

Evidence Check

The NSW Genetic Counselling Workforce

An **Evidence Check** rapid review brokered by the Sax Institute for the NSW Ministry of Health.
May 2015.

This report was prepared by:

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NSW Ministry of Health. May 2015.

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List of abbreviations and definitions

Scope of practice	The procedures, actions and processes that a healthcare practitioner is permitted to undertake in keeping with the terms of their professional license
Genetic testing	Testing of one or more of the 20,000 or so genes known to directly cause a condition affecting growth, development or health or lead to susceptibility to a genetic condition
Genomic testing	Testing of all of the genes for a variant in the sequence
Whole Exome Sequencing	WES Testing all of the genes known to code for a protein
Whole Genome Sequencing	WGS Testing all of the DNA (not only the genes)
Next Generation Sequencing	NGS describes the raft of new DNA testing technologies
Human Genetics Society of Australasia	HGSA
Australasian Society of Genetic Counsellors	ASGC
Board of Censors	BOC
National Society of Genetic Counselors	NSGC
American Board of Genetic Counseling	ABGC
Accreditation Council for Genetic Counseling	ACGC
Activity Based Funding	ABF
MOH	Ministry of Health
GSAC	Genetics Services Advisory Committee

1 Executive summary

Introduction

The purpose of this Evidence Check review is to inform understanding of the current Genetic Counselling workforce in NSW and to provide an evidence-based resource for planning the composition and strategic development of the Genetic Counselling workforce into the future. It will contribute to development of a state-wide workforce plan, aligned with the NSW Health Professionals Workforce Plan 2012-2022.

This review aims to examine the evidence in both the peer-reviewed and grey literature regarding the:

- Current roles and scope of practice of genetic counsellors in the NSW Health Service, other Australian jurisdictions, and internationally
- Current training and certification pathways regarding genetic counsellors in NSW
- Challenges facing the NSW genetic counselling workforce now and in the future.

Genetic Counselling

Genetic counsellors are non-medical professionals working in the field of human genetics. The Human Genetics Society of Australasia (HGSA) defines genetic counselling as a "*communication process that 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*".

This process includes both medical practitioners (clinical geneticists and medical specialists with genetics expertise) and non-medical practitioners (genetic counsellors).

In Australia, genetic counsellors are professionals who have either been awarded HGSA certification (Certified Genetic Counsellor) or have completed the first part of their training and who may or may not, be undertaking certification (Associate Genetic Counsellor).

The process of genetic counselling integrates:

- 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.

Results

The report identified both peer-reviewed and grey literature. Grey literature included government and professional association reports, policy statements and discussion papers.

The majority of the evidence found was from North America, followed by Australia, the UK and Europe. The largest evidence source identified was expert opinion.

Findings

Current roles and scope of practice in the NSW, other Australian jurisdictions, and internationally

A search for relevant peer review literature found only one paper (James et al. 2003) related to current roles and scope of practice of genetic counsellors in NSW, a questionnaire study of 76 (34 in NSW) non-medical genetic health professionals. Therefore, this review has drawn upon policies and data from the HGSA and its Special Interest Group, the Australasian Society of Genetic Counsellors (ASGC).

Workplace setting and service configuration

The majority of NSW genetic counsellors work in the public sector. They work in a variety of clinical areas including general paediatric and adult genetics, and specialised areas such as cancer genetics and reproductive genetic counselling.

Clinical genetics services can be categorised as:

- Main Units within a teaching hospital with onsite clinical geneticists or non-clinical genetic specialist physicians e.g. cancer genetics specialists/cardiologists/endocrinologists
- Metropolitan outreach units where the genetic counsellor is the sole practitioner linked to a Main Unit with a Clinical Geneticist or medical specialist regularly attending clinics or providing consultation by telehealth
- Rural outreach units where the genetic counsellor is the sole practitioner usually based in a community health setting linked to a Main unit with a Clinical Geneticist or medical specialist regularly attending clinics or providing a consultation by telehealth.

These services are governed independently within eight NSW Local Health Districts (LHDs) and specialty networks.

James et al. reported that the majority (44%) of NSW genetic counsellors work in Main Units followed by rural outreach (23%) and metropolitan outreach (1%). It also reported that 27% worked in specialty services, specialising in the management of people affected by particular genetic conditions.

A 2012 census conducted by the ASGC found that the majority of Australian genetic counsellors (71%) work in a public hospital as part of a team while 21% work as a sole genetic counsellor linked to a main unit.

There is no published data regarding the current clinical settings of NSW genetic counsellors as a group. However, NSW Health's Centre for Genetics Education (CGE) lists NSW general genetic counselling services, family cancer services, and prenatal testing services.

Also listed are specialty services which provide carrier and predictive testing such as the Genetics of Learning Disability (GOLD) and familial cancer services where a genetic counsellor works in conjunction with a clinical geneticist.

Family Cancer Clinics also often operate with a cancer genetics specialist from a medical oncology and surgery unit. Genetic counsellors working in cancer genetics operate in parallel with medical clinicians, involving the latter in complex cases.

Two recent studies describe genetic counsellors working in conjunction with a cardiologist outside a clinical genetics unit in NSW.

Private practice

In NSW genetic counsellors in private practice work mainly in private ultrasound practices and IVF centres.

One genetic counsellor works in sole private practice in NSW. In 2015 a private company offering direct to consumer preconception and pharmacogenetic testing employed a genetic counsellor.

Client population

NSW genetic counsellors' client population includes adults and children, individuals and families. Data generated from the NSW Statewide Information System Database (Kintrak) shows that in the last four years:

- Services regarding predisposition for cancer were the most commonly provided services
- Client groups attended for consideration of genetic issues in the family and, where appropriate, regarding genetic testing in the settings of: prenatal; carrier screening; newborn screening follow-up; diagnostic; predictive; community genetic screening result reporting; and research.

Scope of practice

The scope of practice of NSW genetic counsellors includes direct patient care (including pre- and post-clinic contact), and administrative duties. Daily practice is largely dictated by service structure, most importantly the level of Clinical Geneticist and administrative support. Scope of practice is influenced by level of certification and experience. Those with Board Certification are qualified to work with more autonomy and less clinical supervision, manage more complex cases and exercise independent professional judgement.

In relation to direct patient care, genetic counsellors provide:

- Interpretation of family and medical histories to facilitate risk assessment of disease occurrence or recurrence in offspring
- Education about inheritance, testing, disease management and prevention, resources and research
- Counselling to aid informed decision making and psychosocial adaptation to the risk or condition.

In relation to pre- and post-clinic consultations, genetic counsellors perform tasks related to triage of referrals (including assessing appropriateness and urgency, allocation to clinical geneticist or genetic counsellor).

A genetic counsellor may conduct the consultation solely or in consultation with a clinical geneticist or other medical specialist depending on their level of experience and the complexity of the case. Tasks include but are not limited to:

- Pre-consultation contact including outlining the purpose of the consultation, identifying client needs and expectations, gathering pedigree information, identifying necessary documentation, providing emotional support
- Preparation for the consultation including retrieval and review of health records and literature, facilitating tumour testing if applicable, discussion with colleagues, preparing information for the client.

The consultation may be face-to-face or via phone or telehealth. It includes but is not limited to:

- Provision of information, including patterns of inheritance and natural history
- Risks to the client and/or other relatives
- Discussion of the medical, emotional and social implications for the individual and family
- Presenting options including genetic testing, reproductive options and aiding informed decision making
- Arranging and ordering appropriate genetic tests; depending on level of experience and service configuration.

Tasks in the post-consultation include:

- Return of results; depending on level of experience and service configuration
- Follow up to provide an opportunity to review information, answer questions and provide support.

Changing scope of practice

Certified and highly experienced genetic counsellors are increasingly conducting clinics without a clinical geneticist present. Where there is no clinical geneticist on site, telehealth is an increasing useful option. Genetic counsellors are also encouraged to pursue more telephone counselling and case management with the arrival of new cancer treatments and treatment-focused (rapid) genetic testing for cancer genes.

While there is no published data specific to NSW on the impact on scope of practice of new genetic and genomic technologies, the international literature emphasises this as a current and future challenge. The complexity of the genetic counselling process is increasing with new developments in technology making genetic and genomic testing cheaper and faster. The return of the results and addressing their personal familial impact is becoming a more time-consuming and difficult process.

Unmet needs and gaps to patients and genetic counsellors

Australian data regarding unmet client need is limited. International data outlines barriers to patients accessing genetic counselling including:

- Non-genetics health care providers' lack of awareness of patient risk factors
- Lack of obtaining adequate family history information
- Lack of knowledge of genetics and genetic conditions
- Lack of awareness of genetics services
- Inadequate coordination of referral
- Lack of genetic workforce.

There is no published data to support the anecdotal reporting of long waiting lists to attend genetic clinics following referral to a genetics service in NSW. There is also limited data on Indigenous people's access to genetic health services in Australia.

A number of barriers may result in reluctance to undertake Board Certification in NSW and lead to challenges to increasing the qualified workforce. These include limited availability of places in the Masters programs; limited availability of training supervisors; employment requirements for Board Certification and limited employment opportunities that meet requirements for Board Certification. Further to this, the NSW Award does not specifically recognize the attainment of certification.

Genetic counselling is not a registered health profession in Australia. There is currently no prohibition on any person offering genetic counselling services and no formal sanctions for breach of ethical or professional standards. This potentially places patients at risk of sub-standard care.

The Medicare Benefits Schedule does not cover genetic counselling by certified genetic counsellors but an increasing number of genetic tests are receiving an MBS number. This means more tests will become accessible outside the current model of service provision and patients may or may not be offered appropriate genetic counselling.

The ASGC census (2012) found a lack of diversity in the workforce. Two other studies have suggested that attracting more men to the profession may increase patient choice and satisfaction.

Scope of practice in other jurisdictions and internationally

Very little evidence was found about the roles and scope of practice of genetic counsellors in other jurisdictions. The evidence found did not suggest significant variation in the scope of practice of genetic counsellors across jurisdictions. Training and certification for the Australasian workforce occurs at a national level, therefore it is reasonable to expect that practice in the public hospital system will not vary widely in different jurisdictions. However, there are varying levels of organisation of genetics services in different jurisdictions, impacting on the work setting of genetic counsellors.

There is consistency between the Australian guidelines and those from the UK, Europe and North America indicating a similar scope of practice. Genetic counselling is a relatively new profession in South-East Asia and the Middle East.

Gaps informing future scope of practice for NSW

Registration of the profession, remuneration for services provided by genetic counsellors and service delivery models are major issues that represent gaps informing the future scope of practice for the NSW genetic counselling workforce.

A literature review looking at service delivery models in Europe, North America and Australia identified two complementary models: a multidisciplinary model with genetic and non-genetic specialists; and the other where genetics is integrated in primary care. However the latter model has not been implemented in NSW.

There is emerging evidence internationally supporting the benefit of genetics/genomics laboratory genetic counsellors to identify and clarify genetic tests, as well as interpretation of results and patient education. There is no evidence in the literature that these developments are occurring in NSW, although anecdotal data suggest that such opportunities are developing.

Current training and certification pathways for genetic counsellors in NSW

Genetic counsellors are certified by the HGSA Board. Currently, Board Certification is offered following completion of a two-year Masters degree accredited by the HGSA, attainment of Board Eligibility, and a minimum period of supervised employment. The HGSA offers only general certification; certification in specialty areas (e.g. oncology, prenatal) is not offered. Two levels of qualification are offered:

- Certified Genetic Counsellor: certified by the HGSA Board in genetic counselling. May use the title FHGSA (Fellow, HGSA Genetic Counselling).
- Associate Genetic Counsellor: determined by the HGSA Board to fulfil the eligibility requirements to undertake HGSA Board certification in genetic counselling. May use the title MHGSA (Member, HGSA Genetic Counselling).

To date 128 genetic counsellors have been awarded Board Certification and can use the title FHGSA.

The Masters program

The two-year clinical Masters program has been the only entry to the genetic counselling profession since 2011. The Masters degree is offered by two universities in Australia, The University of Sydney and The University of Melbourne. The Masters program is limited to 12-14 students per year.

Entry requirements are:

- An undergraduate degree in a related field
- Experience (voluntary or paid) in counselling and/or genetics
- Experience in a care role.

Both Masters programs comprise approximately one third each of:

- Coursework
 - scientific knowledge (genetic and genomic basis of health conditions and their clinical and psychosocial impact, current and developing genetic testing regimes and requirements, associated ethical, legal and social issues)
 - counselling skills
 - genetic counselling specific theories and models and research methodologies
- Clinical practice (min. 15 weeks in a range of supervised clinical placements, formal assessments of skills and competencies, formative assessments mirroring the requirements for certification)
- A practice-based research project and dissertation.

Board eligibility

Graduates of the Masters program who are financial members of the HGSA may apply for Board eligibility following 12 months employment at a minimum 0.4 FTE in a genetic counselling role. Those who completed a one-year Graduate Diploma or Masters prior to 2015 may also apply for Board Eligibility.

Board certification

Once Board eligibility has been accepted, candidates commit to training for HGSA Board Certification that takes a minimum of 2 years FTE employment under supervision. Assessment is based on competencies and skills demonstrated by submission of a portfolio as well as other tasks comprising:

- Five long cases
- Logbook of 50 cases
- Published article, or alternatively a literature review
- Two reflective essays (transcript analyses)
- Supervisor reports
- Interview with the Board.

Maintenance of Professional Standards

The HGSA administers a voluntary maintenance of professional standards program (MOPS) for Board Certified practicing genetic counsellors.

Cross training following removal of cancer genetic counselling as a sub-specialty

Cancer genetic counselling as a sub-specialty was removed by the HGSA in 2011. Those with cancer genetic counselling certification must cross train to achieve general certification and the HGSA has produced guidelines covering the cross training requirements.

Challenges facing the NSW genetic counselling workforce now and into the future

Evidence in the literature relating to challenges to current and future roles and practice is largely provided from North America. However, given that many of these challenges are generated by new genetic and genomic technologies similarly used in Australia, it is apparent that they may also be relevant to the NSW genetic counselling workforce. Key challenges identified for the workforce now and into the future include:

- The impact of new genomic technologies on genetic counselling practice
- A changing clinical paradigm
- Challenges related to building the workforce
- Increased awareness of genetic and genomic testing leading to increased demand
- Training and certification
- Challenges related to key partners.

The impact of new technologies on genetic counselling practice

The most widely reported challenges relate to new developments in technology known as next generation DNA testing. New technologies are making genetic and genomic testing more widely available, cheaper and faster. In turn, this is making the genetic counselling process more complex due to:

- More available tests and subsequent number of diagnoses
- Complex result analysis and interpretation
- Results of uncertain significance
- Incidental findings.

In particular, there is a need for careful pre-test counselling for unexpected findings. Researchers from the Centenary Institute note that the basic principles of pre-test counselling essentially remain unchanged however the inherent uncertainty of the gene result must be conveyed to the patient.

In NSW, the literature also notes the lack of an appropriate database of known human genotype-phenotype associations with clinical decision making currently relying on the knowledge of individual practitioners and genetic counsellors. In addition to this, the broader literature notes a lack of guidelines and protocols around the genomic testing process and a need to manage the enormous amounts of electronic data generated.

A changing clinical paradigm to meet increasing demand

Mainstreaming of genetic and genomic tests

Introduction of next generation DNA testing into mainstream medicine is changing the clinical genetics paradigm. The literature notes that:

- As the cost of testing continues to drop there will be increasing demand on those able to interpret results in the clinical context and make appropriate management recommendations
- Genetic counsellors will play a key role in multidisciplinary teams and primary health care networks as educators and interpreters of results
- There will be increased pressure on the workforce / increased workload.

While the employment of laboratory-based genetic counsellors is a model to overcome the complex nature and volume of reported results, there is an increased need for genetic counsellors to be employed in the NSW Health Service to ensure that patients and the broader NSW population can reap the benefits from the changing clinical paradigms in the genomic era.

Impact in clinical genetics services and increased demand in specialty areas

As the referral rate continues to rise in cancer genetics, the current workload is unsustainable. Innovative solutions to support service delivery are required. In general genetics, there is an increase in paediatric cancer referrals, with the identification of new syndromes. Challenges include education of paediatric staff to recognise cases for referral, cross credentialing of genetic counsellors to work across LHDs, activity-based funding barriers where funding does not return to the site providing the service, and availability of outpatients' rooms.

There is likely to be a similar need in other specialty areas such as cardiac and neurological genetics, as these services grow with the identification of more genes.

Challenges specific to genetic counselling as a small but critical health professional workforce **Challenges to building the workforce**

There are a number of challenges to building the genetic counselling workforce.

- Limited places in the Masters programs
- Limited clinical placement opportunities and availability of senior supervisors for these (required as part of the Masters)
- Limited employment opportunities which meet the requirements for Board Certification
- Employment requirements for starting and obtaining certification
- Limited numbers of certified genetic counsellors who can provide supervision for training
- The NSW Health Service Health Professionals (State) Award does not include recognition for attainment of Board Certification.

Diversity and gender balance

To respond to the needs of the whole population diversity of health professionals is important. According to the ASGC census ethnic and racial minorities are under-represented in the genetic counselling profession. The census also found that women made up 95% of its membership. Two US studies found in the literature note that attracting more men to the profession may increase patient choice and satisfaction.

Increased demand on genetics services and counselling due to increased awareness of genetic testing

Increased demand on genetics services due to increased awareness has resulted from:

- An increase in the number of tests available and awareness of these
- The time needed for interpretation, analysis and follow up
- An increase in the volume of referrals (both appropriate and inappropriate)
- Lack of health professionals' knowledge in this area to manage community concern.

This continues to highlight the critical role played by genetic counsellors in meeting patient and service needs and inform priorities for future workforce planning.

With increasing provision of population genetic screening, through both public and private sector modalities, the demand for genetic counsellors to support individuals and families faced with decisions emanating from the results of the tests will inevitably increase.

