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SUMMARY OF RECOMMENDATIONS

1. Research is a fundamental part of the delivery of clinical genetics services and should be accounted for in role descriptions and service planning, in addition to patient care.
2. Service planning should account for clinical geneticists, genetic counsellors, and any other relevant trainees, including: ● supervision and resources for students and trainees on clinical placement ● training positions with appropriate supervision and educational opportunities
3. During national census data collection, the HGSA encourages all of those qualified and trained as clinical geneticists to use the title of 'clinical geneticist' to enable the HGSA in advocating for specific Australian and New Zealand Standard Classification of Occupations (ANZSCO) codes.
4. Employers can help to uphold the voluntary regulatory system for genetic counsellors through employer Job Description Form (JDF) and position descriptions, multiemployer collective agreements (MECA)[1] , enterprise bargaining agreements (EBA) and pay scales.
5. Services should plan for and support the CPD requirements for each genetic counsellor according to the requirements of the HGSA Continuing Professional Development Policy for Genetic Counsellors, including provisions for educational leave.
6. Services should plan for, support, and fund adequate supervision for genetic counsellors according to the requirements of the HGSA Supervision Policy for Genetic Counsellors, and arrangements for supervision should be included in the genetic counsellor's employment agreement.
7. National or state position descriptions and awards for genetic counsellors should be consistent. They should provide for and include: ● recognition of HGSA Registration and the regulatory process for genetic counsellors ● recognition of FHGSA Clinical Certification, with protected time and support to achieve certification according to the HGSA Clinical Certification Policy for Genetic Counsellors ● protected time and funding for continuing education according to the HGSA Continuing Professional Development Policy for Genetic Counsellors ● protected time and funding for genetic counselling supervision according to the HGSA Supervision Policy for Genetic Counsellors ● time for relevant research within clinical roles ● recognition of senior roles across services
8. During national census data collection, the HGSA encourages all of those in the table above to use the title of 'genetic counsellor' to enable the HGSA in advocating for specific Australian and New Zealand Standard Classification of Occupations (ANZSCO) codes.
9. Individual services or health regions can get a more accurate estimate of their specific staffing needs using the service capacity data above in combination with their own referral numbers. The HGSA publishes an Excel template that can be used to make these calculations based on the WHO WISN model[2] on the HGSA website (WISN template at HGSA Policies and Position Statements). If services

[1] Te Whatu Ora, Health New Zealand. (2023). Multi-Employer Collective Agreements (MECAs) and Policies.

[2] World Health Organization. (2023). Workload indicators of staffing need. WHO Press.

have their own data for service delivery metrics (e.g. time on task) these should be used for more accurate estimates of their specific needs.
10. Adequate administrative support of at least 0.66 FTE administrative staff per FTE of clinical staff should be prioritised to enable clinical staff to practice at top of scope and minimise the current workforce gaps. Capabilities of genetic counsellors should be considered to make the most effective use of their clinical skills and scope of practice to help bridge workforce gaps.
11. In addition to mandated health recordkeeping requirements, services should account for: ● the value of information for future generations ● the importance of individual information and family information as a whole for blood relatives
12. Clear guidance and up-to-date costing studies from IHACPA are needed to support national consistency in the use of ABF codes and equity in genetic service delivery.
13. Funding models for genetic services should account for: ● Genetic test, pathology, and other necessary investigations. ● Obligations to provide services across regional or state boundaries. ● Medical and socioeconomic cost reduction and the flow-on financial benefits of genetic services to a range of hospital, health, community and social services. ● Funds for genetic counselling and clinical genetics trainees, as well as trainees from other health disciplines and medical specialties. ● Resources to teach genetic counselling, medical and other undergraduate health students. ● Sufficient funding to support adequate supervision, continuing education, and professional development ● Capacity to undertake clinical, biomedical, psychosocial and service-related research and evidence-based service improvement.

1. Purpose

The purpose of the HGSA Clinical Genetic Services Framework is to provide general information about and guidance for the structure and staffing of clinical genetic services in Australia and Aotearoa New Zealand. This document aims to inform service development and support appropriate human and financial resourcing by institutions, regional health services, and state and federal governments.

The HGSA recognises that there is great variability in many aspects of genetic service delivery across services and jurisdictions, and the content of this document is not intended to be prescriptive. The primary intention of the Framework is to provide support for services advocating to promote equitable access to quality genetic health care across jurisdictions, while remaining adaptable to local needs and a rapidly changing service delivery environment. It also aims to provide guidance around the variables for defining/measuring service delivery based on data required and requested by state and federal governments.

Clinical Standards should address the three main aspects of clinical service delivery:

- Structure: The attributes of those providing the care and the setting in which care is provided
- Process: What takes place during the provision of care
- Outcome: Capturing "if the goals of service provision have been achieved"

The Framework should evolve over time in response to changing service needs and models of service delivery and the Framework will be subject to two yearly review by the HGSA Clinical Genetic Services Committee.

2. Scope and limitations

The Framework is intended to provide general and consistent guidance across a number of regional, state, and national jurisdictions. Individual service providers will need to ensure that their standards also comply with local legislative and service delivery requirements.

The majority of clinical genetic services are based in the public health setting, which is the main focus of this document. However, there are an increasing number of private services becoming available that employ both clinical geneticists and genetic counsellors for which much of the content may be relevant.

While this document addresses the relationship between clinical genetics and laboratories providing genetic pathology services, the delivery of laboratory genetic/genomic services and genetic pathology are beyond the scope of this document.

The role of the general practitioner (GP) is central to the care of patients and their families. They manage many common genetic conditions in the population (also see 4.3 Mainstream genetics). They support individuals and families after a genetic diagnosis including follow up and coordination of care with multiple specialists, and facilitating cascade testing in the family. They are a primary source of referrals to genetic services, but there is unfortunately limited access to genetic counsellors in primary care. The delivery of primary care is beyond the scope of this document.

3. Ethical and professional standards

The ethical and professional standards set by the HGSA for its members are detailed in the HGSA Policies and Position Statements, including the HGSA Code of Ethics. As relevant, the following also apply:

For clinical geneticists and other medical providers in Australia, the Health Practitioner Regulation National Law is enacted in each state and territory, as administered by the Australian Health Practitioner Regulation Agency (AHPRA). The AHPRA National Medical Board publishes *Good medical practice: a code of conduct for doctors in Australia*[3] complementing the Australian Medical Association *Code of ethics*[4], and consistent with the World Medical Association *Declaration of Geneva*[5] and *International code of medical ethics*[6]. In New Zealand, the Medical Council of New Zealand (MCNZ) publishes a Good medical practice standard.

For genetic counsellors in Australia, the National Code of Conduct for Healthcare Workers sets out minimum standards for health practitioners not regulated by AHPRA and is enacted at the state and territory level. The HGSA publishes a specific Code of Ethics for Genetic Counsellors, which is a requirement of HGSA membership of the National Alliance of Self Regulating Health Professions (NASRHP) for the regulation of genetic counsellors.

The Australian National Safety and Quality Health Service (NSQHS) Clinical Governance Standard[7] provides a clinical governance framework to ensure safe and high-quality health care for patients and consumers. This standard aims to ensure that there are systems in place within health service organisations to maintain and improve the reliability, safety and quality of health care. This standard, together with the Partnering with Consumers Standard, set the overarching requirements for the effective implementation of all other standards. The Clinical Governance Standard recognises the importance of governance, leadership, culture, patient safety systems, clinical performance and the patient care environment in delivering high quality care.

For services engaged in research, the National Health and Medical Research Council Australian Code for the Responsible Conduct of Research[8] sets out principles and responsibilities for both researchers and institutions. Good Clinical Practice (GCP) is an international ethical and scientific quality standard for clinical trials that relates specifically to research involving human participants and is distinct from good clinical practice applied by clinicians in the care of their patients.

[3] Medical Board of Australia. (2020). Good medical practice: a code of conduct for doctors in Australia.
[4] Australian Medical Association. (2017). Code of ethics.
[5] World Medical Association. (2017). Declaration of Geneva.
[6] World Medical Association. (2022). International code of medical ethics.
[7] Australian Commission on Safety and Quality in Health Care. (2017). National Model Clinical Governance Framework.
[8] National Health and Medical Research Council. (2018). Australian Code for Responsible Conduct of Research.

In New Zealand, clinical genetic services are bound by the health sector principles in the Pae Ora (Healthy Futures) Act 2022 to embed the principles of Te Tiriti o Waitangi in service delivery[9], as articulated by the courts and the Waitangi Tribunal.

Common law and consumer protection law provide avenues of redress for aggrieved consumers and apply to all healthcare staff in the jurisdiction, including those in the private sector. The Australian Health Complaints Commissioner, Australian Competition and Consumer Commissioner, or Patient/Consumer Liaison Officer at the genetic counsellor's place of employment receive complaints, enforced under state and territory specific Health Complaints Acts. In New Zealand, the Code of health and disability consumers' rights applies under the Health and Disability Commissioner Act 1994[10]. Complaints may be directed to the individual or individuals who provided the services complained of; and any authorised to receive complaints about that provider; an independent advocate provided; or the Health and Disability Commissioner.

[9] Ministry of Health. (2022). Position statement and working definitions for racism and anti-racism in the health system in Aotearoa New Zealand.

[10] Health and Disability Commissioner Act 1994 (NZ).

4. DEFINITIONS

For more detailed definitions and information about qualifications, training, and regulation of genetic health professionals, see section 5. WORKFORCE.

4.1. Genetic and genomic health

For the purpose of this document, the term *'genetic'* is inclusive of single genes (genetics), multiple genes (genomics) and the interactions between genes and environmental or non-genetic factors that have health implications.[11] These terms may be used interchangeably in this document.

4.2. Genetic healthcare providers

Genetic healthcare providers (clinical geneticists, genetic counsellors, and other healthcare providers with specialised training in genetics) play an important role in the provision of genetic counselling and testing, and the interpretation and communication of genetic information and test results. The following are general descriptions of the types of healthcare providers working in clinical genetic services. Genetic pathologists and Advanced trainees in genetic pathology also work in laboratory settings.

