genetic and genomic counselling. Further research to identify exactly what genetic counsellors do and how they do it, through clinical audit and other quantitative methodologies, would strengthen the knowledge base and assist with future workforce planning and the development of strong clinical governance and accountability structures.

1. INTRODUCTION

1.1. WHAT IS GENETIC COUNSELLING?

In 1947, Sheldon Reed, the director of the Dight Institute of Human Genetics in Minnesota coined the term 'genetic counselling' and described the primary function of genetic counselling as providing *people with an understanding of the genetic problems in their family* (Resta, 2006, p. 269). In Reed's view, health care professionals working as community physicians would most likely provide genetic counselling while relying on support from geneticists at speciality heredity clinics.

While Reed's view dates back nearly 70 years, his perspective on genetic counselling is still relevant today. In Australia, genetic counselling is defined as *a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions* (Human Genetics Society of Australasia, 2012).

The process of genetic counselling typically integrates the following elements:

- interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
- education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research
- counselling to promote informed choices in view of risk assessment, family goals, ethical and religious values
- support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (Resta 2006 et al in: Human Genetics Society of Australasia (2014).

The HGSA description of genetic counselling is adapted from that developed by the National Society of Genetic Counsellors (NSGC) in the United States. Based on the notion that genetic counselling can be performed by medical and non-medical professionals other than individuals specifically trained as genetic counsellors, the NSGC deliberately describes the activity or process of genetic counselling, rather than the role of a genetic counsellor (Skirton, Cordier, Ingvoldstad, Taris, & Benjamin, 2015).

In NSW, public sector genetic services are governed by LHDs and speciality networks, as there is currently no formal or informal state-wide clinical governance for genetic counselling services in NSW. Genetic services, including genetic counselling, are most often offered through clinics located within tertiary hospitals, or as outreach units (e.g. community health centres) in regional and rural areas linked with a primary unit located in a city hospital (Centre for Genetics Education, 2016).

1.2. THE CHANGING LANDSCAPE

Existing academic and 'grey' literature suggests that genetic counsellors are currently practicing in a rapidly changing 'genetics landscape', in which their future role scope and practice, including within NSW Health, is uncertain (See for example Ormond, 2013). For example, while DNA techniques have allowed individuals to be tested for different genetic conditions such as Huntington's disease, familial cancer syndromes, and Lynch syndrome since the mid-1980s, genetic testing has expanded over the past three decades to include testing for a much wider range of disorders. This evolution has also impacted the workforce (including genetic counsellors), and genetic testing-related services are now offered not only by health professionals working for specialist genetic centres, but also by other health services, such as cardiac, diabetes and cancer clinics. This change has meant that genetic counsellors are working more independently of clinical geneticists, and tend to be integrated within multidisciplinary teams, bringing new challenges to ensuring consistent service delivery and sound clinical governance (Skirton et al., 2015).

The development of new technologies, especially next-generation sequencing and increased computer capacity has also facilitated the increased generation and widening accessibility of genomic information (although to a limited extent in Australia).¹ According to a growing literature, the wealth of new information

¹ Note that in the context of clinical and research settings, genetic testing refers to the examination of specific bits of DNA that have a known function, usually in a protein-coding gene. Genetic testing requires that an investigator know which gene or genes to look at, based on some prior understanding of the underlying biological contribution to a trait or disease. Genomic testing, in contrast, looks for variations within large segments across the entirety of genetic material, both within and outside known functional genes.

made available through genomic testing provides numerous and varied opportunities to advance medical care, improve health outcomes, and cut healthcare costs through more effective and individually tailored management of complex conditions (i.e. 'personalised medicine'). This may also impact the knowledge and skills required by genetic counsellors to successfully assist patients and consumers to understand test results in a safe and ethical manner (Ormond, 2013; Shelton & Whitcomb, 2015).

Finally, the increasing availability of direct-to-consumer genetic testing (DTC GT) including in NSW, means that consumers are now able to access genetic testing products independently through the internet. This growing market has been a source of concern for healthcare providers and relevant organisations, particularly in relation to consumers who will then need to understand, interpret, and cope with genetic information without professional assistance (Shelton & Whitcomb, 2015). There is a general consensus, however, that genetic counsellors are well placed to assist consumers, but that this assistance will require an updating of service delivery models (and associated knowledge and skills), so that a much larger cohort of community members are able to receive assistance (through, for instance, video counselling or other changes to services access) (Shelton & Whitcomb, 2015).

Within this context, the Ministry commissioned Urbis to undertake a stakeholder consultation process to gather evidence on the changing landscape in relation to the NSW genetic counselling workforce. Critical questions that the Ministry is seeking to answer comprise:

- What is the relationship of the genetic counselling workforce and the private sector?
- What models of care exist, or may emerge, where the genetic counselling workforce may be impacted?
- What regulatory or policy drivers may impact on this workforce?
- What are the key challenges facing the workforce now and in the future?
- What are the key drivers that are expected to influence the genetics counselling workforce in the future? (technology, science, policy & costs)
- To what extent do the current roles and education pathways meet the needs of health services and opportunities for future reform?

This report provides an analysis of the research findings, and is structured as follows:

- Chapter 2: the NSW genetic counselling workforce
- Chapter 3: contextual influencers on the role of genetic counsellor
- Chapter 4: emerging challenges
- Chapter 5: the future workforce
- Chapter 6: conclusion.

Before discussing the research findings, the following section provides an overview of the research methodology that has been undertaken for this project.

1.3. METHODOLOGY

Table 1 below outlines the approach taken to complete the research, including the key activities relevant to each phase of the project.

Table 1 – Research structure

Research phase	Timeline	Research activities
Phase 1: inception and project planning	June 2016	Inception meeting with the Ministry Project plan and consultation strategy Key informant interviews
Phase 2: initial analysis	June-July 2016	Literature review
Phase 3: stakeholder consultation	August-September 2016	Stakeholder interviews with a total of 60 professionals working within the public and private health systems in NSW and Victoria, and with representatives from relevant peak bodies and education institutions
Phase 4: analysis and reporting	September-October 2016	Preparation of draft and final reports

1.3.1. Literature review

Urbis conducted a literature review to inform the research project, with a focus on issues in relation to the provision of genetic counselling, and the role of genetic counsellors in Australia and internationally.

A search for relevant literature and documents was conducted through the following databases and resources:

- a number of databases provided by EBSCO, including Academic Search Complete, SocINDEX with Full Text, Health Policy Reference Centre and Social Work Reference Centre
- Google, Google Scholar and relevant websites in Australia and overseas, including but not limited to Commonwealth, Australian and state and territory health departments, websites of national associations of genetic counsellors (e.g. the Human Genetics Society of Australasia and the Genetic Counsellor Registration Board in the United Kingdom) and websites reporting on employment data (e.g. the Bureau of Labor Statistics in the United States)
- journals to access relevant peer reviews, including the European Journal of Human Genetics, Journal of Genetic Counselling, Journal of Community Genetics, NPJ Genomic Medicine, Clinical Genetics, British Medical Journal, Medical Journal of Australia, Australian Journal of Rural and Remote Health and the Journal of the American Medical Association.

The search mainly included literature and reports published after 2010, but where relevant, literature and reports published prior to 2010 have been included in the review.

Appendix A of this report provides the full literature review, and many of the findings of the review are included throughout this report.

1.3.2. Stakeholder consultations

Stakeholder consultations took place in August and September with 60 stakeholders, including:

- representatives of Local Health Districts (LHDs) and Specialty Networks
- staff working within NSW Health cancer clinics
- private sector providers

- primary healthcare professionals
- representatives of peak bodies/patient organisations
- staff of the University of Sydney and University of Melbourne.

A particular focus of this research was to seek perspectives from professionals with an interest in genetic counselling *other than genetic counsellors*, to identify alternative service and workforce models for the provision of genetic counselling. More specifically, the consultation explored the roles of medical and non-medical professionals in the provision of genetic counselling, as well as the variety of settings in which genetic counselling can occur.

For this reason, staff from LHDs and Specialty Networks included mainly medical and non-medical staff other than genetic counsellors, such as oncologists, paediatric physicians, medical cardiologists, clinical geneticists, pathologists, specialist nurses and social workers.

Representatives from cancer clinics, private sector providers, peak bodies/patient organisations and educators included executive directors, associated professors and chair persons.

General practitioners were included in the stakeholder consultations as primary healthcare professionals.

A mixed method was applied for recruitment, which comprised:

- appointment of LHD contact persons through the Ministry, who were then contacted by Urbis
- ‘cold calling’ of representatives from peak bodies, patient organisations, private sector providers, cancer clinics and education institutions
- the use of professional recruiters for recruitment of general practitioners.

All interviews were guided by a semi-structured interview guide and were approximately 45 minutes in duration. General practitioners were offered a \$130 incentive in recognition of the time they contributed to the research.

The majority of the interviews were conducted by telephone. An Urbis consultant visited the John Hunter Hospital, St George Hospital and the Prince of Wales Hospital to meet with LHD representatives for the Hunter New England and the South Eastern Sydney Local Health District, respectively, and a focus group was conducted separately with general practitioners.

A full breakdown for each stakeholder cohort is provided in Table 2.

Table 2 – Stakeholder consultation list

Stakeholder group	Number of stakeholders	Organisation
Local Health Districts and Specialty Networks	23	<ul style="list-style-type: none"> • Hunter New England LHD • Mid North Coast LHD • Northern NSW LHD • Murrumbidgee LHD • Southern NSW LHD • South Eastern Sydney LHD • South Western Sydney LHD • St Vincent's Health Network • Western NSW LHD
Cancer clinics	4	<ul style="list-style-type: none"> • Westmead Breast Cancer Institute • St Vincent's Hereditary Cancer Clinic • Hereditary Cancer Clinic

Stakeholder group	Number of stakeholders	Organisation
		<ul style="list-style-type: none"> • Mid North Coast Cancer Institute
Private sector providers	6	<ul style="list-style-type: none"> • The Genetic Clinic • Sydney Cancer Genetics • Sonic Genetics • Genea – World Leading Fertility • My DNA • Genea
Primary healthcare professionals	15	<ul style="list-style-type: none"> • General practices/medical centres (Westmead area)
Peak bodies/patient organisations	6	<ul style="list-style-type: none"> • Australian Society of Genetic Counsellors • Genetic Alliance Australia • Human Genetics Society of Australasia • HGSA/RANZCOG Prenatal Diagnosis & Screening Committee • Melbourne Genomics Health Alliance • Sydney Children’s Hospital Randwick
Educators	2	<ul style="list-style-type: none"> • University of Sydney • University of Melbourne
Other	4	<ul style="list-style-type: none"> • Cancer Institute NSW • Labtests online/ SEALS Genetics • RPAH Medical Genomics
Total	60	

1.3.3. Analysis

The data sources for this research comprised the following:

- key findings of the literature review
- stakeholder consultations (interviews and focus groups).

Key findings from the literature review are included throughout this report where appropriate, and provided in full in Appendix A.

All stakeholder consultations were audio-recorded and transcribed for analysis. The interview transcripts were analysed thematically and iteratively, and informed the emerging themes and the structure for this report.

1.4. NOTE ON THE TEXT

- 1) For the purpose of this research, this report follows the description of genetic counselling as described by the Australian Human Genetics Society of Australasia (HGSA), which states that genetic counselling integrates the following elements:
 - interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
 - education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research
 - counselling to promote informed choices in view of risk assessment, family goals, ethical and religious values
 - support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder (Resta 2006 et al in: Human Genetics Society of Australasia, 2014).
- 2) When referring to 'genetic counsellors' throughout this report, we refer to professionals who are formally appointed in the health workforce as genetic counsellors, and who are involved in the conduct of genetic counselling as described at point 1.

2. THE NSW GENETIC COUNSELLING WORKFORCE

2.1. INTRODUCTION

As noted above, public sector genetic services are governed by LHDs and speciality networks. Genetic services, including genetic counselling, are offered through clinics located within tertiary hospitals, or as outreach units (e.g. community health centres) in regional and rural areas linked with a primary unit located in a city hospital (Centre for Genetics Education, 2016).

Two other main providers of genetic counselling within the NSW public health sector include:

- clinics that offer speciality health related services, such as cancer clinics, prenatal clinics, genetic heart disease clinics and neurological clinics
- specialised genetic services that involve *services that specialise in the management for people affected by particular genetic conditions and risk assessment for concerned family members* (Health Centre for Genetics Clinics, 2013).²

Private service providers also offer genetic counselling in NSW; however, there is little information available in this area. According to the literature, ultrasound practices, IVF centres and familial cancer centres are most likely to provide genetic counselling in private settings (Human Genetics Society of Australasia, 2016; Mann, Taylor, James, & Gaff, 2014).

In 2011, NSW employed the largest proportion of genetic counsellors (41%, or 60 in total), of a total Australian workforce of 145 (Sillence & Barlow-Stewart, 2011). These counsellors are primarily employed within NSW Health, and work in genetic clinics as well as within specialist teams focussing on neurology, paediatrics, obstetrics and maternal health, cardiology, oncology, and gynaecology.

The remainder of this chapter will summarise education and certification requirements, typical referral pathways, and role scope for genetic counsellors currently working in NSW, with a focus on the public sector. While most of this report is devoted to the changing landscape of the genetic counselling workforce, this snapshot of the current workforce as the title suggests will ensure that conclusions drawn upon, and suggestions made about, potential future scenarios consider existing skills and capabilities at both whole-of-workforce (e.g. current number of genetic counsellors) and individual counsellor (e.g. required skills) levels.

2.2. EDUCATION AND CERTIFICATION

The University of Sydney and the University of Melbourne are the only two universities in Australia that have offered a postgraduate course for genetic counselling (Master's of Genetic Counselling) and, in 2016 the University of Melbourne decided not to offer the course in 2017. The course is accredited by the HGSA Board of Censors for Genetic Counselling and is the only program in the country to enable entry to the HGSA certification program (Human Genetics Society of Australasia, 2014).

Enrolment in the postgraduate course requires:

- an undergraduate degree in a related field (i.e. genetics, psychology, social work, nursing/midwifery, science)
- experience in counselling and/or genetics
- experience in a care role (NSW Ministry of Health, 2015b).

The Universities of Sydney and Melbourne have each taken a maximum of 12-14 students annually into the Master's of Genetic Counselling, and the courses have, according to education stakeholders and program data, frequently been oversubscribed. For instance, in 2014, the University of Sydney received a total of 85 applications for the 2015 intake (Barlow-Stewart et al., 2015). After completion of the Master's course, postgraduate students can seek employment as genetic counsellors, and enrol in the HGSA certification program. This certification program is not mandatory, but is recommended by the HGSA (Human Genetics Society of Australasia, 2011). Genetic counsellors who are applying for HGSA certification are required to have formal arrangements in place for supervision and oversight (Human Genetics Society of Australasia, 2014). The HGSA genetic counselling certification training comprises a two-year full-time certification

² Examples of these services located in NSW include the Genetics of Learning Disability (GOLD) Service, Medications in Pregnancy and Lactation Service, and community screening for people with Eastern European (Ashkenazi) Jewish ancestry.

program in which supervision for candidates is mandatory. In general, candidates are expected to have a minimum of two clinical supervisors:

- a clinical geneticist certified by the HGSA or international equivalent
- a health professional with the requisite counselling skills and experience (including HGSA-Certified Genetic Counsellors, at least two years' post-certification; accredited social workers with at least three years of experience in hospital or community settings; accredited clinical psychologists with experience in hospital or community settings).

Completion of the program enables candidates to use the title Fellow of the Human Genetics Society of Australasia, and become Certified Genetic Counsellors. The total duration from undergraduate student to Certified Genetic Counsellor takes approximately nine years in total (NSW Ministry of Health, 2015b).

In Australia, Certified Genetic Counsellors are expected to continuously engage in education and training through the Maintenance of Professional Standards (MOPS) program (Human Genetics Society of Australasia, 2014). This program is developed and offered by the HGSA, and involves a five year cycle of resubmission for Certified Genetic Counsellors (Human Genetics Society of Australasia, 2014). At the time of writing, the HGSA Board of Censors (BOC) is currently reviewing the MOPS program as part of the Australasian Genetic Counsellor Training and Certification standards revision (Human Genetics Society of Australasia, n.d.).

Even with the high levels of education and training experienced by genetic counsellors, many of the clinical geneticists and other stakeholders interviewed for this research were of the view that on-the-ground training is essential to help junior genetic counsellors learn how to apply their skills in practice. This may be particularly applicable to genetic counsellors working autonomously with little supervision in rural areas. The weight of stakeholder feedback suggests that due to the shortage of genetic counsellors and the great need for their services, the ability to have an 'internship' period after the certification period is limited by the lack of clinical geneticists or senior genetic counsellors to provide supervision. Potential alternative pathways to certification – all of which consider the changing landscape and the perceived shortage of genetics practitioners – will be considered through this report.

2.3. REFERRAL PATHWAYS

Based on the available evidence (including literature review and stakeholder consultation), it appears that NSW patients and consumer typically access genetic counselling services, at least in the public sector, after referral from a general practitioner (GP), a specialist, or from another member of a multi-disciplinary team. Each of these referral pathways is discussed in turn below.

Referral from general practitioners

GPs who participated in this review did not consider themselves to be closely involved with genetic counselling, other than the provision of information for patients who might be subsequently referred to a genetic clinic for testing. Generally, GPs noted that they had little control over whether a patient undergoes a consultation with a clinical geneticists or genetic counsellor upon referral to a clinic, as this decision is in the hands of clinic staff. GPs did note, however, that there are some genetic tests that they are comfortable managing themselves, including some pregnancy tests and diagnostic testing for haemochromatosis. At the same time, GPs typically indicated that they knew their limits regarding the provision of genetic information or counselling (especially given the speed of technological advancement), and were comfortable with referring more complex cases to specialist genetic clinics for information, advice and testing. GPs generally expressed themselves as satisfied with the avenues for referral, as well as the ability to speak directly with a geneticist or a genetic counsellor when required.

Several GPs reflected that in the past ten years they have been referring patients more often for genetic testing, particularly for the BRCA1 gene following high levels of publicity regarding this gene and its link with breast cancer. Much of this, according to GPs, is driven by patients who come to the GP with concerns regarding their own family history, seeking further information and predictive testing to help them understand their potential to develop a particular disease in the future. GPs did note that additional information regarding genetic services and genetic testing in general would be helpful, either through Royal Australian College of General Practitioners (RACGP) professional development activities or local education sessions, or through providing web-based resources or leaflets. The potential for expanding the role the GPs play in the provision of basic genetic counselling, especially if demand for testing increases, is considered later in this report, with a focus on implications for clinical governance.

Referral from specialist to genetics clinic

Most of the referrals to genetic clinics appear to be provided within the NSW Health system, from other clinical specialities such as neurology, obstetrics, or cardiology. These referrals are generally discussed at a clinic intake meeting and are then allocated either to the geneticist or directly to the genetic counsellor, depending on the complexity of the issue (and whether the referral is for diagnostic purposes). Where the genetic counsellor is working in a genetics clinic under the direct supervision of a clinical geneticist, generally the referral will come to the clinic and the clinical geneticist and the genetic counsellor will generally confer at an intake meeting and determine which health professional is best placed to undertake the initial engagement with the patient, based on the complexity of the case (for instance, whether the referral is for diagnostic purposes) and current workload.

Referral within a multi-disciplinary speciality team

As noted above, depending on the clinical service and the service model, genetic counsellors can form part of a multi-disciplinary team within a non-genetic specialist clinic, such as a foetal medicine clinic. In these instances, genetic counsellors will receive referrals from colleagues within their own team, and work directly with the referring clinician to provide advice and information about the appropriate test and risks or limitations of the test. A clinical geneticist may only be peripherally involved in the process, depending on the delegated relationship with the genetic counsellor.

Based on stakeholder feedback and existing Australian literature, it appears that a similar process is undertaken in rural services where a genetic counsellor may be working autonomously under the direction of a clinical geneticist based in a regional or metropolitan centre. In this instance, the genetic counsellor typically works within a multi-disciplinary team, often among a range of allied health professionals, but will have sole responsibility for the genetic testing referral, and will liaise with a clinical geneticist as required.