Traditional models of genetic counselling provision will not meet the needs of new paradigms of care

The scope of practice of genetic counsellors has traditionally included delivery and explanation of results. However, with the advent of new genetic testing technologies, it is proposed that the future scope of practice will include:

- Involvement in the interpretation of genetic variants as part of a multidisciplinary team
- Working as laboratory-based genetic counsellors
- Working in a proactive risk-reduction clinic role in the public health arena.

Challenges/changes related to key partners

Challenges for key partners, such as clinical geneticists, are similar in the workplace to genetic counsellors in the analysis, interpretation and management of genomic results and their limitations and the impact this will have on services.

Continuing education needs of genetic counsellors and genetic counsellors as educators of non-genetics-trained health professionals

Genetic counsellors have a large future role in assisting the translation of genomic information into mainstream medicine through the education of non-genetics health professionals.

Urgent attention, however, is needed to upskill the genetic counselling workforce in the genomic era, as this technology and its interpretation is not part of current training. Given that interdisciplinary teams are becoming increasingly commonplace in Australian healthcare, there is also a need for interdisciplinary education for genetic counsellors as a means of improving patient care.

Specialty genetics areas (cancer, cardiac, neurology) are becoming integrated into primary care. As part of this integration, time is needed to educate traditional specialists in the interpretation of genetic information.

Challenges resulting from changes in technology, science, policy, costs and service delivery models, and regulatory frameworks

The impact of new technologies on genetic counselling practice and a changing clinical paradigm is described above.

Existing models of consent need to evolve

The duration and content of the consent process will need to evolve to accommodate:

- The shift from single-gene testing to multiple-gene testing
- Limitations of genomic testing
- Increased generation of results of uncertain significance
- Integration into mainstream medicine
- Increase in population-screening-related tests, where time for pre-test counselling may not be prioritised.

Registration of the profession

Genetic counselling is not a registered health profession in Australia. The profession continues to seek licensure with the Australian Health Practitioner Regulation Agency (AHPRA) but has been unsuccessful to date.

The World Health Organization notes that in Australia there are no specific provisions in any state to prohibit any person, regardless of qualifications, claiming to be a genetic counsellor, or providing genetic counselling. There are also no formal sanctions in place for breach of ethical or professional standards in genetic counselling. There is a need to protect the public from substandard genetic counselling services and, in particular, from persons who are not appropriately trained, qualified and supervised.

The Australian Government established an inquiry into the ethical issues surrounding the protection of human genetic information, which was led by the Australian Law Reform Commission (ALRC) and the

Australian Health Ethics Committee (AHEC) of the NHMRC. The 2003 report, 'Essentially Yours' (Recommendation 23–2) stated "The Commonwealth, States and Territories should examine options for the further development of genetic counselling as a recognised health profession, including the use of certification, accreditation or registration systems for genetic counsellors".

As genetic testing becomes cheaper and more tests receive a Medicare Benefits Schedule (MBS) number, greater interest will emerge from private sector pathology laboratories. This may put the patient at greater risk of sub-standard genetic counselling care.

Costs

The MBS does not cover genetic counselling by certified genetic counsellors and levels of practice do not influence funding to units. With increasing likelihood that genetic counsellors will practice outside of genetics clinics, there are major financial implications to specialty units and the genetic counsellors themselves. In addition, the cost of increasing education for non-genetics health professionals by genetic counsellors is currently being investigated by the clinical genetics departments in the LHDs.

Service delivery models

It is not feasible to set up a clinical genetics department in every LHD or within multiple hospitals in every LHD. However, the literature suggests that with the use of videoconferencing proving to be an acceptable and safe way to deliver telemedicine it is likely more genetic counsellors will use this as a service modality in the future. Nevertheless, there are issues with costs being attributed to where the patient is seen and not back to the site where the service is being delivered.

Need for national harmonisation in policy governing delivery of genetic counselling in the genomic era

Families don't always respect state boundaries in their need for care and it is essential that the information and service they receive is of the same standard and governed by the same protocols. NSW Health's Genetics Service Advisory committee (GSAC) has been in place since 1989, and there are a number of other state-based organisations addressing their population's needs, but it will be essential to have harmonisation. The HGSA, as the main professional organisation of genetic specialists in Australia, encompasses several special interest groups (SIGs), including the ASGC and needs to be part of any policy development by state or federal governments. The HGSA has not published guidelines or policy statements pertaining to counselling in the genomic era. In Australia, as of 2009, a fully coordinated national approach to public health genomics did not exist.

Conclusion

Much of the evidence of the numerous current and future challenges facing the genetic counselling workforce documented in this review is international. It is clear that the developments over the past 10 years in genetic and genomic technologies are driving changes in international clinical practice. It is expected that NSW genetic counsellors are, and will be, facing similar challenges.

The evidence has shown that the current Australian system of training and certification for genetic counsellors is of high academic rigor. It is workplace competencies based and assessed, and is internationally benchmarked. However, this review has also documented many barriers and limitations to growing a fully qualified workforce. These limitations include: the small cohorts of Masters graduates generated annually, the requirement to be employed in a setting that can meet the HGSA guidelines to undertake the full certification, and limited employment opportunities.

While employment settings are currently largely in the public sector, private-sector employment opportunities could enable growth of the Australian genetic counsellor workforce. However, these opportunities are limited by the lack of registration of the profession and associated remuneration issues.

The current workforce is small but of increasing importance in ethically delivering the benefits of faster and cheaper genetic and genomic tests to the NSW population. The workforce provides appropriate support to enable informed decision making and adaptation to the impact of the familial results. It is essential for workforce planning to not only investigate the current roles and scope of practice of the NSW genetic counselling workforce in the era of genomics and how the many challenges are being addressed, but also to plan for and support their training, wider roles and scope of practice.

2 Introduction

Genetic counselling

The definition of genetic counselling as used in Australia is that it is a communication process that 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; Process of Genetic Counselling).

This definition is based on recommendations from the US's National Society of Genetic Counselors (NSGC) Task Force (2006) following a peer-review process with input from the NSGC membership, genetic professional organisations, the NSGC legal counsel, and leaders of several national genetic advocacy groups.⁵

The non-medical professionals who implement this process are genetic counsellors, whose role has been recognised and formalised in the US since the early 1970s.

Developments in Australia

Up until the mid-1980s no formal training program or certification process was in place for health professionals working in genetic counselling roles in different Australian settings.

It was not until 1986, when a number of clinical geneticists, who had trained in the US and worked with graduates of the US genetic counselling programs, returned to Australia and proposed to the Human Genetics Society of Australasia (HGSA) that training of genetic counselors was necessary in Australia to support the needs of the families dealing with the increasing ability to diagnose the genetic basis of health conditions they were experiencing.

Kenen⁶ describes the development of the genetic counselling profession in Australia as occurring in three stages: the emergent, consolidating and institutionalised stages.

She notes that the 'emergent phase' of the profession was relatively brief.⁷ From the 1970s, clinical genetics units in public paediatric hospitals were established within the State Government-funded public healthcare system in New South Wales, South Australia, and Victoria. A multidisciplinary team staffed the units, with a clinical geneticist in charge and professionals including nurses, social workers and laboratory scientists working within them. These professionals offered diagnosis, information, counselling, research and support.

The 'consolidation phase' occurred in the mid-to-late 1980s, with recognition by state health departments such as NSW Health in reviewing and funding genetics services and creating employment opportunities for genetic counsellors.^{8,9}

At the same time, the Human Genetics Society of Australasia (HGSA) recognised the need for a health professional dedicated to working with individuals and families with genetic conditions, and therefore developed training guidelines and a certification process for a new professional group of genetic counsellors. The first Australian genetic counsellor received HGSA certification in 1991. This began the

'institutionalised phase', during which the Australasian Society of Genetic Counsellors (ASGC) was formed in 1993 and incorporated in 2002 as a Special Interest Group of the HGSA.¹⁰ The group generated policies and codes of practice as well as providing governance of training and certification of genetic counsellors.

Edwards et al¹¹ noted that the rapid understanding of the genetic basis of many health conditions and the increasing ability to predictively identify inherited risk for many complex conditions, combined with the impact of such genetic conditions on individuals and families – particularly in ethical, legal and psychosocial arenas – required specially trained professionals to work in this unique and growing dimension of healthcare.

This demand, as predicted in the 2003 federal report 'Essentially Yours: The Protection of Human Genetic Information in Australia', noted that *"as genetic medicine and testing technology develop, there will be an inevitable increase in the need for genetic counselling services in Australia. Strategies should be developed now to assess and respond to this increased need"*.³ This is a global issue and the Transnational Alliance for Genetic Counseling (TAGC) represents 15 countries currently providing genetic counsellor education across five continents.¹¹

The familial aspect of genetic testing means that it is traditionally preceded by counselling to discuss its advantages and disadvantages with individuals, so they can make informed decisions. New genetic testing technologies that enable faster and cheaper genetic testing and broader examination of a person's DNA, generate similar ethical challenges but with additional factors of management of enormous amounts of data with current limitations on interpretation, increased generation of uncertainty and implications for informed consent regarding unsought or unwanted results.^{12, 13}

This review will explore current practices, the development of the profession in Australia, current and future challenges, and describe the evidence for the workforce needs of this small but increasingly important professional group.

3 The research questions

Question 1: What are the current roles of genetic counsellors in the NSW Health Service?

- Describe roles performed in both the public and private sectors
- Include information on the service configurations where these roles are undertaken
- Provide information about the client populations served by these roles
- Include information about the scope of practice of genetic counsellors in NSW
- Identify any existing gaps or unmet client needs related to the current scope of roles.

Question 2: What are the current training and certification pathways for genetic counsellors in NSW?

- Include information about entry professions
- Provide information about relevant regulatory frameworks
- Make comment on the content of training courses, in particular the relative balance between scientific knowledge and counselling.

Question 3: What does the evidence tell us about the scope of practice of genetic counsellors in other Australian jurisdictions or internationally?

- Describe the scope of practice in these jurisdictions
- Identify any gaps that exist between the scope of practice in NSW compared with other jurisdictions that would be important for the future/workforce planning.

Question 4: What are the challenges facing the NSW genetic counselling workforce now and into the future?

- Identify challenges specific to the field of genetic counselling, also the broader relevant challenges facing small but critical health professional workforces
- Detail the challenges/changes related to key partners, such as specialist medical practitioners
- Include new and emerging challenges, as well as current and ongoing challenges that relate to this area
- Comment on the challenges resulting from changes in technology, science, policy, costs and service delivery models, and regulatory frameworks.

4 Methodology

The search strategies were specific for each question. The terms used and the sources searched for each question are detailed in Table 1. Studies and literature were evaluated according to inclusion (English and full text) and exclusion criteria (non-developing countries). Both peer-reviewed and grey literature was searched.

The terms cited in Table 1 were used individually or combined to search the following databases to identify relevant peer-reviewed literature: Embase; Scopus; PubMed; PsycINFO; Medline via Ovid; and Google Scholar. These databases were chosen for their ease of access and presentation of literature relevant to the proposed review.

The titles of all papers were perused and the abstracts of those deemed relevant were read. The reference lists of all selected articles were also scanned for articles of related interest. For Question 2, searches covered the period 1990–present. For Questions 1, 3, and 4, original research papers published in the past 15 years were of greatest overall preference due to the impact of technology change generated by the completion of the Human Genome Project in 2003 on genetic counselling practice and workforce demands, but older articles and policy documents from governing bodies were also included (due to necessary information provided).

The following strategies were also used to identify reports and policies:

- Focused search on Google Scholar (scholar.google.com.au) during August 2015
- Correspondence with experts in the field with reference to genetic services and genetic counselling reports
- Analysis of policies and statements of professional societies and institutions known to the authors
- Analysis of Australian state and commonwealth reports.

Table 1. Search terms in the peer-reviewed and grey literature

	Terms	Sources
Q1	Individually or combined: genetic counselor/counsellor, genetic counseling/counselling, role, task, scope, responsibilities, workforce, challenges, barriers, service(s), New South Wales, Australia(n) and Australasia	Embase; Pubmed; PsycINFO; Google Scholar
Q2	Individually or combined: genetic counselor/counsellor, genetic counseling/counselling, education, training, certification, genomics, Australia Australasia; USA; UK	PubMed; Medline via Ovid; Scopus PubMed; Google Scholar
Q3	Legislation and jurisprudence, OR manpower, OR organization and administration, OR standards	Medline via Ovid
Q4	Individually or combined: genetic counseling Australia models OR access "service delivery"; informed consent; whole genome sequencing; whole exome sequencing; incidental findings; ethical issues; clinical setting; workforce, challenges, barriers, service(s), New South Wales, Australia(n) and Australasia	Medline via Ovid; PubMed; Google Scholar

5 Quality of the evidence

There were 146 sources identified: 114 articles in the peer-reviewed literature, and 25 reports, 5 policy statements and 2 discussion papers from the grey literature.

Table 2 provides an overview of the categories of papers and documents in the peer-reviewed literature. In most of the reported studies, the samples were genetic counsellors or other genetic clinicians.

While 35% of sources were from Australasia (including one from New Zealand), a large proportion of the published data is international: 47% from North America; 9% from the UK and 6% from Europe.

Many of the themes are significant, as models of genetic counselling practice in the UK and Europe reflect current practice in Australia. While there are similarities in genetic counselling training in the US, there is wide variation in models of genetic counselling practice providing insight in future challenges.

Questions 1 and 3 (Appendix 1)

Appendix 1 includes the 16 of 34 studies or reviews that generated evidence. Of the 16 studies, 8 were from Australia, 5 were from North America, and 4 from the UK/Europe. The evidence from 6 of the 34 of the studies was considered high quality.

Question 2 (Appendix 2)

Appendix 2 includes the 7 of 8 studies or reviews that generated evidence. Appropriately, 4 of the 7 studies were from Australia. The evidence from all was considered high quality.

Question 4 (Appendix 3)

Appendix 3 includes the 25 of 72 studies or reviews that generated evidence. Of the 25 studies, only 4 were from Australia; 16 were from North America, and 5 from the UK/Europe. The evidence from 16 of the 25 of the studies was considered high quality.

Table 2. Categories of sources identified for each research question

	Overall %	Questions 1 and 3 (n=34)	Question 2 (n=8)	Question 4 (n=72)
Expert opinion	28	3	3	31
Audit and evaluation	12	7	-	8
Reviews	11	4	2	10
Reports	11	4	1	7
Quantitative studies	10	10	1	5
Qualitative studies	9	5	-	8
Commentary	2	1	1	1
Policy statements	2	-	-	2

6 Results

Question 1: What are the current roles of genetic counsellors in the NSW Health Service?

Roles and scope of practice in the NSW public sector

Genetic counsellors in NSW work in a variety of clinical areas, including general paediatric and adult genetics, and specialised areas including cancer genetics and reproductive genetic counselling.

The majority of NSW genetic counsellors work in the public sector, either in metropolitan units or in outreach settings.

The scope of practice of NSW genetic counsellors mainly involves duties related to direct patient care, including pre- and post-clinic contact, as well as administrative duties.

General scope of practice of genetic counsellors

The scope of practice of genetic counsellors as described by the HGSA⁴ includes:

- Triage of referrals
- Assessment of appropriateness and urgency of referrals, and allocation to clinical geneticist or genetic counsellor
- Pre-consultation contact; including outlining the purpose of the consultation, identifying the needs and expectations of the client, gathering pedigree information, identifying necessary documentation and providing emotional support
- Preparation for consultation; including retrieval and review of relevant health records and literature
- Facilitating tumour testing
- Discussion with colleagues as necessary
- Preparing information to provide to the client.

The genetic counselling consultation

The counselling consultation includes but is not limited to:

- Provision of information, including patterns of inheritance and natural history
- Risks to the client and/or other relatives
- Discussion of the medical, emotional and social implications for the individual and family
- Presentation of options including genetic testing and reproductive options, and assisting with informed decision making
- Arrangement of genetic tests.

The role of conveying test results may be delegated to a genetic counsellor depending on the level of experience of the counsellor and the service configuration.

Additionally, follow up provides an opportunity to review information, an opportunity for questions and enables provision of support.

Data on NSW genetic counsellors

The only data identified in the literature that delineates the role of NSW genetic counsellors in the public sector is that by James et al¹ in a questionnaire study of 76 non-medical genetic health professionals in Australasia who described themselves as genetic counsellors.

Thirty-four of the respondents were located in NSW. While the specific roles of NSW respondents were not isolated from the whole data set, the range of roles identified mirrors the scope described by the HGSA above.

Daily practice for genetic counsellors in NSW genetic services is largely dictated by the structure of the genetic counselling service, most importantly the level of clinical geneticist and administrative support.

Direct patient care role

- Interpretation of family and medical histories to facilitate risk assessment of disease occurrence or recurrence in offspring
- Education about inheritance, testing, management and prevention of disease, resources and research
- Counselling to facilitate informed decision making and psychosocial adaptation to the risk or the condition
- Depending on the setting, genetic counsellors may have ordered tests and returned genetic test results.

Non-patient care roles

- Extensive administration tasks such as filing and data entry.

Clinical settings

The majority of NSW genetic counsellors worked in one of three types of clinical settings in the public hospital system with the majority in main units (Table 3):

- Main units within a teaching hospital with clinical geneticists or non-clinical genetic specialist physicians (e.g. cancer genetics specialists/cardiologists/endocrinologists) on site
- Metropolitan outreach units where the genetic counsellor is the sole practitioner based in a city but is linked to a main unit with a clinical geneticist or medical specialist regularly attending clinics or providing a consultation by telehealth
- Rural outreach units where the genetic counsellor is the sole practitioner usually based in a community health setting but is linked to a main unit with a clinical geneticist or medical specialist regularly attending clinics or providing a consultation by telehealth.

Table 3. Data from James et al (2003) regarding workplace setting of NSW genetic counsellors

Setting	% (n=34)
Main Unit	44
Metropolitan Outreach	1
Rural Outreach	23

Specialty areas

James et al¹ also reported that 27% of genetic counsellors worked in speciality services, which are services that specialise in the management of people affected by particular genetic conditions.