4.2.1.Genetic health professionals

Genetic Health Professionals working in a clinical setting include:

- Genetic counsellors
- Clinical geneticists
- Advanced trainees in clinical genetics

4.2.2.Other genetic healthcare providers

A range of other genetic healthcare providers (medical providers/specialists, nurses, and allied health care providers with expertise and/or training in genetics) may also be employed/embedded in a genetic service to support the activities of a clinical genetic service. This includes but may not be limited to:

- Medical practitioners
    - with a primary qualification in another specialist area (e.g. oncologist), who have significant experience and training in genetic healthcare, and provide a range of genetic services (consent, test ordering and result disclosure) relevant to that subspecialty.
    - in an advanced training program (other than clinical genetics) working in a training position in a clinical genetics service.
- Genetic nurses
- Social workers

[11] Commonwealth of Australia Department of Health. (2017). National Health Genomics Policy Framework.

4.3. Mainstream genetics

The role of the clinical genetic service has changed as testing becomes more accessible and familiar to non-genetics healthcare providers and the public. Mainstream genetic testing, or "mainstreaming", is when preliminary pre-test counselling, informed consent and delivery of genetic testing and results is provided by healthcare practitioners who are not genetic healthcare professionals (including medical specialists and general practitioners), outside of a clinical genetics service, with support from genetic services as required.

However, the resources required of genetic services to provide support to mainstream programmes are significant. The current funding models for Australasian services (e.g. ABF or equivalent, see Section 9. FUNDING) may not account adequately for this workload, and therefore the capacity to provide the same level of support for the long-term is unclear.

4.4. Clinical genetic services

For the purpose of this document, clinical genetic service refers to a multidisciplinary team of genetic healthcare providers with administrative support. Clinical genetic services provide healthcare for people and families who are affected by, or are at risk of developing, a genetic health condition. This includes:

- Diagnosis (clinical and laboratory) of health conditions with a genetic basis
- Specialist genetic counselling
- Family history and risk assessment
- Advice about treatment, management, screening, surveillance, and reproductive options
- Training of genetic counsellors and clinical geneticists
- Teaching of undergraduate and postgraduate health professionals
- Resources for clients and families, health professionals and the public
- Support and advocacy for people and families impacted by genetic health conditions
- Liaison with genetic laboratories
- Participation in national and international networks/collaborations
- Contribution to expert consensus committees regarding diagnosis, management, and treatment
- Audit and participation in clinical governance as per hospital and legislative requirements
- Continuing professional development
- Research – clinical, biomedical, psychosocial and service-related
- Management and service development
- Support of local universities
- Representing the specialty of genetics on local, national, and international committees
- Providing advice and participating in the establishment, development, and delivery of screening programs

Services may provide a general genetic service and/or have several subspecialty interests and conduct clinics or research in areas such as cancer genetics, other specialty clinics, or a combination. Specialty clinics may have a jurisdictional or national referral base. Roles, responsibilities, clinic structures, and additional clinical components can vary significantly between these disciplines. Some units provide or participate in specialised

multidisciplinary clinics for specific conditions for example: familial cancer, bone dysplasias, neurodegenerative disorders, and inborn errors of metabolism.

In addition, and possibly in contrast to other areas of healthcare, clinical genetic services have a responsibility to families/whānau as well as individual clients. Extended family members may be at risk or have concerns about their own risk of developing or passing on a genetic health condition. Family involvement, with the consent and co-operation of the index client/family is central to service provision.

The breadth of service is dependent on the resources and capacity available to each service. Elements of delivering genetic services, which may be addressed over more than one consultation, include:

- Developing a shared agenda (what the clinician plans to discuss, and the client's expectations, questions, and concerns)
- Gathering specific health and family history information
- Informing the client about how their information will be stored and who will have access to it
- Making or verifying a diagnosis by history taking, physical examination and use of information obtained before or during the consultation, including genetic and non-genetic test results
- Providing information about the condition, which may include its cause, pattern of inheritance, natural history, complications, and treatment options
- Providing information about the risk of a condition affecting the client, their children and/or relatives
- Discussing the medical, emotional and social implications for the individual and family
- Considering, and discussing implications for genetic relatives
- Facilitating client and family adaptation to a genetic health condition or risk
- Presenting options, including genetic testing and reproductive options and assisting with informed decision making in a non-judgmental/patient-centred manner
- Arranging genetic tests after obtaining informed consent, which may include carrier testing, predictive testing, screening, and research testing, as well as diagnostic testing
- Conveying and explaining test results
- Referring to other health professionals as needed
- Addressing the interests of third parties
- Providing educational material and/or appropriate references
- Endeavouring to deliver and support culturally safe/equitable care to diverse groups (including Indigenous, culturally and linguistically diverse (CALD), LGBTQIA+, and disability communities).
- Offering contact with community-based support groups or persons
- Arranging a mutually agreed plan for future contact, review, and follow-up as indicated

4.4.1.Genetic counselling

The HGSA has endorsed the following definition of Genetic Counselling as a process:[12]

[12] National Society of Genetic Counselors' Definition Task Force, Resta, R., Biesecker, B. B., Bennett, R. L., Blum, S., Hahn, S. E., Strecker, M. N., & Williams, J. L. (2006). A new definition of Genetic Counseling: National Society of Genetic Counselors' Task Force report. *Journal of genetic counseling*, *15*(2), 77–83. https://doi.org/10.1007/s10897-005-9014-3

Genetic counselling is a communication process, which aims to help individuals, couples and families understand and adapt to the medical, psychological, familial and reproductive implications of the genetic contribution to specific health conditions. This process integrates the following:

- *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.*

More detail about the process of genetic counselling is available in the current literature, including:

- Psychosocial Genetic Counseling, (Weil, 2000)
- A Guide to Genetic Counseling, 2nd Edition (Uhlmann, 2009)
- Practical Genetic Counselling, 8th Edition (Harper, 2020)
- Genetic Counselling: A Psychological Approach (Evans, 2006)

4.4.2.Ongoing support and services for extended families

Genetic services are often provided in the acute setting over the short term. However, for some clients, medium to long term follow up is relevant as genetics input informs future diagnosis and/or long-term case management for individuals and their relatives. Some specialised services (for example metabolic genetics services) may provide life-long clinical care in both an inpatient and outpatient setting. They may undertake a coordinating role in health surveillance and care pathways, especially if linked to a specialised service or research interest. Ongoing care may otherwise be referred back to a specialist clinic or physician, or to the primary health care provider.

Genetic services support families at times of distress and facilitate decision making. However, this does not act as a substitute for formal psychological care or other supportive community services. Genetic services have a responsibility to refer to psychologists and social workers when indicated, and play a role in helping to connect their clients with community-based groups or appropriate individuals or families according to their needs for support.

4.4.3.Education

The application of genetic information and knowledge is now required across almost all branches of healthcare and at all professional levels. Genetic information also impacts other disciplines including, but not limited to, ethics, social science, law, politics, media and insurance. The public also now have access to a large volume of genetic health information that is quickly growing, changing, and may be of variable accuracy. Genetic healthcare providers are well suited to translate this information to be more understandable and applicable for different audiences. Genetic healthcare providers have the skills to deliver presentations or develop educational resources to support clients, families, and other healthcare providers, as well as deliver

medical/nursing training and lectures in relevant disciplines to upskill the next generation of the health workforce.

4.4.4.Research

In a rapidly developing speciality, research is an inherent part of clinical genetic service delivery, to enhance the understanding, prevention or treatment of genetic health conditions and their psychosocial consequences. Research is essential to clinical practice and is not restricted to grant funded or laboratory projects.

Recommendation:
Given the significant overlap between research and clinical care, research is considered a fundamental part of the delivery of clinical genetics services and should be accounted for in role descriptions and service planning, in addition to patient care.

5. WORKFORCE

Although supported by a range of healthcare and other professionals, core clinical genetic services are provided by genetic healthcare providers supported by administrative staff. Genetic healthcare providers may also work in other areas (research, academia, education, policy, industry and pathology). In clinical practice they provide comprehensive, person and family centred, multigenerational care across a range of multidisciplinary healthcare settings.

Recommendation:
To support building the future workforce capacity to meet increasing demand, service planning should account for clinical geneticists, genetic counsellors, and any other relevant trainees, including:

- supervision and resources for students and trainees on clinical placement
- training positions with appropriate supervision and educational opportunities

5.1. Clinical geneticist

A clinical geneticist is a medical practitioner trained in the application of the principles of human genetics to the diagnosis and management of genetic disorders. They are physicians who have undergone speciality training in genetics after completing RACP Basic Training as a physician or paediatrician (or equivalent as determined by RACP).

Clinical geneticists work with genetic counsellors and scientists to provide genetic services to the population of a defined geographical region. They also work closely with clinicians outside of the genetics service, to ensure coordinated care for the client.

Clinical geneticists assess patients who may have a genetic condition. They coordinate appropriate genetic tests and interpret the results in the context of the patient and family. They provide information about any genetic diagnoses, including their natural history, genetic basis, implications for future generations, and available options for treatment and/or reproductive intervention. They interpret complex medical information, and ensure that it is understandable and useful for decision-making in light of the person's expectations, beliefs, psychological state, and family relationships. Implications for other family members are also considered. They may be involved in or recruitment to medical therapies, treatments, and clinical trials. They may also arrange subsequent medical care in collaboration with other specialists once a genetic condition is identified.

5.1.1.Training

Most clinical geneticists are consultant physicians or paediatricians trained by the Royal Australasian College of Physicians, or others trained equivalently overseas.

Training in Clinical Genetics for physicians is accredited and supervised by the Advanced Training Committee in Clinical Genetics of the Royal Australasian College of Physicians (RACP). It requires a minimum of three years (36 months) of full-time equivalent (FTE) training that meets the training guidelines of the RACP[13].

Training includes genetics theory, assessment of patients, genetic counselling theory and practice, genetic laboratory experience and research. Specialty training covers a broad range of sub-specialties such as genetics of adult and paediatric onset genetic disorders such as inborn errors of metabolism, cancer, dysmorphology, prenatal genetics, cardiac genetics and neurogenetics.