2.4. CURRENT ROLES OF GENETIC COUNSELLORS IN NSW

You need a combination of supporting clinicians clinically in doing the right tests for the right reasons and making sure the family along the way are not harmed. (Clinical Geneticist)

Genetic counsellors versus genetic counselling

International literature suggests that different medical and non-medical professionals are commonly involved in the provision of genetic counselling, and key elements of 'the act of providing genetic counselling' appear to be widely utilised in various health settings (Battista, Blancquaert, Laberge, Van Schendel, & Leduc, 2011; Middleton, Hall, & Patch, 2015). For instance, when an obstetrician needs to discuss a genetic test result with patients, it is likely that she or he will use strong, empathic communication skills to help the patient understand and appreciate the significance of the genetic information (Arnold and Self, 2012 in: (Middleton et al., 2015, p. 80). As noted by Middleton et al. (2015), this example demonstrates the fine line between having a conversation based on genetic information and providing genetic counselling. Understanding this distinction is important in determining the necessity for a trained genetic counselling workforce and planning for future workforce requirements. It should be noted that all stakeholders could distinguish between providing genetic information and providing genetic counselling or, even more specifically between genetic counselling and being a genetic counsellor. As one clinical geneticist noted, *I provide genetic counselling but I am not a genetic counsellor*, conveying that the role of genetic counsellor is, at least in their mind, clearly defined.

Overall, evidence suggests that there are two types of professionals involved in genetic counselling:

- professionals involved in genetics (e.g. genetic counsellors and clinical geneticists)
- other healthcare professionals using genetics in routine care, such as general practitioners, specialists (e.g. cardiologists, obstetricians), nurses, psychologists, and social workers (Battista et al., 2011; Centre for Genetics Education, 2016).

For the most part, it appears that genetic counsellors are uniquely placed to assist patients to comprehend and adapt to new genetic information due to their combined expertise in clinical genetics (and sometimes genomics) and counselling/psychosocial engagement. Genetic counsellors, when adequately trained, are able to communicate complex information on genetic testing with sensitivity to the very personal circumstances of the individual patient or family. Geneticists and genetic counsellors interviewed for this research stressed the time required in many cases to prepare the medical and family histories and to undertake initial consultations before any tests are even ordered. They also noted that genetic tests do not always provide a definitive answer to a diagnostic question, and in many instances the answer may be of uncertain significance for the individual and for a family. Many non-genetic medical specialists, reportedly,

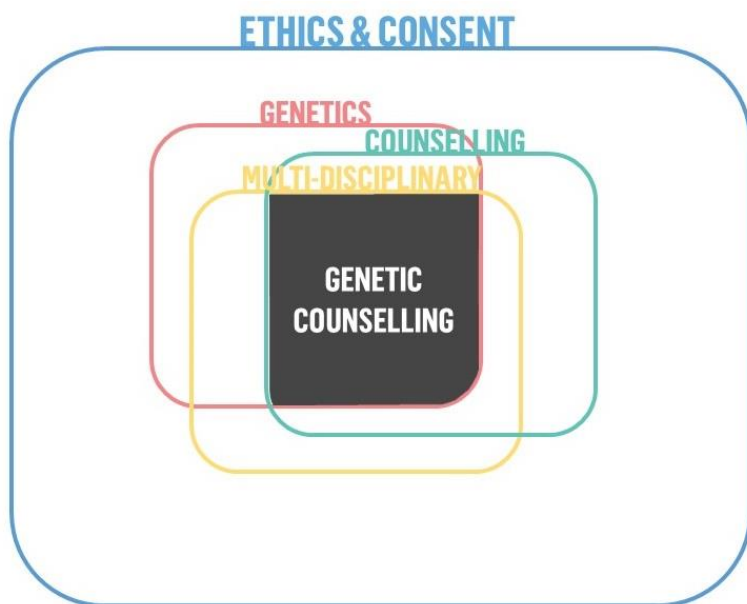
are not fully aware of this lack of certainty, which can be very hard for patients or families to accept. The role of the genetic counsellor in these instances, it was reported, is to spend time with the family to determine what level of information will be beneficial for the family and what may be harmful, including assessing the capacity to tolerate uncertainty. This role, according to stakeholders, may increase as genomic testing allows for increasing uncertainty in results, a scenario which is discussed further in Chapter 4 of this report.

Role scope

Consistent with the results of the literature review, stakeholder feedback suggests that the tasks routinely undertaken by genetic counsellors, and the associated clinical governance and supervisory arrangements, vary significantly across the NSW health system. Models of service delivery for genetic counsellors in the private sector noted by stakeholders, for example, included working as a sole practitioner, working in partnership with a specialist physician, or working as part of a multidisciplinary team with prenatal specialists, cancer specialists, allied health professionals, and so on. To demonstrate the extent of the variation in services provided by genetic counsellors, one stakeholder noted that laboratory based genetic counsellors often do not have any contact with patients, instead acting as a liaison between physicians and specialist pathologists. This stakeholder suggested that varying the role title accordingly may assist clinicians (and other stakeholders) to understand the services provided by laboratory and non-laboratory based genetic counsellors.

Despite variation in the service delivery models reported by stakeholders, the specific tasks performed by genetic counsellors appear to rarely fall outside of the domains of genetics (including ethics and informed consent), counselling, and multidisciplinary team care, and most public sector counsellors seem to work across all three domains.

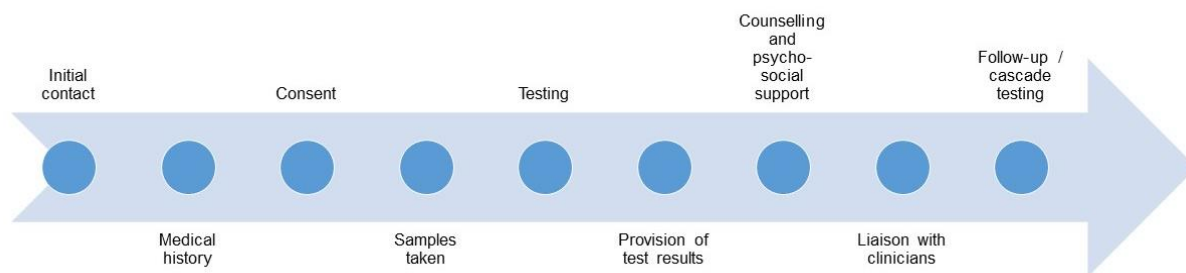
Figure 1 – Role scope



The extent to which genetic counsellors work *within* the domains of counselling, genetics, and multidisciplinary team care, and the nature of the tasks performed, does, however, vary across service delivery models, with some genetic counsellors working primarily as scientific assistants (i.e. mostly genetics related tasks) and others performing more of a therapeutic role (i.e. mostly counselling-related tasks). Levels of supervision, and consequently the capacity for genetic counsellors to work independently, also appear to vary across service delivery models. Most notably, some genetic counsellors reportedly work in partnership with an onsite clinical geneticist (or other specialist physician), while others may have only limited contact with geneticists.

It is not possible to quantify the many and varied tasks undertaken by genetic counsellors in NSW using the available evidence. However, stakeholder feedback and the results of the literature view suggest that there are a number activities routinely undertaken by genetic counsellors across settings, the most common of which are shown in Figure 2.

Figure 2 – Routine activities



A more substantive mapping study, including collection of quantitative data, would provide more robust evidence of the tasks routinely undertaken by genetic counsellors, either when working in a delegated role or within a multi-disciplinary team.

Geneticists and genetic counsellors alike noted the importance of the genetic counsellor role at two particular points in the clinical process: at the beginning of the process, for the *detective* work and initial engagement with the patient or family; and once the test results have been received, when individuals may require *counselling* and assistance in making difficult decisions, and families may require follow-on or ‘cascade’ testing to ascertain the extent of the genetic disorder within the family tree.

Supervision and clinical governance

As noted by the HGSA (2012), medico-legal responsibility for genetic consultations can vary between services (public and private), resting variously with a medical specialist (including clinical geneticist), an institution, or the individual’s private liability cover. In contrast to the public sector, genetic counsellors working in the private sector do not always work under the supervision of a geneticist, and in these instances counsellors are not able to order tests, instead providing counselling and interpretation of test results directly to patients. Stakeholder feedback suggests that in some research centres and clinics (including in the public and private sector), operational supervision for genetic counsellors falls to a non-genetics specialist (e.g. cardiologist), while formal clinical supervision is provided by a clinical geneticist, typically offsite. Similarly, while a genetic counsellor practicing in a rural area may have remote supervision from a clinical geneticist, it often happens that operational supervision will come from a local health service manager, potentially an allied health manager or a managerial level supervisor. This impact of these various supervisory arrangements on genetic counsellor role scope is discussed in the following chapter.

2.5. CHAPTER CONCLUSION

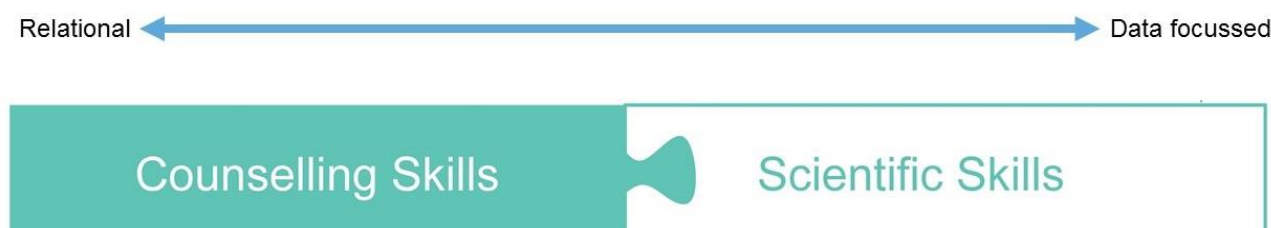
While genetic counsellors are currently working in many and varied ways across the NSW health system, most appear to be employed in a delegated role under the supervision of a geneticist. The genetic counsellor can, under this model of care, be the first point of contact for a patient or family, taking a detailed medical history, providing initial information regarding the pertinent medical issue and outlining the risks and implications of any proposed or potential tests, and even ordering tests in select instances. The genetic counsellor and the geneticist will sometimes meet with a patient or family, either before or after a test, and provide co-counselling to the patient; alternatively, the geneticist may provide the results to a patient and the genetic counsellor then provides the subsequent counselling and assistance to the patient in understanding the results and their implications. Genetic counsellors also commonly work collaboratively with other clinical staff within specialist clinics (e.g. cancer clinic or an obstetrics and maternal medicine department). The role for genetic counsellors in these settings appears to be two-fold, in that they provide internal advice and expertise to other clinicians as well as provide counselling and support to patients and families. Key drivers of variation in the role scope for genetic counsellors (including adaptability to a changing landscape) are covered in the following chapter of this report.

3. CONTEXTUAL INFLUENCES ON THE ROLE OF GENETIC COUNSELLOR

3.1. INTRODUCTION

Genetic counsellors are working in various ways across the NSW health system, partially in response to an ever-changing landscape. Most notably, genetic counsellors appear to differ substantially in the extent to which they undertake relational or data focussed activities, underpinned by variation in the necessity for counselling and scientific skills (see Figure 3).

Figure 3 – Variation in role domains



Stakeholders, especially experienced clinicians, academics, and health managers, tended to suggest that this considerable variation in role scope is enabled by one, or all, of the following health system characteristics:

Public and private sector service delivery models: Many stakeholders pointed out that there is significant variation in models of service delivery for genetic counsellors across the public and private sector in NSW. As noted above, genetic counsellors can work as sole practitioners, even when not certified, in the private sector; however, this is not possible for genetic counsellors employed within NSW Health. Clinical governance arrangements and supervision practices also tend to be less formalised and embedded in the private sector, meaning that the clinical scope of practice for genetic counsellors can vary across the sectors, even when the same service delivery model is being adopted (e.g. working as part of a multidisciplinary team with prenatal specialists in a private sector clinic versus a public sector hospital).

Non-centralised provision of public sector genetic services: Stakeholders with experience working in, or with, genetic services in other jurisdictions suggested that there is less variation in the services provided by genetic counsellors, and greater consistency in clinical governance and supervisory practices, in states and territories with centralised genetic services (e.g. Victoria and Queensland). Reportedly, genetic counsellors in these jurisdictions tend to work out of a single metropolitan site, with regional areas serviced through periodic clinics. Counsellors in these jurisdictions are also required to adhere to consistent protocols and procedures, including scope of practice and governance arrangements. In contrast, the non-centralised approach to genetic services adopted in NSW, it was suggested, has meant that there is variation in genetic counselling services models across, and even within, LHDs.

Lack of formal clinical oversight of public sector genetic services: The results of the literature review and stakeholder feedback suggests that it is possible to promote consistency in role scope (and subsequent service delivery), at least in the public sector, through the provision of mandated clinical guidelines or, at minimum, some form of centralised clinical oversight. However, there is currently no formal or informal state-wide clinical governance for genetic counselling services in NSW, allowing for variation in service delivery across the state. Some stakeholders further suggested that the level of centralised clinical governance and guidance provided at LHD and hospital levels can be inconsistent; however, there is no objective evidence to support this view. This possibility of developing a centralised governance mechanism for genetic (and genomic) counselling is canvassed in Chapters 4 and 5 of this report.

Status as a non-registered health profession: Genetic counselling is not currently a registered health profession in Australia, meaning that there are no legal ramifications for uncertified practitioners working as a genetic counsellor (and defining their own scope of practice), at least in the private sector. It should be noted, however, that genetic counsellors (certified or otherwise) are not able to order genetic tests from registered laboratories, so their capacity to practice in a private capacity is very limited.

The remainder of this chapter will consider the impact of the following personal and contextual factors on the role (and responsibilities) of genetic counsellors working in NSW:

- employer and supervisor
- education and training
- medical specialty
- location.

3.2. EMPLOYER AND SUPERVISOR

Employer

As noted above, stakeholder feedback, in addition to the results of the literature review, suggests that the scope of practice for genetic counsellors varies across the NSW public and private health sectors. For example, LHD clinicians and managers working in large hospitals tended to stress that in their experience, genetic counsellors mostly provide direct or indirect clinical and administrative support to onsite clinical geneticists or other onsite specialist medical practitioners (e.g. setting appointments, data entry, developing family pedigrees). In these settings, the role of the genetic counsellor (including scope, governance, and supervision) tends to be very well defined, even in instances where a counsellor's operational supervisor sits in a different clinical stream or department. These genetic counsellors also tend to work either in partnership with a clinical geneticist (i.e. in a delegated clinical role), or as part of a specialist multidisciplinary team. Even when working in regional and remote areas, public sector genetic counsellors are required to have regular contact with a clinical geneticist, and are typically integrated into a multidisciplinary team, either in a community health or hospital setting. In the literature, genetic counsellors working in clearly defined, delegated clinical roles (typically in the public sector) are sometimes referred to as 'physician extenders' (Shelton & Whitcomb, 2015).

In contrast, there was little consistency in the role scope reported by stakeholders (including physicians and genetic counsellors) working in the private sector, with some suggesting that clinic-based genetic counsellors work in partnership with onsite clinical geneticists, and others suggesting that these counsellors have little or no contact with clinical geneticists. There was, however, a general agreement that clinical governance arrangements and supervisory practices are less well developed, and less consistently implemented, in the private sector when compared to the public sector. This inconsistency has, reportedly, led to greater variation in service provision, including occasional examples of genetic counsellors working outside of their clinical scope of practice (e.g. diagnostic assessment and medical management).

Despite this, stakeholders mostly agreed that genetic counsellors can, and indeed do, work independently in private sector clinics. To be successful in the role, one needs to be highly skilled and experienced, with a focus on understanding the clinical scope of practice. When asked about the ways in which service delivery in the private sector could be improved, stakeholders tended to suggest that governance arrangements may naturally grow more formalised, possibly without any intervention or prompting from government, as the sector develops and diversifies.

Supervisor

There was confusion amongst stakeholders as to whether operational supervision for genetic counsellors in the public sector always falls to an onsite (or offsite) clinical geneticist, with some suggesting that, in their experience, genetic counsellors are always supervised by clinical geneticists and others listing a multitude of supervisory arrangements, none of which included a clinical geneticist. Taken together, however, stakeholder feedback and the results of the literature view suggest that operational supervision for genetic counsellors can fall to any number of health professionals, including:

- clinical geneticist
- clinical nurse manager
- allied health manager
- community health manager
- other medical professional
- radiation therapy manager
- senior genetic counsellor.

Variations in operational supervisory arrangements appear to impact the service delivery model, and subsequently, the scope of clinical practice for genetic counsellors in several ways. For example, genetic counsellors who are supervised by clinical geneticists tend to spend most their time either working in partnership with the geneticist (as is the case with most junior counsellors), or working in proximity in the genetics department or clinical stream. In contrast, genetic counsellors who are managed by another allied health professional (or a manager of allied health professionals) tend to have their practice integrated within a multidisciplinary team, and will commonly have more day-to-day contact with nurses, social workers or psychologists than with clinical geneticists. These counsellors – who are typically located in regional and remote areas – also tend to be more heavily engaged in counselling activities than genetic counsellors managed by clinical geneticists. A few genetic counsellors expressed their frustration at having an operational supervisor located outside of their everyday working environment, and some reported a preference for closer alignment – including through operational supervision – with allied health.

There is evidence to suggest that the scope of practice for genetic counsellors, both in the private and public sector, can also be influenced by their operational manager's style of supervision, and the level of trust that the manager has in the genetic counsellor. One clinical geneticist, for example, noted that because he can supervise the genetic counsellors in his team quite closely, he allows the counsellors a degree of independence, and a slightly expanded clinical role. This service delivery model, he further suggested, is only possible in situations where the genetic counsellor understands, and adheres to, their clinical scope of practice, and the geneticist trusts that the scope of practice will be observed at all times. As discussed further below, expansion of this expanded model of care may assist in meeting current and potentially growing demand for genetic counselling services.

3.3. EDUCATION AND TRAINING

People who become very specialised in an area, people who have been doing cancer genetics for years and years and years mostly can run with cases on their own and don't really need a great deal of input from the geneticist or the oncologist (Genetic Counsellor)

There was a consensus amongst clinical geneticists and other clinicians responsible for the employment and management of genetic counsellors that fully certified genetic counsellors, especially those with extensive experience, can work more independently and broadly within the scope of clinical practice than counsellors who are either unregistered or are currently completing the registration process. Stakeholders consistently suggested that certified genetic counsellors are typically able to manage their own cases from referral through to consultation and case management, and then to the eventual closure of the case (including management of subsequent referral, data entry, and finalisation of case notes). Clinicians further suggested that certified, experienced genetic counsellors tend to be fully cognisant of their clinical scope of practice, and will draw readily upon the expertise of specialist physicians for the management of complex cases, especially those that include a rare genetic variant. This point was also made by genetic counsellors working in the public sector.

As noted above, even the most experienced genetic counsellors are not able to order genetics tests from accredited laboratories, and therefore rely on clinical geneticists, or other specialist physicians, for the conduct of this task. Accredited genetic counsellors are also required to have at least one hour of clinical supervision with a certified clinical geneticist per week; however, physicians and genetic counsellors alike tended to agree that this did not necessitate weekly contact between a geneticist and a senior counsellor, as supervision did not always occur on a weekly basis.

Stakeholder feedback and the existing literature suggests that genetic counsellors with nursing qualifications and experience can work most independently, as their scope of clinical practice allows for the ordering of certain genetic tests. One Victorian-based nurse practitioner, who works both with nurses and genetic counsellors, was of the view that genetic counsellors, in her experience, generally possess clinical knowledge and capability to safely order basic genetic tests, and that the inclusion of this task in their clinical scope of practice would assist in ensuring timely provision of test results to patients and community members. This point is discussed further in Chapter 4 of this report. It should be noted that the NSW Health Service Health Professionals (State) Award specifies that genetic counsellors must possess an undergraduate degree in a **non-nursing, non-medical discipline** and in addition hold a post graduate qualification in genetic counselling or have attained Part I certification in genetics counselling from the HGSA, a requirement that is also discussed further in Chapter 4.

3.4. MEDICAL SPECIALITY

Consistent with the results of the literature review, the qualitative research findings suggest that the role scope for genetic counsellors can differ substantially depending on medical specialisation. Stakeholders typically suggested this is driven by a combination of variation in common sites for service delivery (e.g. private sector clinics, public hospitals), the types of services provided (e.g. familial cancer testing, prenatal testing), and typical team structures (e.g. single geneticist versus multidisciplinary team). For example, generalist genetic counsellors tend to be exposed to a wide range of relatively simple cases, and may refer very complex cases to specialist genetic counsellors, while specialist genetic counsellors tend to focus on a specific condition or mutation, and will sometimes have long-term relationships with clients. As discussed in the following chapter, there may be a need for genetic counsellors to increasingly specialise in the future, as the types of genetic information available to consumers becomes more complex and multilayered. A summary of the most common service delivery models for different service settings is outlined in Table 3 .