These services provide carrier and predictive testing; such as the Genetics of Learning Disability Service (GOLD) and familial cancer services where a genetic counsellor works in conjunction with a clinical geneticist.

Family Cancer Clinics also often operate with a cancer genetics specialist coming from medical oncology and surgery. Genetic counsellors working in cancer genetics work in parallel with medical clinicians, involving the latter in complex cases.

Two more recent studies by Ingles et al^{14, 15} describe genetic counsellors working in conjunction with a non-genetics specialist (cardiologist) outside of a clinical genetics unit in Sydney NSW but the focus was on the health economic benefits of genetic testing in a specialised service rather than delineating the role that the genetic counsellor played.

When a clinical geneticist is not present on site, telehealth is an increasing useful option. Telehealth provision has been set up at several sites and this model of care has been reviewed favourably by patients¹⁶ and genetic counsellors.^{17, 18}

Genetic counsellors are also encouraged to pursue more telephone counselling and case work up with the arrival of new cancer treatments and treatment-focused (rapid) genetic testing for cancer genes.¹⁹

Guidelines for testing for genetic disorders

NSW Health's policy directive on genetic testing²⁰ outlines guidelines for genetic testing and sets out the Ministry of Health's requirements for testing for genetic disorders. It particularly addresses counselling issues (and laboratory requirements associated with genetic testing) but does not specify that genetic counsellors carry out the counselling.

The scope of clinical genetic testing includes:

- Newborn screening
- Diagnostic and carrier testing for inherited disorders
- Predictive and pre-symptomatic testing for adult onset disorders
- And more recently, pharmacogenetic testing to guide individual drug management.^{21, 22}

Professional genetic counselling is considered an essential element of Level 2 genetic testing²³ although this does not specify the professional group providing such genetic counselling.

Service configuration

There is no published data regarding the current clinical settings of NSW genetic counsellors as a group. However NSW Health’s Centre for Genetics Education (CGE) website (www.genetics.edu.au) lists NSW general genetic counselling services, family cancer services, prenatal testing services and speciality services such as cardiac genetics services.

Public sector genetics services

NSW public sector genetics services are governed independently within eight local health districts (LHD) and specialty networks²⁴ (see Figure 1).

Genetic services are placed within LHDs and Specialty Health Networks servicing large populations.

Designation of genetic services

The Health Workforce Productivity Commission²⁵ has recommended that services be designated according to staffing levels.

- Level 1 units do not have an on-site genetic counsellor, however staff are able to arrange on-site clinics with clinical geneticists/genetic counsellors
- Level 2 units have services provided by a genetic counsellor with a visiting clinical geneticist
- Level 3 services include clinical genetics services provided by a less-than-full-time clinical geneticist
- Level 4 and 5 services operate as separate units providing outreach referral services with full-time clinical geneticist activity (level 4) and may provide statewide or national expertise in a specific disorder or disorders (level 5).

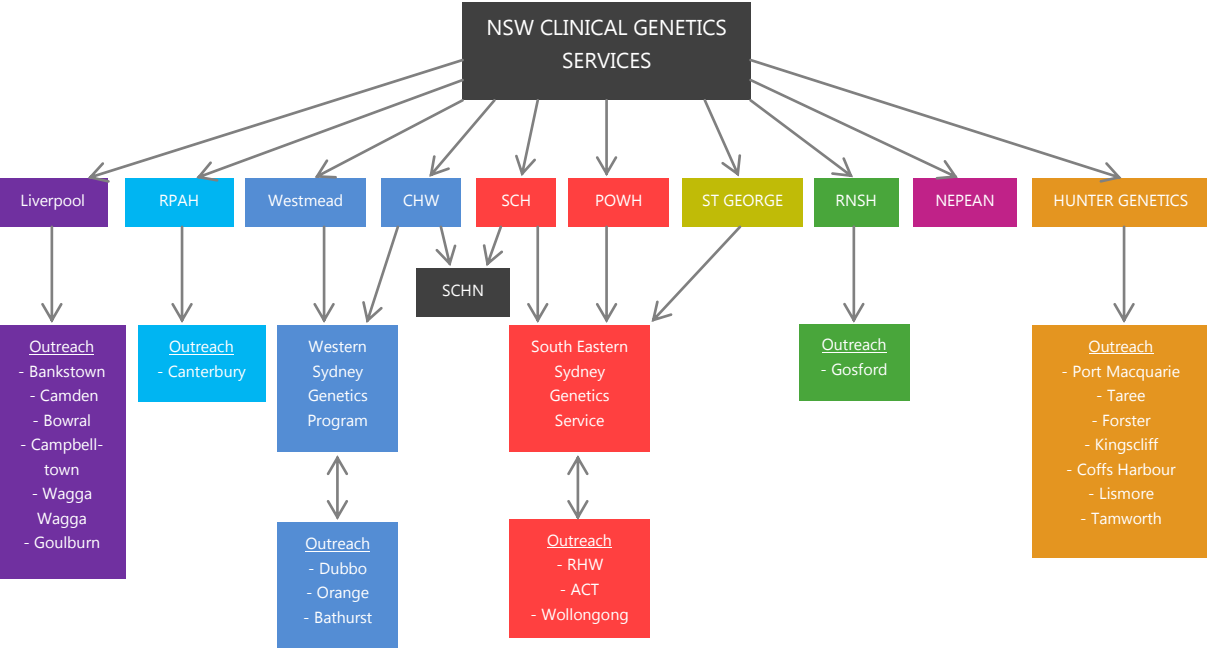


Figure 1. NSW Clinical Genetics Services in NSW Health

Source: Sydney Children’s Hospitals Network Clinical Services Plan 2012–2016)
 RPAH = Royal Prince Alfred Hospital, CHW = Children’s Hospital Westmead, SCH = Sydney Children’s Hospital,
 SCHN = Sydney Children’s Hospitals Network, POWH = Prince of Wales Hospital, RNSH = Royal North Shore Hospital,
 RHW = Royal Hospital for Women, ACT = Australian Capital Territory

Client populations

NSW genetic counsellors' client population includes adults and children. Main services are cancer and general genetics.

Figure 2 provides a graphical representation of adult and children populations seen in general genetics and cancer genetics services across the state, although comparison of actual numbers between cancer and general (adult and children combined) need to be evaluated with caution as data entry may differ across the populations.

The data generated from the NSW statewide information system database (KinTrak) by the system administrator (B Culling, Application Support Specialist, NSW Health's Genetic Information System, personal communication) shows that in the past four years:

- Services regarding inherited predisposition for cancer were the most commonly provided
- Client groups attended for genetic counselling for consideration of genetic issues in the family and, where appropriate, regarding genetic testing in the settings of
 - Prenatal
 - Carrier screening
 - Newborn screening follow-up
 - Diagnostic
 - Predictive
 - Community genetic screening result reporting
 - Research.

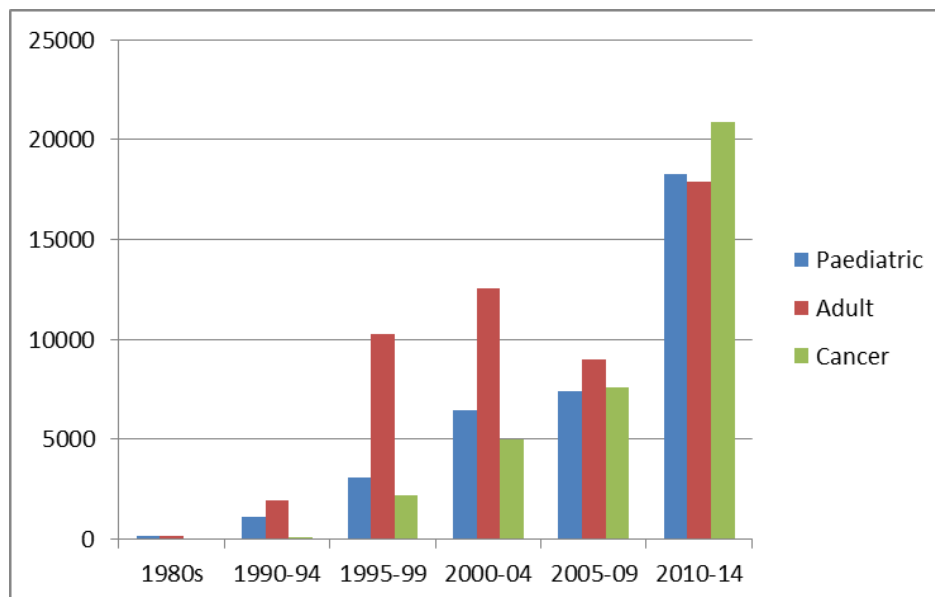


Figure 2. Client population groups in NSW

The three client groups represent patients attending general and cancer genetics services. The general genetic clinic patients include adult and paediatric populations. Source of information: KinTrak, the NSW genetic information system database; B Culling, personal communication.

Genetic counsellors in the private sector in NSW

NSW genetic counsellors in private practice work mainly in private ultrasound practices and IVF centres. Currently, there is no published service directory for NSW private genetic counsellors. The NSW Centre for Genetics Education has recently surveyed the NSW clinical genetics membership for information regarding clinical geneticists and genetic counsellors working in private practice, and found that NSW private genetic counsellors mainly exist within private ultrasound practices and private IVF facilities (K Dunlop, Director CGE, personal communication).

There is only one NSW genetic counsellor in sole private practice, specialising in pre-pregnancy, prenatal and adult genetic counselling and cancer family history risk assessment.

Most recently in 2015, Life Letters, a private Australian genetic company, offering direct to consumer preconception and pharmacogenetic testing to the public, has also employed a genetic counsellor. Private clinical geneticists operate within NSW, without the assistance of genetic counsellors.

An online survey of members of the ASGC explored the perceived interest in development of private genetic counselling services in collaboration with primary care physicians, a model that has yet to be realised.²⁶

Unmet needs and gaps to patients and genetic counsellors

The literature outlines barriers to patients accessing genetic counselling services internationally; however there is limited Australian data.

The NSW genetic counselling workforce faces many barriers that include clear delineation of the diversity of roles and scope of practice required to meet emerging patient care needs within various models of care. There is ambiguity between the essential skills required to meet service needs and associated remuneration as per the current award.

Barriers to patients accessing genetic counselling services

The roles and scope of practice underscore the need for high standards and practitioners with appropriate training and qualifications, and are described in response to Research Question 2.

The systematic review conducted by Delikurt et al²⁷ identifies barriers that all contribute to patients' not accessing genetics services. These include:

- Non-genetics health-care providers' lack of awareness of patient risk factors
- Lack of obtaining adequate family history information
- Lack of knowledge of genetics and genetic conditions
- Lack of awareness of genetics services
- Inadequate coordination of referral and lack of genetic workforce.

There is no published data to support the anecdotal reporting of long waiting lists to attend genetic clinics following receipt of referral to a genetics service in NSW.

It is recognised that Indigenous Australians have lower levels of healthcare access than non-Indigenous Australians.²⁸ There is limited specific data on Indigenous people's access to genetic health services in Australia.²⁹

Barriers for the NSW genetic counselling workforce achieving the qualifications needed for the developing scope of practice

Achievement of a full professional qualification (Board Certification) means that genetic counsellors are qualified to work with more autonomy and less clinical supervision, manage more complex cases and exercise independent professional judgment. In addition to new clinical duties, certified genetic counsellors may supervise associate genetic counsellors.

Following Board Certification, genetic counsellors are also expected to expand their scope of practice in keeping with their experience, skills and knowledge by participating in the MOPS program of the HGSA.

In NSW, most Board Certified genetic counsellors practice at level 3–4 of the NSW Health Service Health Professionals (State) Award³⁰, although no recognition of that certification or additional training that enables skills required of the role is provided in the award. By comparison, genetic counsellors in Victoria are employed under an award that explicitly recognises and rewards certification.³¹

These barriers in NSW may result in reluctance to undertake the Board Certification in NSW, creating a workforce limited in the skills required to work at higher levels, or meet patient and family needs emanating from applications of new genetic and genomic technologies.

There is no Medicare item number for a clinical service provided by a genetic counsellor, and all services are charged according to the supervising clinical geneticist or other medical specialist the referral is made to. While a number of private genetic counselling services exists in Australia (list available from the Centre for Genetics Education) this service provision is limited by the lack of Medicare rebate and coverage by major private health funds.

Question 3: What does the evidence tell us about the scope of practice of genetic counsellors in other Australian jurisdictions or internationally?

Other Australian jurisdictions

There is limited evidence about the genetic counsellors' scope of practice in Australia.

It is expected that core practice in public hospitals will not differ widely by jurisdiction but by the structure of the service they work in.

The ASGC census in 2012 identified the majority of genetic counsellors work in public hospitals as part of a team, have been employed as a genetic counsellor for 5–9 years, and that NSW has one third of the genetic counselling workforce in Australia.

Certified genetic counsellors and those who are very experienced are increasingly conducting clinics without a clinical geneticist present.

The only two publications identified from the literature that examined the roles of Australasian genetic counsellors are the questionnaire study by James et al¹ and the evaluation of genetics services provided in Queensland between January 1998 and December 1999 by Kromberg et al.³² There is no evidence from these studies to suggest that the scope of practice of genetic counsellors across Australasia varies significantly.

Given that training for the Australasian workforce predominantly occurs at only two centres and certification occurs at a national level, it is reasonable to expect that practice in genetics services based in the public hospital system will not vary widely in different jurisdictions.

It is clear from both these studies that the scope of practice of genetic counsellors throughout Australian Genetics Services tends to be determined by the structure of the service to which they belong, including access to supervision by clinical geneticists and administrative support, as well as the experience of the genetic counsellor. Most Australian public sector general genetic counsellors work in partnership with clinical geneticists and in specialised clinical genetics services that are typically located in large hospitals.³³

Structure of Australian genetics services

As in NSW, Australian clinical genetics services can be categorised as Main Units (within a teaching hospital with medical geneticists on site), Metropolitan outreach (counsellors based in cities without a geneticists on site) or Rural/Remote outreach (where a genetic counsellor is a sole practitioner usually in a community health setting).³² However, there are varying levels of centralisation of genetics services provided in different states and territories of Australia.

The states with small and centralised populations – Western Australia and South Australia – each have one central service in the capital city, from which they also provide outreach services to the rest of the state via telehealth and/or fly-in- fly-out clinics (Government of Western Australia, Genetics Services; Government of South Australia, Genetics Services).

The South Australian service is also centralised at the women’s and children’s hospital but is separated into three discrete units: reproductive and paediatric, metabolic and adult.

Queensland has a primary service (Genetic Health Queensland) with satellite services elsewhere in the state.

The ACT, Tasmania (Tasmanian Government) and Northern Territory have services staffed by local genetic counsellors who receive clinical geneticist support on a fly-in- fly-out basis. Services in Victoria were decentralised in 2009 and there are now a number of metropolitan and regional services across the state (Victorian Clinical Genetics Service).

Each of these models dictates different processes for genetic counselling service delivery. However the core day-to day duties of assessment, education, communication and support are consistent across services throughout Australia.

ASGC Professional Status Survey

Other data regarding roles of genetic counsellors have been provided by the Australasian Society of Genetic Counsellors (ASGC), which was established in 2002 as a special interest group of the HGSA. The ASGC represents those providing genetic counselling or members of the HGSA working in areas related to genetic counselling.

In 2012, an ASGC census was conducted of its 270 members and included information regarding the employment of Australian genetic counsellors. The data did not identify specific information about NSW but revealed Australian genetic counselling jobs are equally located in NSW and Victoria (34% each) while 14% are in Queensland. The rest of the Australian jobs are spread across other states and territories. The workplace setting is described in Table 4.

Most commonly, Australian genetic counsellors work as part of a team (71%), while 21% of genetic counsellors work as a sole genetic counsellor, linked to a main unit in a tertiary hospital. Thirty-nine per cent of the membership has been employed in genetic counselling for fewer than 5 years, while 28% have been employed for 5–9 years.

Table 4. Workplace setting in Australia for genetic counsellors (ASGC Survey, 2012)

Workplace	%
Hospital-based	64
Private provider	10
University-based	8

International jurisdictions

The scope of practice of genetic counsellors in Australia is similar to that of other OECD countries, with our national guidelines being highly consistent with guidelines from UK, Europe and North America.³⁴⁻³⁸

In regions such as South-East Asia and the Middle East, genetic counselling as a profession has only emerged relatively recently³⁹⁻⁴¹ and cultural factors influence the genetic counsellor role.

Battista et al² conducted a literature review on the current structure of genetics services in Europe, North America and Australia. They examined the basic units of genetics services, integrated services and core professional resources including counselling, focusing their examination on the basic structures of existing genetics services, including those traditionally belonging in medical genetics units as well as those developed for more common diseases.

They concluded that two types of complementary service delivery models exist: one that encompasses multidisciplinary teams comprising genetic and non-genetic specialists and the other where genetics is integrated into primary care. As noted by Sane et al²⁶ such a model of integration into primary care has not been implemented in NSW, but it is one that may need to be developed in the future. The lack of Medicare funding described above for a genetic counsellor consultation may limit the implementation of this model.

Gaps informing future scope of practice for NSW

Ambiguity between the roles and skills required to meet service needs and associated remuneration

Increasingly, genetic counsellors are able to work autonomously in the US. Hannig et al⁴² demonstrate the utility and competence of experienced genetic counsellors in their establishment of a counsellor-led clinic in Tennessee, in the US. Through careful triage, patients requiring a clinical exam and/or complex medical management were seen by a geneticist, with the remaining patients seen by the genetic counsellor.

Following their genetic counsellor visit(s), more than 80% of patients did not need additional appointments with the clinical geneticist.

Certified genetic counsellors are similarly qualified to work autonomously in such situations. Certification is transparent evidence of skills attained – skills increasingly needed to address the impact of genetic and genomic technologies on patients and their families. There are however differences in recognition of this qualification within the Award in the public sector between states. Genetic counsellors in Victoria are employed under an award that explicitly recognises and rewards certification.³¹ The NSW Award does not specifically recognise the attainment of certification.