Once training is completed the applicant must be recommended by the Advanced Training Committee (ATC) in Clinical Genetics for admission and be invited by the College to apply for Fellowship of the Royal Australasian College of Physicians (FRACP). The ATC informs HGSA of those who have achieved FRACP (Clinical Genetics) and the HGSA grants the title 'Fellow of the HGSA' (FHGSA) in Clinical Genetics.

[13] Royal Australian College of Physicians (RACP). (2023). Advanced Training in Clinical Genetics Handbook.

FHGSA (Clinical Genetics) may also be awarded to other medical providers who have not completed the FRACP Advanced Training to qualify as a specialist clinical geneticist or specialist paediatric clinical geneticist[14], but who have additional specialised training in clinical genetics. However, this **does not** entitle them to AHPRA specialist registration in clinical genetics.

5.1.2.Regulation

In Australia, a clinical geneticist will be registered with the Australian Health Practitioner Regulation Agency (AHPRA) as a medical specialist in Clinical Genetics under the National Registration and Accreditation Scheme (NRAS) and the Health Practitioner Regulation National Law (the National Law), as in force in each state and territory.

In New Zealand, a clinical geneticist will be registered with the Medical Council of New Zealand (MCNZ) with the Vocational Scope of Practice for Clinical Genetics under the Health Practitioners Competence Assurance Act.

Clinical geneticists in Australia and New Zealand are required to meet annual Continuing Professional Development (CPD) requirements through the Royal Australasian College of Physicians (RACP) in order to maintain their annual registration.

5.1.3.Industrial awards

In New Zealand, clinical geneticists, trainees, and registrars working in the public setting fall under national Multi-Employer Collective Agreements (MECA).[15]

There is no uniform award for clinical geneticists in Australia. Clinical geneticists fall under the relevant workplace, state or national award for Medical Officers/Medical Providers. There can be multiple industry awards and enterprise agreements in each state/territory that may apply to medical officers and medical providers, depending on the employer, specific role, and jurisdiction. Registered awards can be found through the Fair Work Ombudsman and registered enterprise agreements through the Fair Work Commission. There may be particular disparities for trainees and/or registrars, as there is limited capacity for negotiation. This can disadvantage services and individuals and reduce mobility between jurisdictions.

5.1.4.Title structure

Job or position titles are defined by employers and may incorporate qualifications, professional experience, or management structure. Job titles and title structures vary. The following are some of the titles currently in use by clinical geneticists and trainees:[16]

[14] Medical Board of Australia. (2018). Medical List of specialties, fields and related titles Registration Standard.
[15] Te Whatu Ora, Health New Zealand. (2023). Multi-Employer Collective Agreements (MECAs) and Policies.
[16] 2022 HGSA and Australian Genomics census of the Australasian Professional Genetic Workforce.

Clinical Geneticist	Trainee
Clinical Geneticist/Clinical Geneticist (Cancer) Geneticist/Cancer Geneticist/Metabolic Geneticist General Clinical Geneticist Staff Specialist - Clinical Genetics/Cancer Genetics/Genetics Visiting Medical Officer (VMO) - Clinical Genetics/Cancer Genetics	Advanced Trainee in Clinical Genetics Clinical Genetics Advanced Trainee Clinical Genetics Fellow Clinical Genetics Trainee Genetics Fellow Genetics Registrar

Recommendation:

The HGSA is advocating for specific Australian and New Zealand Standard Classification of Occupations (ANZSCO) codes for clinical geneticists and genetic counsellors. ANZSCO requires 300 or more individuals on the Australia Bureau of Statistics census before appointing a specific code. The 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce had 75 clinical geneticist respondents, with an estimated workforce of approximately 200 clinical geneticists. During national census data collection, the HGSA encourages all of those qualified and trained as clinical geneticists to use the title of 'clinical geneticist' to enable total numbers to be recorded with consistency and accuracy.

5.2. Genetic counsellor

Genetic counsellors combine counselling and communication skills with clinical skills in the care of their patients. They also often play expert consultative roles in multidisciplinary teams and supporting healthcare providers outside of the genetic clinic. In addition to genetic counselling as a process (4.4.1 Genetic counselling), the following definition of Genetic Counsellors as a profession was developed through a consultative process for the ASGC Strategic Plan 2023 - 2026:[17]

Genetic counsellors are postgraduate trained allied health professionals who support people and families impacted by, or at increased risk of having a health condition with a genetic basis. They integrate clinical genomic knowledge, health communication, and counselling skills to identify and meet clinical, informational, and psychological needs and promote informed decision-making.

Genetic counsellors work across all areas of health, in clinical care, research, academia, education, policy, industry and pathology. In clinical practice, they provide comprehensive, client centred, multigenerational care for families across a range of multidisciplinary healthcare settings. While genetic counselling may be part of the role of many different health professionals, trained genetic counsellors have specialist skills in genetic counselling. The HGSA uses the term 'professional genetic counselling' to distinguish genetic counselling provided by genetic counsellors from that provided by other professions (NHMRC Medical Genetic Testing: Information for health professionals, 2010). The Human Genetics Society of Australasia (HGSA) administers the training, certification, and regulation of genetic counsellors in Australia and New Zealand.

5.2.1.Training

Genetic counsellors have completed a tertiary qualification in genetic counselling. The current entry level postgraduate qualification for employment as a genetic counsellor is completion of a Master of Genetic Counselling from a program accredited or recognised by the HGSA. As part of their training, student genetic counsellors are required to undertake clinical placements in clinical genetic services. Upon successful completion of a recognised qualification, a genetic counsellor can apply to become a Member of the HGSA (MHGSA). Genetic counsellors trained, certified, or working overseas can also apply for MHGSA. MHGSA acknowledges the minimum qualification requirements to practice as a genetic counsellor. For the purposes of regulation, the HGSA Board of Censors for Genetic Counselling has adopted the National Alliance of Self Regulating Health Professions (NASRHP) definition of practice as follows:

"[Practice]…indicates that an individual is drawing on their relevant professional skills and knowledge in the course of their work to contribute to safe and effective delivery of services within the profession…. It is not restricted to the provision of direct clinical care and may also include working in a direct nonclinical relationship with clients; working in management, administration, education, research, advisory, regulatory or policy

[17] Australasian Society of Genetic Counsellors. (2023). ASGC Strategic Plan 2023 - 2026.

development roles, for example. This work can be of a paid or formal volunteer nature on a full or part-time basis."[18]

MHGSA genetic counsellors practicing in Australia or New Zealand can undertake further training towards clinical certification. Submissions over at least two years of supervised clinical practice are assessed by the HGSA Board of Censors for Genetic Counselling (BOCGC), which recommends successful candidates to the HGSA. Clinical certification is granted by the HGSA Board and certified genetic counsellors are granted the title 'Fellow of the HGSA' (FHGSA) in Genetic Counselling. FHGSA certification is a professional expectation addressed in the HGSA Code of Ethics and Scope of Practice for Clinical Genetic Counsellors.

5.2.2.Regulation

Professional regulation helps support the integrity and credibility of health professionals and health services, whilst also protecting the public from harm. It demonstrates that individual genetic counsellors have the relevant professional skills and knowledge to provide safe and effective delivery of services within their profession. The HGSA provides governance, a concerns and complaints process, and assessment for certification as administered by the Board of Censors for Genetic Counselling, whose members have suitable education, qualifications, and expertise.

In Australia and New Zealand, genetic counsellors are a self-regulating health profession under the HGSA. Self-regulation is where members of a profession establish a voluntary regulatory system through a professional association, where members are bound by the standards and codes of ethics set by the association, rather than an external licensing body or legislation. This means that genetic counsellor regulation under the HGSA is not statutory. It is intended to be complementary to and not in contradiction to other ethical and professional standards (2. Ethical and Professional Standards), or to existing legislative requirements. Detailed information about Genetic Counsellor Regulation is available on the HGSA website.

In Australia, the HGSA is a member of the National Alliance of Self Regulating Health Professions (NASRHP), the national peak body that sets benchmarked standards for self-regulating allied health professions in Australia. The Commonwealth recognises self-regulation under the *National Code of Conduct for health care workers* as an alternative for professions that do not come under the *Health Practitioner Regulation National Law*.[19]

In New Zealand the Ministry of Health Clinical Genetics Service Specification[20] requires that genetic counsellors hold HGSA certification in genetic counselling (or equivalent) and ongoing registration (maintenance of proficiency). While there is no equivalent of NASRHP, genetic counsellors are recognised as a self-regulating profession and benefit from the framework established by NASRHP membership. Work is in progress to embed

[18]National Alliance of Self Regulating Health Professions. (2016). Self Regulating Health Profession Peak Bodies Membership Standards.

[19]Australian Health Ministers' Advisory Council. (2018). AHMAC information on regulatory assessment criteria and process for adding new professions to the National Registration and Accreditation Scheme for the health professions.

[20] Manatū Hauora Ministry of Health. (2018). Specialist medical and surgical services – National Services – Clinical Genetics Service Tier 2 service specification.

HGSA Registration and FHGSA Certification into the requirements of employment within the public national genetic service and to identify and pursue additional local pathways for visibility as a self-regulated profession.

Recommendation:
Employers can help to uphold a voluntary regulatory system through employer Job Description Form (JDF) and position descriptions, multiemployer collective agreements (MECA)[21] , enterprise bargaining agreements (EBA) and pay scales.