Table 3 – Scope of practice by medical speciality

Speciality	Main provider	Sector	Model summary and key points to note	Supporting quotes
General clinical genetics	<p>*Clinical genetics departments (and services)</p> <p>*Private sector clinics</p>	Public and private sector	<p>*Genetic counsellors typically work in a delegated clinical role, in which they provide direct and indirect clinical support to one or several clinical geneticists.</p> <p>*Common tasks performed by genetic counsellors working as part of clinical genetics service include (a) setting appointments, (b) initial triage, (c) counselling of patient and family members prior to consultation with clinical geneticists, (d) collation of data, and (e) developing family pedigrees.</p> <p>*The genetic counsellor is also commonly called upon to counsel patients after examination by a clinical geneticist or after genetics test results have been received.</p> <p>*There was a general consensus amongst clinical geneticists that, under this service delivery model, geneticists are able to focus on clinical examination and diagnosis during their consultation with a patient.</p> <p>*Genetic counsellors working in regional and remote areas tend to have less face-to-face contact with clinical geneticists; however, their role scope (e.g. delegated clinical role) is relatively consistent. Genetic counsellors working remotely may also have less administrative support, and operational supervision tends to fall to a local allied health professional.</p> <p>*There is little consistency in private sector service delivery models, including tasks performed and supervisory arrangements.</p>	<p><i>So the genetic counsellor's role is one of bringing efficiency and effectiveness to the actual appointment with the doctor as well as providing the actual psycho-social support counselling. (Clinical Geneticist, LHD)</i></p> <p><i>These [Genetic counsellors] are people who are integrally part of the information giving to the clients where they're actually talking about what are genes, what are chromosomes and they're taking the role in actually helping to consent for genetic tests and organising these new genetic tests, with the doctor, obviously not on their own, but with the doctor they're doing a lot more of the actual information giving and a lot more of the knowing the signs and the genetics and the testing protocols. (Clinical Geneticist, LHD).</i></p>

Speciality	Main provider	Sector	Model summary and key points to note	Supporting quotes
Cancer genetics	Familial cancer services	Public and private sector	<p>*Typically accept referrals from individuals with a personal or family history of cancer or associated tumours.</p> <p>* Genetic counsellors typically work in a delegated clinical role, in which they provide direct and indirect clinical support to a specialist cancer geneticist. Some counsellors may support an oncologist or other medical specialist, in which case they typically have less contact with a clinical geneticist (e.g. weekly rather than daily).</p> <p>*Typically each clinical geneticist is supported by two-three genetic counsellors.</p> <p>*Common tasks are similar to counsellors working in general genetics clinics; however, patient management tends to be more systemised (i.e. a similar approach for all patients) and the turnaround on tests tends to be faster, meaning that there is limited lag between initial and follow-up consultations.</p> <p>* Due primarily to clinic size, clinical supervision and governance tends to be highly formalised in familial cancer services, and genetic counsellors are rarely required to perform administrative tasks.</p>	<p><i>Now in [our LHD] we divorced the general service from the cancer service because we wanted to focus on cancer; they're very different approaches to managing patients, decisions that need to be made in a timely fashion, genetic testing that needs to be done in a timely fashion. (Clinical Geneticist, LHD)</i></p> <p><i>While I started with about one genetic counsellor I [now] have three genetic counsellors; not nearly enough I have to say and we provide a service to over 800, between 800 and 900 new families each year and of course we have 20 years' worth of families already so we have a very large number of mutation carriers that we see, people at high risk and we run a risk management clinic for those individuals with BRCA1 or BRCA2 mutation. (Oncologist, LHD)</i></p> <p><i>And look I think because we're in cancer, a completely different area I think that it's the genetic counsellor who obviously sees the patient, talks to them about family history, looks at that side of things I would say that very much whatever the result is if they need further counselling, until you actually mentioned that I kind of didn't see that [counselling] as the role of the genetic counsellor.</i></p>

Speciality	Main provider	Sector	Model summary and key points to note	Supporting quotes
Reproductive medicine and paediatric genetic services	<p>*Public hospital obstetrics department</p> <p>*Public hospital paediatrics departments</p> <p>*Private sector hospitals and clinics</p>	Public and private sector	<p>*Genetic counsellors working in the public sector tend to work as part of a multidisciplinary team, either in obstetrics or paediatrics departments. This team typically includes at least one clinical geneticist in larger metropolitan hospitals and a part-time (or on-call) geneticist in smaller regional hospitals.</p> <p>*Genetics counsellors typically work in a delegated clinical role; however, counsellors may also have close contact with other specialist physicians.</p> <p>*Tasks performed by genetics counsellors working in obstetrics centre on interpretation and communication of test results (including assisting women to decide whether to test a foetus) and counselling through termination processes, as required.</p> <p>*Tasks performed by genetic counsellors working in paediatrics are primarily ordering specialist tests and counselling families.</p> <p>*Private sector reproductive health clinics typically offer more comprehensive genetics testing (and sometime genomic testing), requiring an additional level of technical expertise and skills from counsellors.</p> <p>*IVF was generally considered by stakeholders to be a highly specialised field, with expertise (including skilled genetic counsellors) primarily found in the private sector.</p> <p>*Genetic counsellors working in the public sector are more likely to be supervised (both operationally and clinically) by clinical geneticists; however, this arrangement is also common in private sector clinics, with many geneticists working across the sectors.</p>	<p><i>So I can obviously speak from my experience, we have two genetic counsellors who perform a lot of our genetic counselling services plus we have several geneticists who perform obviously genetic consultation. In addition to that medical staff and midwifery staff will provide a degree of genetic counselling to women who are trying to decide whether or not to have tests and so on but we do rely quite heavily upon our genetic counsellors for their genetic counselling services. (Maternal Foetal Medicine Specialist, LHD)</i></p> <p><i>We also have counsellors as part of IVF, that's a mandatory part of IVF. You can't be an accredited IVF unit without having access to appropriate counselling services (Oncologist, Private Sector)</i></p> <p><i>So in the public, they are supervised by geneticists who have inspired their work and they are required to meet with the group of genetic counsellors from [another hospital] on a regular basis where they're looking at their work, their workload and their performance and that's how their supervision works so although I'm head of the department I would not be the person doing their performance reviews, it's the geneticists who are doing that. (Obstetrician, LHD).</i></p>

Speciality	Main provider	Sector	Model summary and key points to note	Supporting quotes
Other specialities	<p>*Public hospital specialist units (e.g. neurology, cardiology)</p> <p>*Private sector hospitals and clinics</p>	Public and private sector	<p>*Genetic counsellor works in specialist unit outside of clinical genetics service.</p> <p>*Common tasks performed by genetic counsellors differ depending on the unit of employment; however, tasks tend to centre on interpretation of specialist test results, and communication to and education of patients and family members. Physicians typically order genetics tests, and liaise with clinical geneticists as required.</p> <p>*Genetic counsellors are integrated into the multidisciplinary team (including for operational supervision), and may only have contact with clinical geneticists for clinical supervision.</p> <p>*This model of care, which some stakeholders suggested is becoming more common, highlights the importance of comprehensive education and training for genetic counsellors, as counsellors require adequate clinical expertise and nous to work independently of geneticists.</p> <p>*Stakeholders occasionally cited examples of new graduates working outside of their clinical scope of practice, and giving incorrect or impractical advice to patients and family members (see final supporting quote).</p>	<p><i>I think we are going to see [private practice] a little bit more but that underlines the importance of genetic counsellors having certification because what you want to do is ensure that there is a core competency that any person [has] who is practising independently of a clinical genetic service. (Genetic Counsellor, Victoria)</i></p> <p><i>A certified genetic counsellor does have the skills to be able to work with and the experience to work with medical specialists that have expertise in their management, diagnosis and clinic genetic testing of these type of patients. (Genetic Counsellor, Victoria)</i></p> <p><i>So we have a [junior] genetic counsellor who told a woman at 21 weeks that we would be terminating her pregnancy and we'd do it surgically. Well, no way. (Obstetrician, LHD).</i></p>

While there was no evidence of genetic counsellors currently working in GP surgeries, several stakeholders (including GPs) suggested that there is potential for the development of a primary care genetic counselling model, in which GPs are responsible for communication (and associated counselling) of basic test results, and a part-time onsite genetic counsellor takes on more complicated cases. This service delivery model, it was reported, works well for specialist nurses and allied health professionals, especially in larger practices. There was general agreement amongst clinical geneticists that GPs typically do not possess the requisite knowledge and skills to take on more complex cases, and that, at minimum, primary care models for genetic counselling should incorporate a certified genetic counsellor, one who has regular, formal supervisory contact with a clinical geneticist. This link to a geneticist would, of course, be necessary for the ordering of specialist tests from accredited laboratories. GPs also tended to acknowledge that while they had, in their opinion, the knowledge and skills to order and interpret simple genetics tests, there was a necessity to refer more complex cases to a genetic expert – most commonly a clinical geneticist – for diagnosis and treatment.

Location

In bigger units, you're getting genetic counsellors who are specialising in different areas. So at [one regional centre] for example, they have got one genetic counsellor who keeps it up-to-date and maintains and is the go to person for cystic fibrosis. You've got another person whose main area of interest is cardiac genetics. In rural settings like [we] are in, we're supposed to be experts on everything. So it is really hard. (Clinical Geneticist)

Location was typically acknowledged to be another factor which can influence genetic counselling service delivery models, with genetic counsellors working in regional or remote LHDs (or parts of LHDs) tending to work more independently than those working in metropolitan LHDs, especially if the counsellor is employed outside of the clinical genetics stream. The most common non-metropolitan service delivery model outlined by stakeholders (including genetic counsellors working in regional and remote areas) included the following key elements:

- referrals and consults with patients managed by the genetic counsellor prior to telehealth or face-to-face consultation with clinical geneticist
- face-to-face clinics at local hospitals provided periodically by a clinical geneticist
- operational supervision for the genetic counsellor provided by local team leaders – typically an allied or community health manager – and medical back-up provided by a metropolitan-based clinical geneticist, as required
- clinical supervision by a clinical geneticist provided via telehealth, and mostly in a group environment.

In contrast, genetic counsellors working in metropolitan hospitals, as noted above, mostly work in delegated clinical or integrated allied health roles, and tend to have daily face-to-face contact (including formal and informal clinical supervision) with a clinical geneticist. Despite several stories of well-functioning, clinically sound regional/remote service delivery by genetic counsellors, stakeholders (especially clinical geneticists) cited several potential areas for improvement to ensure state-wide equity in access and quality; the most common of which are listed below.

Training and experience: As noted above, there was consensus amongst stakeholders that genetic counsellors ideally should not work independently of a clinical genetics unit until they have extensive on-the-job experience and a comprehensive understanding of their clinical scope of practice. However, stakeholder feedback suggests that genetic counsellors working in regional and remote areas tend to be new graduates or early career counsellors – primarily due to the difficulty of recruiting people to the country – leading to inexperienced practitioners working in isolation.

Support: After suggesting that genetic counsellors working in regional and remote areas possess variable training and experience, some stakeholders went on to point out that these counsellors are commonly also provided with only limited support, further increasing the likelihood of subpar clinical care, and sometimes prompting early career genetic counsellors to seek employment in metropolitan sites or even to leave the profession.

Expertise: Stakeholders (including genetic counsellors working in regional areas) tended to stress that while genetic counsellors working in metropolitan units have an opportunity to specialise, or at least to work with specialists, genetic counsellors working in regional and remote areas were typically required to consult across numerous specialities, and therefore possess a minimum level of expertise across a range of clinical disciplines, resulting in difficulties in keeping up to date with new tests, mutations, technologies, and so on.

3.5. CHAPTER CONCLUSION

The service delivery model and scope of practice for genetic counselling in NSW vary considerably depending on:

- employer and supervisor
- education and training
- medical specialty
- location.

Variation in service delivery models is enabled through (a) inclusion of public and private sector services, (b) non-centralised provision of public sector genetic services, (c) lack of centralised clinical oversight of public sector genetic services, and (d) current regulatory arrangements (i.e. genetic counselling is currently a non-registered health profession in Australia).

Genetic counsellors tend to work more independently of clinical geneticists when they are (a) employed in the private sector, (b) employed by medical specialties other than clinical genetics, and (c) work in a regional or remote location (including hospital and community based clinics). The extent to which current service delivery models for genetic counselling will be able to ensure safe and sufficient services for consumers in light of emerging challenges is considered in the following chapter of this report.

4. EMERGING CHALLENGES

4.1. INTRODUCTION

Chapters 2 and 3 provided analysis of the role of genetic counsellors and current service delivery models operating in NSW. This chapter further explores the challenges the genetic counselling workforce is expected to face in the coming years, and suggests ways in which these challenges could be addressed.

The literature review identified several emerging challenges currently affecting the delivery of genetic care in NSW, many which were also identified by stakeholders. Most of these challenges relate to the growing field of genomic testing, increased demand for genetic services and the related workforce issues. The common factors that stakeholders expect will influence the future delivery of genetic counselling services typically fell under three interrelated categories:

- the transition from genetic counsellor to ‘genomic counsellor’
- the current (and anticipated) mismatch between demand for and supply of genetic counselling services
- challenges associated with ensuring consistent service provision and adequate governance of an evolving workforce.

Each of these areas is consistent with challenges identified in the literature, and will be explored in detail in the following sections, with discussion focused on options identified by stakeholders and the literature to overcome these challenges.

4.2. FROM GENETIC COUNSELLING TO ‘GENOMIC COUNSELLING’

I think we're just about to enter an era of technological advancement that is going to challenge our profession quite a bit, because we've been used to dealing with one condition at a time and single gene disorders and with advances in technology we're going to need to start getting used to conditions that are more complex, caused by much more complex genetic architecture.
(Genetic counsellor)

It is widely recognised, including in the Australian and international health literature, that the field of genetics is expanding and will become more complex due to the rapid technological advances in genetic and genomic testing (Manolio et al., 2013; Ormond, 2013; Shelton & Whitcomb, 2015). In the past decade, testing for a range of genetic related conditions has become more accessible, and it is expected that the field of genomic testing will grow exponentially in the coming years (Middleton et al., 2015; Shelton & Whitcomb, 2015). These technological developments have practical applications and clear benefits for diagnosis and treatment across a range of diseases. Genetic advances are expected, for example, to lead to an increase in the number of diagnostic laboratory tests available, and in the case of genomics, more complex result analysis and interpretation (Ormond, 2013; Shelton & Whitcomb, 2015). Genomic advances also, according to stakeholders and the literature, represent a paradigm shift to personalised and predictive health care, as more laboratories will have the capability to test for multiple variations across the entire genome; information which can then be incorporated into a patient’s clinical care planning.

While cost is currently considered to be a barrier to widespread genomic testing, almost all stakeholders were of the view that the Australian health system will see a substantial increase – even a ‘genomic explosion’ – over the next five to ten years. While acknowledging the benefits that will stem from these genomic advances, many stakeholders were cautious. These stakeholders typically suggested that without clearer referral guidelines and greater communication between specialties, genomics could become a risk to the effective delivery of genetic services due to the increased demand on services and the difficulty of correctly interpreting increasingly technical and complex test findings. This view is consistent with commentary in Australian and international literature, with researchers and clinicians alike keen to point out the shift in medical paradigms (e.g. from Germ Theory of Disease to Personalised Medicine) allowed by genomic advances requires a transformation in both medical practice patterns and the education of health professionals (See for example Shelton & Whitcomb, 2015). Regarding genetic counsellors, commentators in the literature were especially concerned about whether genetic counsellors are prepared (either through education or training) to play an expanded role in health promotion and in technology-assisted service delivery models (Ormond, 2013; Shelton & Whitcomb, 2015). Each of these potential roles is discussed in turn below.

Preventative health: As noted in the literature, predictive disease modelling in personalised medicine requires the integrating of multiples types of information – genomic, environmental, physiological, and disease biomarkers – to manage health and prevent disease (Shelton & Whitcomb, 2015). There is, therefore, potentially an increased role for genetic counsellors to play in preventative healthcare as the genomic technology becomes more accessible to Australian consumers (Manolio et al., 2013; Ormond, 2013; Shelton & Whitcomb, 2015). This could include using, or assisting a non-genetic specialist to use, an individual patient’s genotypic information in his or her clinical care (Manolio et al., 2013). Prior to taking on this role, however, there is a need for genetic counsellors in NSW to familiarise themselves with health promotion models, an area of study which is not, according to stakeholder feedback and the literature, currently the focus of counsellors’ training or practice.

New service delivery models: International research suggests that there may not be enough genetic counsellors or clinical geneticist to address the challenge of providing counselling for (potentially) widespread genomic testing, with comprehensive pre-counselling for whole gene sequencing estimated to take approximately 2-3 hours (Shelton & Whitcomb, 2015). Educational videos and interactive technologies have been suggested as one way of overcoming this obstacle, through increased efficiency and a reduced need for one-on-one counselling of every detail. As noted by stakeholders, technology-assisted distance genetic counselling may also improve access to counselling for patients in remote locations or when face-to-face counselling is not feasible. This approach should be considered with caution, however, as according to stakeholders and the literature, genetic counsellors in NSW have not been systematically trained in technology-assisted counselling, with most counselling in NSW currently occurring face-to-face, even in regional and remote locations.

Ormond (2013) more broadly suggests that the routine incorporation of genomics into medicine (if it eventuates) will likely induce differences in the scope, approach and process of genetic counselling (i.e. the transition from genetic to genomic counselling). Drawing upon the literature and stakeholder feedback, the remainder of this section of the report will critically consider Ormond’s suggestions regarding likely scope expansion of genetic counselling, with a focus on implications for the NSW health system.

Increased number of conditions included in testing: Genomic testing allows for a wider range of possible diagnoses to be explored through a single test and for additional information regarding unrelated pathologies or risks determined (Ormond, 2013; Shelton & Whitcomb, 2015). Genetic counsellors, therefore, must be able, under a genomic model, to address the scientific and psychosocial issues associated with complex diagnosis, prognosis, and reproductive planning, as well as continuing to perform similar roles for simple Mendelian disorders (i.e. rare single gene disorders). However, stakeholders working in public genetic services were concerned about the increased workload associated with understanding, analysing, interpreting, and reporting the complex variants enabled by genomic testing. A solution offered by the literature, and supported by stakeholders, was increasing specialisation of the genetic counselling workforce, and the genetics workforce, more broadly. Increasing specialisation would, however, necessitate additional training and targeted supervision for counsellors, to ensure ongoing sound clinical governance and consistent patient care, regardless of test focus. Importantly, this service delivery model is currently working successfully in a limited number of large NSW metropolitan hospitals with genetic counsellors developing disease-specific specialist interests, such as cardiology or Huntington’s Disease.

Increased number of ‘positive’ and uncertain results, and overall increased number of disclosed results: A personalised medical paradigm, according to Shelton and her colleagues, requires the transformation of many practice patterns to guard against ethical grey zones and violation (including informed consent) (Shelton & Whitcomb, 2015). Ethical standards must, according to Shelton et al (2015), be addressed and operationalised to protect both from unexpected or unintentional consequences of genetic testing, including protocols for updating patients on complex, unclear, and potentially clinically meaningless results. There is support for the development of such a protocol amongst the stakeholders interviewed for this research, with genetic counsellors in particular noting a need to develop more comprehensive guidelines for informed consent, ensuring that patients are made fully aware of the implications of this scale of testing, and setting realistic expectations as to the nature of the results expected from next generation testing.

Increased time spent with clinicians: Medical specialists focussing on complex diseases are not typically genetics experts, and the rapid advancement of genomics is likely to increase the number of specialists reliant on clinical geneticists and counsellors for interpreting and using genetic information, especially as this information is utilised for prevention and care planning, rather than treatment. According to Shelton & Whitcomb (2015:4) genetic counsellors, especially specialist counsellors, are well-placed to play a role in ‘managing the influx of genetic information both in clinical and laboratory settings’, including communication with and education of non-genetic specialists. The researchers were concerned, however, that there was an inadequate number of genetic counsellors, at least in North America, to meet this expanded role. This assertion is supported by the results of the current research study, with stakeholders expressing concern

that genetic departments will receive an increasing number of genomic test requests from non-genetic specialists, who lack an adequate understanding of the practice and protocols of clinical genetics.

... you do a whole genomic test on somebody, it might pop up with tens or hundreds of changes, we will be involved with helping which of those changes are significant, which ones can we put aside as being not relevant based on clinically what we know about the patient, which ones are going to be likely to be our candidate genes that actually are having a role so there will be a big push for getting counsellors involved in that aspect, more so than ever before. (Genetic Counsellor)

It should be noted that the Australian Genomics Health Alliance is currently undertaking a program of policy-related research to support the development of a framework for translation of genomic medicine to policy. The research will examine the following questions:

- How will health technology assessment need to evolve to allow governments to judge the relative value of genomics in health care?
- What changes will be necessary in the Australian health system to deliver this type of health care in a safe, ethical and cost-effective way?
- What approach to implementation is best suited to manage the development and introduction of genomic medicine into the health care system?
- How can existing infrastructure and electronic platforms be used to maximise the benefits of genomics?