Further, Feero et al⁴³ note that *“clinicians and health-care teams [in the US] cannot deliver an infinite number of time-intensive services to patients without adequate reimbursement”*. In 2006, the USA National Society of Genetic Counselors (NSGC) working with the American Medical Association’s Health Care Professional Advisory Committee developed a Medical Genetics and Genetic Counseling Services CPT® code, 96040, specifically for use by non-physician, trained genetic counsellors.⁴⁴ The 96040 code does not require the presence of a physician to qualify for payment and is a time-based code, meaning services are covered for every 30 minutes of the genetic counsellor’s time spent face-to-face with the patient. The code covers services that, *“... include obtaining a structured family genetic history, pedigree construction, analysis for genetic risk assessment, and counseling of the patient and family.”*

Laboratory-based genetic counsellors

Swanson et al⁴⁵ describe the emerging trend in the US for the employment of genetic counsellors within genetic/genomic laboratories. They describe various roles for a laboratory genetic counsellor, including providing patient-independent education (general information about the risks, benefits and limitations of genomic testing technologies) and patient-specific education (pertaining to a particular patient’s medical and family histories and test results/ interpretation). In particular, they note that patient-specific education is also necessary post-testing, stating *“Although some physicians may be able to order genetic tests appropriately, they may not have the ability to appropriately understand the interpretation and counsel patients about their test results”*.

These roles and future roles of laboratory genetic counsellors are further examined by Zetzsche et al⁴⁶, Christian et al⁴⁷, and Scacheri et al.⁴⁸ The benefit of laboratory genetic counsellors was demonstrated by Waltman et al⁴⁹, where they noted that the laboratory genetic counsellor was instrumental in producing significant costs savings by identifying and clarifying genetic tests that were incorrectly or unnecessarily requested.

Question 2: What are the current training and certification pathways for genetic counsellors in NSW?

Current entry

As described in the information provided by both the University of Sydney and the University of Melbourne applicants for acceptance in the Master’s courses – and therefore entry into the profession – must have:

- An undergraduate degree in a related field
- Experience (voluntary or paid) in counselling and/or genetics
- Experience in a care role.

The courses take a maximum of 12–14 students each year. There were 85 applications to the University of Sydney in 2014 for the 2015 intake (K Barlow-Stewart, Director, Master of Genetic Counselling Program, personal communication).

Applicants can already have experience or qualifications in a variety of different areas, such as genetics, psychology, social work, law, nursing/midwifery, science etc.; but it is an academically rigorous course that they are applying to enter.

Interest in genetic counselling

Data from the ASGC 2012 census indicated that more than three-quarters of the membership were attracted to the field because of the mix of science and psychology (genetics and working with people). A number mentioned their interest in science but not wanting to be a 'lab rat'.

Before becoming a genetic counsellor, the most common occupation was as a scientist or researcher. Next most common was student, indicating that many have entered the profession directly from university.

The current regulatory framework for certification governed by the HGSA – Board eligibility and certification

The HGSA has been responsible for oversight of training and awarding professional certification in genetic counselling in Australasia through a Board of Censors since 1990. Governance is now the responsibility of the ASGC, a Special Interest Group of the HGSA.

Genetic counsellors working in NSW will have undertaken training and certification via different pathways, dependent on when that training was initiated.

A major change to the training and certification system that had been in place since 1990 was introduced in 2011.

Rationale for change – need for international reciprocity

Sahhar et al⁷ compared the training program in Australia to that in the US, Canada and the UK, all of which required candidates to have completed a two-year clinical Master's degree. It was noted that this difference in academic standing limited international reciprocity in recognition of Australian-trained genetic counsellors.

McEwen et al¹⁰ describe that, following formal consideration of this need for reciprocity, the ASGC and the BOC in genetic counselling began work in 2007 to consider development of guidelines for a two-year Master-level training program that would be minimum eligibility for Part 1, followed by a modification of the Part 2 requirements.

These guidelines were completed in 2010 and are to be reviewed in 2016 (see HGSA Guidelines for Training and Certification in Genetic Counselling⁵⁰). HGSA guidelines for the accreditation of University Master of Genetic Counselling programs were also developed as part of the review process so that the university programs were internationally benchmarked (see HGSA Accreditation Guidelines⁵¹).

Changes to terminology

McEwen et al¹⁰ also note that in the process of updating the guidelines, the training process previously described as Part 1, was renamed as Board Eligible (i.e. working as an associate genetic counsellor, having previously completed a Master's degree, or until 2015, having a Graduate Diploma in Genetic Counselling, engaged in regular supervision and working towards certification) and Part 2 was renamed as Board Certified (i.e. having successfully completed their training).

Specialist cancer genetic counselling certification removal

A further change was removal of a specialty certification option introduced in 1998 for cancer genetic counselling, when there were concerns about workforce demands. This option meant completion of half the requirement for the award of Part 2 in terms of long cases and logbook cases, but did not require the evidence of skills and competencies with the broad range of conditions required for full certification. These genetic counsellors were awarded Cancer Genetic Counselling and could still use the title FHGSA.

The 2010 Guidelines recognised the importance of developing competence in 'genetic counselling', rather than developing counselling skills in specific content or knowledge areas.¹⁰

This change has caused some difficulties for those with Cancer Genetic Counselling certification as described below (under Challenges).

Board eligibility requirements – two-year Master of Genetic Counselling

From 2011, graduation from a two-year clinical Master of Genetic Counselling from an accredited program is the only entry to becoming Board Eligible (or a Graduate Diploma awarded prior to 2015).^{10, 52}

As a result:

- The University Graduate Diploma programs were discontinued; Newcastle in 2009 and Charles Sturt in 2010
- Three clinical Master of Genetic Counselling programs were implemented: the University of Melbourne program (2008), Griffith University (2011) and the University of Sydney (2011)
- The Griffith University program was discontinued in 2013.

There are therefore only two universities in Australia accredited by the HGSA Board of Censors for Genetic Counselling, to provide training for Board Eligibility: The University of Sydney and the University of Melbourne.

Content of training courses, in particular the relative balance between scientific knowledge and counselling

As described in the HGSA Accreditation Guidelines, the requirements mean that the curricula of both Universities' Master's programs are similar. Underpinning the curricula is preparation in terms of skills and competencies that are included as Appendix 2 (see HGSA course accreditation). Both Master's programs comprise approximately one third each of:

- Coursework (scientific knowledge, counselling skills and genetic counselling specific theories and models and research methodologies)
- The scientific knowledge encompasses the genetic and genomic basis of health conditions, their clinical and psychosocial impact, the current and developing genetic testing regimes and requirements, and the ethical, legal and social issues associated

- Clinical practice (minimum of 15 weeks in a range of supervised clinical placements across Australia and internationally with formal assessments of skills and competencies gained, as well as formative assessments mirroring the requirements for certification)
- A genetic counselling practice-based research project and dissertation.

Towards Board certification

Graduates of the Master's Degree can make an application to the HGSA to be considered Board Eligible and initiate the pathway to Board certification.

- Candidates must be financial members of the HGSA
- They can only commence 12 months after being employed a minimum of 0.4 FTE in a genetic counselling role, in a service that can meet the requirements for genetic counselling training.
- Board certification takes a minimum of two years FTE employment under supervision.

The competencies and skills described in Appendix 2 underpin the portfolio of assessment tasks required by the BOC¹⁰. Assessment is based on the competencies and skills described in Appendix 2 demonstrated by submission to the BOC of a portfolio as well as other tasks that must be undertaken over a minimum of two years comprising:

- Five long cases
- Logbook of 50 cases
- Published article, or alternatively a literature review
- Two reflective essays: one based on the transcript of a recorded consultation and one based on a simulated consultation with an actor
- Annual reports documenting continuing education, counselling supervision and genetics supervision
- Interview with the BOC.

The current BOC is composed of 10 genetic counsellors.⁵⁰

Maintenance of certification

The HGSA has developed a program for audit of maintenance of professional standards (MOPS) for Board Certified practicing genetic counsellors given their ethical and professional obligation to their clients, to themselves and to their clinical team members (currently under review). The MOPS program is administered by the HGSA through its BOC for genetic counselling and submissions are expected every five years (HGSA Guidelines for Training and Certification). Evidence must be provided of continuing professional education through the enhancement and maintenance of existing skills, and the development of new learning directions to meet individual professional needs. Candidates must have fulfilled the following criteria to be eligible for the MOPS program:

- Worked as a genetic counsellor or made a contribution that is of direct relevance to the practice of genetic counselling (e.g. clinical, education or research) for a minimum of 1500 hours (approximately 40 weeks' FTE) in the previous five years
- Participated in genetic counselling supervision if working in a clinical role
- Completed a minimum of 125 hours (i.e. 25 hours per year) of learning activity relevant to the area of practice, for both full-time and part-time genetic counsellors.

Training and certification of genetic counsellors 1990–2010

The majority of associate and certified genetic counsellors working in NSW entered the profession, and then undertook training and certification by a different pathway to that currently in place.

Development of Australasian certification 1980–1990

Sahhar⁵³ provides the history of the training of genetic counsellors in Australia. Genetic counsellors working in NSW Health will have become qualified in the profession by the various pathways available at the time. Although health professionals worked in genetic counselling roles in different Australian settings since the mid-1980s¹, no formal training program or certification process was in place. It was not until 1986 when a number of clinical geneticists who had trained in the US and worked with graduates of the US genetic counselling programs (see below – Other jurisdictions) returned to Australia and proposed to the HGSA that training of genetic counsellors was necessary to support the needs of the families dealing with the increasing ability to diagnose the genetic basis of health conditions they were experiencing.⁵³

A steering committee was formed comprising the president of the HGSA, a PhD scientist working as a genetic counsellor, two clinical geneticists and a social worker with experience working with families with genetic conditions, with the terms of reference to develop a proposal considering:

- The logistics of academic training in Australia
- 'Ownership' of this new profession – scientists, nurses, others
- Development of guidelines governing qualifications and process for the certification of this new profession from an Australasian perspective
- Recommendations for a wage structure.⁵³

Given the small numbers of individuals who would initially pursue training, a system of training and certification overseen by a professional Board of Censors (BOC) within the HGSA was proposed. This was a process similar to that in place for genetics professionals in other disciplines in Australia.

The HGSA accepted the recommendations of the steering committee in 1990, at which time the genetic counselling profession was officially launched in Australia. Two genetic counsellors were awarded certification in 1991, the first in Australasia.

Certification pathways 1990–2010

Further description of the training and pathway to certification, proposed as a two-step process that would be in place for the following 20 years, is provided by McEwen et al¹⁰ and Sahhar et al.⁷

Part 1 comprised development of theoretical knowledge of the science and counselling skills, and Part 2 (certification), the development of practical/clinical skills and knowledge.

Part 1 of the pathway

- Completion of a checklist of core knowledge in aspects of genetics and counselling obtained by enrolling in existing university programs and/or through additional training, such as professional development courses within the clinical genetics unit where the trainee genetic counsellor was employed⁷
- Formal postgraduate academic training programs of a one-year Graduate Diploma or Master's Degree were introduced from 1995: Newcastle University (1995), and University of Melbourne (1996), Griffith University (Brisbane), and Charles Sturt University (by distance learning), in 1997.

Part 2 of the pathway

- A genetic counselling BOC of the HGSA was created to examine a portfolio of evidence of practice
- As there were no certified genetic counsellors, the inaugural working party members became the inaugural BOC.

General Genetic Counsellors

- Development of practical/clinical skills and knowledge 'while employed' for a minimum of two years FTE under supervision in a clinical genetics service with evidence provided by submission of a portfolio to the HGSA Board of Censors in genetic counselling
 - 20 case reports (written under supervision while employed) covering a wide range of genetic conditions and diagnostic/predictive testing in the following settings: prenatal; paediatric; adult including cardiac, cancer and neurological conditions
 - Logbook of 100 cases representing this wide range of conditions.

Cancer genetic counsellors

- 1998–2010 Cancer genetic counsellor certification was introduced to meet the demands of the workplace
 - 10 case reports (written under supervision while employed) addressing genetic cancer cases only and diagnostic/predictive testing
 - Logbook of 50 cases representing these conditions.

Professional titles

In 2002 the HGSA ratified the professional titles of associate genetic counsellor for those with Part 1 certification, and certified genetic counsellor for those with both Parts 1 and 2.

Further, associate genetic counsellors were formally awarded the title, Member HGSA (MHGSA) and certified genetic counsellors were awarded Fellowship of the HGSA (FHGSA) (McEwen et al 2013).

Entry professions/qualifications

1990–2011

When genetic counselling was developing in Australia pre-1990, health professionals from a range of backgrounds (see above – consolidation phase) were considered to be providing 'genetic counselling' and moved into the position of genetic counsellors.

However, the development of standards for training and certification has provided credibility and integrity to a new and internationally recognised profession, to which entry to this is now an academic issue.

Australasian society of genetic counsellors governance

The Australasian Society of Genetic Counsellors (ASGC) was formed in 1993 and in 2002 was formalised as a Special Interest genetic counselling professional group within the HGSA.

By 2013 there were more than 270 members of the ASGC, which has developed policies such as a Code of Ethics and Standards of Best Practice for Genetic Counsellors, which are now well accepted within the Australasian context, and the ASGC is responsible for the governance and review of the training and certification guidelines.¹⁰

The majority of the members are either training or certified genetic counsellors: in 2012, just over 50% of members were associate genetic counsellors, 31% board certified and 11% currently undertaking a Master degree in genetic counselling.

The ASGC has documented the certification pathways taken to date (Figure 4). The first genetic counsellor was awarded Board certification in 1991; to date 128 genetic counsellors have been awarded HGSA Board certification and can use the title FHGSA.

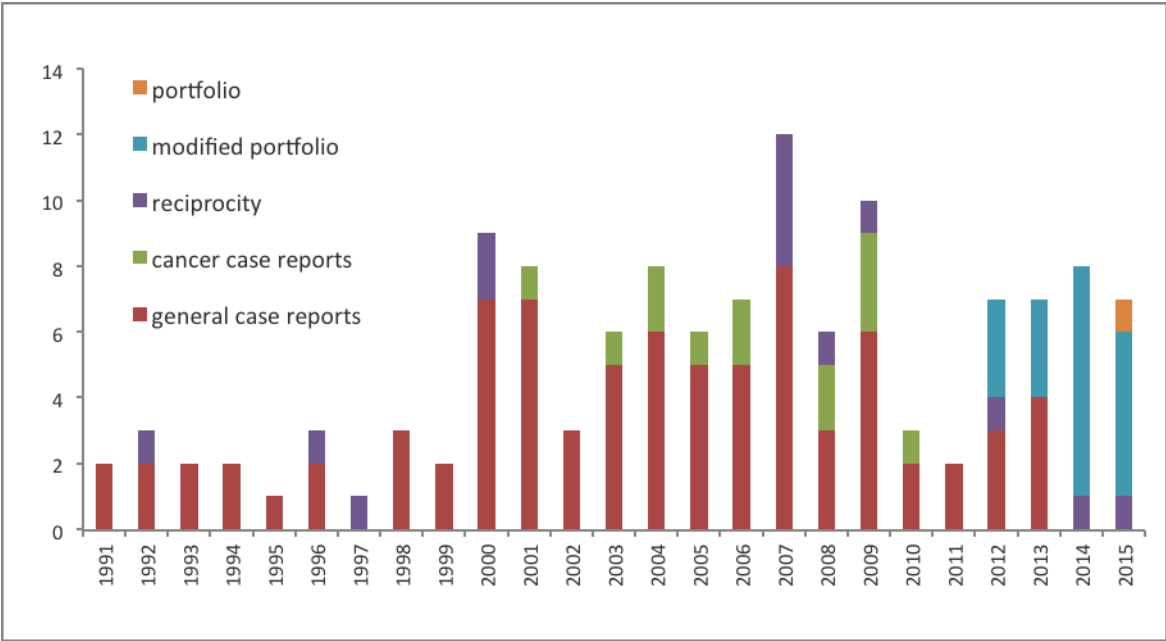


Figure 4. Number of genetic counsellors certified by the HGSA (1991–2015)

Source: Minutes of the ASGC Annual Meeting, Perth, August 2015, personal communication)

Comparisons with other jurisdictions

The UK

The UK and Australasian training programs are much aligned. In the UK, the certification is referred to as registration.⁵⁴ There is reciprocity of recognition of training between the UK and Australia. Prior to the 1990s, UK genetic counsellors were either ‘genetic nurses’ or ‘genetic social workers’.

Training and registration (certification)

Formal training with a Master’s degree in genetic counselling was established in 1992 at the University of Manchester and then in 2000 at Cardiff University. This shift was reflected in the change of professional title for non-medically qualified genetic counsellors from ‘genetic nurse’ or ‘genetic associate’ to ‘genetic counsellor’, with the establishment of the Genetic Counsellor Registration Board (GCRB) in 2001.

Voluntary registration (certification) of genetic counsellors in the UK began in 2002 under the auspices of the GCRB. Trainee genetic counsellors working towards registration require a Master’s degree in genetic counselling qualification.

- A training period of at least two years, full time (or part- time equivalent), is necessary prior to becoming eligible to apply for registration
- Training must take place in a centre meeting criteria set by the GCRB as a suitable training environment, with a named registered Genetic Counsellor as a training supervisor/mentor^{54, 55}
- Assessment for admission to the register is based upon the applicant providing a portfolio of evidence demonstrating that he/she has attained the core competencies related to the full spectrum of genetic counselling.

Maintenance of professional standards

Genetic Counsellors have to renew their registration with the GCRB every five years, which requires the demonstration of continuing professional development and evidence that the practitioner is upholding the standards of practice.