The HGSA Board of Censors for Genetic Counselling (BOCGC) sets policies, and convenes and oversees Committees for:

- Accreditation of Masters of Genetic Counselling Programs
- Certification of Members and Fellows in Genetic Counselling
- Continuing Professional Development (CPD) for Genetic Counsellors
- Professional Practice for Genetic Counsellors

To ensure genetic counsellors have relevant professional skills and knowledge to provide safe and effective delivery of services within their profession, the BOCGC has developed a set of guidelines and policies by which to model best practice:

- The Scope of Practice for Genetic Counsellors
- The Code of Ethics for Genetic Counsellors
- English Language Proficiency for Genetic Counsellors
- Competencies for Genetic Counsellors
- Mandatory Declarations for Genetic Counsellors
- Indemnity Insurance for Genetic Counsellors
- Supervision Policy for Genetic Counsellors

The HGSA publishes an online Register of Genetic Counsellors that includes:

- ACTIVE genetic counsellors who are qualified to practice in any area of practice
- PROVISIONAL genetic counsellors who are qualified to practice in any area of practice and undertaking further training specific to clinical practice
- REGISTERED genetic counsellors who have achieved FHGSA certification for clinical practice
- Lapsed: Failed to meet or submit the requirements for annual registration
- Expired: Failed to meet or submit the requirements for annual registration over a period of three consecutive years
- Suspended: Registration suspended for a defined period of time
- Terminated: Registration has been terminated

[21] Te Whatu Ora, Health New Zealand. (2023). Multi-Employer Collective Agreements (MECAs) and Policies.

- Retired: Retired from practice

In addition to workplace and government complaints processes (see 2. Ethical and professional standards) the HGSA has established a Professional Concerns and Complaints Committee (PCCC) for receiving, overseeing, managing and resolving complaints regarding MHGSA or FHGSA genetic counsellors.

5.2.2.1. Continuing professional development (CPD)

The HGSA Code of Ethics for Genetic Counsellors states that genetic counsellors must meet current standards of practice and continue education and training, demonstrating these through engaging in the CPD program and annual CPD submissions. CPD activity is mandatory for all MHGSA and FHGSA genetic counsellors listed on the HGSA Register of Genetic Counsellors, and for FHGSA Certification.

Recommendation:

Services should plan for and support the CPD requirements for each genetic counsellor according to the requirements of the HGSA Continuing Professional Development Policy for Genetic Counsellors, including provisions for educational leave.

5.2.2.2. Supervision

The term supervision for genetic counsellors refers to both reflective practice supervision and professional supervision. Supervision encompasses educational and supportive functions, development of self-awareness, and may encompass case management functions. Supervision is included in the HGSA Competency Standards for Genetic Counsellors and Code of Ethics for Genetic Counsellors. Many healthcare organisations have a professional supervision policy for allied and mental health professionals that supports regular, individual supervision with a trained supervisor, and serves functions that include professional practice, practice development, and support for practitioners.

A Genetic Counselling Supervisor should be an FHGSA Registered genetic counsellor with sufficient experience and training to provide supervision that encompasses both genetics and counselling. In addition to supervision with an FHGSA Registered genetic counsellor, genetic counsellors should have access to workplace appropriate genetic case review and clinical consultation with clinical geneticists, other medical specialists, MHGSA or FHGSA laboratory scientists, or experienced researchers with appropriate qualifications (usually a PhD) who provide expertise in the workplace.

It has been common practice for some genetic counsellors to obtain specific counselling supervision from other appropriately qualified mental health professionals (social workers and psychologists), who meet supervisor requirements for their profession and have a working knowledge of the genetic counsellor's role. It is important to ensure access to appropriate clinical case review and consultation in these circumstances.

Recommendation:

Services should plan for, support, and fund adequate supervision for genetic counsellors according to the requirements of the HGSA Supervision Policy for Genetic Counsellors, and arrangements for supervision should be included in the genetic counsellor's employment agreement.

5.2.3.Industrial awards

Information about the industrial awards for genetic counsellors has been included as most genetic counsellors fall under a single employment award or agreement in each jurisdiction. Specific disparities across these have been identified as key professional issues for genetic counsellors, including lack of support for or recognition of certification, professional development, and supervision.

Recommendation:

National or state position descriptions and awards for genetic counsellors should be consistent. They should provide for and include:

- recognition of HGSA Registration and the regulatory process for genetic counsellors
- recognition of FHGSA Clinical Certification, with protected time and support to achieve certification according to the HGSA Clinical Certification Policy for Genetic Counsellors
- protected time and funding for continuing education according to the HGSA Continuing Professional Development Policy for Genetic Counsellors
- protected time and funding for genetic counselling supervision according to the HGSA Supervision Policy for Genetic Counsellors
- time for relevant research within clinical roles
- recognition of senior roles across services

In New Zealand until February 2022, there was a national multiple employer collective agreement (MECA)[22] specific to genetic counsellors, with a nationally standardised pay scale. From March 2023, genetic counsellors fall under the PSA Allied, Public Health, Scientific & Technical Multi Employer Collective Agreements 2022-2023 with specific variations and will be included in these MECA following the next bargaining cycle in 2023.

There is no national or uniform state award for genetic counsellors in Australia. Consequently, there is great disparity between remuneration and the recognition of training and prior experience. This could impinge upon mobility of genetic counsellors between states or area health services to avoid being disadvantaged by the lack of comparability.

[22] Te Whatu Ora, Health New Zealand. (2023). Multi-Employer Collective Agreements (MECAs) and Policies.

	Enterprise Bargaining Agreement/Award	CPD allowance (hours or funding)	Recognition of FHGSA Certification	Protected funding for Supervision	Higher degree recognition for MGC[23]
ACT	Public Sector Health Professional Enterprise Agreement	Yes-hours and funding 3 days per annum Up to 2% of base pay			
NSW	Health Industry Status of Employment (State) Award NSW Health Service Health Professionals (State) Award Public Hospitals (Professional and Associated Staff) Conditions of Employment (State) Award				Employees who have completed an undergraduate degree and a Master's degree, or other approved equivalent qualifications requiring more than four years of combined full-time study shall commence on the Level 1, Year 3 salary.
QLD	Health Practitioners and Dental Officers (Queensland health) Certified Agreement	3 days' professional development leave per annum that will accrue for up to two years. Professional development allowance paid directly as part of normal salary and included in gross earnings before tax		Supervision (professional, clinical, and operational) is recognised as a requirement for the execution of the role. regardless of level.	
SA	South Australian Modern Public Sector Enterprise Agreement: Salaried	Yes-hours and funding 5 days over 2 years Reimbursement			Yes-increment Start at AHP2 instead of AHP1

[23] Master of Genetic Counselling (MGC)

TAS	Allied Health Professionals Public Sector Unions Wages Agreement	Yes Counted as being on duty, with cover as appropriate. $1000 per annum (cumulative for up to 5 years)			
VIC	Public Health Sector (Medical Scientists, Pharmacists & Psychologists) Enterprise Agreement	Yes- hours and funding 5 days per year plus reimbursement of reasonable expenses.	Yes-required for entry into Grade 3 and above		Increment and allowance Base rate is Grade 1 year 2. MGC Sum of 7.5% of base rate. PhD (Genetic Counselling) Sum of 10% of base rate.
WA	HSUWA - PACTS Industrial Agreement	Yes-hours 16 hours per year			
NZ	PSA Allied, Public Health, Scientific & Technical Multi Employer Collective Agreements 2022-2023[24]	Variable by health region	Yes- required for progression beyond step 5. Professional association fees (HGSA and Special Interest Group Membership) are reimbursed.		Dedicated pay schedule for genetic counsellors.

[24] Correspondence "Te Whatu Ora - Health New Zealand and the Public Service Association (PSA) Te Pūkenga Here Tikanga Mahi Genetic Counsellors Variation to the Allied, Public Health, Scientific & Technical MECAs". 13 March 2023.

5.2.4.Title structure

Job or position titles are defined by employers and may incorporate qualifications, professional experience, or management structure. Job titles and title structures vary. The following are titles are in use by genetic counsellors, and some general information about how they may be defined[25].

The below are general descriptions of the use of titles for clinically practicing genetic counsellors based on the levels to which they apply in the above industrial agreements. There is some overlap, and they may not reflect specific requirements for qualifications or levels of experience, which vary between roles, services, and states. These are not intended to reflect title structures for genetic counsellors working outside of clinical genetic services.

Clinical titles in use in Australasia	Minimum qualification	HGSA Certification	HGSA Registration	Clinical experience	Description
Head of Service/Department Director Principal Genetic Counsellor Clinic Lead Team Lead Manager	Master of Genetic Counselling May have additional qualifications relating to health service management or delivery.	Fellow of the HGSA (FHGSA) Clinical Certification	HGSA Registered	5+ years	A Genetic Counsellor who also is the head of a clinical genetic service. Has responsibility for service delivery and operations including human, physical and financial resources. Used in conjunction with other Genetic Counsellor titles (see below).

[25] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.

Principal Genetic Counsellor Senior Genetic Counsellor Team Lead Manager	Master of Genetic Counselling	FHGSA Clinical Certification	HGSA Registered	5+ years	The most senior Genetic Counsellor in a team or service. Provides significant clinical leadership within the service and multidisciplinary teams. Takes a management and supervisory role for a team of genetic counsellors. Makes significant contribution to the management and strategic direction of the service.
Senior Genetic Counsellor	Master of Genetic Counselling	FHGSA Clinical Certification	HGSA Registered	>4 years[26]	Works at an advanced expert specialist or broad generalist level. Extensive clinical expertise and knowledge. and provides clinical leadership. Has the skills and experience to provide genetic counselling supervision. May have additional supervisory, educational management, or advisory responsibilities reflective of their experience.
Specialist Genetic Counsellor	Master of Genetic Counselling	FHGSA Clinical Certification	HGSA Registered	>4 years	Works in a specialty area [27] that requires clinical expertise and specific knowledge. May perform a consultative role within their professional field of specialty.

[26] A minimum of two years post-FHGSA Certification is required to be a genetic counselling supervisor

[27] For example, including but not limited to cancer, cardiac, paediatric, prenatal, etc.