The results of the research study should assist in providing a foundation for the incorporation of genomic testing into the Australian healthcare system (including more concrete answers to some of the questions discussed in this section). However, an ongoing research program will be crucial to ensuring that policy and other guidelines are in line with rapidly changing technological advancements.

4.3. MATCHING SUPPLY AND DEMAND

As the field of genetic becomes more complex it may be necessary for genetic counsellors to become more specialised. This specialisation may follow physician specialisation, which is divided by organ systems such as gastroenterology, hepatology, cardiology, pulmonary, etc. (Shelton & Whitcomb, 2015, p. 5).

Almost all stakeholder reported that demand for genetic testing is increasing and, as a result, the need for genetic counsellors, or at least clinicians with clinical genetics and counselling skills, is also increasing. There was also a consensus amongst stakeholders that, at least within NSW Health, there is not currently an adequate number of clinical genetics staff (including counsellors) to meet the increasing demand. This assertion is supported by stakeholder reports of local wait lists for LHD genetic services from six months to two years, depending on geographical location and perceived urgency of the test. In many locations, the continuing receipt of urgent test requests (for instance, for pre-natal abnormalities) regularly mean that other patients fall further and further back on the waiting list. There is a need, however, to further examine whether demand for genetic counsellors (and indeed clinical geneticists) in NSW is currently outstripping supply, as the current evidence available to the report authors (including the evidence collected for this research project) is reliant on qualitative data. This examination should consider current wait-lists for genetic services, and a clinical audit to ensure that genetic testing (and associated counselling) is only being undertaken when clinically necessary.

It should, however, be noted that in 2015 there were only 27 clinical genetic physicians employed in NSW. According to the data, these physicians primarily work in NSW public sector, mainly in metropolitan Sydney and Hunter New England. The average age of the workforce is 50.5 years of age and working hours 28.5 hours per week (NSW Ministry of Health, 2015a).

Stakeholders typically attributed changing demand patterns to:

- increasing knowledge among non-genetic specialists (e.g. cardiologists, neurologists, endocrinologists) regarding the availability of genetic testing (and sometimes genomic testing) for diagnostic purposes, what one stakeholder referred to as an *explosion of technology*
- increasing information available on the internet and growing public awareness of the links between health disorders and genetic anomalies, including high profile medical stories
- increased availability of unregulated genetic testing available on the internet.

A limited number of stakeholders further mentioned the growing number of private clinics, such as research or IVF clinics, which are providing genetic testing. Some of these clinics, according to stakeholders, employ geneticists or genetic counsellors; however, the level of support for individuals and families reportedly varies considerably between these facilities. In addition, genetic tests can be very expensive, leading some patients to seek further support through the public system following initial engagement with the private sector, increasing demand for genetic services across LHDs. Relatedly, other stakeholders noted that the availability of genetic testing over the internet means that often people receive results without any preparation or follow-up, leading people to then seek assistance through the public system (including NSW Health) to help them understand the test results and their implications.

The attributions made by stakeholders for increased demand are consistent with the results of the literature review, with national and international research consistently showing that increased awareness (both amongst health professionals and the public) coupled with a largely unregulated DTC GT market has meant that more and more consumers are seeking genetic (and sometimes genomic) testing. For example, NSW Health Pathology experts have seen a two-fold increase in demand for genetic testing associated with breast and ovarian cancers, following the public plight of actress Angelina Jolie who chose to have a double mastectomy after learning she carried a mutation in the BRCA1 gene (NSW Ministry of Health Pathology, 2013). Unlike in decades past, this cohort of consumers, importantly, includes community members with a family history of genetic abnormalities, like BRCA1 gene, or symptomology consistent with abnormalities, and those who are simply keen to learn more about their genetic makeup, sometimes so that preventative health measures and meditations can be more targeted (Manolio et al., 2013).

Stakeholders were consistent in suggesting that employment of additional genetic counsellors within NSW Health could reduce wait times for genetic testing, given that one geneticist with several counsellors in his/her team could routinely treat more patients and families by delegating counselling tasks. Stakeholders also commonly suggested that the 'bottleneck' in service delivery is being caused by the lack of counsellors who have the *time* and *capability* to undertake the lengthy tasks of medical and family history taking, gathering of tests and other evidence, and meeting with individuals and families to discuss options, risks and implications, so that these tasks sometimes fall to, already busy, clinical geneticists. Genetic counsellors, it was argued across stakeholder groups, can provide essential background research and preparation for the geneticist, and ensure that the time spent with patients is as efficient as possible.

Stakeholders further suggested that a shortage of qualified clinical geneticists – which was typically attributed to the combined impact of increased demand for genetic services coupled with long training pathways and limited degree placements – is also negatively impacting the number of genetic counsellors working within NSW Health. It is very difficult, if not impossible, for genetic counsellors to practice without a clinical geneticist or other medical professional. Therefore, the lack of qualified clinical geneticists has, according to stakeholders, the potential to result in decreased employment opportunities for genetic counsellors, and consequently increased waiting times for service provision. However, as noted above there is a need to undertake further research before reaching firm conclusions about the availability of genetic service (including clinical geneticists) across NSW Health.

Two further themes emerged in stakeholder feedback on the perceived current (and perceived *increasing*) mismatch between demand for, and supply of, genetic counselling services in the NSW public system: professional registration and education pathways. Each of these themes is discussed in turn below.

Health profession registration: Stakeholders commonly suggested that registration of genetic counselling as a health profession would not only promote consistent service provision, it would also assist in ensuring that NSW patients and consumers are able to access genetic services in a timely manner. These stakeholders viewed registration as a first step in having genetic counselling services covered by the Medicare Benefit Scheme (MBS), which would in turn give genetic counsellors the ability to directly order genetic tests (many of which are MBS items), and to charge patients directly for genetic counselling services in private settings. This, potentially, would both reduce wait times for genetic services in the public sector and also increase the capacity for patients and consumers to access services (including the ordering of basis genetic tests and counselling), especially given that patients could be directly referred to a genetic counsellor for a complete service (e.g. test ordering, interpretation, and counselling).

Because at the moment we don't seem to have any billing processes whereby if they see a patient and they do a fantastic job all by themselves, I'm [clinical geneticist /oncologist] more or less coming in as token and bill [the patient]. And the reason we bill them is that provides for our budget, it pays for the tests, so the emphasis is on billing. (Clinical Geneticist)

Private genetic providers most frequently raised the issue of registration and lack of Medicare classification as an ongoing challenge for the private provision of genetic care, one that would become more evident over

the next few years. These stakeholders reiterated that as the public becomes more aware of genetic testing, there may be an increase in those seeking testing privately; however, without the ability to register for a Medicare provider number, there remains limited incentive or capacity for genetic counsellors to work in private practice.

Education pathways: Stakeholders were quick to point out that education pathways and opportunities for subsequent certification for genetic counsellors in NSW, and more broadly across Australia, are extremely limited and need to be refined to meet (potentially) growing demand (e.g. increased Master's placements; increased opportunities for supervision) or completely re-imagined (e.g. alternative pathways for certification). These measures could, according to some stakeholders, include allowing nurses, psychologists, social workers, and others with appropriate scientific and psycho-social skills to provide genetic counselling with appropriate governance. Other stakeholders suggested the funding of additional training positions for associate genetic counsellors could result in an increase in the number of genetic counsellors achieving certification. It should be noted that the national provision for education and training for genetic counsellors is not under the control of NSW Health. However, it may be possible for the Ministry to initiate discussions with HGSA, with a particular focus on changing entry requirements for certification. Currently, the weight of evidence suggests that the small number of students accepted into the accredited Master's degree each year will continue to act as a bottleneck for the workforce unless alternative pathways are developed.

4.4. GOVERNANCE OF AN EVOLVING ROLE

When you're working in a very well established environment like we are all the genetic counsellors work very closely with the geneticist, clearly we've made that quite clear so there's this two way stream happening all the time. The concern is where there are genetic counsellors that are displaced from genetic units where they are functioning more autonomously and I guess that exists a bit more in the research roles there are some examples of that and that's more of a worry. (Genetic Counsellor)

As noted in the literature review, as genetic testing has become more common, so too has the development of national and international guidelines to regulate the practice of genetic counselling (Rantanen et al., 2008a, 2008b). A recent review of international guidelines found agreement that an ideal counselling session should include 'an appropriately trained professional who understands genetics and its ethical implications well, providing relevant and objective information, promoting its understanding by the patient, offering proper psychological support, respecting the confidentiality of genetic information, dealing adequately with familial implications and potential discrimination, and ensuring informed consent and autonomous decision-making by the counsellee' (Rantanen et al., 2008a, p. 1208).

Despite this consensus, the literature examining governance of the genetic counselling workforce also acknowledges some unique challenges associated with the development of guidance, whether, it be law, written guidelines, or generally applied practice, for this workforce. Put simply, the role scope of genetic counsellors is largely driven by (sometimes rapidly changing) technological advancements and consumer demand, and it is difficult for legislation, and to a lesser extent clinical guidelines, to keep pace with emerging practice. Despite this, most stakeholders acknowledged a need for greater governance of the genetic counselling workforce within NSW Health (and possibly the NSW health system more broadly), with some explicitly providing evidence of inconsistent service provision:

The desire for guidelines amongst stakeholders, and commentators in the literature, was typically driven by one, or both, of the following factors:

Concern about an uncertain future: As noted above, stakeholder and commentators alike are uncertain about whether and how an increase in genomic testing will impact the practice of genetic counselling. There was a general consensus, however, that government and other professional bodies need to 'lead the way' understanding the technology, and ensuring that genetic counsellors continue to practice in a safe and ethical manner, rather than making ad hoc decisions about how new types of information can be used, including through legislative and non-legislative means.

Concern about inconsistency in practice: As noted throughout this report, genetic counsellors employed within the public system in NSW are working in varied ways across location, specialty, and so on, leading some stakeholders to cite examples of inconsistent service provision and a desire for increased accountability. That said, the current (reported) inconsistencies in service provision are less to present significant harm to health consumers, as genetic counsellors are only able to provide ill-informed or incorrect *advice*, rather than perform *procedures* outside of their clinical scope of practice. This assertion is supported

by the results of the research, with no stakeholders recounting situations in which a (currently unregistered) genetic counsellor had significantly harmed a consumer. Further, it should be noted that genetic counsellors, as with all unregistered health professions, are required to adhere to the *Health Care Complaints Commission's Code of Conduct for Unregistered Health Practitioners*.

The recent review of international guidelines suggests that governance for the genetic counselling workforce is most productively managed through a combination of broad legislation (which may not be specific to the counselling workforce) and professional guidelines (Rantanen et al., 2008a). The main advantage of professional guidelines over legislation, according to the literature, is their flexibility to respond quickly to emerging topics (e.g. susceptibility testing, recontracting patients, and counselling ethnic minorities), with allowances for precise wording, fast enforcement, and timely review (Rantanen et al., 2008a). As noted above, the certification program for genetic counsellors (including ongoing clinical supervision) in Australia is not mandatory, but recommended by the HGSA (Human Genetics Society of Australasia, 2011). In addition, there are no legal or other disciplinary ramifications for failing to comply with *HGSA Guidelines on the Process of Genetic Counselling*. There is, therefore, potential for NSW Health to improve governance within the NSW public sector by developing a Policy Directive (if deemed necessary after policy review) and flexible but mandated practice guidelines, for the delivery of genetic counselling services. This potential approach, which is discussed further below, is in line with evidence of current best-practice for regulation of the genetic counselling workforce.

4.5. CHAPTER CONCLUSION

The NSW genetic counselling workforce is facing, and expected to face, several challenges over the coming years, including:

- the transition from genetic counsellor to 'genomic counsellor'
- the current (and anticipated) mismatch between demand for and supply of genetic counselling services
- consistent service provision and adequate governance of an evolving workforce.

Workforce planning and service delivery re-design will assist in ensuring that these challenges are overcome, and genetic counsellors can continue to safely service NSW patients and consumers. Elements of planning and re-design could include:

- increased training in preventative health for genetic counsellors
- increased training in technology-assisted service delivery models for genetic counsellors
- health profession registration
- alternative pathways to registration and practice
- developing of state-wide guidelines (or some other form of centralised clinical governance) for genetic counsellors.

These, and other suggestions for workforce planning, are considered in the full in the following chapter of this report.

5. THE FUTURE WORKFORCE

5.1. INTRODUCTION

This research has examined the role of the genetic counsellor within the NSW Health system, and the settings in which genetic counsellors conduct their activities. Through the literature review and consultation with informed stakeholders – including genetic counsellors themselves, clinical geneticists, and medical specialists, general practitioners and allied health professionals – the research team has sought to answer the questions posed by the Ministry with regard to the current models of care and scope of practice of the genetic counselling workforce, education and training pathways, as well as drivers and challenges which may influence the workforce now and in the future. The findings of this research suggest that there are four major areas which will influence future planning for the genetic counselling workforce:

- the growth of genomics and personalised medicine
- workforce capacity and the need for additional education and training pathways
- flexible models of service delivery to meet rising demand
- clinical governance and accountability structures.

Each of these areas is discussed in turn below.

5.2. GENOMICS AND PERSONALISED MEDICINE

National and international evidence suggests the range and complexity of genetic testing is rapidly evolving. This evolution includes the development of genomic testing, and a more general trend towards personalised medicine, in which medications and treatments are increasingly tailored to the unique genetic and other physical characteristics (e.g. blood type, tumour type, metabolism) of the individual. These developments are occurring so rapidly compared to previous evolutions in medicine that it is difficult to predict exactly how genetic and genomic technology will influence future services.

At the same time, the literature and the consultations have identified an increasing demand by consumers for predictive testing which is likely to increase demand from the public sector for genetic counselling and interpretation. Based on stakeholder feedback and literature, it appears that this demand trend is driven by several factors. First, the commercial availability of DTC GT, at times without the corresponding pre- and post-test counselling to help consumers understand the risks or interpret the results, leading consumers to seek follow up within public health services. Second, advances in genomic testing for specialist medical services (e.g. cardiology, obstetrics, neurology), which will increase the need for genetic counselling; and finally a growing population with heightened awareness of health and disease, leading to requests for predictive screening or preventative health services.

Genomic testing, due to its complexity and the challenge of making sense of the data, will require greater time in interpretation and in imparting results. The literature suggests that this will mean more time required for post-test counselling (Shelton & Whitcomb, 2015).

The unique contribution of the genetic counsellor role to the larger health system lies in the combination of both detailed knowledge of genetics and well-developed skills in counselling. The proportion of skills and knowledge utilised in any one exchange may vary although overall, according to some stakeholders, the need for both elements (scientific knowledge and counselling skills) has been roughly equal to date. In the future, however, the need to interpret ever more complicated test results may mean that the proportion of time spent interpreting tests increases while the amount of time available for providing information, conducting medical histories, and other tasks decreases.

It is likely that the increase in personalised medicine will lead to the need for more health professionals to be able to provide genetic counselling rather than this being the role of only genetic counsellors or medical geneticists (Ormond, 2013; Shelton & Whitcomb, 2015). Such a scenario will require new means of providing education and training to upskill health professionals in the practice of genetic counselling.

5.3. EDUCATION AND TRAINING PATHWAYS

The decision of the University of Melbourne to withdraw the genetic counselling training program for 2017 has highlighted the limited opportunities in Australia for formal training as a genetic counsellor. The requirements for entry to the certification program, as well as the long period of training, also limit the potential of the genetic counsellor workforce to expand to meet expected future demand.

As noted in the literature review, while there is a recommended education pathway to certification, and certification is required to practice in most jurisdictions, the education pathway is not supported by regulation or legislation. Thus, currently it is possible for anyone to undertake genetic counselling as the role is not protected. The lack of regulation, while potentially limiting the development of a career path for the specified genetic counsellor role, offers an opportunity to expand the opportunities for training in genetic counselling, so that other existing health professionals can undertake genetic counselling within the scope of their own roles, for instance, as a nurse practitioner, oncologist, general practitioner, or clinical geneticist.

Increasing the workforce could also be achieved through the inclusion of alternative prerequisites to entry to the genetic counselling certification program, including entry through nursing and other health professional training or individuals with existing appropriate scientific and psychological qualifications. This may also allow for an expanded scope of clinical practice in certain circumstances; for example, to accommodate nurse practitioners who are legally able to order basic diagnostic and surveillance tests. It should be noted that while this may increase the number of already-qualified individuals who could then enter the training program, it does not address the limited number of educational pathways for those seeking to *enter* the genetic counselling workforce without previous health professional training.

The pathway to genetic counselling is currently limited not just by the small numbers accepted into the academic training course, but also, reportedly, by the number of clinical geneticists available to provide supervision. Addressing this shortage will require consideration of the projected future geneticist workforce, as well as whether services could be restructured to allow geneticists to supervise more training positions.

Stakeholders consulted for this project also pointed out that the title 'genetic counsellor' is used in a variety of settings, including some without any patient contact at all; it has been suggested that differentiating roles by additional descriptors such as 'clinical genetic counsellor' or 'laboratory genetic counsellor' might be useful to designate roles. There is not enough evidence to suggest that this is a priority; however, if a structure for career progression were considered, it could be useful to articulate the pathways and opportunities for genetic counselling across service settings. This may be especially useful if the specialisation of genetic counsellors continues to increase with the move toward genomics and personalised medicine.

It may be possible to develop alternative pathways to certification, as well as alternative forms of accreditation to ensure that other health professionals could be trained to provide genetic counselling without claiming the title of 'genetic counsellor'. Potentially, this could include allowing nurses, psychologists, social workers, and others with appropriate scientific and psycho-social skills to provide genetic counselling in the NSW public system with appropriate governance. This will be the most immediate mechanism for increasing the capability of the health system to provide genetic counselling, given that the small number of university training places for genetic counsellors will continue to limit the number of genetic counsellors entering the workforce.

The research team does consider that, on balance, some form of accreditation should be a requirement for providing genetic counselling, whether one is qualified specifically as a genetic counsellor or as some other health professional. In addition, certification should be required in order to use the title 'genetic counsellor'. While protecting the title through formal registration may make the role more attractive for some, and create a career trajectory for genetic *counsellors*, registration potentially may limit the capacity of the health system to create greater capability for genetic *counselling* to be provided by a range of health professionals.

The range of evidence available to the researchers to date does not lead to a conclusion regarding the change of the role to a registered health profession. However, the research team suggests the following avenues could be explored further:

- creating additional specialised training avenues for qualified health professionals to add genetic counselling to their scope of practice, potentially through some form of accreditation indicating that one has completed additional training
- continuing to promote certification as the required standard for employment as a genetic counsellor

- articulating the career opportunities for genetic counsellors, potentially including career progression pathways (such as nominating junior and senior roles)
- initiating discussion between the Commonwealth Government and jurisdictions regarding the potential for increasing the number of places available for formal education as a genetic counsellor
- undertaking further analysis as to the benefits and limitations of registration.

5.4. FLEXIBLE MODELS OF CARE

As identified in the literature review, a range of service models already exist, summarised in the table below.

Table 4 – Flexible models of care

Service model	Key elements
Genetic counselling by a multidisciplinary team	<ul style="list-style-type: none"> • Delivery of genetic counselling by a multidisciplinary team of medical and non-medical staff • Genetic counsellors form an integral part of the team.
Genetic counselling integrated within primary care	<ul style="list-style-type: none"> • Primary care as the gate-keeper for genetic counselling • Greater involvement of general and nurse practitioners in the provision of genetic counselling.
Genetic counselling in collaboration with primary care professionals	<ul style="list-style-type: none"> • Provision of genetic counselling service through close collaboration between genetic counsellors and primary care professionals • Genetic counsellors would be involved in educating and supervising primary care professionals.
Genetic counselling within a linkage model	<ul style="list-style-type: none"> • Centralisation of genetic counselling services through regional centres • Regional centres coordinate services and resources across the health care system.
Genomic counselling through reverse phenotyping	<ul style="list-style-type: none"> • Genomic testing aimed at phenotyping would replace pre-test assessments with post-test assessments with patients • The role of genetic counsellors needs to be explored further due to greater complexity of available information that needs to be shared with patients.

Future models can develop in at least two ways:

- focussing on the role of genetic counsellor
- focussing on the activity of genetic counselling

Each of these possibilities is discussed in turn below.