North America

The profession of genetic counselling has been recognised in the US since 1947 and the first Master's Degree in Genetic Counselling training program was initiated at Sarah Lawrence College, California in 1969.^{7, 56}

The National Society of Genetic Counselors (NSGC) was formed in 1979 and the American Board of Medical Genetics certified the first genetic counsellors in the USA in 1980, with the American Board of Genetic Counseling (ABGC) active since 1993.^{7, 56}

Training and certification

The Accreditation Council for Genetic Counseling (ACGC) is the specialised program accreditation board for the 32 educational training programs granting clinical Master's degrees or higher in genetic counselling in the US. The ACGC also accredits the three Canadian programs. Certification is by examination supervised by the American Board of Genetic Counseling (ABGC). The ABGC certifies and recertifies genetic counsellors based on a five-year certification period.⁵⁷

This system means that there is no reciprocity with the US in terms of access to training and certification.

Question 4: What are the challenges facing the NSW genetic counselling workforce now and into the future?

New and emerging challenges, as well as current and ongoing challenges that relate to this area

Impact of new technologies on genetic counselling practice

The most widely reported challenge described was the increasing complexity of the genetic counselling process with new developments in genetic testing and genomic testing technologies. These new developments make it a more time-consuming and difficult process.

These technologies are referred to as next generation DNA testing and may lead to an increase in the number of tests available, complex result analysis and interpretation, results of uncertain significance, and incidental findings in an environment that lacks guidelines and adequate databases.

Genomic testing

Until recently, the molecular genetic testing approach to the diagnosis of symptomatic individuals was carried out by testing one or a few genes at a time. The advent of new technologies, termed genomic testing, which includes whole exome sequencing (WES) and whole genome sequencing (WGS), has allowed genetic variation to be detected down to single nucleotide differences across the genome.

Genomic testing has resulted in an exponential increase in the number of possible laboratory tests available and subsequently the number of diagnoses.⁵⁸ In the past 15 years, the cost has drastically reduced (see Figure 5), as has the capacity to generate results. Results from genetic testing are available now in days rather than many months.

Concomitantly, major analytical and interpretative challenges have emerged, including validation of a large number of genomic variations identified in individuals, the economics of these new genomic approaches and management of the enormous amount of electronic data that accompanies genomic testing.²¹ Additionally, incidental variants in unexpected genes may be identified and deemed medically actionable.

Impact on genetic counselling

While the benefits of these advances are clear, and enable an increase in the diagnostic capacity for many genetic conditions, the benefits consequently impact on health care management, disease progression and family planning.²¹

- The meaning of the variants in the DNA sequence as to their pathogenicity is still in the early stages (National Institute of Health, National Human Genome Research Institute)
- Results may be of uncertain impact or may generate unwanted and unsought-after information, known as incidental findings
- Result interpretation is not straightforward^{38, 59}, the limitations of new sequencing methods need to be understood, interpretation of multiple results requires data that is difficult to obtain or still developing, and sequencing reveals many previously unknown variants of uncertain significance⁶⁰
- The lack of published and clinical guidelines and protocol around the genomic testing process is reported as adding to the complexity of the process⁶¹
- The return of these results to the patient will require careful pre-test counselling for unexpected findings.⁶²

Therefore result interpretation and how best to counsel and disclose incidental findings and variants of unknown significance, including the social and ethical issues associated, is an area that is evolving and is often difficult and inconsistent.^{61, 63-67}

In NSW, Mattick et al⁶⁸ note the lack of a purpose-built, well-curated and continuously updated evidence-based database of human genotype–phenotype associations where clinical decision-making relies heavily on the knowledge of individual practitioners and genetic counsellors. Ingles et al⁶⁹ noted in their current experience in the Centenary Institute, Sydney that the basic principles of pre-test counselling essentially remain unchanged however the inherent uncertainty of the gene result must be conveyed to the patient.

Genomic testing is becoming increasingly available for many groups of diseases, including cancer, and neurological and psychiatric. Thus, genetic counselling will be sought by these specialties.^{62, 70} Direct-to-consumer testing is yielding significant data of unknown significance. The layperson and their GP's inability to accurately interpret this data are resulting in an increased number of referrals and demands on service.⁷¹

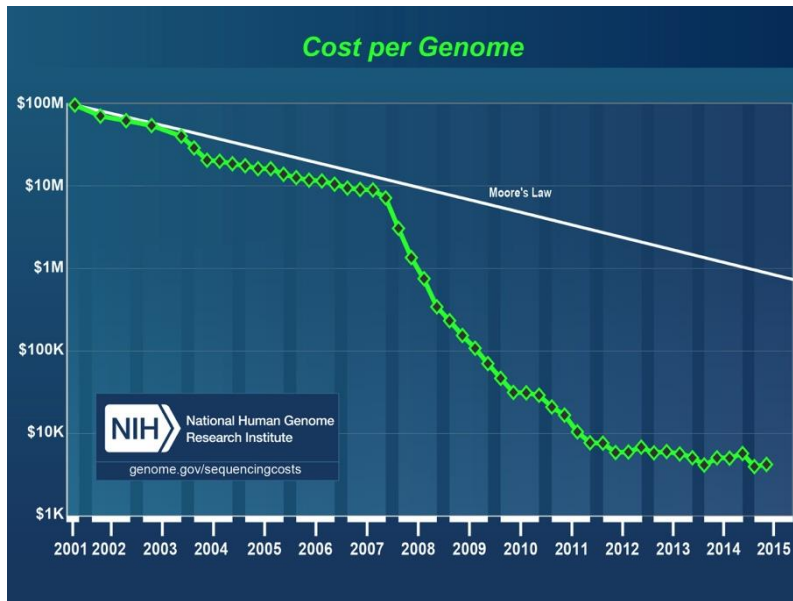


Figure 5. DNA sequencing costs over time. Source: National Institute of Health, National Human Genome Research Institute. DNA sequencing costs. Retrieved from: www.genome.gov/sequencingcosts/ [August 2015]

A changing clinical paradigm to meet increasing demand

The introduction of next generation DNA testing into mainstream medicine is changing the current clinical paradigm as health professionals outside of clinical genetics services seek an understanding of complex genetic results and how to communicate them. Genetic counsellors will play a key role in multidisciplinary teams and primary health care networks as educators and interpreters of these results.

Genetic counsellors working in cancer genetics work in parallel with medical clinicians, involving the latter in complex cases. As the referral rate continues to rise in cancer genetics, the current workload is becoming unsustainable.

There is an increase in paediatric cancer referrals, with the identification of new syndromes. Challenges include education of paediatric staff to recognise cases for referral, cross credentialing of genetic counsellors to work across LHDs, activity-based funding barriers where funding does not return to the site providing the service, and availability of outpatients' rooms. General genetics service to adults is underserved because of delayed implementation of genetics services in adult hospitals.

Mainstreaming of genetic and genomic testing

Radford et al⁶³ note that next generation sequencing (NGS) will force a change in the conventional clinical genetic practice paradigm as the cost of testing continues to drop.⁷² The authors suggest that while this will improve broader access to genetic testing, there will be an increasing demand on those with the proficiency to interpret results in the clinical context and make appropriate management recommendations.

The widespread adoption of NGS technology requires a heightened importance in understanding variations in laboratory practice with regard to their coverage and analytical sensitivity.⁷³

Manolio et al⁷⁴ cite *"costs concerns and institutional inertia"* as major challenges in implementation of genomic medicine into the clinic. Despite these challenges, their recommendations include *"integrating*

genetic counsellors and/or geneticists in non-genetics clinical services through primary institutions and affiliates” in order to ensure that the benefits of genomic testing are used to the maximum benefit of patients. In addition to this, Feero et al⁴³ emphasise that education of non-genetic health professionals is key, stating that, “advancements in genomic science... have opened potential opportunities for improved patient care and disease prevention that can only be fully capitalised on by an educated health professional, and in particular, physician, workforce”. They also note the challenge of funding the implementation of genomic medicine, stating “clinicians and health-care teams cannot deliver an infinite number of time-intensive services to patients without adequate reimbursement”.

As genomic testing infiltrates into routine clinical care, there is an emerging view that genetic counsellors will be the primary professionals called upon to educate and assist primary care providers and other specialists with the requisition of genomic testing, as well as interpretation and understanding of results.^{75, 76}

Additionally, genetic counsellors may well be integral members of many more multidisciplinary teams.⁷⁰ The NSGC has established a workforce-growth working group, with the aim of developing a strategic plan for increasing the number and diversity of trained genetic counsellors.⁷⁰

Increased demand requires increased workforce

NSW public family cancer clinics often operate with a cancer genetics specialist coming from medical oncology and surgery. A dual training program is now recognised by the Royal Australasian College of Physicians, with new medical staff likely to come from medical oncology in the future. With 2.8 FTE of the 4.1 FTE cancer genetics physicians in NSW approaching retirement age in the next five years, this has been a critical need identified and highlights the need to have fully and highly trained genetic counsellors to continue to work autonomously, but part of a wider team. Genetic counsellors working in cancer genetics work in parallel with medical clinicians, involving the latter in complex cases. As the referral rate continues to rise in cancer genetics, the current workload is becoming unsustainable. The Draft Model of Cancer Genetics Services in NSW was developed in 2010, identifying the need for attracting new cancer genetics non-geneticist professionals to the area.

Autonomous genetic counsellors were an important part of this model, working in these wider teams to provide the majority of the routine care. The specialist physician would see cases of complexity, mutation positive and variants of unknown significance. There is a need to increase the number of genetic counsellor positions in cancer services.

There is likely to be a similar need in the area of other specialty areas, such as cardiac and neurological genetics, as these services grow with the identification of more genes. Consultations for indications such as neurological disease, dementia and cardiovascular risk have been observed to have increased and will likely to continue in the future with increasing availability of testing and demand from the population (personal communication, Australasian Association of Clinical Geneticists). It is also likely that there will be increases in the need for counselling and genetic testing for susceptibility to common disorders related to public health.⁷⁷

There is an increase in paediatric cancer referrals, with the identification of new syndromes. Challenges include education of paediatric staff to recognise cases for referral, cross credentialing of genetic counsellors to work across LHDs, activity-based funding barriers where funding does not return to the site

providing the service and availability of outpatients' rooms. General genetics service to adults is underserved because of delayed implementation of genetics services in adult hospitals.

Challenges specific to the field of genetic counselling and broader relevant challenges facing small but critical health professional workforces

Challenges and barriers to training and certification, and building the workforce

Challenges in building the workforce include limited availability of student clinical placements required for accreditation of the course, limited availability of senior supervisors due to small numbers, and workload and limited employment opportunities that meet requirements of Board certification.

Limited student numbers in the Master's courses

The challenge to increasing the workforce is the small intake annually in the enrollment in the two Master's programs in Australia and the entry to the profession, which is limited by the availability of clinical placements that are required for HGSA accreditation of the courses. This intake is similar to that in the US programs with the availability of placements also the limiting factor.⁵⁷

The course accreditation requires clinical practice training as part of the academic course and in the certification training to be supervised by experienced genetic counsellors. The limited availability of such supervisors and their clinical workload contributes to this barrier.

Training places to provide opportunities for Board certification

Barriers to completing Board certification for Australian Master's graduates include the limitation in employment that meets the requirements for Board certification. As noted, a candidate must complete two years FTE employment as an associate genetic counsellor while the employment-based training takes place.

This barrier was overcome in the UK by the creation of specific two-year training places for genetic counsellors.^{54, 78, 79} Although the current registration system for genetic counsellors is voluntary, the UK Health Professions Council made a recommendation to the government in 2009 that the profession be statutorily regulated. The current UK government proposes a form of regulation involving quality assurance of voluntary registers, and this is currently under consideration.

Approved training centres were allocated funding for between one to four posts over the course of the scheme.⁵⁴ The most important outcome of the training post scheme is the benefit to patients and families.

A trained genetic counsellor workforce means shorter waiting times for patients (who would otherwise join the queue for appointments in the limited number of consultant clinical geneticist clinics) and access to skilled counselling to facilitate decision-making and adjustment to genetic diagnoses and testing.⁵⁴

Reciprocity of training internationally

Guidelines have been developed for HGSA Board certification for genetic counsellors trained in other countries (HGSA certification eligibility for overseas trained and/or certified genetic counsellors).

As Board eligibility in the US requires graduation from a US-accredited Master's program (American Board of Genetic Counseling), no reciprocity in training for certification is possible for Australian graduates in the US. This is not the case for Canada and the UK, and international exchanges in training during the clinical Master's degree and in working towards certification are encouraged.^{80, 81}

Cancer genetic counsellors and cross training

A further barrier exists for genetic counsellors awarded cancer genetic counselling certification in the period 1998–2010 who now wish to attain full certification for professional recognition or wider employment opportunities. Given the high workload in the family cancer services, release of the employees to enable meeting the cross training requirements is not easily facilitated.^{10, 82}

Clarifying the ambiguity in relation to certification and the award

The NSW Health Service Health Professionals (State) award that covers genetic counsellors states that they must have an undergraduate degree in a non-nursing, non-medical discipline and in addition hold a postgraduate qualification in genetic counselling or have attained Part 1 certification in genetics counselling from the Human Genetics Society of Australasia. This is the minimum requirement to be employed as a genetic counsellor in NSW.

While it is accepted that a higher level of skill and knowledge is attained through the completion of the full certification process, there remains some ambiguity in relation to the range of essential skills and knowledge required by health services of genetic counsellors working across a variety of models of care and how this relates to remuneration as per the award. This may become clearer over time as emerging technologies, new models of care and the associated scope of practice of genetic counsellors are further developed.

Need for increased diversity within the profession

Mittman and Sullivan (2011) propose that a diversity of health professionals is important to respond to the needs of the whole population. They note that ethnic and racial minorities are under-represented in the genetic counselling profession and this has also been found in the Australasian genetic counselling workforce (ASGC status survey³⁰).

Similarly, Kopesky et al⁸³ and Schoonveld et al⁸⁴ note that attracting more male genetic counsellors to the profession may increase patient choice and satisfaction.

The increased demand on genetics services and genetic counselling workload as a result of increased awareness about genetic testing

An increase in the number of tests available, time needed for interpretation, analysis and follow up, volume of referrals both appropriate and inappropriate, and a lack of health professionals' knowledge in this area to manage community concern, has significantly increased demand on genetics services. This continues to highlight the critical role played by genetic counsellors in meeting patient and service needs and inform priorities for future workforce planning.

Demand for genetic counselling and genetic testing increased following the initiation of the mapping of the human genome.⁵⁶ However there has been a marked recent increase in workload due to the increase in the volume of tests⁷⁰ and the time necessary for interpretation and the genetic counselling process accompanying new testing technologies. These include the burden of follow up of clients related to changing and increasing knowledge and understanding of variants^{60, 85}, which are well documented internationally⁸⁶ but limited reports are available in Australia.

The Angelina effect

James et al⁸⁷ and Evans et al⁸⁸ report on the Australian and UK experience respectively relating to the increase in referrals to family cancer services following media celebrity Angelina Jolie reporting her diagnosis of a *BRCA1* gene in The New York Times (May 2013), resulting in increased waiting times for assessment. An increasing volume of referrals related to Angelina Jolie and other celebrities is bringing attention to cancer genetics. The number of referrals increased significantly, with the adjusted levels still significantly increased from pre-Angelina Jolie announcement times.^{87, 88}

However, there has been no increase in genetic counselling services.⁸⁷ Ms Jolie's announcement saw a change in public awareness and generated a phenomenon of worried-well clients. Mechanisms are being developed to deal with this increase in referrals, such as online information, but are still in the research setting. This has potential to be integrated into cancer genetics practice in the future. The introduction of reflex pathology testing is increasing the identification of individuals with potential hereditary cancer conditions, thus increasing referrals to cancer genetics units.⁸⁹

Community awareness and expectations

Other reported reasons for increased demand included general increase in inappropriate referral by other health professionals to genetics services due to a lack of knowledge in this area.⁶⁵

High public and patient expectations related to increased community awareness about genetic testing has created challenges for genetic counsellors to educate about limitations. Bernhardt, 2014^{60, 87} and James⁸⁷ noted that this reality is at odds with the expectations of genetic testing created through media interest. Presently, clinical genetic tests can often provide a patient with meaningful information about risk for one particular disorder, whereas very little of the whole-genome sequence data will have any associated meaning⁶⁰ and patients need to be warned.

Demand may also outstrip available supply of genetic counsellors^{63, 74} as a consequence of these factors.

Traditional models of genetic counselling provision will not meet the needs of new paradigms of care

The scope of practice of genetic counsellors has traditionally included delivery and explanation of results. However, with the advent of new genetic testing technologies, it is proposed that the future scope of practice of genetic counsellors will include involvement in the interpretation of genetic variants as part of a multidisciplinary team, as laboratory-based genetic counsellors and in a proactive risk reduction clinic role in the public health arena.

With increasing provision of population genetic screening, through both public and private sector modalities, the demand for genetic counsellors to support individuals and families faced with decisions emanating from the results of the tests will inevitably increase.

Expanding the role of genetic counsellors

In the UK, it is reported that the clinical geneticist is more likely to be involved in the delivery of results than the genetic counsellor.⁷² However, genetic counsellors in some speciality clinics in the UK and the USA are already involved in the process of interpreting the significance of genomic findings.^{65, 66} The employment of laboratory-based genetic counsellors is a model to overcome the complex nature and volume of reported results.

Expanding the roles and responsibilities of genetic counsellors requires changes to genetic counselling training program curriculum, in order to effectively and efficiently deliver results to patients and health care providers.⁴⁵

Swanson's laboratory-based genetic counsellor's model, where the genetic counsellor is key to the interpretation of results and patient education, as well as the expanded role of education of providers, additionally introduces a shift in focus for genetic counsellors' from the clinical setting and thereby challenging traditional genetic counselling goals.⁴⁵

A future proactive risk-reduction model where genome sequencing is seen as a screening tool for future health is suggested by Middleton⁶⁵ and Mills.⁹⁰ It highlights the need for adapting the consent and genetic counselling process, where pre-test counselling is highly unlikely and where an upskilling of genetic counsellors in health promotion and behavioural change practices is likely.

Several experts in Australia⁶⁹ and internationally^{65, 75, 91, 92} have expressed the view that the multidisciplinary team model where the genetic counsellor works directly with non-genetic-trained medical specialists outside of the traditional clinical genetic service to provide recommendations about disclosure of genomic results and clinical follow up, has introduced significant challenges in role clarification and responsibilities in education of other multidisciplinary team members.