Genetic Counsellor Genetic Counsellor (FHGSA)	Master of Genetic Counselling	FHGSA Clinical Certification	HGSA Registered	>2 years[28]	Has completed FHGSA Clinical Certification. Expected to perform the usual work of a professional genetic counsellor, to a more complex/sophisticated level. May be expected to play a greater role in clinical and professional activities and service development.
Genetic Associate (NZ only)	Master of Genetic Counselling	Member of the HGSA (MHGSA)[29] FHGSA Clinical Certification	HGSA Registered HGSA Provisional[30] HGSA Active[31]		The title Genetic Associate has historically been in use in New Zealand and may be used interchangeably with the title Genetic Counsellor (above) or Associate Genetic Counsellor (below).
Associate Genetic Counsellor Genetic Counsellor (MHGSA) Genetic Associate	Master of Genetic Counselling	Member of the HGSA (MHGSA)[19]	HGSA Provisional[20] HGSA Active[21]	Entry level	Has completed a two-year Master of Genetic Counselling tertiary qualification recognised by the HGSA, but has not yet completed FHGSA Clinical Certification Qualified to perform the usual professional work of a genetic counsellor. Usually practices under the clinical and professional supervision of a Senior genetic counsellor(s).

[28] A minimum of two years clinical practice is required to obtain FHGSA Clinical Certification

[29] MHGSA replaces the terms “Part 1 Certification” for genetic counsellors.

[30] Provisional status on the HGSA Register applies to genetic counsellors who have not yet made their first submission for FHGSA Clinical Certification.

[31] Genetic counsellors who have not yet made their first submission for FHGSA Clinical Certification can hold Active status on the HGSA Register of Genetic Counsellors. MHGSA or FHGSA Genetic Counsellors who are not practicing clinically may also hold Active status on the Register. Clinically practicing genetic counsellors are expected to make a first submission as soon as practicable after completing 12 calendar months of clinical practice AND >250 hours of direct clinical contact. See the HGSA Clinical Certification Policy for Genetic Counsellors for more detail.

5.3. Genetic assistant

Genetic assistants are allied health or clinical assistants who complete tasks delegated by a qualified genetic healthcare professional to support the delivery of genetic services. Genetic assistant roles exist in the United Kingdom and the United States of America, and have more recently been introduced in Australia and New Zealand. Internationally, genetic assistants are employed across a variety of work settings (including clinical, laboratory, and research).

When tasks do not require the specialised skill set of a genetic counsellor (such as coordination and administrative responsibilities), use of genetic assistants can enable genetic counsellors practice at the top of their professional scope[32]. This process of role substitution supports clinical staff across the service and helps to make the most effective use of limited genetic health professional workforce and resources.

5.3.1. Titles

Titles used may include genetic assistant, (genetic counselling) intake assistant, intake worker, intake officer, allied health assistant, clinical assistant, or genetic counselling assistant.

In the UK this role is titled “Genetic Associate”. This should not be confused with the use of the titles Genetic Associate and Associate Genetic Counsellor in Australasia, which denote qualified genetic counsellors. There are allied health assistant roles in other professions including:[33] dietetics, exercise physiology, occupational therapy, physiotherapy[34], podiatry, social work[35], and speech pathology[36].

5.3.2.Qualifications and training

Genetic assistants are often students or graduates who have completed some or all of their training in genetic counselling. They may also be healthcare professionals or administrative staff who have appropriate skills in clinical genetics and/or counselling (for example nurses or social workers with an interest in genetics and genetic health care). As part of on-the-job training, genetic assistants may benefit from observing intake calls, appointments, and other tasks to help inform their understanding of the role.

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[32] Krutish, A., Balshaw, R. F., Jiang, X., & Hartley, J. N. (2022). Integrating genetic assistants into the workforce: An 18-year productivity analysis and development of a staff mix planning tool. *Journal of genetic counseling*, *31*(5), 1183–1192. https://doi.org/10.1002/jgc4.1589

[33] Allied Health Assistants' National Association Ltd (AHANA) https://www.ahana.com.au/

[34] Australian Physiotherapy Association. (2022). Statement from the Australian Physiotherapy Association on Unregulated Allied Health Assistants. Australian Physiotherapy Association.

[35] The Australian Association of Social Workers. (2016). Position statement: Social Work Assistants. Australian Association of Social Workers.

[36] The Speech Pathology Association of Australia. (2014). Working with Support Workers. The Speech Pathology Association of Australia Limited.

5.3.3. Industrial awards

The genetic assistant role is relatively new and emerging in Australasia. Due to the variability in genetic assistant roles across clinics and jurisdictions, an overview of awards is not included in this Framework.

5.3.4.Supervision

The genetic service employing the genetic assistant should provide clear documentation and line management to guide the genetic assistant. The service should establish the competency of the genetic assistant and ensure adequate training and supervision.

Genetic assistants should receive appropriate training and supervision from clinical staff, who retain the duty of care to the client(s), remain ethically and professionally accountable, and are responsible for ensuring the genetic assistant has the necessary skill level to perform the task. The genetic service should ensure that the clinical staff supervising a genetic assistant receive adequate professional support and supervision to support appropriate delegation of tasks and supervision of the genetic assistant.

5.3.5.Scope of role

A genetic assistant is not a substitute or replacement for a professional genetic counsellor. Qualified genetic counsellors undertake training that equips them with the skills necessary to deal with the complexities of individuals, couples and families in the context of genetic assessment, risk calculation and the provision of complex genetic information.

Tasks **NOT** suitable for genetic assistants include:

- Undertaking genetic counselling appointments
- Undertaking and/or providing risk assessment
- Providing risk management information
- Clinically or psychosocially complex cases

What is considered in scope will vary between roles, services, and jurisdictions, and based on the individual's level of experience. The following task list (modified from Thoms et al, 2023)[37] is not exhaustive, but reflects some of the tasks that could be performed by an appropriately qualified, experienced, and supervised genetic assistant. An individual genetic assistant will not usually perform all of these tasks, and there may be other tasks deemed appropriate based on the service context and the genetic assistants' individual competency.

[37] Tohms, A., Krutish, A., & Hartley, J. N. (2023). Refining the activities of genetic assistants: Development of task statements applicable across practice settings. *American journal of medical genetics. Part A*, 10.1002/ajmg.a.63487. Advance online publication. https://doi.org/10.1002/ajmg.a.63487

Category	Genetic Assistant Tasks[38] [a] *Denotes tasks that may not be appropriate for entry-level genetic assistants*
Academics and Professional Development	● Participate in professional development activities (e.g., rounds, journal clubs, webinars, health and safety) ● Act responsibly and ethically in all aspects of work ● Review and synthesise literature (e.g., guidelines and new developments)
Administrative	● Scan, copy, and file documents ● Manage telephone, fax, and email inquiries ● Manage the distribution of patient records (e.g., test results, requests for records) ● Enter data ● Prepare meeting agendas, minutes, and presentations ● Create and update forms
Care Coordination	● Obtain information, records, and reports from patients, providers, and institutions ● Assist with patient flow, scheduling, and appointment coordination ● Coordinate appointment follow-up tasks (e.g., ancillary tests, referrals, and services) ● Assist with genetics documentation (e.g., test results, chart notes, consult letters) ● Attend and participate in case review meetings ● Assist with referral triage (e.g., gathering information, records, and reports)
Case Preparation	● Prepare and review genetics charts (e.g., gathering and summarising information) ● Perform literature and scientific reviews ● Enter data in risk assessment tools ● Collect and document patient and family history information ● Identify testing options and specimen requirements
Education and Outreach	● Create and revise educational materials for a variety of audiences (e.g., patients, health professionals, public) ● Collaborate with partners to disseminate educational materials ● Promote health, safety, and wellness in the workplace
Liaison	● Liaise with patients, laboratory staff, providers, and with relevant professionals, agencies, and teams on behalf of the genetics service ● Represent the genetics service on committees and at meetings
Research	● *Recruit and enrol patients in research studies (e.g., obtain informed consent)* [a] ● Maintain regulatory documentation (e.g., protocols, ethics board forms) ● Assist with quality monitoring/improvement projects ● Collect research data and track participant progress ● Coordinate activities with research collaborators (e.g., case reports, gene discovery, clinical trials, grant applications) ● Contribute to research publications/presentations
Test Results	● Assist with the curation of data and preparation of test reports ● *Share results with patients and providers* [a] ● Follow-up on, coordinate, and document the (re)classification of variants
Testing	● Track testing and follow-up on late results ● Complete paperwork and coordinate specimen collection ● Assist with sample workflow (e.g., samples/requisitions for testing and add-ons) ● Prepare and curate resources describing available tests ● Assist with offering additional tests and obtaining consent for testing ● Assist with genomic data transfer

[38] Modified from Tohms, A., Krutish, A., & Hartley, J. N. (2023). Refining the activities of genetic assistants: Development of task statements applicable across practice settings. *American journal of medical genetics. Part A*, 10.1002/ajmg.a.63487. Advance online publication. https://doi.org/10.1002/ajmg.a.63487

5.4. Other Genetic Healthcare Providers

Many clinical genetic services employ other medical, nursing, and allied health providers who have expertise and/or training in genetics but do not have AHPRA registration as a specialist clinical geneticist, or HGSA recognised training or FHGSA certification as a genetic counsellor. For example, this may include but is not limited to oncologists, cardiologists, genetic nurses, psychologists and social workers.

Job or position titles are defined by employers and may incorporate qualifications, professional experience, or management structure. Job titles and title structures vary. The following are some of the titles in use by other medical specialists working in genetics[39].

Other Medical Specialists with advanced training in Clinical Genetics
Senior Medical Practitioner Staff Specialist – Genetics/Cancer Genetics Career Medical Officer – Genetics/Cancer Genetics

5.5. Administrative

The administrative team may include (but are not restricted to) the following. Some of these roles will be client facing and require the interpersonal skills to communicate with clients and families who may be in distress.

- Administrative assistant
- Administrative officer
- Administrator
- Clinic coordinator
- Data manager
- Data entry clerk/Data officer
- Medical typist
- Personal assistant (PA)
- Secretary
- Service manager
- Receptionist

5.6. Other supporting staff

Other supporting staff may be employed within some clinical units and are an important part of service delivery. Some may be in combined roles that may overlap with clinical work.

- Researcher (e.g. clinical trial coordinator)
- Data scientist (e.g., bioinformaticians, software developers)
- Data/Database management
- Registry management

[39] 2022 HGSA and Australian Genomics census of the Australasian Professional Genetic Workforce.