5.4.1. Expanding the role of genetic counsellor

On the whole, the evidence suggests that genetic counsellors tend to work more independently of clinical geneticists in the private sector, when working under medical specialties other than clinical genetics, or when working in a regional or rural location (including hospital and community-based clinics). The service delivery model for genetic counsellors in these settings is closely aligned with that of allied health professionals such as psychologists and social workers, who typically work independently within a multi-disciplinary team with discipline-specific supervision arranged separately from the work team or setting. The findings of this research project suggest that genetic counsellors work best in this model when their clinical scope of practice is clearly defined and understood, and there are clear avenues for clinical governance and supervision, whether locally or by distance.

In contrast, genetic counsellors working in the genetics departments of metropolitan hospitals tend to work collaboratively with clinical geneticists in a delegated role, even when the geneticist is not always located onsite. Clinical geneticists often spoke of co-counselling and considered that their team-based approach allowed the allocation of cases based on the level of complexity and the requirement for clinical diagnosis, with the clinical geneticist providing the medical expertise and the genetic counsellor providing the ability to synthesise complex data and attend to the informational and emotional needs of patients and families.

Both models appear to work across NSW, and have flexibly evolved to meet the logistical, financial and workforce challenges of settings. Within both models, stakeholders have identified that genetic counsellors are unable to match the increasing demand for services due to the numbers of existing and new families that they treat each year. As noted throughout this report, service expansion is limited both by the number of genetic counsellors and by the number of clinical geneticists who can provide supervision.

In the future, should workforce shortages persist, alternative models may need to be developed, including models which widen the scope for genetic counsellors to work in additional settings, potentially with less direct oversight by clinical geneticists. This is similar to the current practice in which genetic counsellors are employed in private clinics and research institutes but may not be under the direct supervision of a geneticist. Such a model would need to have a clearly defined scope of practice and strong clinical governance, and may require registration as a health profession (or some other form of legislated governance) to ensure the safety of patients and families.

In response, consideration could be given to state-wide centralisation of genetic counselling clinical governance, so that genetic counsellors can work in a wider range of settings with central oversight. Such a structure would help to increase consistency of scope of practice and clinical standards across the state, and could potentially assist to reduce waiting lists by providing a mechanism for balancing supply and demand on a state-wide basis (rather than at an LHD level). A more centralised model, as demonstrated in other Australian jurisdictions, may also increase supply by increasing the capacity of a single geneticist to supervise several genetic counsellors.

5.4.2. Expanding the activity of genetic counselling

While genetic counsellors are considered experts, it is widely acknowledged that other clinicians routinely undertake genetic counselling. For instance, general practitioners provide genetic information and basic genetic counselling for conditions commonly seen in general practice, such as diseases identified through routine pre-natal testing, and conditions such as hemochromatosis. Most GPs consulted for this research were clear about their limitations in this regard, and commonly reported referring to public sector genetics clinics for genetic counselling. As demand for genetic counselling continues to increase over the next decade or so, there could be potential for GPs to play a more significant role in conducting counselling, especially as it pertains to basic diagnostic tests. This could potentially decrease the demand for public sector genetic services, but it would require additional training and education of GPs, many of whom do not, according to research findings, currently possess the requisite knowledge and experience (or indeed desire or capacity) to routinely undertake genetic counselling.

An alternative model for the provision of services within primary care could be considered, however. Under this model, which was proposed by clinical geneticists and received some support from GPs, GPs could employ genetic counsellors (or other health professionals such as nurses or social workers with appropriate additional training) within the GP surgery to provide information, education and support to other health professionals as well as providing genetic counselling directly to patients. This service delivery model would be best suited to large general practice surgeries, to ensure an adequate demand for services, and would be dependent upon (a) general practitioners possessing requisite knowledge and experience to safely order and interpret basic genetic tests, and (b) the availability of appropriate clinical supervision of those undertaking genetic counselling, including direct links with clinical geneticists.

Developments in genomic testing have the potential to increase the need for post-test counselling given the complexity and uncertainty inherent in current understanding of the genome and its implications. Increasing the avenues by which consumers may access such counselling, whether through private practice at a GP surgery or a private genetics clinic, or through the public system in hospital or community-based clinics, may also assist in meeting projected future demand for genomic testing for preventative rather than diagnostic purposes.

As noted above, the research team considers that it could be useful to conduct a clinical audit to determine the actual scope of practice of genetic counsellors across the state and the proportion of time spent on the spectrum of activities conducted from patient engagement through clinical liaison and interpretation of results.

5.5. CLINICAL GOVERNANCE AND QUALITY OF CARE

Across Australia, most jurisdictions have a level of centralised service provision and governance, whether at a state-wide level or through a regional hub-and-spoke model. NSW's model is more varied, with devolved governance to the service level or, in some instances, an outreach model combining genetic supervision from a metropolitan hospital with delegated genetic counselling provided at regional and rural services. Some of the other distinctions between Australian jurisdictions include whether the service accepts self-referrals or whether genetic counselling is designated as a specialist service requiring a referral from another health professional.

Service delivery models also vary for genetic counselling in private medical clinics, with genetic counsellors sometimes operating in close collaboration with a clinical geneticist and sometimes operating relatively independently and with no direct supervision by a geneticist. The existing *Guidelines for Genetic Counsellors Working in Private Practice in Australasia* provide guidance and a framework for clinical governance in the private sector. It is likely that these will need to be monitored and refined as patterns of private practice evolve with increasing technological developments and consumer demand.

Within the public sector, stakeholders were varied in their views but generally supported closer supervision and greater consistency in clinical governance to manage the emotional and other risks for patients that may arise through the results of genetic testing. This could be enhanced through the development and state-wide promotion of clinical guidelines for use in both the public and private sectors. Variations in service access across the state might also be addressed through greater centralisation of referrals and intake, allowing allocation of referrals where capacity exists across the state. Increased use of telephone and video conferencing could facilitate access to genetic counsellors for patients who otherwise would be faced with long waiting lists locally. Such a state-wide system would require cooperation across LHDs and would require negotiation and the development of agreed protocols to be implemented consistently across LHDs.

An alternative to the development of a centralised service system for genetic counsellors could be the enhancement of regional hub-and-spoke models within LHDs with an increased number of generalist genetic counsellors working under a clinical geneticist to ensure clear clinical governance structures.

The research team did not hear many concerns regarding the quality of care provided by genetic counsellors, and those that were expressed primarily concerned instances where newly-trained genetic counsellors made assertions beyond their level of knowledge and experience. This speaks to the need for close supervision of junior genetic counsellors by clinical geneticists or senior genetic counsellors, something which reportedly is compromised when newly-trained counsellors are employed in rural or remote settings where positions are difficult to fill and staff often work autonomously or with little direct supervision. Ensuring that the scope of practice is clearly defined and monitored is likely to address the risk of poor information or advice being provided to patients.

The use of agreed national or state-wide guidelines for practice combined with clearly defined scope of practice and well-articulated clinical governance structures should ensure that risks to patients are minimised. As noted above, it is also possible for guidelines to be flexibly updated in response to emerging topics and challenges, including those related to new technology.

5.6. CHAPTER CONCLUSION

The finding of the consultations and literature, when taken together, suggest that there is a need for the NSW genetic counselling workforce to increase and expand to meet current and expected service demand (including *types* of service), while maintaining strict clinical governance. Future models can develop in at least two ways:

- focussing on the *role* of genetic counsellor – increasing the types of settings in which genetic counsellors may work, increasing the delegated scope of the role, creating a career structure along the lines of nursing through which one may progress to more senior roles (e.g. from genetic counsellor to senior to consultant)
- focussing on the *activity* of genetic counselling – increasing the training available to other health professionals so that genetic counselling may be provided by a range of clinicians.

6. CONCLUSION

In summary, the weight of the literature and the consultation evidence suggested that the future genetic counselling workforce will need to be 1) expanded, with new education and training pathways, 2) more specialised, with additional training in genomics, and 3) flexible, with new models of care and defined governance structures. Consideration of educational pathways is especially pressing, as the University of Melbourne's decision to withdraw the genetic counselling training program for 2017 has highlighted the limited opportunities in Australia for formal training as a genetic counsellor. The requirements for entry to the certification program, as well as the long period of training, also limit the potential of the genetic counsellor workforce to expand to meet expected future demand.

The literature has shown that genetic counselling has positive impacts on individuals and families such as increased understanding of genetic information, and lower levels of anxiety. The value of genetic counsellors lies particularly in their ability to adapt to the needs of the patient, provider and clinic across any healthcare setting.

It is also clear that greater specialisation will be required in the future as genetic and genomic testing become more specialised, requiring the increase in education and training pathways for both genetic counsellors and other health professionals providing genetic counselling. Given the expected increase in demand for genetic and genomic testing, it is likely that the number and type of qualified professional providing genetic counselling will also increase. Recognising this distinction between the role of genetic counsellor and the activity of genetic counselling, the need for clearly defined scopes of practice, clinical guidelines and clinical governance structures will increase.

Emerging service models have been identified in which genetic counselling may be integrated within primary care, or provided in a growing range of settings including genetics clinics, other speciality clinics, and private sector health services such as IVF clinics. Given the increasing specialisation of genomic science, with the need for highly trained professionals to interpret complex and uncertain data, the role of primary care professionals may be less clear. Nevertheless, it is likely that there would be a number of standard and common genetic tests that could be managed entirely within the primary care setting with an appropriately qualified professional providing genetic counselling.

The literature is not expansive with regard to best practice and workforce models for genetic and genomic counselling. Further research to identify exactly what genetic counsellors do and how they do it, through clinical audit and other quantitative methodologies, would strengthen the knowledge base and assist with future workforce planning and the development of strong clinical governance and accountability structures.

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APPENDIX A KNOWLEDGE REVIEW

THE CHANGING LANDSCAPE OF THE GENETICS COUNSELLING WORKFORCE

DRAFT KNOWLEDGE REVIEW
PREPARED FOR NSW MINISTRY OF HEALTH

URBIS

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Report Number	Knowledge Review



Urbis' Public Policy team has received ISO 20252 Certification for the provision of Social Policy Research and Evaluation

Template version 2016.1.0

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ACRONYMS

ASGC	Australasian Society of Genetic Counsellors
ASR	Australian Survey Research
BOC	Board of Censors
CGS	Clinical Genetic Society
DTC GT	Direct-to-consumer Genetic Testing
GCRB	Genetic Counsellor Registration Board
GHQ	Genetic Health Queensland
GOLD	Genetics of Learning Disability
GP	General Practitioner
GSWA	Genetic Services Western Australia
HGSA	Human Genetic Society of Australasia
IVF	In-vitro fertilisation
LHDs	Local Health Districts
MBN	Metastatic Breast Care Nurse
MOPS	Maintenance of Professional Standards
MSc	Master of Science
NHS	National Health Service
NSGC	National Society of Genetic Counsellors
QFCR	Queensland Familial Cancer Registry
RBWH	Royal Brisbane and Women's Hospital
TCGS	Tasmania Clinical Genetic Service
VCGS	Victorian Clinical Genetic Service

EXECUTIVE SUMMARY

INTRODUCTION

Background

Urbis has been commissioned by the NSW Ministry of Health (the Ministry) to gather evidence on the changing landscape of the NSW genetics counselling workforce. This literature review was conducted to inform the research project, with a focus on issues in relation to the provision of genetic counselling, and the role of genetic counsellors in Australia and internationally.

Methodology

A search for relevant literature and documents was conducted through the following databases and resources:

- a number of databases provided by EBSCO, including Academic Search Complete, SocINDEX with Full Text, Health Policy Reference Centre and Social Work Reference Centre
- Google, Google Scholar and relevant websites in Australia and overseas, including but not limited to Commonwealth, Australian state and territory health departments, websites of national associations of genetic counsellors (e.g. the Human Genetics Society of Australasia and the Genetic Counsellor Registration Board in the United Kingdom) and websites reporting on employment data (e.g. the Bureau of Labor Statistics in the United States)
- journals to access relevant peer reviews, including the European Journal of Human Genetics, Journal of Genetic Counselling, Journal of Community Genetics, NPJ Genomic Medicine, Clinical Genetics, British Medical Journal, Medical Journal of Australia, Australian Journal of Rural and Remote Health and the Journal of the American Medical Association.

The search mainly included literature and reports published after 2010, but where relevant, literature and reports published prior to 2010 have been included in the review.

FINDINGS

Genetic counselling

In Australia, genetic counselling is defined as *a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions* (Human Genetics Society of Australasia, 2012).

Overall, evidence suggests that there are two types of professionals involved in genetic counselling:

- professionals involved in genetics (e.g. genetic counsellors and clinical geneticists)
- other healthcare professionals using genetics in routine care, such as general practitioners, specialists (e.g. cardiologists, obstetricians), nurses psychologists and social workers
- (Battista et al., 2011; Centre for Genetics Education, 2016)

This finding applies to Australia, where genetic counselling can be provided by professionals other than genetic counsellors, such as clinical geneticists, other medical specialists and social workers (Centre for Genetics Education, 2016)

The literature suggests that general genetic clinics are the primary providers of genetic counselling services in Australia, and that they are commonly located within tertiary hospitals in metropolitan areas (Mann et al., 2014).

Genetic counsellors in Australia

The role of genetic counsellors is diverse and multidisciplinary. Genetic counsellors can work across a range of specialty areas, and their work can involve a significant workload associated with direct patient care in the clinical environment (Cordier, Taris, Moldovan, Sobol, & Voelckel, 2016; Skirton et al., 2015). In general, genetic counsellors are viewed as specialists who have completed education and training in the core competencies and skills of genetic counselling (European Board of Medical Genetics, 2015). They have

expertise in genetic conditions, but are also trained professionals in psychosocial counselling. This latter element distinguishes a genetic counsellor from a clinical geneticist, who is specialised in the diagnosis and clinical management of patients with genetic disease (Middleton et al., 2015). On the other hand, the genetic counsellor is specifically trained in genetics as well as counselling, which distinguishes them from a social worker (Alabek, Mohan, & Raia, 2015).

At the time of writing, genetic counselling is not a registered health profession in Australia, which makes it possible for any person to offer genetic counselling services (Australian Law Reform Commission, 2003). Within this context, the Australasian Society of Genetic Counsellors (ASGC) highly recommends that genetic counsellors in Australasia complete an approved post-graduate program and practice according to the ASGC professional code of ethics, but this education pathway is not underpinned by legislation (Human Genetics Society of Australasia, 2012).

Variations are evident in the provision of genetic counselling across Australia, mainly depending on the type of clinic or service provider, and the geographic location. However, it is clear from the literature that in Australia, the provision of genetic counselling predominantly involves:

- a multidisciplinary team comprising medical and non-medical professionals, and a genetic counsellor as the centre point of contact for the client
- close collaboration between the genetic counsellor and other members of the team
- public service providers with strong linkages to a tertiary hospital
- private service providers that often operate as IVF clinics, ultrasound clinics and familial cancer clinics.

Emerging service delivery models

A review of the literature shows that genetic counselling can be delivered under different service models. A summary of the most common service models is shown in the table below.

Service model	Key elements
Genetic counselling by a multidisciplinary team	<ul style="list-style-type: none"> • Delivery of genetic counselling by a multidisciplinary team of medical and non-medical staff • Genetic counsellors form an integral part of the team
Genetic counselling integrated within primary care	<ul style="list-style-type: none"> • Primary care as the gate-keeper for genetic counselling • Greater involvement of general and nurse practitioners in the provision of genetic counselling
Genetic counselling in collaboration with primary care professionals	<ul style="list-style-type: none"> • Provision of genetic counselling service through close collaboration between genetic counsellors and primary care professionals • Genetic counsellors would be involved in educating and supervising primary care professionals
Genetic counselling within a linkage model	<ul style="list-style-type: none"> • Centralisation of genetic counselling services through regional centres • Regional centres coordinate services and resources across the health care system
Genomic counselling through reverse phenotyping	<ul style="list-style-type: none"> • Genomic testing aimed at phenotyping would replace pre-test assessments with post-test assessments with patients • The role of genetic counsellors needs to be explored further due to greater complexity of available information that needs to be shared with patients

CONCLUSIONS

- **The value of genetic counselling:** Research has shown that genetic counselling has positive impacts on the patient and the patient's family members, such as increased understanding of genetic information, and lower levels of anxiety. The value of genetic counsellors lies particularly in their ability to adapt to the needs of the patient, provider and clinic across any healthcare setting.
- **A need for clearer governance of genetic counselling:** It is essential that when genetic counselling occurs without dedicated genetic counsellors, the core skills of genetic counsellors should be addressed by other healthcare professionals. In light of this, clear governance and identification of best practice is needed to ensure a high quality of care and consistency of care.
- **Integrating genetic counselling in primary care:** There is a shared view that if genetic counselling is provided by professionals other than genetic counsellors, primary healthcare providers (e.g. practitioners and nurses) are considered as highly suitable for this role. This view is translated into emerging service models where genetic counselling is integrated within primary care to meet a growing consumer demand. The role of primary care professionals appears to be less appropriate amid the growing field of genomics, which highlights the need for specialised professionals who are able to interpret and communicate more complex genetic/genomic information to patients.
- **Moving forward:** It is widely recognised that the field of genetic counselling is expanding and will become more complex due to advanced technology in genomics. In this context, efforts should be made to:
 - define and promote best-practice models focused on high-quality and efficient delivery of genetic counselling
 - identify existing and needed tools and technology to support the efficiency of genetic counsellors.

Seeking direct feedback from stakeholders involved in the provision of genetic counselling will be essential, particularly when considering the limited available literature around best practice and workforce models that address the expanding work area of genetics and genomics. Given the variety of models in which genetic counselling is provided, and the fact that currently there are other medical and nursing professionals who undertake some form of genetic counselling or provision of genetic information, there is a need to clarify the qualifications for undertaking this role and for defining more clearly the place of genetic counselling within and across the health system.

1 INTRODUCTION

1.1 SCOPING THE CHANGING LANDSCAPE OF THE GENETIC COUNSELLING WORKFORCE

Urbis has been commissioned by the NSW Ministry of Health (the Ministry) to gather evidence on the changing landscape of the NSW genetics counselling workforce. Critical questions that the Ministry is seeking to answer include:

- What is the relationship between the genetics counselling workforce and the private sector?
- What models of care exist, or may emerge, where the genetics counselling workforce may be impacted?
- What regulatory or policy drivers may impact on this workforce?
- What are the key challenges facing the workforce now and in the future?
- To what extent do the current roles and education pathways meet the needs of health services and opportunities for future reform?

This literature review was conducted to inform the research project, with a focus on issues in relation to the provision of genetic counselling, and the role of genetic counsellors in Australia and internationally.

1.2 METHODOLOGY

A search for relevant literature and documents was conducted through the following databases and resources:

- a number of databases provided by EBSCO, including Academic Search Complete, SocINDEX with Full Text, Health Policy Reference Centre and Social Work Reference Centre
- Google, Google Scholar and relevant websites in Australia and overseas, including but not limited to Commonwealth, Australian state and territory health departments, websites of national associations of genetic counsellors (e.g. the Human Genetics Society of Australasia and the Genetic Counsellor Registration Board in the United Kingdom) and websites reporting on employment data (e.g. the Bureau of Labor Statistics in the United States)
- journals to access relevant peer reviews, including the European Journal of Human Genetics, Journal of Genetic Counselling, Journal of Community Genetics, NPJ Genomic Medicine, Clinical Genetics, British Medical Journal, Medical Journal of Australia, Australian Journal of Rural and Remote Health and the Journal of the American Medical Association.

The search mainly included literature and reports published after 2010, but where relevant, literature and reports published prior to 2010 have been included in the review.

This review is structured as follows:

- Chapter 2 examines national and international definitions of the provision of genetic counselling and genetic counsellors, highlighting some examples of international practice
- Chapter 3 explores emerging service delivery models in the provision of genetic counselling and challenges impacting the genetic counselling workforce in Australia and internationally
- Chapter 4 provides a summary and identifies key trends to be considered when scoping a new workforce model for the provision of genetic counselling.

1.2.1 Notes on the text

Please note that the international literature shows different forms of words and terminology used by different countries, such as 'clinical geneticist' and 'medical geneticist', and 'genetic service' and 'genetics service'.

For consistency, we have followed the terminology used by the Human Genetics Society of Australasia (HGSA), and thus refer to 'clinical geneticist' when the literature also refers to 'medical geneticist', and use the adjective 'genetic', such as 'genetic testing' and 'genetic service'.

2 GENETIC COUNSELLING

2.1 DEFINITIONS OF GENETIC COUNSELLING

In 1947, Sheldon Reed, the director of the Dight Institute of Human Genetics in Minnesota coined the term 'genetic counselling' and viewed the primary function of genetic counselling as providing *people with an understanding of the genetic problems in their family* (Resta, 2006, p. 269). In Reed's view, health care professionals working as community physicians would most likely provide genetic counselling while relying on support from geneticists at speciality heredity clinics.

While Reed's view dates back nearly 70 years, his view of genetic counselling is still relevant today.