Public health impact

Population screening studies in the NSW Jewish community have been well established in general genetics, for conditions such as Tay-Sachs disease. This is likely to increase into the area of hereditary breast and ovarian cancer. It has been documented that population screening programs are effective, as in the case of Ashkenazi Jews where more than 50% of carriers were identified that would have been missed using family history assessment alone. This will result in an increase in referrals for mutation-positive individuals. Population screening programs are also likely to increase the workload with the need to provide pre-test counselling in a cost-effective manner.⁹³

Proposed models for general population screening for other genetic conditions, such as cystic fibrosis (CF) have been considered in Australia.⁹⁴ In Australia, 1 in 25 people of European ancestry are carriers of CF, the most common, life-limiting genetic disease.⁹⁵ Currently, there is inequality of access both to information about CF and about access to carrier testing. Modra et al⁹⁴ examine models of government-funded, nationwide CF-carrier screening to ensure a fair system of access.

Challenges/changes related to key partners

Challenges for key partners, such as clinical geneticists, are similar in the workplace to genetic counsellors in the analysis, interpretation and management of genomic results and their limitations and the impact this will have on services. Geneticists are also concerned about the shortage of trainees.

Non-genetics trained health professionals are reported as ill-prepared for the genomic era.

Clinical geneticists

Clinical geneticists share many of the challenges reported for genetic counsellors, including the burden in obtaining, interpreting and managing genomic results where there is limited evidence and conflicting interpretation of benefit and value. This includes incidental findings and variants of uncertain significance⁹⁶; cost of testing and the psychological impact of the information on patients or their families⁷⁴, concerns

about the consent process⁹⁷, and the time-consuming demands of medical genetics in the genomic era.⁹⁸ Additional challenges include concerns that the number of trainees choosing the primary specialty of medical genetics is not increasing.⁹⁹ An Australian study¹⁰⁰ cited result interpretation and communication were challenges identified by clinical geneticists following the introduction of chromosomal microarray testing (CMA), a genomic test.

Non-genetics trained medical specialists

The main challenge reported consistently in the literature and across all specialties for this group is that medical specialists are ill-prepared for the genomic era and there is a need for sustained education as described above.^{43, 74, 101-103} Gaps identified for particular groups include essential knowledge of genetics and communication skills for effective counselling of patients around genetics and legal implications¹⁰⁴, performing adequate prenatal genetic counselling in a busy practice by Obstetricians¹⁰⁵, ensuring comprehensive phenotyping and clinical evaluation of first-degree relatives before genetic testing⁶⁹ and managing the genomic data and its interpretation, particularly as part of personalised medicine as its expands and changes the established treatment paradigms in oncology.¹⁰⁶

Continuing education needs of genetic counsellors and genetic counsellors as educators of non-genetics trained health professionals

Genetic counsellors have a large future role in assisting the translation of genomic information into mainstream medicine through the education of non-genetics health professionals. Ongoing education of practicing genetic counsellors to fulfill this role is needed.

Upskilling the genetics workforce in the genomics era

Urgent attention is needed towards upskilling of the genetic counselling workforce in the genomic era, as this technology and its interpretation was not part of any of the early training regimes.¹⁰⁷

Upskilling the genetics workforce in the genomics era Given interdisciplinary teams are increasingly commonplace in Australian healthcare, there is also a need for interdisciplinary education for genetic counsellors as a means of improving patient care.¹⁰⁸ Specialty genetics areas (cancer, cardiac, neurology) are becoming integrated into primary care. As part of this integration, time is needed to educate the traditional specialists in the interpretation of genetic information.^{2, 109}

Multidisciplinary team meetings increasingly require the presence of a genetics clinician in cancer genetics. This has allowed for more appropriate and timely referrals, especially in the setting of treatment-focused decision-making. However, with this integration and treatment focused aspect, comes an increasing demand for rapid service, putting a strain on existing service.

There are proposals to mainstream such genetic testing, with genetics services only becoming involved for medically actionable results or variants of uncertain significance.^{19, 110}

Educating non-genetics health professionals and genomics

The critical importance of education of non-genetic health professionals to the successful integration of genomics into health care has been emphasized.^{43, 68, 102, 103} A number of authors reported that genetic counsellors have a large future role in supporting other health professionals^{65, 90} and will be called upon to educate them.⁷⁰

Radford et al⁶³ noted that genetic counsellors may be leveraged to facilitate incorporation of genomic information into mainstream medicine serving as a hub of information, instrumental in facilitating new models to deliver genetics-based care through promoting academic-community partnerships and interfacing with non-genetics health professionals.

Challenges resulting from changes in technology, science, policy, costs, and service delivery models and regulatory frameworks

Increasing impact of new technologies on genetic counselling practice and a changing clinical paradigm

A number of challenges have been previously outlined in relation to the impact of new technologies on genetic counselling practice and the changing clinical paradigm of genetic counselling practice. Additional challenges have been identified and are outlined in the following section.

Existing models of consent need to evolve

The duration and content of the consent process will need to evolve to accommodate the shift from single-gene to multiple-gene testing, limitations of this new testing, an increased generation of results of uncertain significance, and the integration into mainstream medicine and increase in population screening related tests where time for pre-test counselling may not be prioritised.

The familial aspect of genetic testing means that testing is traditionally preceded by counselling to discuss its advantages and disadvantages with individuals, so that informed decisions can be made.

The medical and ethical significance of genetic testing is dependent on the context in which the testing is being considered. Consent may be verbal for a DNA test designated as Level 1 or if the results are associated with complex issues, it is designated as level 2.^{23, 111} Level 2 DNA testing must be accompanied by pre- and post-testing professional genetic counselling.²³ The complex issues have ethical, legal and psychosocial dimensions: including autonomy, beneficence and non-maleficence, privacy and confidentiality and justice, just as any medical test but with the added complexity of the shared nature of genetic information within families and the potential for genetic discrimination.^{3, 111}

New genetic testing technologies (that enable faster and cheaper genetic testing and broader examination of a person's DNA) generate similar ethical challenges but with additional factors of management of enormous amounts of data with current limitations on interpretation, increased generation of uncertainty and implications for informed consent regarding unsought or unwanted results.^{12, 13}

The duration and content of the consent process⁶³ needs ongoing revision to manage potential for genomic testing-related incidental findings and variants of uncertain significance. Also, integration in mainstream medicine and an increase in population-screening-related testing will require existing models of risk communication and results delivery to be reviewed⁶⁵ where patients will have not have extensive pre-test counselling.

The complexity of information provision and division of responsibilities between clinicians and the lab requires close collaboration between clinician and lab, and further development of guidelines.⁹⁷ Obtaining informed consent for concurrent testing of multiple genes requires that genetics professionals modify their discussions with patients regarding the potential cancer risks and the associated implications to medical management.⁶⁴

Need for registration of the profession

Genetic counselling in Australia is not a registered health profession. The profession continues to seek licensure with the Australian Health Practitioner Regulation Agency (AHPRA) but has been unsuccessful to date.

There is no prohibition on any person offering genetic counselling services that puts patients at risk of sub-standard care.

The Australian Government established an inquiry into the ethical issues surrounding the protection of human genetic information, which was led by the Australian Law Reform Commission (ALRC) and the Australian Health Ethics Committee (AHEC) of the NHMRC. The 2003 report, *Essentially Yours*³ (Recommendation 23–2) stated “*The Commonwealth, States and Territories should examine options for the further development of genetic counselling as a recognised health profession, including the use of certification, accreditation or registration systems for genetic counsellors*”. It was noted that unlike medical practice or nursing or psychology, genetic counselling is not a registered health profession.

There is, therefore, no equivalent of provisions in medical practice acts that make it an offence for persons who are not registered medical practitioners to claim to be, or hold themselves out as, medical practitioners. 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.

There is a need to protect the public from substandard genetic counselling services and, in particular, from persons who are not appropriately trained, qualified and supervised yet hold themselves out to be genetic counsellors. As genetic testing becomes cheaper and more tests receive a Medicare Benefits Schedule (MBS) number, greater interest will emerge from private sector pathology laboratories. This will put the patient at greater risk of sub-standard genetic counselling care or even no care.

The ALRC report³ also stated that the Commonwealth, State and Territory governments need to examine closely all options for the further development of genetic counselling as a health profession. This examination should focus on measures to recognise the importance of genetic counsellors in building a bridge between emerging genetic science and the psychosocial needs of individuals who are, or may come to be, affected by genetic disorders. In this context, the development of clear standards of professional ethics would be desirable.

Proponents of registration cite reasons including protecting the public from unqualified “genetic counsellors”: WHO¹¹² notes that in Australia there are no specific provisions in any state to prohibit any person, regardless of qualifications, claiming to be a genetic counsellor, or providing genetic counselling. There are also no formal sanctions in place for breach of ethical or professional standards in genetic counselling.

The genetic counselling profession seeks to not only ensure public safety but also to maintain competency standards of the profession and seeks statutory regulation. The genetic counselling profession in Australia is not governed by statute although registration of the profession in Australasia has been discussed within the ASGC for over a decade. The ASGC has and continues to endeavour to achieve licensure with the Australian Health Practitioner Regulation Agency (AHPRA) and has been unsuccessful to date, with the response being

that genetic counsellors do not pose a threat to the public's safety (ASGC Secretary, personal communication).

International comparisons

UK

Genetic counselling has been a registered profession in the UK since 2001.⁵⁵ Additionally, in the UK, a 2003 Government White Paper recognised genetic counselling as an emerging profession that may warrant statutory regulation. The Health Professionals Council (HPC) regulatory body subsequently recommended statutory regulation for UK genetic counsellors, recognising that genetic counselling was a stand-alone profession with defined education and training standards, evidence-based practice and a code of conduct. The UK genetic counselling profession however continues to work towards attaining regulation following the UK coalition government's shift from statutory regulation in favour of a new system for quality assurance through voluntary registers.¹¹³

US

Fifteen states in the US are currently issuing licenses for genetic counsellors (www.nsgc.org). This supports the views of US genetic counsellors surveyed by Mester et al¹¹⁴, who believed licensure would legitimise genetic counselling as a distinct allied healthcare profession, increase the public's protection, and allow genetic counsellors to practice independently. In this study, 95% of respondents were supportive of licensure.

Costs

Medicare Benefits Schedule does not cover genetic counselling by certified genetic counsellors and levels of practice do not influence funding to units.

With increasing likelihood that genetic counsellors will practice outside of clinical genetics clinics, there are major financial implications to specialty units and the genetic counsellors themselves.

In addition, the cost of increasing education for non-genetics health professionals by genetic counsellors is generally borne by clinical genetics departments.

Medicare funding

At present, genetic counselling is predominantly provided within the state and territory public health system, for example, by genetic counsellors working in public hospital-based clinical genetics services or family cancer clinics and in some specialised units such as family cancer clinics, fetal medicine departments and cardiology departments. There are few private, practicing genetic counsellors. The ALRC Report³ notes that the Medicare Benefits Schedule (MBS) does not cover genetic counselling by HGSA-certified genetic counsellors.

Currently, the levels of practice – defined as Level 1–5 genetics services – do not influence funding to units, but if this does become the case specialised units may be disadvantaged where the unit is genetic counsellor-led and a clinical geneticist does not work with the unit, despite the unit functioning as a level 5 service. This may have major financial implications to these specialty services in the future if budget is attached to this, impacting on Genetic Counsellor employment and sustainability of an otherwise expert service.

With the increasing likelihood that cancer genetics will mainstream in the near future¹¹⁰, there will be further contention on pathology budget to pay for the tests ordered. Reluctance will come from non-genetic specialists to order these tests if the pathology budget is to come from their own service. However, it is not sustainable for genetic counsellors to see every patient diagnosed with ovarian cancer to order genetic testing, nor is it sustainable for the department pathology budget.

Educating non-genetics professionals

The integration of genetic testing into mainstream medicine, as well as the identification of new genes and genetic syndromes, will see a greater need for genetic counsellors to educate other health professionals in regard to appropriate use of such testing.¹¹⁵ This is a cost borne by the genetics departments, and often provided by genetic counsellors.¹¹⁰

Board certification

Finally, costs are borne by the specialty clinics when genetic counsellors are training for their Part 2 certification. The cross training requirement has proven to be extremely difficult in NSW and a costly exercise when units are not able to 'swap' a genetic counsellor for the purpose of cross training. While this is not linked to the NSW Health Professionals Award, it is a very difficult undertaking with limited support. Although highly valued in improving the quality of genetic counsellors' work, the training of these genetic counsellors is also labour intensive with very few senior genetic counsellors or other appropriate allied health professionals available to supervise a large junior workforce.

Health Education & Training Institute (HETI) has rolled out clinical supervision in all allied health areas of work, lending support to not only the training process but beyond certification. Due to the lack of appropriate and available supervisors, a number of both junior and senior genetic counsellors pay privately to access supervision via psychologists in the private sector.

Service delivery models

It is not feasible to set up a genetics service at every LHD or within multiple hospitals in every LHD. However, with the use of Webex proving to be an acceptable and safe way to deliver telemedicine, it is likely more genetic counsellors will use this as a service modality in the future.^{116, 117}

Telehealth provision has been set up at several sites and proved integral and well accepted for rural communities.¹⁶ There are issues with costs being attributed to where the patient is seen and not back to the site the service is being delivered from.

Need for national harmonisation in policy governing delivery of genetic counselling in the genomic era

Families do not respect state boundaries in their need for care and it is essential that the information and service they receive is of the same standard and governed by the same protocols. There are a number of other state-based organisations addressing their population's needs but it will be essential to have some harmonisation.

The HGSA as the main professional organisation of genetic specialists in Australia and encompassing several special interest groups (SIGs), including the ASGC, needs to be part of any policy development by the State and Commonwealth governments.

The HGSA has an important role in informing practice and policy and has published a document entitled *Process of Genetic Counselling*⁴, which is currently under review. The HGSA has not published guidelines or

policy statements pertaining to counselling in the genomic era. In Australia, as of 2009, a fully coordinated national approach to public health genomics did not exist.¹¹⁸

The NSW Ministry of Health's (MOH) Genetics Services Advisory Committee (GSAC) comprises health professionals, including genetic counsellors, and since 1989 has been at the forefront of developments of policies and protocols in Australia. One of the aims of GSAC is to provide expert advice to NSW Health on the health implications associated with the new genetic technologies. A cancer genetics sub-committee of GSAC was initially in existence until the introduction of the NSW Oncology Group (cancer genetics), under the Cancer Institute NSW (CINSW) in 2005 to advise the MOH of the same around cancer genetics issues. Genetic counsellors are representative on this committee. A representative of GSAC is a member of this group.

Several other Australian states have established genetic health networks or advisory bodies.¹¹⁸ Three Australian states, NSW, WA and Victoria have fully or partially funded units with a remit covering genetic policy, service delivery, research and education. In NSW, the Centre for Genetics Education (CGE) provides genetics information for professionals and for individuals and family members affected by genetic conditions (www.genetics.edu.au). The WA Office of Population Health Genomics plans, monitors and evaluates genetics services provided through the public health system (www.genomics.health.wa.gov.au). In Victoria, the Murdoch Children's Research Institute includes a research group addressing Public Health Genetics and Genetics Education and Health Research.¹¹⁸

The Australian Government's National Health and Medical Research Council (NHMRC) published a document entitled, Principles for the translation of 'omics'-based test from discovery to healthcare (<https://www.nhmrc.gov.au/guidelines-publications/g102015>). The aim of this document is to assist researchers and clinicians in translating 'omics' based discoveries, including genomics, into properly validated tests that are clinically useful.

6 Conclusion

This rapid review has identified that there is a dearth of evidence regarding the current role(s) and scope of practice of genetic counsellors in NSW and more broadly in Australia. The report has therefore relied heavily on policies and documents produced by the HGSA, the body representing Australian professionals in the field of human genetics, and international experience and perspectives. However, the evidence provided more than 10 years ago demonstrates that the role(s) and practice of NSW genetic counsellors matched best practice as described by the HGSA and that the practice was similar to that in other Australian jurisdictions.

Much of the evidence of the numerous current and future challenges facing the genetic counselling workforce documented in this review is international and it is clear that the developments over the past 10 years in genetic and genomic technologies are driving changes in clinical practice. It is expected that NSW genetic counsellors are, and will be, facing similar challenges, highlighting the need for attention to be paid to their workforce needs.

The evidence has shown that the Australian current system of training and certification for genetic counsellors under the governance of the HGSA and its special interest group the ASGC, is of high academic rigor, is workplace competencies based and assessed, and is internationally benchmarked. These qualifications enable genetic counsellors to work autonomously. However this review has also documented that there are many barriers and limitations to growing a fully qualified workforce including the small cohorts of Masters' graduates generated annually and the requirement to be employed in a setting that can meet the HGSA guidelines to undertake the full certification, with limited employment opportunities.

While employment settings are currently largely in the public sector, private sector employment opportunities could enable growth of the Australian genetic counsellor workforce. However, these opportunities are limited by the lack of registration of the profession and associated remuneration issues.

The current genetic counsellor workforce is small but of increasing importance in ethically delivering the benefits of faster and cheaper genetic and genomic tests to the NSW population with appropriate support to enable informed decision-making and adaptation to the impact of the familial results. It is essential for workforce planning to not only investigate the current roles and scope of practice of the NSW genetic counselling workforce in the era of genomics and how the many challenges are being addressed, but also to plan for and support their training and wider roles and scope of practice.