6. SERVICE STRUCTURE

6.1. Geographical distribution

Genetics Services are most often based in public tertiary (or higher) health facilities located in state or regional capitals. They are closely affiliated, in both research and clinical service, to teaching hospitals, medical schools and specialised laboratory services including cytogenetics, molecular and biochemical genetics, and newborn screening. Most patients are seen as outpatients and are referred by general practitioners or specialists.

The staff of a clinical genetics service may be based in one institution or spread over multiple sites within the region served. For example, genetic counsellors working in regional centres may be supervised by a clinical geneticist and/or genetic counsellor based in the central service of that region.

Australia: Modified Monash classification. This can be searched by location using the online Health Workforce Locator.	**New Zealand: Interactive maps are available at Stats NZ urban-rural geographies (arcgis.com)**
MM1 Metropolitan areas	Major urban area – 100,000 or more residents
MM2 Regional centres: Areas that are in, or within a 20km drive of a town with over 50,000 residents.	Large urban area – 30,000–99,999 residents
MM3 Large rural towns: Areas that are in, or within a 15km drive of a town between 15,000 to 50,000 residents.	Medium urban area – 10,000–29,999 residents
MM4 Medium rural towns: Areas that are in, or within a 10km drive of a town with between 5,000 to 15,000 residents.	Small urban area – 1,000–9,999 residents.
MM5 Small rural towns	Rural settlement Rural other
MM6/7 Remote/Very Remote	N/A [note none of NZ is classified as remote due to its geographical size]

While only 10% of clinics are based in regional areas and 5% in remote areas, over 70% of clinics provide services regionally and rurally, and nearly 40% provide services to remote areas[40]. Public health funding models may not account adequately for travel, workload and funding to support services that are delivered interjurisdictionally or outside of a designated health region. This disadvantages and introduces inequities for people living in non-metropolitan areas, particularly those for whom telehealth is not appropriate, for example, in cases where a physical examination is required.

[40] 2022 HGSA and Australian Genomics census of Australasian Professional Genetics and Genomics Services.

6.1.1.National services

The New Zealand (NZ) National Service has three regional hubs in Auckland, Christchurch, and Wellington. Services are provided to the entire country through a network of metropolitan and regional outreach clinics by a specialised team of genetic healthcare professionals based in, or affiliated with, tertiary level medical services. Telehealth is used extensively to provide services throughout NZ. Each clinical genetics service is affiliated with, or has access to, the services of laboratories performing genetic tests.

6.1.2.State-wide services

There are single statewide units in Queensland, Tasmania, and Western Australia. The central service in Queensland is based in Brisbane but there are also genetic counsellors permanently stationed in regional locations, with visiting geneticists regularly delivering clinics in regional centres across the state.

South Australia has a state-wide Adult Genetics (adult general and familial cancer) and a state-wide Paediatric and Reproductive Genetics Unit. Although the units collaborate and share resources (e.g. client database), they are distinct services.

6.1.3.Regional services

In NSW, major units support locally established metropolitan and rural outreach units. Most services cover regional and remote areas through telehealth and/or outreach clinics. Major genetics units may conduct outreach clinics in regional areas and may have locally based genetic counsellors in a few regions. Outreach services in regional NSW are staffed by local genetic counsellors. NSW employs a significant number of genetic counsellors in primary health facilities such as community health centres or child and family health centres. These genetic counsellors are employed by Local Health Districts to provide a genetic counselling service, which is augmented by visiting clinical geneticists from larger genetics units.

The ACT service functions similarly to regional NSW. Genetic counsellors are employed by the local health service, which is augmented by visiting clinical geneticists from major NSW genetics units.

Services to the Northern Territory are provided from Victoria.

Clinical genetic services in Victoria are provided on a hub and spoke basis. There are four metropolitan hubs that also provide outreach clinics to other metropolitan, regional and rural hospitals. Some clinics are general, where others are condition-specific, for example cancer, cardiology or neurogenetics.

6.1.4.Outreach services

Many state-wide or regionally based clinics provide services to large geographic areas to facilitate equity of access. Community outreach (rural or metropolitan) refers to genetic services attached to and supported by a major genetics service. Outreach clinics are run differently in different jurisdictions. They may have an onsite or visiting genetic counsellor and regular clinics/consultations and clinical support from a visiting clinical geneticist.

Some outreach centres are visited by a clinical geneticist and genetic counsellor who are based in the main service. Some are staffed by visiting clinical geneticists from the main service with outreach genetic counsellors who live in the region and facilitate local contacts and follow-up.

In NZ the outreach services are provided by visiting clinical geneticist and genetic counsellor from the main centre (hub) serving that region. There are no outreach genetic counsellors in NZ.

6.1.5.Telehealth services

Telehealth, also called telemedicine or "telegenetics", refers to the use of information and communication technologies to deliver health care when patients and care providers are not in the same physical location[41]. Telehealth consultations are provided by video or telephone rather than face to face[42] and can be used for outreach cases that are urgent or do not need examination. Telehealth may be preferred by some patients[43,44] as it can minimise cost, travelling time, risks from travel, and time away from home or off work, and can help to facilitate access to clinical services for people from rural and remote locations.

However, telehealth is not always a suitable alternative for the many clinical genetic assessments requiring physical examination. Physical attendance of genetic health professionals is necessary to provide adequate and equitable service to regional and remote areas.

6.2. Multidisciplinary clinics

While individually uncommon, Mendelian disorders collectively affect a significant proportion of the population. Interactions between genetic health professionals and other medical specialists, GPs, and allied health providers is a cornerstone of multidisciplinary practice, which facilitates development of expertise in the holistic management of the unique complications of rare diseases.

The suitability of disorder specific clinics depends on multiple factors including the population serviced and local research interests. System specific clinics, for example genetic dermatology, genetic ophthalmology, skeletal dysplasia, and genetic cardiac clinics can be tailored to local expertise and facilitate continuity of service provision by genetic and other healthcare providers. A dedicated multidisciplinary facility or clinic can be an optimum way of gaining expertise in the management of rare manifestations and achieving efficiencies, including economies of scale. It may be housed within a dedicated, multidisciplinary clinic, and ideally incorporates a range of healthcare professionals, including allied health services.

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[41] National Telehealth Service. Te Whatu Ora, Health New Zealand.
https://www.tewhatuora.govt.nz/our-health-system/digital-health/national-telehealth-service/

[42]Telehealth. Department of Health and Aged Care, Commonwealth of Australia.
https://www.health.gov.au/topics/health-technologies-and-digital-health/about/telehealth

[43] Toll, K., Spark, L., Neo, B., Norman, R., Elliott, S., Wells, L., Nesbitt, J., Frean, I., & Robinson, S. (2022). Consumer preferences, experiences, and attitudes towards telehealth: Qualitative evidence from Australia. *PloS one*, *17*(8), e0273935. https://doi.org/10.1371/journal.pone.0273935

[44] Savira, F., Robinson, S., Toll, K., Spark, L., Thomas, E., Nesbitt, J., Frean, I., & Norman, R. (2023). Consumer preferences for telehealth in Australia: A discrete choice experiment. *PloS one*, *18*(3), e0283821. https://doi.org/10.1371/journal.pone.0283821

6.3. Private practice

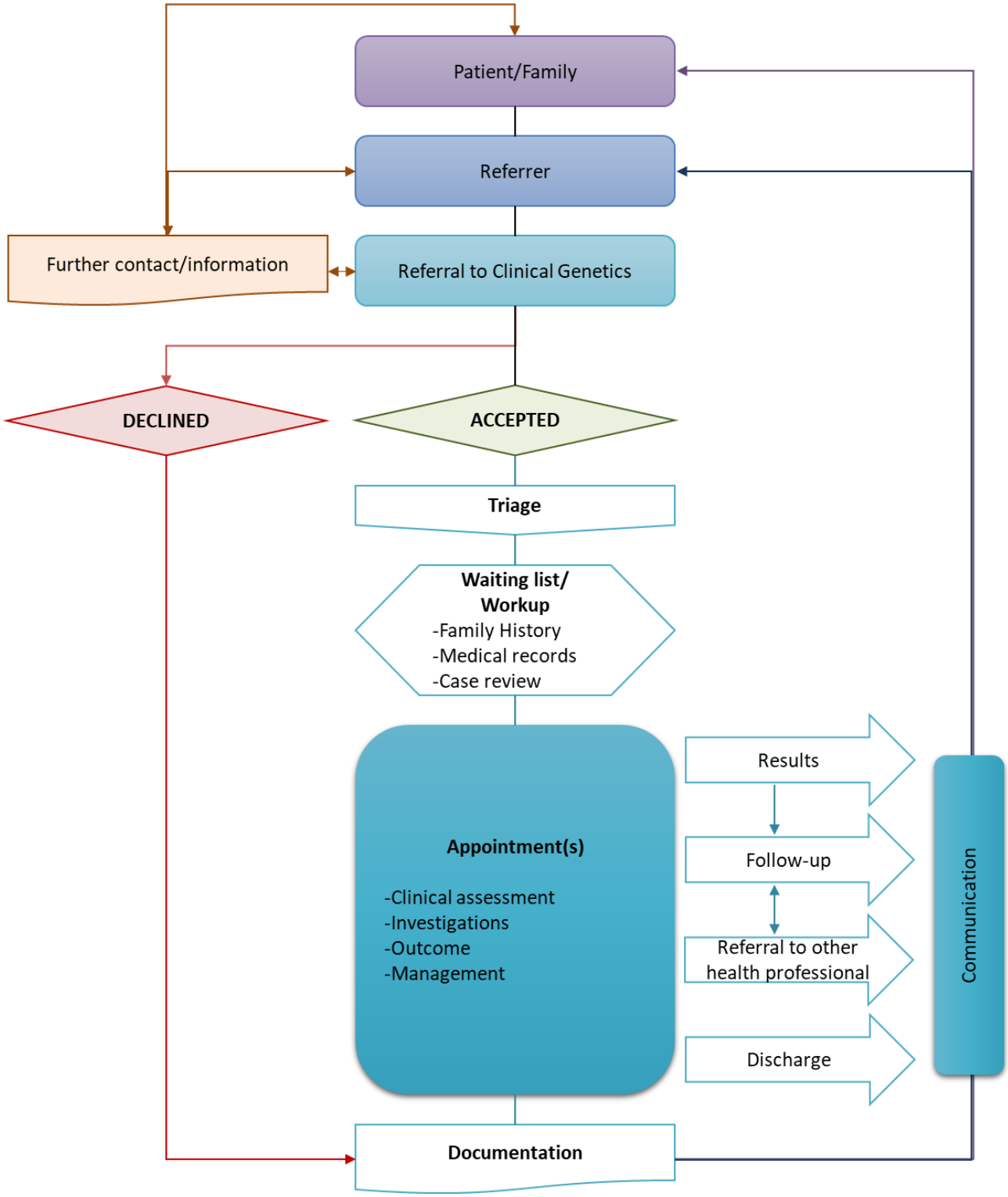
Whilst the majority of clinical genetic services sit within the public health system across Australia and New Zealand, there is increasing opportunity for the delivery of genetic services in the private sector. Models of private service delivery include working as a sole practitioner, working in partnership with other genetic healthcare providers, or working as part of a multidisciplinary team with non-genetics healthcare providers. Genetic counsellors in private practice may or may not work with a medical specialist, namely a clinical geneticist, prenatal specialist, cancer specialist, and/or other professionals.