In Australia, genetic counselling is defined as *a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions* (Human Genetics Society of Australasia, 2012).

The process of genetic counselling integrates the following elements:

- interpretation of family and medical histories to assess the chance of disease occurrence or recurrence
- education about the natural history of the condition, inheritance pattern, testing, management, prevention, support resources and research
- counselling to promote informed choices in view of risk assessment, family goals, ethical and religious values
- support to encourage the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.

Source: Resta 2006 et al in: Human Genetics Society of Australasia (2014)

The Australian Human Genetics Society of Australasia (HGSA) description of genetic counselling was developed by the National Society of Genetic Counsellors (NSGC) in the United States. Based on the notion that genetic counselling can be performed by medical and non-medical professionals other than individuals specifically trained as genetic counsellors, the NSGC deliberately describes the activity or process of genetic counselling, rather than the role of a genetic counsellor (Skirton, Cordier, Ingvaldstad, Taris, & Benjamin, 2015).

The following section will describe the provision of genetic counselling within the Australian context, including the role of professionals involved in this process.

2.2 PROVISION OF GENETIC COUNSELLING IN AUSTRALIA

International literature suggests that different medical and non-medical professionals are commonly involved in the provision of genetic counselling, and key elements of 'the act of providing genetic counselling' appears to be widely utilised in various health settings (Battista, Blancquaert, Laberge, Van Schendel, & Leduc, 2011; Middleton, Hall, & Patch, 2015).

For instance, when an obstetrician needs to discuss a genetic test result with patients, it is likely that the obstetrician will use *strong, empathic communication skills to help the patient understand and appreciate the significance of the genetic information* (Arnold and Self, 2012 in: (Middleton et al., 2015, p. 80). As noted by Middleton et al. (2015), this example shows a thin line in *having a conversation based on genetic information* and providing genetic counselling. This is an important distinction in determining the unique contribution to the genetic counsellor and whether genetic counselling services can be undertaken by other clinicians in the health system.

Overall, evidence suggests that there are two types of professionals involved in genetic counselling:

- professionals involved in genetics (e.g. genetic counsellors and clinical geneticists)

- other healthcare professionals using genetics in routine care, such as general practitioners (GP), specialists (e.g. cardiologists, obstetricians), nurses psychologists and social workers

(Battista et al., 2011; Centre for Genetics Education, 2016)

This finding also applies to Australia, where genetic counselling can be provided by professionals other than genetic counsellors, such as clinical geneticists, other medical specialists and social workers (Centre for Genetics Education, 2016).

For example, the literature reports that in Australia, the provision of genetic counselling by cancer genetic centres generally involves co-counselling sessions by a genetic counsellor and other member(s) of the healthcare team (Mann, Taylor, James, & Gaff, 2014). In these settings, the genetic counsellor will usually be part of a multidisciplinary team, which may include clinical geneticists, medical oncologists, nurses, gastroenterologists, breast surgeons and/or gynaecologists. During interdisciplinary meetings, treatment plans for individual patients will be discussed by team members who represent various healthcare specialty areas (Mann et al., 2014).

2.2.1 Genetic counselling service provision

The literature suggests that general genetic clinics are the primary providers of genetic counselling services in Australia, and that they are commonly located within tertiary hospitals in metropolitan areas (Mann et al., 2014).

In New South Wales, public sector genetic services are governed by local health districts (LHDs) and through speciality networks. Genetic services, including genetic counselling, are offered through clinics located within tertiary hospitals, or as outreach units (e.g. community health centre) in regional and remote areas linked with a primary unit located in a city hospital (Centre for Genetics Education, 2016).

Other two main providers of genetic counselling within the NSW public health sector include:

- clinics that offer speciality health related services, such as cancer clinics, prenatal clinics, genetic heart disease clinics and neurological clinics
- specialised genetic services which involve *services that specialise in the management for people affected by particular genetic conditions and risk assessment for concerned family members* (Health Centre for Genetics Clinics, 2013). Examples of these services located in New South Wales include the Genetics of Learning Disability (GOLD) Service, Medications in Pregnancy and Lactation Service, and community screening for people with Eastern European (Ashkenazi) Jewish ancestry.

Private service providers also offer genetic counselling in New South Wales; however, there is little information available in this area. Ultrasound practices, in-vitro fertilisation (IVF) centres and familial cancer centres are most likely to provide genetic counselling in private settings (Human Genetics Society of Australasia, 2016a; Mann et al., 2014).

2.2.2 Genetic counselling per jurisdiction

According to the HGSA website, all states and territories across the country offer genetic counselling services. New South Wales and Victoria offer the widest range in genetic counselling services, mainly located in metropolitan and regional areas in both states.

Key features of genetic services offered by jurisdictions other than New South Wales include the following:

- In Victoria, public genetic services are provided on a 'hub and spoke' basis. Victoria comprises four metropolitan hubs (Parkville, Heidelberg, Clayton and East Melbourne) which also offer outreach clinics to other metropolitan, regional and rural hospitals (State Government of Victoria, 2015). Individuals can access genetic services through a referral by a GP or specialist, or through self-referral.
- In Western Australia, genetic services are provided through the Genetic Services of Western Australia (GSWA). The services primarily involve genetic paediatric services, familial cancer and obstetrics services, and general genetic services. GSWA is located within the King Edward Memorial Hospital for Women and Princess Margaret Hospital for Children. As stated on the

website of the Western Australian Department of Health (n.d.), GSWA offers genetic services through:

- a network of clinics in metropolitan Perth
 - a network of outreach clinics in country areas (Bunbury, Albany, Port Hedland, Kalgoorlie and Geraldton)
 - telemedicine and telephone counselling where appropriate
 - consultation in hospital wards, neonatal nurseries, outpatients, and after hours on-call cover.
- Access to genetic services in Western Australia is through self-referral and GP/specialist referrals (Department of Health, 2011).
 - In South Australia, genetic services are mainly provided through the Women's and Children Hospital in Adelaide, which comprises a paediatric and reproductive genetic unit, an adult genetic unit and a metabolic clinic (Human Genetics Society of Australasia, 2016a).
 - Queensland offers genetic services through Genetic Health Queensland (GHQ) which integrates the Queensland Familial Cancer Registry (QFCR), Queensland Cardiac Genetics Clinic, and the Royal Brisbane and Women's Hospital (RBWH) Renal Genetics Clinic. GHQ clinics are located in Brisbane, Gold Coast, Toowoomba, Nambour, Bundaberg, Rockhampton, Mackay, Townsville and Cairns. Telehealth consultations are also available to patients. GHQ is considered a specialist service and therefore requires patients to provide a written referral from a GP or specialist; it does not accept self-referrals (Queensland Health, 2012, 2013).
 - The Tasmanian Clinical Genetic Service (TCGS) involves genetics related clinical services, including diagnosis and counselling. Services are centralised at the Royal Hobart Hospital, with genetic clinics operating in the Royal Hobart Hospital, Launceston General Hospital and the Mersey Hospital (Latrobe). The TCGS staff primarily comprises genetic counsellors and clinical geneticists (Tasmanian Health Service, n.d.).
 - In the Australian Capital Territory and the Northern Territory, genetic services are provided through the city/regional town hospitals in both territories: the Canberra Hospital, the Royal Darwin Hospital and the Alice Springs Hospital. The Royal Darwin Hospital administers all referrals in the Northern Territory (Human Genetics Society of Australasia, 2016a; Victorian Clinical Genetics Services (VCGS), 2015).

A notable difference between the jurisdictions is in relation to self-referral. Contrary to New South Wales, Victoria, Western Australia and Tasmania, public genetic services in Queensland are considered as specialist services. As such, individuals in Queensland can only access public genetic services upon referral by a GP or specialist, and not through self-referral.

Queensland, Tasmania and Western Australia describe a similar process once individuals are referred to a genetic service. In all three states, a genetic counsellor will initiate the first contact with the individual by mail or telephone to undertake a first pre-consultation session to screen the patient's history, issue of concern and to assess what further steps needs to be taken (Department of Health, 2011; Queensland Health, 2012; Tasmanian Health Service, n.d.).

2.3 GENETIC COUNSELLORS IN AUSTRALIA

The role of genetic counsellors is diverse and multidisciplinary. Genetic counsellors can work across a range of specialty areas, and their work can involve a significant workload associated with direct patient care in the clinical environment (Cordier, Taris, Moldovan, Sobol, & Voelckel, 2016; Skirton et al., 2015).

In general, genetic counsellors are viewed as specialists who have completed education and training in the core competencies and skills of genetic counselling (European Board of Medical Genetics, 2015). They have expertise in genetic conditions, but are also trained professionals in psychosocial counselling. This latter element distinguishes a genetic counsellor from a clinical geneticist, who is specialised in the diagnosis and clinical management of patients with genetic disease (Middleton et al., 2015). On the other hand, the genetic counsellor is specifically trained in genetics as well as counselling, which distinguishes them from a social worker (Alabek, Mohan, & Raia, 2015).

The literature suggests that genetic counsellors commonly work as part of a multidisciplinary genetic healthcare team, and that their role could be more autonomous depending on the type of specialty area, such as oncology or obstetrics (Heald et al., 2016; Skirton et al., 2013).

Importantly, the involvement of genetic counsellors as part of a multidisciplinary healthcare team has been shown to have a positive impact on the provision of genetic counselling services (Alabek et al., 2015; Cordier, Lambert, Voelckel, Hosterey-Ugander, & Skirton, 2012). In Europe, the inclusion of genetic counsellors or specialist genetic nurses is viewed as an essential element in specialist genetic services (Godard et al., 2003).

To maintain high quality services and consistency, HGSA (2012) recommends that the core provision of genetic counselling is provided by trained professionals, and emphasises the key role that genetic counsellors play in the process of genetic counselling. The HGSA (2014) also notes that genetic counsellors are expected to work in partnership with clinical geneticists and other medical specialists. The importance of collaborating with other health professionals is also reflected in the HGSA Guidelines for Training and Certification in Genetic Counselling (2014), which outlines the establishment of effective working relationships *to function within a multidisciplinary team, and as part of the wider health and social care network* is a core competency for genetic counsellors (Human Genetics Society of Australasia, 2014, p. 15).

At the time of writing, genetic counselling is not a registered health profession in Australia, which makes it possible for any person to offer genetic counselling services (Australian Law Reform Commission, 2003).

Within this context, the Australasian Society of Genetic Counsellors (ASGC) highly recommends that genetic counsellors in Australasia complete an approved post-graduate program and practice according to the ASGC professional code of ethics, but this education pathway is not underpinned by legislation (Human Genetics Society of Australasia, 2012).

2.3.1 Education and certification

To date, the University of Sydney and the University of Melbourne are the only two universities in Australia that offer a postgraduate course for genetic counselling (Master's of Genetic Counselling), and the University of Melbourne chose not to offer the course in 2017. The course is accredited by the HGSA Board of Censors for Genetic Counselling and is the only program in the country to enable entry to the HGSA certification program (Human Genetics Society of Australasia, 2014).

Enrolment in the postgraduate course requires:

- an undergraduate degree in a related field (i.e. genetics, psychology, social work, nursing/midwifery, science)
- experience in counselling and/or genetics
- experience in a care role (NSW Ministry of Health, 2015).

After completion of the Master's course, postgraduate students are able to seek employment as genetic counsellors, and to enrol in the HGSA certification program. As noted previously, this certification program is not mandatory, but is recommended by the HGSA (Human Genetics Society of Australasia, 2011). Completion of the program enables candidates to use the title Fellow of the Human Genetics Society of Australasia, and become Certified Genetic Counsellors. The total duration from undergraduate student to Certified Genetic Counsellor takes approximately nine years in total (NSW Ministry of Health, 2015).

Recent data shows that there is an oversupply of students with an interest in genetic counselling. The University of Sydney and Melbourne each take a maximum of 12-14 students annually, and the courses are frequently oversubscribed. For instance, in 2014, the University of Sydney received a total of 85 applications for the 2015 intake (Barlow-Stewart et al., 2015).

2.3.2 Maintenance of professional standards

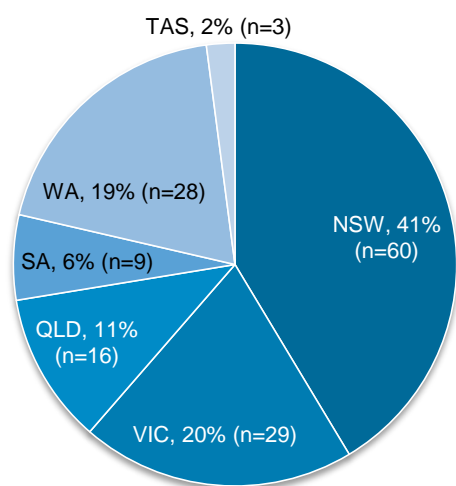
In Australia, Certified Genetic Counsellors are expected to continuously engage in education and training through the Maintenance of Professional Standards (MOPS) program (Human Genetics Society of Australasia, 2014). This program is developed and offered by the HGSA, and involves a five year cycle of resubmission for Certified Genetic Counsellors (Human Genetics Society of Australasia, 2014).

At the time of writing, the HGSA Board of Censors (BOC) is currently reviewing the MOPS program as part of the Australasian Genetic Counsellor Training and Certification standards revision (Human Genetics Society of Australasia, n.d.).

2.3.3 Snapshot of genetic counsellor workforce

Australia-wide employment data for genetic counsellors indicates that the genetic counselling workforce is relatively small. In 2010, a total of 145 genetic counsellors were employed by government health services in Australia (Sillence & Barlow-Stewart, 2011). New South Wales employed the largest proportion of genetic counsellors (41%, or 60 in total), followed by Victoria and Western Australia that both together represented nearly one fifth of the total workforce (20% and 19% respectively).

Figure 1 – Employed genetic counsellors by Government Health Services per state in 2010



Source: Sillence and Barlow-Stewart (2011).

More recent employment data indicates that in 2012, Victoria and New South Wales each employed around one third of the total workforce of genetic counsellors (Australian Survey Research Group Pty Ltd, 2012). This data is based on a survey conducted by the Australian Survey Research (ASR) among members of the ASGC, and included genetic counsellors working in public and private settings.¹ The survey results highlighted other interesting findings related to the work experience and profile of genetic counsellors in Australia. A summary of key findings is provided in the table overleaf.

¹ The data sets from 2010 and 2012 do not allow a comparison over time, given that the 2012 data included genetic counsellors working in the public and private sector, and the 2010 data only comprised genetic counsellors working in the public sector.

Table 1 – Key survey outcomes of members of the ASGC in 2012

Based on census data among 270 members of the Australian Society of Genetic Counsellors (ASGC) in 2012, genetic counsellors were most likely to:

- be female (95%)
- work in New South Wales or Victoria (both states comprised one third of all genetic counsellors)
- work in genetic counselling for a relatively short period of time (39% were employed for five years or less and 28% were employed for 5-9 years)
- work in hospitals (64%), followed by private providers (10%) and universities (8%)
- work as part of a team (71%), compared to 21% that worked as a sole genetic counsellor
- work in general genetic counselling
- feel attracted to the field due to the mix of science and psychology (genetics and working with people)
- enjoy client/patient contact in their profession, followed by being autonomous and being independent and flexible
- have entered the workforce directly from university, or have worked as a scientist or researcher prior to their role as genetic counsellor
- intend to work as a genetic counsellor until retirement (60%), while 21% indicated to move to a different field after working as a genetic counsellor for some time due to limited career progression or job opportunities.

2.3.4 Supervision and accountability

Genetic counsellors who are applying for HGSA certification are required to have formal arrangements in place for supervision and oversight (Human Genetics Society of Australasia, 2014). The HGSA genetic counselling certification training comprises a two-year full-time certification program in which supervision for candidates is mandatory. In general, candidates are expected to have a minimum of two clinical supervisors:

- a clinical geneticist certified by the HGSA or international equivalent
- a health professional with the requisite counselling skills and experience (including HGSA-Certified Genetic Counsellors, at least two years post-certification; accredited social workers with at least three years of experience in hospital or community settings; accredited clinical psychologists with experience in hospital or community settings).

The HGSA (2016b) guidelines for genetic counsellors working in private practice indicate that genetic counsellors are expected to have regular clinical supervision when working in clinical practice. The supervision should entail *educational and supportive functions, development of self-awareness in the genetic counsellor, and may encompass case management functions* (Human Genetics Society of Australasia, 2016b, p. 7).

The HGSA guidelines do not outline more specific management requirements or frameworks for how management of genetic counsellors could take place in practice. This could partly be a result of the various settings in which genetic counsellors can be employed, which makes it harder to have a 'one size fits all' approach to governance and accountability.

As noted by the HGSA (2012), medico-legal responsibility for genetic consultations can vary between services (public and private) and could rest with the medical specialist, the institution or the individual's private liability cover.

2.3.5 Summary

Variations are evident in the provision of genetic counselling across Australia, mainly depending on the type of clinic or service provider, and the geographic location.

Notwithstanding some differences in the context and setting, it is clear from the literature that in Australia, the provision of genetic counselling predominantly involves:

- a multidisciplinary team comprising medical and non-medical professionals, and a genetic counsellor as the centre point of contact for the client
- close collaboration between the genetic counsellor and other members of the team
- public service providers with strong linkages to a tertiary hospital
- private service providers that often operate as IVF clinics, ultrasound clinics and familial cancer clinics.

A summary of various settings in which genetic counselling is provided is shown in Table 2.

Table 2 – Locations of Genetic Counselling in Australia

	Main Provider	Setting	Role of Genetic Counsellor	Geographic Location	Sector
Setting 1	Tertiary hospital	Unit/clinic within hospital	Part of an integrated team	Metropolitan area	Public sector
Setting 2	Tertiary hospital/ community health centre	Outreach unit	Sole practitioner with links to main unit in hospital	Metropolitan/ Regional/rural/ remote area	Public sector
Setting 3	Speciality service	Speciality clinic	Part of an integrated team	Metropolitan/ regional area	Public sector/private sector
Setting 4	Speciality service	Mainly private ultrasound practices, IVF clinics and familial cancer clinics	Part of an integrated team /sole practitioner	Metropolitan/ regional area	Private sector

2.4 INTERNATIONAL COMPARISONS

The definition of genetic counselling as defined by the US-based National Society of Genetic Counsellors (NSGC) is widely accepted internationally (Resta, 2006). In light of this, it is not surprising that the core provision of genetic counselling is comparable across various Western countries. More specifically, there are similarities in where genetic counsellors are employed, and how genetic counselling is provided to patients.

Some differences are notable in relation to legislation, education and how genetic counselling fits within a national healthcare system.

Table 3 overleaf provides a comparison of the role of genetic counsellors and the provision of genetic counselling across Australia, the United Kingdom, France and the United States. Particular consideration has been given to the following:

- legal recognition of genetic counsellors

- certification and qualification of genetic counsellors
- common work areas of genetic counsellors
- main setting of genetic counselling.

Comparisons will be discussed in more detail in the following sections.

Table 3 – International comparison of genetic counsellors and genetic counselling

Country	Legal Recognition of Genetic Counsellors	Certification/Qualification of Genetic Counsellors	Common Work Areas of Genetic Counsellors	Main Setting of Genetic Counselling
Australia	Absence of legal recognition, but formal certification through the HGSA is widely recognised.	Completion of a Master's course in genetic counselling, followed by HGSA certification.	<ul style="list-style-type: none"> Public hospital settings and outreach centres Main work areas include general genetics and oncology 	<ul style="list-style-type: none"> Public service providers with strong linkages to tertiary hospitals (including affiliated clinics and outreach units) Main private providers include IVF clinics, ultrasound clinics and familial cancer clinics
UK	Legal recognition of the title 'Registered Genetic Counsellor' is underway (according to the Genetic Counsellor Registration Board in 2015).	Completion of a Master's course in genetic counselling, followed by registration through the Genetic Counsellor Registration Board (GCRB). A new Master's course 'Genomic Counselling' will commence in September 2016.	<ul style="list-style-type: none"> Regional genetic centres Main work area include general genetics 	<ul style="list-style-type: none"> Regional genetic centres that integrate general genetics, clinical, laboratory services and specialised services Sub-specialty services such as cardiology or oncology share responsibility with clinical genetics
France	Provides a legal framework that protects the profession of genetic counsellors.	Genetic counsellors with a Master's degree in genetic counselling are legally allowed to practice the profession.	<ul style="list-style-type: none"> Regional centres and hospitals Main work areas include clinical genetics, general genetics and oncogenetics 	<ul style="list-style-type: none"> Regional centres in association with tertiary hospitals, including referral centres for rare diseases and cancer, and multidisciplinary clinics for prenatal diagnosis
USA	Legal recognition is partly in place. In 2015, 20 states required genetic counsellors to be licensed, and other states have pending legislation for licensure.	After completion of an accredited Master's degree in genetic counselling, candidates are eligible to apply for certification through the American Board of Genetic Counselling.	<ul style="list-style-type: none"> Hospitals (state, local and private), physician offices and colleges/universities Main work areas include general genetics, prenatal, cancer and paediatrics 	<ul style="list-style-type: none"> Affiliated genetic centres with academic medical centres offering multidisciplinary patient care clinics and laboratory services

Sources: Association of Genetic Nurses and Counsellors (2016); Australian Survey Research Group Pty Ltd (2012); Barlow-Stewart, Dunlop, Shalhoub, and Williams (2015); Barnes, Kerzin-Storarr, Skirton, and Tocher (2012); Bureau of Labor Statistics (2015); Cordier et al. (2013); Doyle et al. (2008); Health Careers and NHS Health Education England (n.d.); Genetic Counsellor Registration Board (2015)

2.4.1 Legal recognition of genetic counsellors

Worldwide, there appears to be a lack of governance that provides legal protection to genetic counsellors. So far, France and Norway appear to be the only countries with legislation that protects genetic counsellors. In France, only professionals with a completed Master's degree in genetic counselling are legally allowed to practice the profession (Skirton et al., 2013).