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8 Appendices

Appendix 1: Peer-reviewed literature identified for Research Questions 1 and 3

Author	Country	Sample and method	Main findings	Quality of evidence
Burgess et al (2015)	US	Online survey with 88 Genetic Counsellors regarding telehealth as a service model	To assess whether the tasks were performed similarly or differently in telehealth compared to in person genetic counselling. A majority of genetic counselling tasks were performed similarly and high satisfaction with the telehealth model of delivery was found	Low to moderate – small response rate
Christian et al (2012)	Canada	Online survey with 43 Genetic Counsellors who work predominately in laboratory settings	To explore the tasks performed in non-clinical roles. The two primary tasks performed by participants, include acting as a customer liaison (95%) and calling out test results (88%). Nineteen participants (44.2%) also reported spending a considerable amount of time signing reports. The most prevalent areas of job satisfaction were support from laboratory directors (76.8%), autonomy (76.7%), interactions with clinicians (69.7%) and other genetics counsellors (67.5%)	Low to moderate – small response rate
Delikurt et al (2015)	UK	Systematic literature review	Investigation of barriers to referral to genetics service included: lack of awareness of patient risk factors, failure to obtain adequate family history, lack of knowledge of genetics and genetic conditions, lack of awareness of genetics services, inadequate coordination of referral and lack of genetics workforce. Those related to individuals affected by or at risk of a genetic condition were: lack of awareness of personal risk, lack of knowledge and/or awareness of medical history of family members and lack of knowledge of genetics services	High

Hannig et al (2014)	US	Audit and evaluation of genetic service delivery in Tennessee of a clinic led by genetic counsellors, who are licensed in Tennessee, and MD geneticists served as medical advisors	To examine the clinic referral sources, reasons for referral and patient dispositions following their clinic visit(s). Genetic counsellors are accepted as health care providers by patients and referring providers for 80% of clinical genetics cases. A Genetic Counseling Clinic can expand genetics services, complement more traditional genetic clinic models and use the strengths of the genetic counsellor	High
Harrison et al (2010)	US	Online survey with genetic counsellors regarding awareness of the billing code for 'Medical Genetics and Genetic Counseling Services'	To identify the impact of having this new code and to identify issues with implementation of the code. Many facilities are not using this code and the reported success of billing using 96040 is highly varied. Continued education may be beneficial to encourage reimbursement for 96040 and follow up is needed to assess the ongoing implementation and impact of the new CPT® code	Low to moderate – small response rate
Ingles et al (2012)	Australia	Health economic evaluation of genetic testing in a specialised genetic cardiology service	To determine the cost-effectiveness of the addition of genetic testing to management of families with hypertrophic cardiomyopathy (HCM), compared with clinical screening alone. The addition of genetic testing to the management of HCM families is cost-effective in comparison with the conventional approach of regular clinical screening. This has important implications for the evaluation of families with HCM, and suggests that all should have access to specialised cardiac genetic clinics that can offer genetic testing	High
James et al (2003)	Australia	Survey of 76 health professionals who identified as genetic counsellors in all Australian States and NZ	To obtain information about the needs, resources, and day-to-day operation of the genetic counsellors. Differences were observed in roles between the 3 settings (main, metropolitan outreach and rural) and included large clinical loads for metropolitan outreach counsellors, varying responsibilities in the clinical setting, and a lack of support and resources for rural outreach counsellors. Australasian genetic counsellors were keenly interested in maintaining credentials and professional role development	High – 71% response rate

Kowal et al (2015)	Australia	Semi-structured interview study with 12 genetic counsellors and clinical geneticists from around Australia who had delivered services to Indigenous Australians	To identify elements of culturally competent genetic health service provision in Indigenous Australian contexts. Participants were reluctant to generalise the needs of Indigenous peoples. Some asserted that Indigenous peoples have needs that differ from the general population, while others felt that there were no collective cultural needs, instead advocating an individualised approach. Being flexible and practical, taking time to build rapport, recognising different family structures and decision-making processes, as well as socio-economic disadvantage were all identified as important factors in participants' interactions with Indigenous clients. Indigenous support workers and hospital liaison officers were seen as valuable resources for effective service provision	Low to moderate
Kromberg et al (2006)	Australia	Audit and evaluation of genetics service delivery in Queensland January 1998 to December 1999	During the study period, 8007 clients were seen in 4817 counselling sessions in urban and regional settings, with general practitioners (GPs) constituting the second largest referral source of clients. Genetic counsellors contributed to 80% of clinical genetic sessions as sole and co-counsellors, and counselled across 79 different disorders. Prenatal diagnosis counselling constituted the greatest workload with demands for cancer counselling increasing. Counsellors also provided educational and information services to individuals, families, general practitioners, health professionals and the community. The ratio of counsellors per head of population was less than national recommendations. However, although the existing model of genetic service delivery in Queensland demonstrates accessibility, the service in general is under-utilised	High

McEwen et al (2013)	Australasia	Review of development of and update in genetic counsellor training in Australasia	This paper presents an overview of the process of developing a robust training and certification program that reflects the social and cultural environment of genetic counsellors working in Australasia. Provides a detailed description of genetic counsellor training and certification in Australasia	High as the lead author led the development and implementation of the recommendations
Meiser et al (2012)	Australia	Interview study with 20 women regarding a model of genetic testing to guide treatment for breast cancer and pilot testing of a resource	To identify young women's information preferences regarding treatment-focused genetic testing (TFGT) and to develop and evaluate a novel educational resource. The potential for more widespread TFGT in the future indicates a need for patient educational materials that enable women to make informed choices about TFGT	Moderate – not generalisable
O'Shea et al (2011)	UK	Interview study with 27 parents of children with galactosaemia and Maple Syrup Urine Disease	The aim was to inform health professionals about the need or role of a genetic counsellor in a specialist setting. While parents are well informed, the majority expressed a wish for more information about the condition and its transmission. Adult patients with galactosaemia and parents from certain ethnic backgrounds could especially benefit from genetic counselling	
Sane et al (2015)	Australia	Online survey of members of the Australasian Society of Genetic Counsellors – 78 responses	To explore the perceived interest in development of private genetic counseling services in collaboration with primary care physicians in the Australasian setting. 4 hypothetical private practice models of professional collaboration between genetic counsellors and primary care physicians or clinical geneticists were proposed. The majority (84.6 %) showed a positive degree of interest and enthusiasm towards potential for clinical work in private practice. Support from clinical genetics colleagues and the professional society was highly rated as a facilitator and, conversely, lack of such support as a significant barrier	Moderate – 40% response rate

Skirton et al (2015)	UK/Europe	Systematic literature review	To evaluate the extent to which genetic counsellors undertake a range of roles, including provision of information and facilitation of psychosocial adjustment of the client to their genetic status and situation. Where genetic counsellors are used in specialist genetic settings, they undertake a significant workload associated with direct patient care and this appears to be acceptable to patients. With the burden on genetics services, there is an argument for the increased use of genetic counsellors in countries where they are under-utilised. In addition, roles undertaken by genetic counsellors in specialist genetic settings could be adapted to integrate genetic counsellors into multidisciplinary teams in other specialisms	
Zetsche et al (2014)	US	Semi-structured interviews with nine genetic counsellors working in laboratory settings	To document how positions were created in a developing role for genetic counsellors and how they have changed with time. Themes identified included that early positions were often part time, laboratory-initiated and had a lack of job definition. Laboratory genetic counsellors commented on their evolving roles and responsibilities, with their positions becoming more technical and specialised over time and many taking on managerial and supervisory roles. All genetic counsellors surveyed reported using core genetic counselling skills in their positions. The expansion of diagnostic testing and quickly evolving technology were common themes in regards to the future of laboratory genetic counsellors, and participants commented on laboratory genetic counsellors having expanding roles with data management, result interpretation and reporting, and guidance of other healthcare providers. This study describes the emergence, and subsequent evolution, of laboratory genetic counselling positions as a significant subspecialty of genetic counselling	Low

Zilliacus et al (2011)	Australia	Evaluation of genetic service delivery using telehealth for hereditary breast and ovarian cancer with 106 women seen by telehealth and 89 women seen face-to-face using self-administered questionnaires before, and one month after, genetic counselling	To compare the effectiveness and acceptability of genetic counselling using telehealth involving a genetic clinician via telegenetics in addition to a local genetic counselor present with the patient. No significant differences were found between telegenetics and face-to-face genetic counselling in terms of knowledge gained ($P = 0.55$), satisfaction with the genetic counselling service ($P = 0.76$), cancer-specific anxiety ($P = 0.13$), generalised anxiety ($P = 0.42$), depression ($P = 0.96$), perceived empathy of the genetic clinician ($P = 0.13$), and perceived empathy of the genetic counsellor ($P = 0.12$). Telegenetics performed significantly better than face-to-face counselling in meeting patients' expectations ($P = 0.009$) and promoting perceived personal control ($P = 0.031$). Telegenetics seems to be an acceptable and effective method of delivering genetic counselling services for hereditary breast and ovarian cancer to underserved areas	High
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Appendix 2: Peer reviewed literature identified for Research Question 2

Author	Country	Sample and method	Main findings	Quality of evidence
Barnes et al (2012)	UK	Audit and evaluation of the Genetic Counsellor Training Post Scheme designed to ensure that both appropriately qualified nurses and Master's level genetic counselling graduates were offered the opportunity to undertake a 2-year training period prior to registration	After a government commitment to increase the genetic counsellor workforce, the national professional organisation for genetic counsellors obtained government funding to expand training capacity for genetic counsellors through a training scheme. The scheme has proved highly successful. Of 43 trainees appointed, 42 went on to work as genetic counsellors, and 36 have already gained their professional registration	High – report of outcome of government workforce intervention
Cooksey et al (2000)	US	Report generated from a survey of the national Society of Genetic Counsellors survey	This report describes the genetic counselor workforce – a professional group specifically trained to counsel individuals and families about genetic risks. The report includes a description of the number, distribution and composition of the profession, the training programs, certification, work setting and professional practice, the job market, and factors expected to influence the future supply and demand. There are about 1800 master's trained genetic counsellors, with most working as clinicians within medical teams in urban academic medical centers and hospitals. Genetic counsellors provide information and counselling to individuals and families at risk for genetic conditions. While counsellors have traditionally worked in prenatal and paediatrics clinical areas, in recent years, their practice has expanded into adult medicine and specialty areas such as cancer centers and neurology, and into commercial genetic testing laboratories. The profession is relatively young, with the first graduates in 1971, and a steady increase in programs and graduates since that time. Currently 24 programs, usually based in academic medical centers, provide a 2-year training program, with 120 to 130 graduates per year. The American Board of Genetic	High

			Counselors offers certification for genetic counsellors, and although no state requires licensure, California has legislation pending that would require state licensure	
James et al (2003)	Australia	Survey of 76 health professionals who identified as genetic counsellors in all Australian States and NZ	To obtain information about the needs, resources, and day-to-day operation of the genetic counsellors. Differences were observed in roles between the 3 settings (main, metropolitan outreach and rural) and included large clinical loads for metropolitan outreach counsellors, varying responsibilities in the clinical setting, and a lack of support and resources for rural outreach counsellors. Australasian Genetic counsellors were keenly interested in maintaining credentials and professional role development	High – 71% response rate
McEwen et al (2013)	Australia	Review of development of genetic counsellor training and certification	An overview of the process of developing a robust training and certification program that reflects the social and cultural environment of genetic counsellors working in Australasia. A brief history of the development of the profession in Australasia is provided, followed by a detailed discussion of the recent development of Master's programs and a portfolio of work required for certification	High
Sahhar et al (2005)	Australia	Review of the Australian experience of genetic counsellor education and the history of the profession. The relevance of local factors, including the healthcare system, the education system and the small population in the evolution of the 1-year training programs are considered as an alternative model for emerging programs	The development of the education and training processes compared to that of other countries namely the US, the UK and Canada is discussed. The importance of international collaborations between the programs, to facilitate academic discussion and possible curriculum innovations, and to maintain professional understanding between genetic counsellors is emphasised. Core genetic counselling competencies have been published for the UK and US and an Australian set is proposed. Recommended that a 2-year Master's program be established to better equip Australian genetic counsellors with increased clinical skills and genetic knowledge and increase the possibility of international reciprocity, thus increasing the employment opportunities for Australian-trained genetic counsellors overseas	High – expert opinion from the Director of the Melbourne University Genetic Counsellor training program

Sahhar et al (2011)	Australia	Review of the development and 2008 implementation of the first 2-year professional Master's program in Australia at the University of Melbourne	In 2008 the first 2-year professional Master's program in Australia began at the University of Melbourne. The model for how this was achieved is discussed, along with the impact of the competencies developed by the Human Genetics Society of Australasia (Australia and New Zealand), on program evolution, teaching styles and clinical supervision. The principal challenges in developing the Master's program are explored and initiatives specific to SE Asia are discussed	High – expert opinion from the Director of the Melbourne University Genetic Counsellor training program
Skirton et al (2003)	UK	Review of the system developed in the UK for genetic counsellor certification	In the UK, a registration system has been developed based on core competencies to ensure high standards of practice	High – expert opinion from the Director of the UK program

Appendix 3: Peer reviewed literature identified for Research Question 4

Author	Country	Sample and Method	Main Findings	Quality of Evidence
Alexander et al (2013)	US	Online survey of 309 genetic counsellors	In terms of international exchange in training the study investigated (1) How prevalent are international genetic counselling experiences? (2) What types are pursued and why? (3) What supports and barriers exist? 3) What are the demographic characteristics of individuals accruing international experience? (5) Does international experience promote professional development? (6) Do genetic counselling students and professionals perceive international experiences as beneficial? Most respondents were Caucasian females born in one of 25 countries. The most prevalent experiences involved either clinical observation or clinical training. Common motivations for pursuing international experience were personal growth, exposure to a different healthcare system and travel opportunities. Outcomes included professionally relevant experience and personal growth. Barriers included finances, limited availability of opportunities, and for those without international experience – family responsibilities	High
Barnes et al (2012)	UK	Audit and evaluation of the Genetic Counsellor Training Post Scheme designed to ensure that both appropriately qualified nurses and Master's level genetic counselling graduates were offered the opportunity to undertake a 2-year training period prior to registration	After a government commitment to increase the genetic counsellor workforce, the national professional organisation for genetic counsellors obtained government funding to expand training capacity for genetic counsellors through a training scheme. The scheme has proved highly successful. Of 43 trainees appointed, 42 went on to work as genetic counsellors and 36 have already gained their professional registration	High – report of outcome of government workforce intervention

Battista et al (2011)	Canada	Literature review of the current organisation of genetics services in Europe, North America and Australia	To review, explore emerging service delivery models and probe challenges inherent in the transition process. Multidisciplinary specialist clinics and coordinated services appeared to be key to delivering proper care in rare genetic disorders. Inter-professional collaboration between geneticists and other specialists seemed to be favoured. On the other hand, there was also a tendency toward the integration of genetic service directly into primary care. Among the most pressing challenges was the morphing of paediatric care into adult care. Barriers to overcome include the redistribution of role, sharing of data and databases, and the lack of preparedness of non-genetics professionals and of the health care system in general	High
Bernhardt et al (2014)	US	Interviews with 10 genetic counsellors experienced with using chromosomal microarray analysis (CMA) in prenatal diagnosis informed a survey of 193 prenatal genetic counsellors	To assess the prevalence of genetic counsellors' attitudes towards, experience and comfort with, and educational needs regarding prenatal CMA. When there is an uncertain CMA result, only 59% would be comfortable providing genetic counselling and only 43% would be comfortable helping a patient make a decision about pregnancy termination. Being less comfortable was associated with seeing fewer patients having prenatal CMA testing. Respondents expressed a high degree of interest in additional education about prenatal CMA and counselling about uncertain results. Further genetic counsellor education and training aimed at improving counsellors' personal comfort with uncertain results and communicating about them with patients is recommended	High

Cichon et al (2014)	US	Two online surveys: medical students and medical program directors to explore why approximately 50% of medical genetics residency positions remain unfilled each year	To evaluate current recruiting efforts and institutional support for programs and to identify factors that helped trainees choose genetics as a career. Program directors identified the most successful recruiting methods as 'direct contact with residents or medical students' and 'word of mouth' (80%). Residents listed having a mentor (50%), previous research in genetics (35%), and genetics coursework (33%) as the top reasons that influenced them to enter the field.	High
Cooksey et al (2000)	US	Report generated from a survey of the National Society of Genetic Counselors survey	This report describes the genetic counselor workforce – a professional group specifically trained to counsel individuals and families about genetic risks. The report includes a description of the number, distribution and composition of the profession, the training programs, certification, work setting and professional practice, the job market, and factors expected to influence the future supply and demand. There are about 1800 master's trained genetic counselors, with most working as clinicians within medical teams in urban academic medical centers and hospitals. Genetic counsellors provide information and counselling to individuals and families at risk for genetic conditions. While counsellors have traditionally worked in prenatal and paediatrics clinical areas, in recent years, their practice has expanded into adult medicine and specialty areas such as cancer centers and neurology, and into commercial genetic testing laboratories. The profession is relatively young, with the first graduates in 1971, and a steady increase in programs and graduates since that time. Currently 24 programs, usually based in academic medical centers, provide a 2-year training program, with 120 to 130 graduates per year. The American Board of Genetic Counselors offers certification for genetic counselors, and although no state requires licensure,	High

			California has legislation pending that would require state licensure	
Delikurt et al (2015)	UK	Systematic literature review	Investigation of barriers to referral to genetics service included: lack of awareness of patient risk factors, failure to obtain adequate family history, lack of knowledge of genetics and genetic conditions, lack of awareness of genetics services, inadequate coordination of referral and lack of genetics workforce. Those related to individuals affected by or at risk of a genetic condition were: lack of awareness of personal risk, lack of knowledge and/or awareness of medical history of family members and lack of knowledge of genetics services.	High
Evans et al (2014)	UK	Audit and evaluation of genetic service delivery (2012 and 2013) with a consortium of over 30 breast cancer family history clinics that have contributed to two research studies on early breast surveillance as well as 10 genetics centres	Followed two events in 2013: publicity regarding Angelina Jolie's decision to have genetic testing for the BRCA1 gene and subsequently undergo risk reducing mastectomy (RRM), and a pre-release of the NICE guidelines on familial breast cancer regarding the potential for use of chemoprevention using tamoxifen or raloxifene in January and their final release on 26th June. A rise in referrals was reported from May 2013 onwards. Referrals were nearly 2.5 fold in June and July 2013 from 1981 (2012) to 4847 (2013) and remained at around two-fold to October 2013. Demand for BRCA1/2 testing almost doubled and there were also many more enquiries for risk reducing mastectomy. Internal review shows that there was no increase in inappropriate referrals. The Angelina Jolie effect has been long lasting and global, and appears to have increased referrals to centres appropriately	High