Ethical, professional, and clinical standards, and regulatory requirements under HGSA, the Australian Health Practitioner Regulation Agency (AHPRA), and the Medical Council of New Zealand (MCNZ) do not differ between public and private services.

The HGSA publishes an opt-in listing of public and private Clinical Genetic Services. Private services are encouraged to list their clinic's details on the HGSA website so that they can be contacted about shared families within the bounds of appropriate consent and legal requirements.

7. SERVICE DELIVERY

The following is a general description of genetic service delivery. Service delivery roles and needs will be highly variable by service type and/or specialty.



7.1. Eligibility

Although there are some general guidelines for referral to genetic services and eligibility for testing (for example eviQ Cancer Genetics Reference Committee Guidelines), referral requirements, eligibility criteria, and management of ineligible referrals will vary by service depending on their scope, capacity, and jurisdictional requirements.

7.2. Triage

In the absence of national standards specifically for genetic services, the NSW Agency for Clinical Innovation triage categories for clinical genomics care (2021) and Queensland Health referral guidelines for specialist outpatient services (cancer and general genetics)[45] provide guidelines for local frameworks specific to genetic service delivery. Category A/1, B/2, and C/3 align with many Australian jurisdictions' general triage frameworks for outpatient appointments. However, there may be variation in service-specific triage models, and state/territory government access policies for specialist clinics.

In New Zealand, Clinical Genetics is classified as a Tier 2 National Service. Standards for service delivery are set out in Service Specifications[46] and underpinned by the more general national Elective Services Patient Flow Indicators (ESPIs).[47]

The descriptions for the following categories are not exhaustive. For example, urgent cases could include any case relating to an ongoing current pregnancy, or to a patient who is palliative or terminal.

Category	**NZ Clinical Genetics Service Specifications**	**Elective Services Patient Flow Indicators**
Triage	A clinical triage process[48] within a maximum of five working days from referral.	Appropriately acknowledge and process referrals in 15 calendar days or less
Urgent	Clinical response/contact within two working days	
Priority	Assessment[49] within six weeks of referral.	

[45]The State of Queensland (Metro North Health). (2022). Refer your patients: Genetics (General) and Refer your patients: Genetics (Cancer).

[46] Manatū Hauora Ministry of Health. (2018). Specialist medical and surgical services – National Services – Clinical Genetics Service Tier 2 service specification.

[47] Te Whatu Ora, Health New Zealand. (2023). Elective Services Patient Flow Indicators.

[48] The service specifications state that "All referrals received are triaged, according to the Human Genetics Society of Australasia (HGSA) standards." The HGSA does not currently publish independent triage guidelines for genetic services.

[49] Assessment includes clinical history, confirmation of family history, examination, clinical diagnosis, risk assessment and appropriate diagnostic investigations. This may include a non-contact assessment (without the patient present) where a written plan of care is developed, provided to, and discussed with the referring clinician, the patient, and their general practitioner.

Routine	Assessment[40] within four months of referral.	All patients accepted for a first specialist assessment (FSA) should be seen within four months of the date of referral.

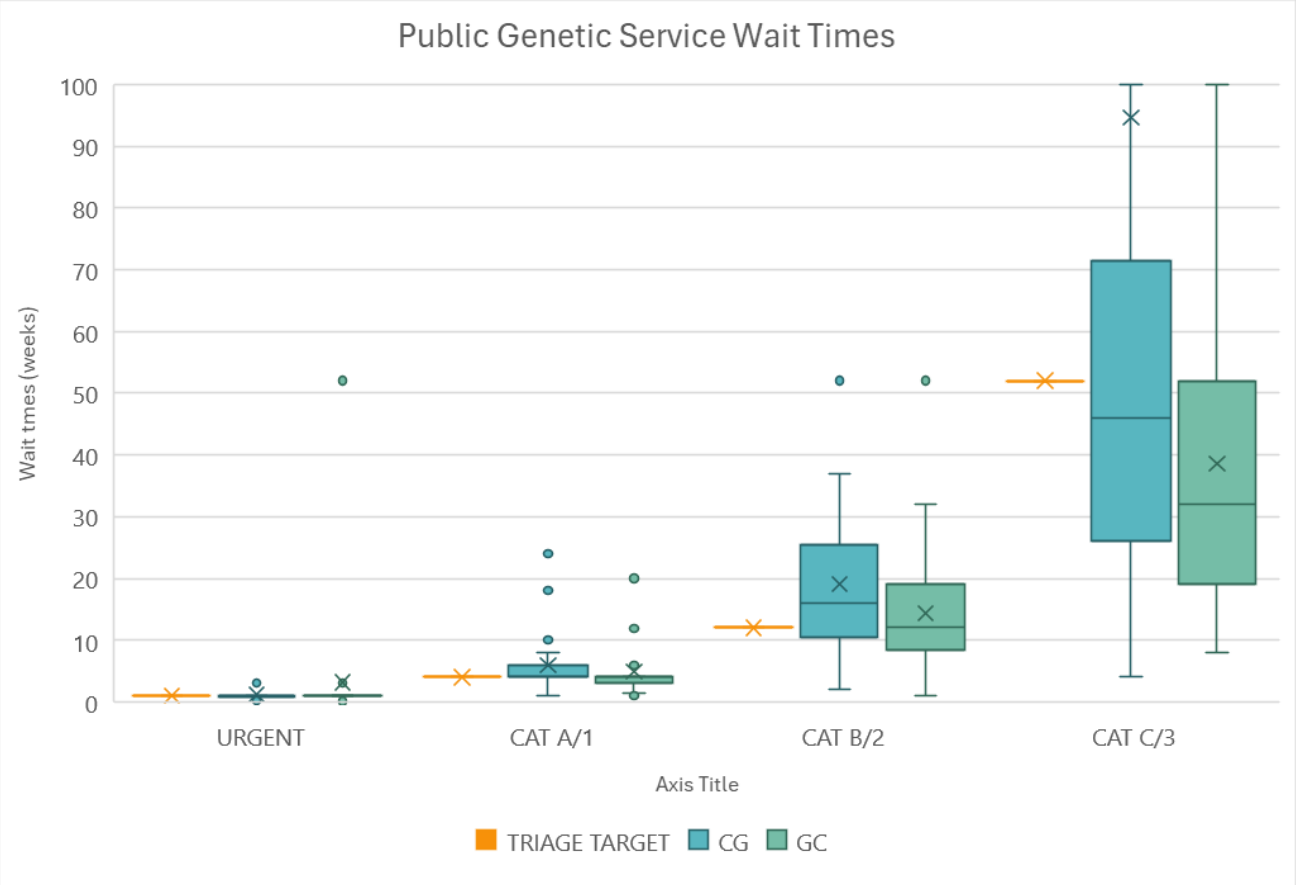
NSW Agency for Clinical Innovation: Clinical genomics prioritisation categories[50]			
Urgent	Urgent	Actioned within one week	optimal for: ● pregnant patients ● cancer patients scheduled for surgery or therapies where genetic testing will be required for immediate surgical or treatment options ● patients where results of the genetic consultation are required for urgent medical management decisions.
Category A/1	Non-urgent and ready	Appointment within 30 days	for conditions that may deteriorate if assessment is delayed, optimal for patients with: • a personal history of a genetic diagnosis and is currently on or about to go onto a palliative care pathway • any child under 12 months of age.
Category B/2	Non-urgent and ready	Appointment within 90 days	optimal for patients with conditions that may require more complex care if assessment is delayed: ● a personal or family history (genetic relatives) of a confirmed genetic diagnosis ● a personal or family history (genetic relatives) of a genetic diagnosis, where a specific gene mutation has been identified on a genetic test ● deteriorating with a genetic disorder ● child affected by a neurodegenerative genetic condition ● preconception counselling of couples who are at an increased risk of having children with a genetic condition because of a personal and or ● any family history (genetic relatives) of a known or suspected genetic condition and or consanguinity ● any child under 6 years of age.
Category C/3	Non-urgent and ready	Appointment within 365 days	for conditions unlikely to deteriorate quickly, optimal for patients with: ● A personal and or any family history (genetic relatives) of a genetic diagnosis, where a specific gene mutation has not been identified on a genetic test.

[50] Based on NSW Agency for Clinical Innovation. (2021). Clinical genomics Model of Care: Organisational Model of Care. Adapted from GL2019_011 Outpatients Services Framework and PD2012_011 Waiting Time and Elective Surgery Policy.