Other countries are currently working towards such a legal framework, including the United States, the United Kingdom, Iceland, Sweden and Turkey.

For instance, in 2015, 20 states within the United States required genetic counsellors to be licensed, and other states had pending legislation for licensure (Bureau of Labor Statistics, 2015). In that same year, the Genetic Counsellor Registration Board in the United Kingdom anticipated legal protection of the title 'Registered Genetic Counsellor' and formal recognition of genetic counselling as a profession (Genetic Counsellor Registration Board, 2015).

Within the Australian context, the Australian Law Reform Commission (2003, p. 600) reported that the absence of legal protection for genetic counsellors means that *there is no prohibition on any person, however qualified, holding themselves out as a genetic counsellor or offering genetic counselling services. Nor are there formal sanctions for breach of ethical or professional standards in genetic counselling.*

It should be noted that countries tend to have more general healthcare legislation and national guidelines in place that affect genetic counselling, such as confidentiality or patient rights. One study reported that across 38 European countries in 2008, genetic counselling in the context of prenatal testing was most often regulated by legislation and guidelines, followed by genetic counselling in the relation to diagnostic testing and carrier testing (Rantanen et al., 2008).

2.4.2 Certification and qualification of genetic counsellors

Although there are few legal standards specific to the conduct of genetic counsellors, it is more common for countries to formally recognise the profession through developed standards and guidelines, and accredited education and training programs (Cordier et al., 2012).

Overall, countries with national boards and associations for genetic counsellors appear to lead the development and maintenance of professional standards by granting certification, recognition and registration to eligible candidates (Skirton et al., 2015).

In Australia, this role is taken up by the HGSA. Other countries with similar boards and certification processes include the United States (American Board of Genetic Counselling) and the United Kingdom (Genetic Counsellor Registration Board). Both boards grant eligible candidates with formal registration and/or certification. Also similar to Australia, the boards in the United States and the United Kingdom require candidates to have completed an accredited or formally recognised Master's program in genetic counselling.

Other European countries with national organisations specifically for genetic counsellors include France (French Association of Genetic Counsellors), Norway (Norwegian National Association of Genetic Counsellors), Sweden (Swedish Association of Genetic Counsellors) and the Netherlands (the Dutch Society of Genetic Counsellors) (Cordier et al., 2012).

Within relation to education programs, undergraduate students in the United States can choose from a vast range of accredited Master's programs in genetic counselling. In 2014, the United States offered 31 Master's programs accredited by the Accreditation Council for Genetic Counselling (Bureau of Labor Statistics, 2015).

Since 2004-2005, France offers a Master's degree specialised in 'Genetic Counselling and Predictive Medicine', which is the only program in the country for genetic counsellors (Cordier et al., 2013). This program has been evaluated by the genetic counsellors' division of the European Board of Medical Genetics, and is considered as a suitable training program for European genetic counsellors. In 2016, it was reported that a total of 122 genetic counsellors had graduated from this program (Cordier et al., 2016).

In the United Kingdom, important changes in genetic counsellor education are currently underway. In September 2016, a new Master's course 'Genomic Counselling' will commence as part of the National Health Service (NHS) Scientist Training Program. This training program involves a Master of Science (MSc) in Genomic Counselling with traineeship in regional genetic centres. It is anticipated that the program will combine education in the science of genomics and counselling theory, and the required work experience to

practice as a 'genetic counsellor'. The program will also meet the requirements to obtain professional registration.

As stated by the NHS Health Careers website, the Master's course is specifically called 'genomic counselling' to reflect *the increased focus on the use of genomic information and technologies in healthcare* (Health Careers and NHS Health Education England, n.d.)

It is noted that professionals currently working in this area (genomic counselling) are considered as 'genetic counsellors', and that candidates who complete the training program will carry the professional title 'genetic counsellor' (Association of Genetic Nurses and Counsellors, 2016; Health Careers and NHS Health Education England, n.d.).

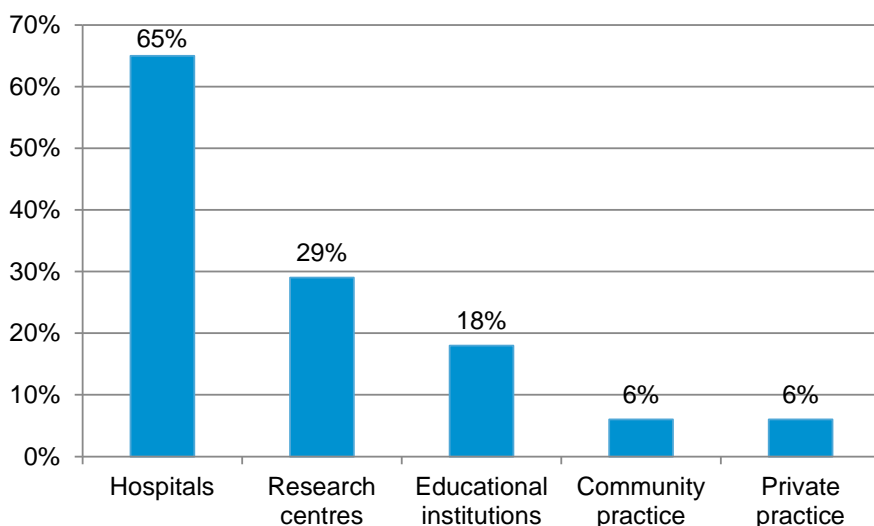
It is anticipated that the new program will run annually, and the appointment of the university that will deliver the MSc program will be released by the start of the academic year 2016-17.

In light of the new program, the Association of the Genetic Nurses and Counsellors (2016) report that the two existing Master's programs in the United Kingdom at the Universities of Cardiff and Manchester will not recruit new intakes of students in 2016.

2.4.3 Common work areas of genetic counsellors

A European study found that across 18 European countries, the most common places of employment for genetic counsellors were hospitals, followed by research centres, educational institutions, community practice and private practice (Cordier et al., 2012).

Figure 2 – Places of employment for genetic counsellors in Europe



Source: Cordier et al. (2012)

Common specialty areas for genetic counsellors include oncology, obstetrics and gynaecology, paediatrics and neurology (Alabek et al., 2015; Centre for Genetics Education, 2016; Cordier et al., 2016).

Due to advances in genetic technology, the need for genetic counsellors is increasing among less traditional settings, such as genetic testing laboratories, insurance companies, and specific multidisciplinary healthcare clinics (e.g. haemophilia treatment centres) (Alabek et al., 2015). Nowadays, genetic counsellors are also more often operating in non-healthcare settings such as education, administration and policy-making (World Health Organization, 2016).

Notwithstanding the expanding work field of genetic counsellors, the 'traditional' settings of genetic counsellors (e.g. hospital settings and general genetic clinics) are still considered as the most common work areas to date.

Available workforce data show that overall, genetic counsellors work across similar speciality areas and settings in Australia, the United States and France². A summary of workforce data for Australia, the United States and France is provided in Table 4, and where possible, proportions are included.

Table 4 – Comparison of workforce data for genetic counsellors in Australia, the US and France

	Australia	United States	France
Workforce data	Workforce data from 2012 among 270 members of the Australian Society of Genetic Counsellors (ASGC).	Workforce data from 2014 which comprised 2,400 employed genetic counsellors across the country	Workforce data from 2011 which comprised 77 employed genetic counsellors.
Most common workplaces	<ul style="list-style-type: none"> • hospitals (64%) • private providers (10%) • universities (8%) 	<ul style="list-style-type: none"> • hospitals (state/local/private) (39%) • physician offices (20%) • colleges/universities/professional schools (12%) 	<ul style="list-style-type: none"> • hospitals (64%) • private providers (10%) • universities (8%)
Most common specialty areas	<ul style="list-style-type: none"> • general genetic counselling 	<ul style="list-style-type: none"> • prenatal • cancer • paediatrics 	<ul style="list-style-type: none"> • clinical genetics (35%) • general genetics (30%) • oncogenetics (29%)

Sources: Australian Survey Research Group Pty Ltd (2012); Bureau of Labor Statistics (2015); Cordier et al. (2013).

2.4.4 Genetic counselling settings

The literature shows that across Western countries, genetic services (including genetic counselling) are primarily provided within tertiary hospital settings and affiliated genetic clinics.

Similar to Australia, each state in the United States manages and delivers its own genetic service system, but throughout the country, genetic centres are primarily affiliated with academic medical centres that offer multidisciplinary patient care clinics, laboratory services, research divisions and outreach clinics (Battista et al., 2011). For example in 2006, the state of Illinois contained over 20 academic medical centres with prenatal or reproductive genetic services, as well as paediatric and adult services, and cancer services (Battista et al., 2011). In addition, 39 local community centres across the state were responsible for implementing screening programs and following up on affected individuals.

In France, genetic services are commonly provided through regional centres in association with university teaching hospitals. This includes referral centres for rare diseases and cancer, and multidisciplinary prenatal clinics. The literature reports that genetic counselling is often integrated within these services (Battista et al., 2011).

A slightly different structure is found in the United Kingdom, where genetic services are mainly provided by regional genetic centres. In 2015, a total of 23 regional centres were established throughout the United Kingdom, with each centre operated by a multidisciplinary team of professionals, including genetic counsellors, clinical geneticists, genetic technologists and scientists, and bioinformaticians (Healthcare UK, 2015).

Interestingly, under the NHS GPs commonly refer patients to a regional genetic centre, where patients are more likely to be seen by a clinical geneticist, rather than a genetic counsellor (Kerr, 2011). It appears that in many cases, the clinical geneticist will determine whether there is a need for a genetic counsellor. For instance, the website of the UK health centre states that once referred to a genetic service, pregnant women *will probably meet with a clinical geneticist who might be able to arrange counselling if necessary* (UKHealthCentre, 2016).

² This section only reports on recent workforce data from Australia, the United States and France. We were unable to find comparable data from other countries.

3 EMERGING MODELS AND CHALLENGES

This chapter examines emerging service delivery models in the provision of genetic counselling, and discusses emerging challenges that may have an impact on the genetic counselling workforce.

3.1 EMERGING SERVICE DELIVERY MODELS

As noted in Chapter 2, a review of the literature shows that genetic counselling can be delivered under different service models. A summary of the most common service models is shown in Table 5, and discussed in more detail below.

Table 5 – Emerging service delivery models for genetic counselling

Service model	Key elements
Genetic counselling by a multidisciplinary team	<ul style="list-style-type: none"> • Delivery of genetic counselling by a multidisciplinary team of medical and non-medical staff • Genetic counsellors form an integral part of the team
Genetic counselling integrated within primary care	<ul style="list-style-type: none"> • Primary care as the gate-keeper for genetic counselling • Greater involvement of general and nurse practitioners in the provision of genetic counselling
Genetic counselling in collaboration with primary care professionals	<ul style="list-style-type: none"> • Provision of genetic counselling service through close collaboration between genetic counsellors and primary care professionals • Genetic counsellors would be involved in educating and supervising primary care professionals
Genetic counselling within a linkage model	<ul style="list-style-type: none"> • Centralisation of genetic counselling services through regional centres • Regional centres coordinate services and resources across the health care system
Genomic counselling through reverse phenotyping	<ul style="list-style-type: none"> • Genomic testing aimed at phenotyping would replace pre-test assessments with post-test assessments with patients • The role of genetic counsellors needs to be explored further due to greater complexity of available information that needs to be shared with patients

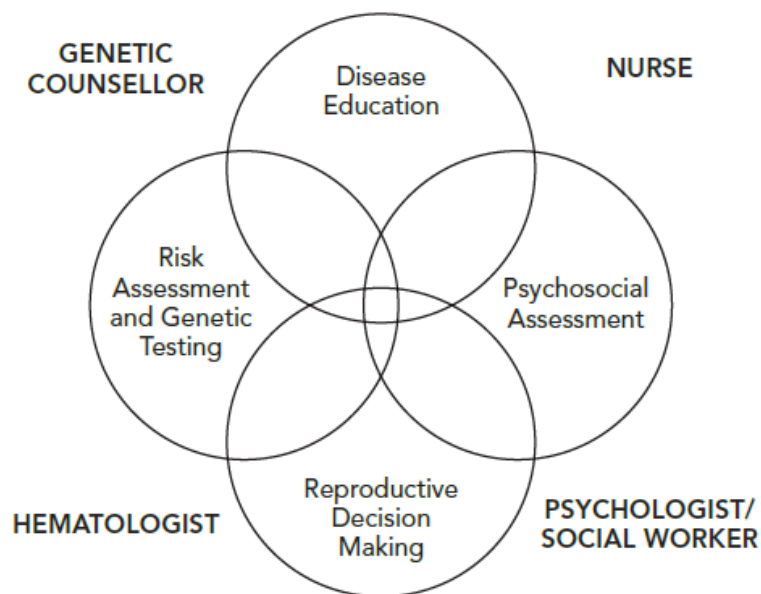
3.1.1 Genetic counselling by a multidisciplinary team

It is clear from the literature that genetic counselling is most commonly provided by a multidisciplinary team of professionals, which resonates with an ongoing trend in healthcare in which interdisciplinary teams are increasingly common (Mann et al., 2014).

An example of how a multidisciplinary team can operate is seen in a service model of genetic counselling in haemophilia treatment and management in the United States (Alabek et al., 2015). As shown in Figure 3, this model depicts the involvement of four different healthcare professionals in the provision of genetic counselling: the genetic counsellor, the nurse, the haematologist and the psychologist/social worker.

In this model, the healthcare providers bring their own specialisations to the team, which enables them to deliver a specific core element in genetic counselling, such as the genetic counsellor who will focus on disease education and risk assessment, and the haematologist who will complement this with counselling relating to reproductive decision making.

Figure 3 – Service model of genetic counselling in haemophilia treatment



Source: Alabek et al. (2015)

The literature emphasises the need for a multidisciplinary model to improve genetic services for children who transition from paediatric to adult care (Battista et al., 2011). Due to improved health care, more children with diseases such as cystic fibrosis, haemophilia and sickle cell anaemia are now able to live longer and transition to adulthood. This requires appropriate continuation of genetic services. A challenging aspect in this transition process includes coordination and inter-professional collaboration, *whether one is dealing with disease-specific models, enhanced primary care or any of the three main categories of clinical genetics: prenatal, paediatric and adult services* (Battista et al., 2011, p. 40). In a transition process, a multidisciplinary approach is needed, which should 'systematically' include paediatricians, general practitioners and/or internists (Battista et al., 2011).

3.1.2 Genetic counselling integrated within primary care

Some literature suggests that there is potential for a model that integrates genetic services within primary care in community settings. This model would dissolve genetic services as a specialty area for clinical geneticists, and would require greater involvement of general and nurse practitioners (Battista et al., 2011).

It has been suggested that this approach could enhance the effectiveness of genetic service provision in terms of comprehensiveness, ongoing care and coordination. However, some considered that this model would *fail to fully recognise the ever-increasing pace of medical genetics, and the resulting alteration in practice in every specialty, including primary care* (Battista et al., 2011, p. 40).

Overall, this model does not specifically envision the role of a genetic counsellor in genetic services, but views GPs and nurse practitioners as potential key professionals in the delivery of genetic counselling services.

3.1.3 Genetic counselling in collaboration with primary care professionals

Contrary to fully integrated genetic services in primary care, an alternative model is suggested which emphasises close collaboration between genetic counsellors and professionals in primary care (Battista et al., 2011). This model would put greater demands on the genetic counsellor's ability to educate and supervise other professionals, and would create new roles and responsibilities for non-genetic professionals in primary and secondary care.

This model would allow GPs and specialist nurses to undertake key elements of genetic counselling, such as identifying at-risk cases, educating patients and recognising specific psychosocial needs. It is expected that this approach would enhance information access for physicians and patients, and improve case management and follow up processes. The literature does not refer to specific countries where this approach has been fully integrated in the health system.

3.1.4 Genetic counselling within a linkage model

Based on the service care system in the United Kingdom, a 'linkage' model has been suggested which links regional centres with primary and secondary services, as well as with specialised care centres (e.g. cancer centres) and other tertiary services (Battista et al., 2011). Regional centres would coordinate services and resources across the health care system. Regional centres would also be well placed to mobilise expertise in their networks to develop public health policy or practice guidelines *while also relying on the experience of user groups* (Battista et al., 2011, p. 38).

This existing structure in the United Kingdom has led to greater interaction between regional centres and other speciality areas due to increased awareness of genetic medicine. It is foreseen that this will also lead to a broader multi-disciplinary genetic team, and specialisation of other health professionals within genetics (Kerr, 2011).

3.1.5 Genomic counselling through reverse phenotyping

Within the context of genomics as a new and growing area of practice, the Clinical Genetics Society (CGS) in the United Kingdom has proposed a different model for a genetic clinic (Clayton-Smith, Newbury-Ecob, & Greenhalgh, 2015). As stated by the CGS, genetic clinics usually follow a traditional model which involves a referral for assessment, followed by investigation tailored to a specific presentation and diagnoses. The growing field of genomic testing could replace pre-test assessment by post-test assessment when it is *aimed at phenotyping to assist in interpretation of genomic results* (Clayton-Smith et al., 2015, p. 8).

An example is a patient with an undiagnosed intellectual disability, who could participate in whole genome sequencing arranged by a paediatrician. The patient will then receive an appointment with a clinical geneticist for discussion and interpretation of the results. This approach is called 'reverse phenotyping'.

In this approach, the CGS does not refer to the specific involvement of a genetic counsellor. It rather highlights that the role and skills of genetic counsellors will need to be explored due to the increased complexity of result reporting and interpretation, and that the need for pre-test counselling would diminish.

3.2 EMERGING CHALLENGES

Besides emerging models in the provision of genetic counselling, the literature also describes emerging challenges that genetic counselling is currently facing. Most of these challenges are in relation to the growing field of genomic testing, and changes in consumer demand.

This section summarises emerging challenges in relation to the profession of genetic counsellors and the provision of genetic counselling. A summary of these challenges is provided in Table 6, and discussed in more detail in the sections below.

Table 6 – Summary of identified challenges

Challenges	Key area	Key description
Challenges in governance and accountability	The role of genetic counsellors in medical examinations and genetic testing	The role of genetic counsellors in medical examinations and genetic testing can vary in practice, and there is no best practice available that provides clear guidelines and processes
	Management and oversight	There is a need for formal guidelines or models that outline: <ul style="list-style-type: none"> governance of the provision of genetic counselling by a multidisciplinary team in the absence of a genetic counsellor supervision/management of genetic counsellors when working in multidisciplinary teams
Challenges in career progression	Professional development of genetic counsellors	A lack of professional development is a key reason why genetic counsellors leave the profession Opportunities are identified to improve this area in a multidisciplinary healthcare setting
Challenges due to consumer demand	Increased consumer demand for genomic testing	The growing field of genomic testing will require specialised and new skills in counselling to interpret and communicate complex genetic and genomic information Genomic testing may not always require genetic/genomic counselling, and primary care professionals could be more involved in counselling services
	Increased Direct-to-Consumer Genetic Testing (DTC GT) Integration of advanced technology to meet consumer demand	DTC GT integrates new ways in which genetic counselling can be provided, and healthcare professionals other than genetic counsellors could play a role in this Increased consumer demand is partly reflected in the use of genetic counselling by telephone and telegenic, which require new skills from genetic counsellors to engage and communicate with patients

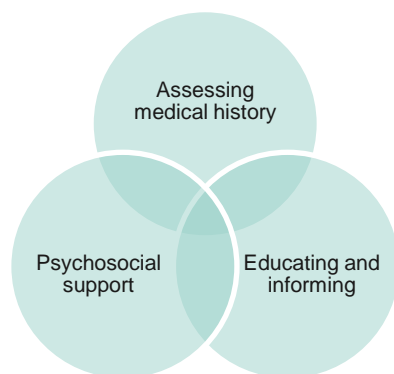
3.2.1 Challenges in governance and accountability

The role of Genetic Counsellors in medical examinations and testing

The literature reports on different views on the roles and responsibilities of genetic counsellors.