Everett et al (2014)	US	Audit and evaluation of reporting of genetic test results by genetic counsellors	Report of experiences as part of a research team implementing a protocol for whole genome sequencing (WGS). Genetic counsellors discuss options for return of results with patients during the informed consent process and document family histories. Genetic Counsellors also review germline findings and actively participate in the multi-disciplinary Precision Medicine Tumor Board (PMTB), providing clinical context for interpretation of germline results and making recommendations about disclosure of germline findings. Genetic counsellors have encountered ethical and counselling challenges with participants. Findings demonstrate that genetic counsellors have important applicable skills to contribute to multi-disciplinary care teams implementing precision oncology. Broader use of WGS in oncology treatment decision-making and American College of Medical Genetics and Genomics (ACMG) recommendations for active interrogation of germline tissue in tumor-normal dyads suggests that genetic counsellors will have future opportunities in this area outside of research settings	Low to moderate
Kopesky et al (2011)	US	Online survey of 190 undergraduates (110 females, 79 males, 1 unknown) in upper division bioscience courses assessing their knowledge and perceptions of and interest in genetic counselling as a career	Genetic counselling is a female-dominated field, with women comprising about 95% of the profession. Greater patient choice and satisfaction may be achieved by increasing the number of male counsellors. Females indicated significantly greater interest than males in pursuing a genetic counselling career, and they rated interpersonal skills as more integral to genetic counselling than males. For females, there were four significant predictors: estimated salary, career characteristics, perceptions of genetic counselling as interpersonally focused, and whether they had already chosen a career. Implications for recruiting males to the profession and research recommendations are presented	High

Machini et al (2014)	US/Canada	Online survey of 221 genetic counsellors, geneticists and nurse practitioners – one third of whom currently offer new genetic technologies (WES/ WGS)	To identify barriers to the implementation of WES/WGS; to provide the first systematic report of current practices regarding the integration of WES/WGS in clinic and/or research across the US and Canada and to illuminate the roles and challenges of genetic counselors participating in this process; and to evaluate the impact of WES/WGS on patient care. Genetic counselling practices with respect to WES/WGS are consistent with the criteria set forth in the ACMG 2012 policy statement, which highlights indications for testing, reporting and pre/post-test considerations. Challenges related to offering WES/WGS, which included billing issues, the duration and content of the consent process, result interpretation and disclosure of incidental findings and variants of unknown significance. In addition, respondents indicated that specialty area (i.e., prenatal and cancer), lack of clinical utility of WES/WGS and concerns about interpretation of test results were factors that prevented them from offering this technology to patients. The aspects of their professional training that have been most beneficial in aiding with the integration of WES/WGS into the clinical setting included molecular/clinical genetics, counselling and bioethics and counselling aids (to assist them when explaining aspects of these tests to patients). Webinars focused on WES/WGS (for genetic counsellors and other health care professionals) were suggested as useful educational tools	High
Mann et al (2014)	Australia	Online survey of genetic counsellors	To investigate implementation of interdisciplinary education for genetic counsellors in oncology settings as a formal continual learning tool for genetic counsellors. Currently most genetic counsellors source knowledge about oncology procedures through indirect means and that, overall, anecdotal descriptions from patients were the most common information source (74%). More	Moderate

			than 95% of respondents expected that interdisciplinary observations would be a beneficial part of their professional development and almost 90% expected the program to be potentially feasible in their workplace	
Matloff et al (2011)	US	Brief review	Review of the role of the certified genetic counsellor in the multidisciplinary endocrinology setting. Certified genetic counsellors assist the endocrinology team by eliciting a detailed pedigree, determining the appropriate genetic test to order, obtaining informed consent, interpreting complex genetic test results, providing psychosocial and family counselling, and assessing which family members are at risk. Many endocrine tumors can be caused by a variety of different genes and investment in the genetic counselling process likely increases the chance that the correct genetic test is ordered, results are accurately interpreted, and adequate informed consent and counselling is offered. Genetic counselling both pretesting and post-testing is essential to accurate, cost-efficient care for the endocrine patient and the entire family	Moderate
McDonald et al (2014)	US	Survey with 149/300 (49.7% response rate) residents at risk for hereditary cancer susceptibility disorders from 11/16 Maine counties – 30.8% were from rural counties	To examine acceptability of cancer genetic counselling models of service delivery among Maine residents. 92.2% indicated that an important/the most important model of care characteristic is provider professional qualifications. Among other characteristics, 65.1% ranked one-on-one counselling as important/the most important. In-person and local counselling were ranked the two least important characteristics (51.8% and 52.1% important/the most important, respectively). Responses did not vary by patient characteristics with the exception of greater acceptance of group counselling among those at perceived high personal cancer risk. Cancer telegenetics services hold promise for access to expert	High

			providers in a one-on-one format for rurally remote clients	
McPherson et al (2008)	US	Audit and evaluation of genetic service delivery. All physician geneticists and genetic counsellors in the medical genetics department used an electronic tool to record their activities in 15-minute increments during clinic hours, evenings and weekends over a 10-week period	To document, in real time, of workflow in a general genetics department including data on patient care, research, and other activities for both clinical geneticists and genetic counsellors. The average work week was 54.1 hours for physicians and 43.5 hours for genetic counsellors. During clinic hours physicians spent about one-quarter of their time on direct patient care, one-quarter on other patient-related activities, one-quarter on research unrelated to individual patient care and the remaining quarter on all other activities. However, after hours and on weekends they spent most of their time on research. Genetic counsellors spent half of their time on patient-related activities, one-quarter on direct patient care and the remainder on all other activities. The total professional time averaged 7 hours per new patient and 3.5 hours per follow-up with nearly 60% of this time devoted to patient-related activities. Conclusions: The labour-intensive nature of clinical genetics, the large amount of time devoted to patient-related activities, and continuing limitations on billing by genetic counsellors all contribute to the financial challenges faced by genetics departments	High
Meiser et al (2012)	Australia	Interview study with 20 women regarding a model of genetic testing to guide treatment for breast cancer and pilot testing of a resource	To identify young women's information preferences regarding treatment-focused genetic testing (TFGT) and to develop and evaluate a novel educational resource. The potential for more widespread TFGT in the future indicates a need for patient educational materials that enable women to make informed choices about TFGT	Moderate – not generalisable

Mester et al (2009)	US	Online survey of 62 genetic counsellors working in Michigan in the US (66% response rate)	To explore genetic counsellors' beliefs and attitudes about licensure. 95% percent of respondents were supportive of licensure, believing licensure would legitimise genetic counselling as a distinct allied healthcare profession (97.5%), increase the public's protection (75%), and allow genetic counselors to practice independently (67%). While 45% felt licensure would increase counsellor involvement in lawsuits, this did not impact licensure support ($p = 0.744$). Opinions were split regarding physician supervision and ordering tests. Even though 28% favoured physician supervision, there was overwhelming support for genetic counsellors performing some components of genetic testing (95%) and ordering some types of genetic tests (82%) independent of a physician	High
Mittman et al (2011)	US	Review of issues impacting health workforce diversity in the US	Ethnic and racial minorities have been under-represented in the genetic counselling profession since its inception, despite vigorous professional initiatives to remedy this situation. The Sullivan Alliance to Diversify the Health Professions is a national catalyst for increasing diversity within the health professions by forging state collaborations among institutions of higher education, health professions schools and other key stakeholders	Low
Reiff et al (2014)	US	Semi-structured interviews with 15 healthcare providers of genome-wide chromosomal microarray analysis (CMA) in paediatric clinical practice (7 genetic counsellors, 4 medical geneticists, and 4 non-genetics providers)	To investigate the utilisation of CMA testing. Most providers reported that genomic testing enhanced their professional experience and was beneficial to patients, primarily due to the improved diagnostic rate compared with earlier chromosomal studies. Other effects on practice included moving towards genotype first diagnosis and broadening indications for chromosomal testing. Opinions varied concerning informed consent and disclosure of results. The duty to disclose incidental findings (IFs) was noted; however concerns were raised about potential psychosocial harms of disclosing pre-symptomatic findings.	Low

			Tensions were revealed between the need for comprehensive informed consent for all families and the challenges of communicating time-consuming and potentially anxiety-provoking information regarding uncertain and incidental findings that may be relevant only in rare cases. Genetic counsellors can play an important role in liaising with families, health professionals and testing laboratories, providing education and guidance to non-genetics providers, and enabling families to receive adequate pre- and post-test information and follow-up care	
Rigter et al (2014)	Netherlands	Semi-structured interviews with 11 professional experts and one professional gave a written response regarding experiences with unexpected findings in genomic testing. The counselling process was also observed in 3 cases where exome sequencing was offered, followed by interviews with the patient (representative) and the genetic counsellor	To explore the first experiences with, and needs for, the informed consent procedure in diagnostic exome sequencing, with the stakeholders involved. The respondents not only preferred an opt-out for unsolicited findings, but also identified many challenges. Context-dependent decision-making was observed and an advisory board for unsolicited findings was considered helpful while doubts were raised about the feasibility and the possibility of undermining patients' autonomy. Finally, respondents brought up the complexity of information provision, and division of responsibilities between clinicians and the lab	Low to moderate
Rosa-Blum et al (2007)	US	Systematic literature review	Review of the future challenges of effectively communicating genetic information to patients by paediatricians. The review indicated that successful communication of genetic information involves a number of important skills and considerations. It is likely that these skills and considerations are universally required for the communication of most complex specialised medical information. In the past, communication skills have not been considered a priority. Today, these skills have become a demanding professional and even legal obligation. However, the challenges involved in communicating complex medical information cannot be successfully addressed with universal, one-size-fits-all	High

			<p>recommendations. Residency training programs require changes to adequately prepare future pediatricians for the growing challenge of communicating genetic information. 4 important skills should be considered in the training of residents to improve the communication of complex information to patients. These skills are (1) discriminating, (2) understanding, (3) simplifying, and (4) explaining information</p>	
Schoonveld et al (2007)	US	Semi-structured telephone interviews with 8 genetic counselling students and 7 practicing genetic counsellors who were ethnic minority and/or male	<p>To explore recruitment and retention efforts to increasing greater ethnic and gender diversity. Introduction to the field tended to be late and accidental. There were several career supports (e.g., field combines science and helping others) and barriers (e.g., lack of information about the field). Participant experiences, although primarily positive, included instances of passive, unintentional discrimination; and there were internal and external pressures to be diversity experts and positive representatives of their group. Participants reported positively impacting colleagues' cultural competency and offering a different presence within clinical settings</p>	Low to moderate
Slade et al (2015)	UK	Report of evaluation of senior representatives from all 24 UK cancer genetic centres in a workshop to discuss the development of cancer genetics within the UK National Health Service	<p>Workshop discussion addressed the opportunities and challenges to increasing cancer gene testing in the National Health Service (NHS). Services vary with respect to population served and models of service delivery, and with respect to methods and thresholds for determining risk and testing eligibility. Almost all centres want to offer more cancer gene testing (82%) and reported increasing demand for testing from non-genetic clinical colleagues (92%). Reported challenges to increasing testing include the complexity of interpreting the resulting genetic data (79%), the level of funding and complexity of commissioning (67%), the limited capacity of current processes and cross-disciplinary relationships (38%), and workforce education (29%). Priorities to address include the</p>	High

			development and evaluation of models of increasing access to gene testing, the optimal process for interpretation of large-scale genetic data, implementation of appropriate commissioning and funding processes, and achieving national consistency	
Turbitt et al (2013)	Australia	Interviews with 15 key informants, 6 clinicians, and 9 laboratory scientists from 4 Australian states who had experience with using chromosomal microarray (CMA) in the clinic.	To explore the process by which CMA was implemented in clinical settings. Challenges included result interpretation and communication. Strengths were also highlighted, including the collaborative approaches of some centers	Low
Zilliacus et al (2011)	Australia	Evaluation of genetic service delivery using telehealth for hereditary breast and ovarian cancer with 106 women seen by telehealth and 89 women seen face-to-face using self-administered questionnaires before, and 1 month after, genetic counselling	To compare the effectiveness and acceptability of genetic counselling using telehealth involving a genetic clinician via telegenetics in addition to a local genetic counsellor present with the patient. No significant differences were found between telegenetics and face-to-face genetic counselling in terms of knowledge gained (P = 0.55), satisfaction with the genetic counselling service (P = 0.76), cancer-specific anxiety (P = 0.13), generalised anxiety (P = 0.42), depression (P = 0.96), perceived empathy of the genetic clinician (P = 0.13), and perceived empathy of the genetic counsellor (P = 0.12). Telegenetics performed significantly better than face-to-face counselling in meeting patients' expectations (P = 0.009) and promoting perceived personal control (P = 0.031). Telegenetics seems to be an acceptable and effective method of delivering genetic counselling services for hereditary breast and ovarian cancer to underserved areas	High

Appendix 4: Skills and Competencies underpinning Genetic Counselling Training and Certification

(McEwen et al 2013)

Standards and competencies	Indicator of competency
A. Communication skills: Competencies 1–3	A. The genetic counsellor establishes and maintains a relationship with clients through effective communication, which promotes autonomy
1. Establishes a mutually agreed upon genetic counselling agenda with the client	Uses both verbal and non-verbal interactions to establish rapport
	Ability to determine the aims of the client from the session
	Ability to explain the process of genetic counselling
	Ability to record a pedigree and take a brief medical and social history
2. Conveys clinical and genetic information to clients appropriate to their individual needs	Ability to give explanations (e.g. about the condition, genes and chromosomes or testing) to clients
3. Explains options available to the client, including the risks, benefits and limitations	Ability to explain inheritance pattern and risk assessment
	Ability to provide options for clients, including the risks, benefits and limitations
B. Reflective practice, counselling and interview skills: Competencies 1–7	B: The genetic counsellor takes a self-aware, client-centred approach to facilitate client support and decision-making
1. Establishes relationship with client and elicit their concerns and expectations	Ability to engage the client and start an interview
	Ability to provide information and support to enable clients to make an informed choice about options presented
2. Elicits and interprets appropriate medical, family and psychosocial history	Ability to identify psychosocial issues in the genetic counselling session.
	Ability to elicit, confirm and accurately interpret the medical, family and psychosocial history
3. Acknowledges the implications of individual and family experiences, beliefs, values and culture for the genetic counselling process	Ability to recognise roles and relationships within families
	Facilitate and support dissemination of information about the condition to at-risk relatives
4. Uses a range of counselling skills to facilitate	Uses a range of appropriate and effective counselling skills to facilitate informed decision-making

clients' autonomy and decision-making	
B. Reflective practice, counselling and interview skills: Competencies 1–7 (cont.)	B: <i>The genetic counsellor takes a self-aware, client-centered approach to facilitate client support and decision making</i>
5. Recognises their own limitations in knowledge and capabilities, and seeks consultation and refers clients when necessary	Recognises their own of strengths and limitations
	Consults other health professionals appropriately
6. Demonstrates reflective skills within the counselling context and participates in genetic counselling supervision	Has self-awareness and can critically reflect on practice
7. Assesses client understanding and response to information and its implications, and modifies the counselling session as necessary	Ability to assess client's understanding and respond appropriately using both verbal and non-verbal skills
C. Critical thinking skills: Competencies 1–4	C: <i>The genetic counsellor identifies, synthesises and organises pertinent information for use in genetic counselling</i>
1. Identifies, synthesises, organises and summarises pertinent medical and genetic information for use in genetic counselling.	Ability to Identify and use appropriate resources to support knowledge of the genetic condition, genetic principles and relevant client information
	Ability to make an appropriate genetic risk assessment on the basis of the pattern of inheritance, laboratory results, databases, medical and genetic literature or clinical presentation
2. Evaluates a psychosocial history.	Demonstrates understanding of the psychodynamics in the family and its impacts on understanding and retention and coping mechanisms
	Demonstrates awareness of the need for intervention and/or referral
3. Makes appropriate and accurate genetic risk assessments	Demonstrates understanding of the required medical, family, personal and other information that needs to be collected to assist in or inform a diagnosis or risk assessment
	Ability to seek clinical or genetic information from other sources to confirm family information and or diagnosis

4. Develops the necessary skills to critically analyse research findings to inform practice development.	Ability to critically analyse the explanations given to clients by experienced genetic counsellors, clinical geneticists and genetics specialists
	Ability to critically appraise and utilise appropriately current evidence that informs practice
D. Case management skills: Competencies 1–3	<i>D: The genetic counsellor facilitates best practice by advocating for clients, referring clients to appropriate services and maintaining comprehensive records</i>
1. Demonstrates ability to organise, prioritise and manage a case load	Ability to identify and appropriately address client’s needs within resource limitations
2. Identifies and supports clients’ access to local, regional and national resources and services	Ability to identify appropriate information and support services either proactively or at the client’s request
3. Serves as an advocate for clients	Ability to offer appropriate information and support services either proactively or at the request of the client
	Demonstrates understanding of the relevant health and social services available to clients
E. Professional and ethical practice: Competencies 1–2	<i>E: The genetic counsellor promotes knowledge and access to genetics services through effective communication and education, maintains professional behaviour and boundaries in keeping with accepted codes of ethical practice, and promotes evidence-based practice for one’s self and others through continual professional development</i>
1. Establishes effective working relationships to function within a multidisciplinary team and as part of the wider health and social care network	Ability to relate to team members
	Ability to participate effectively in team meetings, case discussions
	Demonstrates a willingness to learn and participate in learning experiences provided
	Demonstrates professional behaviour
2. Acts in accordance with the ethical, legal and philosophical principles and values of the ASGC Code of Ethics	Ability to identify ethical issues that may arise in session
	Demonstrates an understanding of the ethical or legal matters related to the exchange, impact and communication of genetic information