As shown in Figure 4, the commonly identified core activities for genetic counsellors include activities in relation to assessing the medical history of an individual and the individual's family, educating and informing the individual about outcomes and options, and providing psychosocial support (Skirton et al., 2015).

Figure 4 – Core activities for genetic counsellors



However, there is discussion in the literature in relation to the role of genetic counsellors in medical examinations and genetic testing.

For example, a French study found that of 126 clinical and laboratory geneticists who worked closely with genetic counsellors, 44% were satisfied with the ability of genetic counsellors to offer genetic testing, compared to 29% that occasionally agreed with this, and 18% that found this task inappropriate for genetic counsellors (Cordier et al., 2016). Views were also mixed in relation to delivering genetic test results, with 46% of physicians who found this appropriate for genetic counsellors. Physicians working in cancer genetics were least likely to share this view.

In addition, physicians viewed the skill 'helping the patient in the decision to carry out a genetic test' as irrelevant for genetic counsellors. They also considered genetic counsellors to be less appropriate for managing pre-symptomatic testing sessions, and rather preferred genetic counsellors to manage risk assessments of genetic abnormality with patients (Cordier et al., 2016). Overall, it was noted that the scope of the genetic counsellor's activities could largely depend on the 'relationship of trust' with the clinical geneticist, underpinned by the genetic counsellor's professional experience, humane qualities and professionalism (Cordier et al., 2016).

In the United Kingdom, an established working party of the national Association of Genetic Nurses and Counsellors and the Clinical Genetics Society noted that the role of genetic counsellors would be less appropriate when medical input is needed (Kerr, 2011). The working party viewed genetic counsellors as most appropriate for counselling intervention such as facilitating decision making in pregnancy or with regard to genetic testing, and in the follow-up phase with patients. However, it was reported that the role of a genetic counsellor could expand, depending on the acquired specialised skills of the individual.

This view is shared by the HGSA (2016b, p. 7) which describes genetic counsellors as allied health professionals, and considers diagnostic assessment and medical management *beyond the scope of the genetic counselling role*. The HGSA recognises that in practice, the role of genetic counsellors can vary, depending on the context of the patient and the level of experience of the genetic counsellor. However, the HGSA emphasises the need for involvement of clinical geneticists and other medical specialists in complex cases, and that clear boundaries need to be set in order to identify these cases.

There is anecdotal evidence that clinical examinations can be undertaken in Australia by genetic counsellors, particularly by genetic counsellors employed in rural outreach areas (James et al., in: Skirton et al., 2015). Although this finding was first reported in 2003, the literature describes this outcome as significant compared to other international findings. It was suggested that the nursing background of some of the genetic counsellors may have played a role in undertaking clinical examination, as well as the rural or remote location where these types of examinations were conducted (Skirton et al., 2015).

Management and oversight

A study among European genetic counsellors found that there was a need for more regulation of genetic counselling, and more uniform practices of education and organisation (Rantanen et al., 2008). Given that genetic counselling can be provided by professionals other than genetic counsellors, clear management and oversight of the provision of genetic counselling is important.

In the absence of a genetic counsellor, the responsibility for the delivery of genetic information is usually carried by other members of a medical team, such as physicians, nurses or psychosocial professionals. However, it has been suggested that the clinical responsibilities of professionals could mean that genetic counselling is considered to be less of a priority (Alabek et al., 2015).

A lack of clear standard practice in the management and oversight of genetic counselling also resonates with the Australian context, where, as noted previously, medico-legal responsibility for genetic consultations in Australia can vary between services, and could rest with the medical specialist, the institution or the individual's private liability cover (Human Genetics Society of Australasia, 2012).

There is also evidence that while genetic counsellors commonly work as part of a healthcare team, the role of genetic counsellors in the team can be rather autonomous (Cordier et al., 2016; Kerr, 2011; Skirton et al., 2013). For instance, in practice, genetic counsellors can manage consultations independently, and they can provide information and sometimes share test results with patients without the presence of a qualified clinical geneticist (Cordier et al., 2016).

This highlights the need for clear processes to ensure the supervision and guidance of genetic counsellors, although the literature does not report on best practice or developed models. The different settings where genetic counsellors can work could be a main reason for a lack of clear models and frameworks, as well as the involvement of various professionals in the provision of genetic counselling.

The development of a workforce model that incorporates clear governance of genetic counsellors could partly be informed by the job statements for genetic counsellors as developed in the United Kingdom. In the United Kingdom, four levels are identified in which genetic counsellors can operate, depending on the level of experience and expertise. Each level comprises a set of key competencies, which includes the provision of guidance and leadership to peers (see Table 7).

Table 7 – Key competencies of genetic counsellors

Level of expertise	Key competencies
1. Genetic counsellor trainee	<ul style="list-style-type: none"> • Undertakes genetic counselling under the guidance of a more senior counsellor and maintains relevant records • Working towards registration with professional body
2. Genetic counsellor	<ul style="list-style-type: none"> • Assesses and provides genetic counselling to patients/clients • Manages a caseload and maintains patient/client records • Provides support and guidance to students and less experienced counsellors
3. Genetic counsellor Principal	<ul style="list-style-type: none"> • Assesses and provides specialist genetic counselling to patients/clients • Manages a specialist caseload and maintains patient/client records • Maintains a knowledge of the scientific medical and psychological aspects of clinical genetics and provides support and guidance to students, genetic counsellors and other health professions • Provides specialist training, and may lead a specialist team
4. Genetic counsellor Consultant	<ul style="list-style-type: none"> • Provides expert professional advice to patients/clients, colleagues and other health professionals • Acts as lead genetic counsellor in field of expertise • Collaborates with academic institutions to lead on education, research and practice development • Provides specialist consultancy within organisation and externally

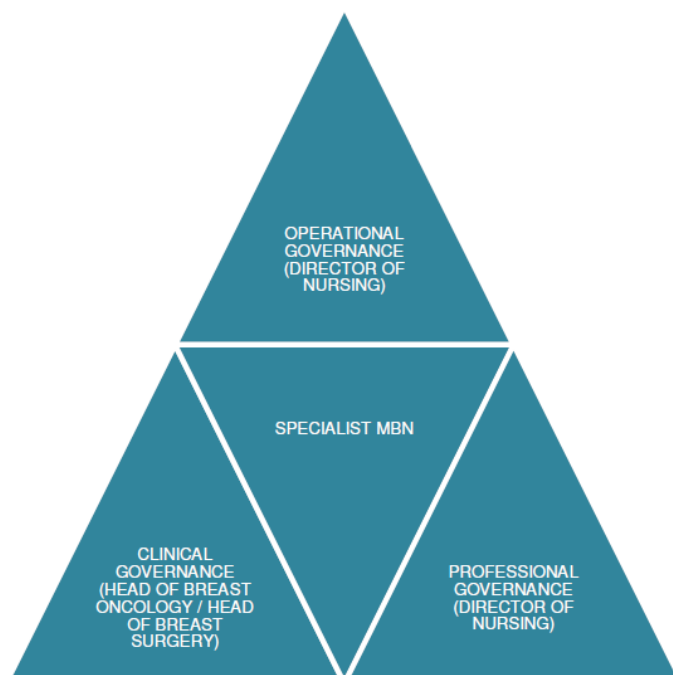
Source: Kerr (2011)

While these job statements outline expected levels of leadership roles of genetic counsellors, it does not reflect how genetic counsellors could be supervised by other professionals when part of an interdisciplinary team. It is suggested that frameworks used for specialised nurses could be useful resources to address this issue.

For example, the Southern Melbourne Integrated Cancer Service proposed a governance framework which reflects the multidisciplinary nature of the team in which a specialist metastatic breast care nurse (MBN)

takes part. It also shows how management and oversight is governed in each healthcare specialty area (Kruss, Macindoe, & Davis, 2014).

Figure 5 – Specialist MBN Governance flowchart



Source: Kruss et al. (2014)

While this model is for breast care nurses, it may provide a useful model for genetic counselling by integrating three levels of governance: clinical governance, operational governance and professional governance. This model implies that a senior leadership role in genetic counselling needs to be created to provide operational and professional governance.

3.2.2 Challenges in career progression

As found in the workforce survey among members of the Australasian Society of Genetic Counsellors in 2012, the most common reason for genetic counsellors to leave the workforce was limited career progression or a lack of job opportunities (Australian Survey Research Group Pty Ltd, 2012).

Another Australian study notes that genetic services are often located in distinct clinical departments, which creates limited opportunities for genetic counsellors to have direct exposure to medical environments while working in an interdisciplinary medical team (Mann et al., 2014). As such, this study reported that Australasian genetic counsellors working in cancer centres mostly gained knowledge through anecdotal descriptions from patients, while professional development through genetic counselling training was the least commonly reported source (Mann et al., 2014).

Interestingly, this study found that a vast majority of respondents anticipated that interdisciplinary observations (e.g. observation of colonoscopy, gastroscopy, chemotherapy) would enhance their professional development. This finding suggests that interdisciplinary education could be considered as an important resource for genetic counsellors to enhance their professional development.

3.2.3 Challenges due to consumer demand

Increased consumer demand for genomic testing

It is widely recognised that the field of genetics is becoming increasingly more complex due to new developments in genetic testing. In the past decade, testing for conditions other than Mendelian diseases has become more accessible, and it is expected that the field of genomic testing will grow exponentially in the coming years (Middleton et al., 2015; Shelton & Whitcomb, 2015). On behalf of the Clinical Genetics Society in the United Kingdom, Clayton-Smith et al. (2015) even foresees that counselling and testing for commoner diseases, and particularly Mendelian disorders will be dissolved.

In general, medical specialists in complex diseases are often not genetic experts, which makes it challenging for medical specialists to inform and guide patients on the interpretation of genetic test results and to provide appropriate psychosocial support associated with specific diagnoses (Shelton & Whitcomb, 2015).

Within the context of genomics as a growing field, the role of specialised genetic professionals who are able to interpret complex genetic and genomic information and guide patients through this process will become more important. Genetic counsellors could play a role in this; however, specialisation will be necessary, given that genetic counsellors would need to be able to address psychosocial issues associated with complex and rare diagnosis, prognosis, reproductive planning, and risk to family members (Shelton & Whitcomb, 2015). This also includes informing patients on appropriate genetic testing approaches such as single gene, sequencing panels, whole-genome sequencing, the costs and benefits of testing, the interpretation of negative results, and the follow-up steps needed based on the results.

Middleton et al. (2015) note that genomic information may not always involve an ‘emotional conversation’, which makes the role of genetic counsellors for the delivery of genomic information less relevant. It has also been argued that primary health care providers could be more involved in genomic counselling, particularly when it involves genomic health risk assessments which relate to decisions about lifestyle choices and appropriate participation in population screening programs (Clarke & Thirlaway, 2011).

Alternatively, it has been suggested that genetic counsellors could play an important role in supporting other health care practitioners in providing generic genomic information, particularly in the interpretation of data, how to incorporate this into their usual practice and appropriate criteria for referral of families to genetic services (Clarke & Thirlaway, 2011; Middleton et al., 2015). In this scenario, genetic counsellors and clinical geneticists are likely to evolve into expert practitioners in genomic counselling, and specialists in the management of genomic information delivery ‘when there is an emotional and familial context’ (Middleton et al., 2015, p. 82).

Increased direct-to-consumer genetic testing

A growing consumer demand in genetic testing is also notable within the area of direct-to-consumer genetic testing (DTC GT). In the last decade, more consumers are able to independently access genetic testing products online. Heald et al. (2016) reported that in February 2016, over 55,500 genetic tests were commercially available for over 4,000 genes and genetic disorders.

This form of genetic testing has created new ways of genetic counselling. As identified by Harris, Kelly, and Wyatt (2013), four representations of genetic counselling provision can be seen within DTC GT (see Table 8).

Table 8 – Types of genetic counselling services within DTC GT

Counselling type	Key description
Integrated counselling	Genetic counselling is marketed as an integral part of the genetic testing product. This form of genetic counselling includes determination of test appropriateness, and personal interpretation, are represented as part of the testing process, following a clinical services model.
Discretionary counselling	Consumers are provided with the option to contact a genetic counselling service, offered by a DTC GT company. There is variation in how companies advertise these services, ranging from positioning a genetic counselling service as a ‘critical part of the genetic testing experience’, to only briefly referring to this service as a potential option.
Independent counselling	Genetic counselling is offered by an external company, and consumers can choose whether they would like to access the service or not.
Product advice	The provision of genetic counselling concerns information provision about the test for sale. In some circumstances, this is similar to ‘traditional’ pre-test counselling. For example, genetic counsellors can assist in the decision making about whether testing is appropriate.

Source: Harris et al. (2013)

Importantly, research has shown that accessing genetic counselling services through DTC GT has a positive impact on consumers' understanding of test results and genetics in general (Darst, Madlensky, Schork, Topol, & Bloss, 2013). In this study, consumers who accessed a genetic counselling service generally viewed the service as informative and beneficial, and that it made them more likely to discuss their results with a physician. Consumers who took the test but did not access a genetic counselling service generally felt comfortable to interpret the test results themselves, without the help of a genetic counsellor (Darst et al., 2013).

The literature includes some discussion of possible roles for genetic counsellors within DTC GT. This new form of genetic testing shows a move towards the provision of preventive health and lifestyle advice associated with disorders. This could include risk calculation and counselling on the influence of genes and the consumer's environment on disease developments, such as type 2 diabetes or kidney disease (Harris et al., 2013; Shelton & Whitcomb, 2015). In light of this, some consider that other genetic experts than genetic counsellors may be more appropriate to provide such services, such as nurses specialised in genetics, who may be perceived as more suitable to provide lifestyle advice and clinical advice regarding common health conditions (Harris et al., 2013).

Integration of advanced technology

The literature describes a shift in genetic counselling practices other than 'in-person counselling' to improve access and efficiency of genetic counselling services, and ultimately, to better meet a growing consumer demand (Cohen, Huziak, Gustafson, & Grubs, 2016).

Based on the methods in which genetic counselling can be delivered, the literature identifies four types of practice models as summarised in Table 9.

Table 9 – Practice models for the delivery of genetic counselling

Practice models	Key description
In-person genetic counselling	Counselling that is provided in-person. Follow up and results disclosure may occur by telephone or other means.
Telephone genetic counselling	Remote counselling by telephone. The telephone call may be supplemented by written, online or other resources.
Group patients	Counselling in a group setting by a genetic counsellor, which may be followed by individual assessment and counselling.
Telegenic counselling	Remote counselling by using videoconferencing.

Source: Cohen et al. (2013)

There is evidence that the provision of genetic counselling is increasingly provided through telegenic and telephone counselling (Cohen et al., 2016). Research in the United States shows that while the 'traditional' in-person counselling approach was most commonly used, a considerable portion of genetic counsellors applied telephone genetic counselling as an alternative method in their communication with patients. Interestingly, telegenic counselling was most commonly utilised by genetic counsellors working for government agencies and diagnostic laboratories, but further investigation is needed to explore the reasons for this (Cohen et al., 2013).

Another study in the United States reported that genetic counsellors used telephone counselling most often for post-testing communication than pre-test communication, and for both cancer, and non-cancer conditions (Bradbury et al., 2011). This finding suggests that genetic testing for hereditary predisposition to cancer and other medical conditions has become more common in clinical settings.

The key benefits of remote counselling, such as telephone genetic counselling, include a decrease in costs, travel time and waiting time for the consumer (Buchanan, Rahm, & Williams, 2016; Cohen et al., 2016). However, with telephone genetic counselling, the inability to see the patient and to read 'physical cues' may not always make this method a suitable resource. Additional training in this method to assess clients' understanding and readiness for genetic information is important to ensure high quality standards of counselling services (Cohen et al., 2016). In light of this, telegenic counselling is an important area for further exploration in the provision of genetic counselling, given that it enables visual contact between the counsellor and patient, while travel and waiting time are significantly reduced.

4 CONCLUSION

The value of genetic counselling

It is clear from the literature that genetic counselling is an important element in the provision of genetic services, but that the need for genetic counselling can depend on the context of the patient and the healthcare professionals involved. Nonetheless, research has shown that genetic counselling has positive impacts on the patient and the patient's family members, such as increased understanding of genetic information, and lower levels of anxiety (Darst et al., 2013; Torrance et al., 2006). The value of genetic counsellors lies particularly in their ability to adapt to the needs of the patient, provider and clinic across any healthcare setting (Alabek et al., 2015).

A need for clearer governance of genetic counselling

Given the variety of ways in which genetic counselling is currently provided in Australia and other international jurisdictions, it is clear that a 'one size fits all' approach does not apply. Genetic counselling is provided across a broad range of healthcare settings by genetic counsellors and other healthcare professionals from various specialty areas.

This multidisciplinary element enables a wide public to access genetic counselling services, but it also appears to lead to a lack of clear governance processes, particularly in relation to accountability and management of genetic counselling.

While genetic counsellors are considered as experts providing genetic counselling, it is widely accepted that genetic counselling can occur without genetic counsellors. However, it is suggested that without the presence of a genetic counsellor, the prioritisation of the psychosocial element with regard to the provision of genetic information could diminish, which ultimately may have a negative impact on the quality of care to the patient. Therefore it seems essential that when genetic counselling occurs without dedicated genetic counsellors, the core skills of genetic counsellors should be addressed by other healthcare professionals. In light of this, clear governance and identification of best practice is needed to ensure a high quality of care and consistency of care.

Integrating genetic counselling in primary care

There is a shared view that if genetic counselling is provided by professionals other than genetic counsellors, primary healthcare providers (e.g. GPs, nurses and nurse practitioners) are considered to be highly suitable for this role. This view is translated into emerging service models where genetic counselling is integrated within primary care to meet a growing consumer demand. Bringing genetic counselling services more to the forefront in community settings is particularly relevant for the expanding field of direct-to-consumer genetic testing, where the appropriateness of accessing a genetic counselling service is only assessed by the consumer.

On the other hand, the role of primary care professionals appears to be less appropriate amid the growing field of genomics, which highlights the need for specialised professionals who are able to interpret and communicate more complex genetic/genomic information to patients. Some view that genetic counsellors would be appropriate for this role, but that upskilling and specialisation would be essential to transfer to this new and highly complex field.

It is envisioned that primary care professionals could play a role in genomic testing when there is greater demand for genomic health risk assessments, which could lead to informing and advising patients on healthy lifestyle choices and the influence of the consumer's environment on disease developments, such as type 2 diabetes or kidney disease. Within this context, the genetic counsellor can take up the role as the educator and supervisor for primary care professionals to provide them with the necessary skills in interpreting and sharing information.

Given that the area of genomic testing is still in its infancy, further exploration of the role of the clinical geneticists and genetic counsellors is needed, as well as assessing the appropriateness and relevance of genomic counselling in this area.

Moving forward

It is widely recognised that the field of genetic counselling is expanding and will become more complex due to advanced technology in genomics. This trend is reflected in workforce data in the United States, where it is estimated that the profession of genetic counsellors will grow by 29% from 2014 to 2024 (Bureau of Labor Statistics, 2015). This growth rate was significantly higher than the growth rate anticipated for 'other healthcare practitioners and technical occupations' (10%) and the national average (7%). In light of this, the

National Society of Genetic Counsellors in the United States identified the growing workforce of genetic counselling as one of the key strategic initiatives for the period 2015-2017 with the aim to:

- define and promote best-practice models focused on high-quality and efficient delivery of genetic counselling
- identify existing and needed tools and technology to support the efficiency of genetic counsellors.

(Heald et al., 2016)

These two key areas are also critical for the Australian health system. Seeking direct feedback from stakeholders involved in the provision of genetic counselling will be essential, particularly when considering the limited available literature around best practice and workforce models that address the expanding work area of genetics and genomics. Given the variety of models in which genetic counselling is provided, and the fact that currently there are other medical and nursing professionals who undertake some form of genetic counselling or provision of genetic information, there is a need to clarify the qualifications for undertaking this role and for defining more clearly the place of genetic counselling within and across the health system.

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This report has been prepared with due care and diligence by Urbis and the statements and opinions given by Urbis in this report are given in good faith and in the reasonable belief that they are correct and not misleading, subject to the limitations above.

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