7.3. Waitlists

The 2022 HGSA census[51] saw a range of wait times for Australasian clinical genetic services. The survey was not limited by clinical specialty and included clinics delivering services in cancer, cardiac, cleft, dermatological, hearing loss, neurological, metabolic, paediatric, personalised, pharmacological, prenatal, population, endocrine, renal, reproductive, skeletal dysplasia, and thalassaemia as well as general genetics clinics. This was a voluntary survey and therefore there may be gaps in the dataset. Averages are presented below and may not reflect individual or specialised service wait times. Private services for both genetic counsellors and clinical geneticists have much shorter wait times, on average approximately four weeks.

The wait time for a triage Category C/3 patient to see a clinical geneticist had an average of 95 weeks (over 23 months or nearly 2 years) in the public system. Thirteen services had a wait time of 12 months or longer, and three services with a wait time of two years or longer. The wait time to see a genetic counsellor in the public clinical genetic services had an average of 39 weeks (about 9 months) for a triage Category C/3 patient (see 7.2 Triage). Ten services reported a wait time in excess of one year.



The services with greater than one year wait times are across numerous jurisdictions, and cover rural, regional and metropolitan areas.

[51] 2022 HGSA and Australian Genomics census of Australasian Professional Genetics and Genomics Services.

7.4. Staffing

Historically, the HGSA considered staffing for various levels of service as outlined below.

Level 1	● Staff able to arrange on site clinics with visiting clinical geneticists/genetic counsellors. ● Phone consultation with a clinical geneticist/genetic counsellor is available.
Level 2	● Services provided by a genetic counsellor. ● Counselling and diagnostic services provided by visiting clinical geneticists. ● Phone consultation with a clinical geneticist/genetic counsellor is available. ● Access to interpreters. ● Access to pathology services for diagnostic purposes.
Level 3	Services as level 2 *plus* ● Clinical genetics services provided by a less than full-time clinical geneticist. ● Clinical geneticist and/or genetic counsellor on call service usually available.
Level 4	Services as at level 3 *plus* ● Service operates as a separate service providing outreach referral services. ● Full-time clinical geneticist with after-hours availability. ● Access to relevant specialists e.g. oncology, neurology, gastroenterology and cardiology as required by disease diagnosis. Additionally, a level 4/5 service may provide positions for: ● Trainee clinical geneticists, trainee genetic counsellors and trainee medical specialists. ● Other supporting staff as outlined in Section 5.5
Level 5	Services as at level 4 *plus* ● May provide statewide or national expertise in a specific disorder or disorders ● Usually co-located with genetics laboratory services.

Provider-population estimates in the literature for genetic health professionals are limited, out of date, or based on anecdote[52,53]. This has been compounded by increasingly rapid developments in genetics, and changes in demand and models of care. Calculating and presenting workforce needs as staffing requirements (X number of providers per 10 000 population), facility-based staffing standards (X number of providers for a type of health centre), or based on health needs or health objectives, all have limitations.

The following estimates for clinical geneticists, genetic counsellors, and administrative staff are based on the World Health Organisation (WHO) Workload indicators of staffing need (WISN). The WISN method has been developed to assess both workforce needs and workload pressure to estimate staff requirements.[54]

[52] Dragojlovic, N., Borle, K., Kopac, N., Ellis, U., Birch, P., Adam, S., Friedman, J. M., Nisselle, A., GenCOUNSEL Study, Elliott, A. M., & Lynd, L. D. (2020). The composition and capacity of the clinical genetics workforce in high-income countries: a scoping review. *Genetics in medicine: official journal of the American College of Medical Genetics*, *22*(9), 1437–1449. https://doi.org/10.1038/s41436-020-0825-2

[53] Hoskovec, J. M., Bennett, R. L., Carey, M. E., DaVanzo, J. E., Dougherty, M., Hahn, S. E., LeRoy, B. S., O'Neal, S., Richardson, J. G., & Wicklund, C. A. (2018). Projecting the Supply and Demand for Certified Genetic Counselors: a Workforce Study. *Journal of genetic counseling*, *27*(1), 16–20. https://doi.org/10.1007/s10897-017-0158-8

[54] World Health Organization. (2023). Workload indicators of staffing need. WHO Press.

7.4.1.Service capacity

Both genetic counsellors and clinical geneticists estimated approximately 4.4 hours on average spent on clinical activity per patient over their entire patient journey.[55]

This includes but is not limited to:

- Preparation for new referral (triage, intake call, retrieving records, determining options, appointment booking, etc.)
- First appointment (telehealth or face-to-face)
- First appointment follow-up (coordinating testing, completing forms, case notes and letters, referrals, psychosocial follow-up, etc.)
- Preparation for Review/Results appointment (reviewing/interpreting results, reviewing records, etc.)
- Review/Results appointment (telehealth or face-to-face)
- Further follow up activities (case notes and letters, referrals, psychosocial follow-up, coordinate further testing/management, summary letters, etc.)

Limitations of this data are that it is based on average and estimates that may be subjective, and would be variable depending on patient needs, complexity and type of referral, clinical specialty, experience of the practitioner and/or type of test. Service capacity calculations also need to account for the amount of time spent on non-clinical activities, including but not limited to meetings, administration, data entry, research, management, and professional development. This would also include activities like supporting mainstreaming programs (see 4.3 Mainstream Genetics) that require significant service resources but are often not adequately accounted for in funding models (such as ABF or equivalent, see Section 9. FUNDING) or service and workforce planning.

Based on the workforce survey responses,[56] clinical staff spend approximately half their time on clinical activities (46% for genetic counsellors and 50% for clinical geneticists) and spend an average of 4.4 hours for clinical activities per new patient referral (including all appointments, preparation, review and follow-up). Accounting for weekends, holidays, and leave allowances, there are approximately 1618 working hours across approximately 46 working weeks per year. Using this data, genetic counsellors have 744 available clinical hours and clinical geneticists 809 available clinical hours per year per full time equivalent (FTE). This equates to a capacity for 170 new patient referrals per FTE genetic counsellor and 186 new patient referrals per FTE clinical geneticist per year, or about 4 new referrals per FTE per working week for both genetic counsellors and clinical geneticists. This capacity will depend on other factors specific to the service, like the availability of administrative support, physical space and scheduling for appointments, and other resources.

Service capacity calculations also need to account for additional activities that may be undertaken by only some individuals on the team, such as supervising (students, trainees, other staff), management, research, and additional training/qualification (e.g. certification or advanced training). For staff delivering clinical and support activities, an additional 0.12 FTE per genetic counsellor FTE and 0.13 FTE per clinical geneticist FTE need to be included to account for these activities:[57]

[55] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.
[56] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.
[57] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.

7.4.2.Workforce needs

Based on reported referral numbers and population catchment, the estimated referral load for is 0.003 genetics referrals per year per capita.[58] Limitations of this data are that population catchments were estimated, and referral numbers could be variable depending on the type of service and clinical specialty.

The number of patients seen[59] and number of appointments delivered[60] were not used due to the potential artifice (for example as a result of fixed clinic schedules, referral allocation, or eligibility criteria resulting in patients not being provided an appointment).

Based on service data of the appointments delivered[61] 59% of referrals are seen by a genetic counsellor and 41% by a clinical geneticist. While this factor is necessary to consider the workforce needs for each type of professional, confounders including clinic scheduling, referral allocation, and billing models may differ between services and may not be an accurate reflection of the number of referrals that may be appropriately seen by each type of professional.

Estimated staffing needs have been calculated below based on the reported service capacity data, population data[62, 63] and estimated referral load per population.

	AUS	*ACT*	*NSW*	*NT*	*QLD*	*SA*	*TAS*	*VIC*	*WA*
Projected referrals PY	**78,805**	*1,383*	*24,716*	*750*	*16,135*	*5,503*	*1,715*	*20,113*	*8,476*
CG FTE	**173.6**	*3.0*	*54.4*	*1.7*	*35.5*	*12.1*	*3.8*	*44.3*	*18.7*
GC FTE	**349.6**	*6.1*	*109.6*	*3.3*	*71.6*	*24.4*	*7.6*	*89.2*	*37.6*
Admin FTE	**346.6**	*6.1*	*108.7*	*3.3*	*71.0*	*24.2*	*7.5*	*88.4*	*37.3*

	NZ	*Northern*	*Te Manawa Taki*	*Central*	*Te Waipounamu*
Projected referrals PY	**15,471**	*5,765*	*3,073*	*2,928*	*3,606*
CG FTE	**34.1**	*12.7*	*6.8*	*6.4*	*7.9*
GC FTE	**68.6**	*25.6*	*13.6*	*13.0*	*16.0*
Admin FTE	**68.0**	*25.4*	*13.5*	*12.9*	*15.9*

It is also important to consider the distribution of service, and the workforce needed to serve remote and rural populations. Telehealth appointments are not adequate to provide services in many circumstances. Nearly one third (27%) of the clinical workforce are needed to serve and provide equitable access to these areas based on Australian Bureau of Statistics data on regional and remote populations:[64]

	Total regional/ remote	*Regional*	*Remote*
Projected referrals PY	**21,660**	*20,166*	*1,494*
CG FTE	**47.7**	*44.4*	*3.3*
GC FTE	**96.1**	*89.4*	*6.6*
Admin FTE	**95.3**	*88.7*	*6.6*

[58] 2022 HGSA and Australian Genomics census of Australasian Professional Genetics and Genomics Services.

[59] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.

[60] 2022 HGSA and Australian Genomics census of Australasian Professional Genetics and Genomics Services.

[61] 2022 HGSA and Australian Genomics census of Australasian Professional Genetics and Genomics Services.

[62] Australian Bureau of Statistics (June 2022). National, state and territory population.

[63] Stats NZ Tatauranga Aotearoa (30 June 2022). National population estimates.

[64] Australian Bureau of Statistics (2021-22). Regional population.

Recommendation:

Individual services or health regions can get a more accurate estimate of their specific staffing needs using the service capacity data above in combination with their own referral numbers. The HGSA publishes an Excel template that can be used to make these calculations based on the WHO WISN model[65] on the HGSA website (WISN template at HGSA Policies and Position Statements). If services have their own data for service delivery metrics (e.g. time on task) these should be used for more accurate estimates of their specific needs.

7.4.3.Workforce gap

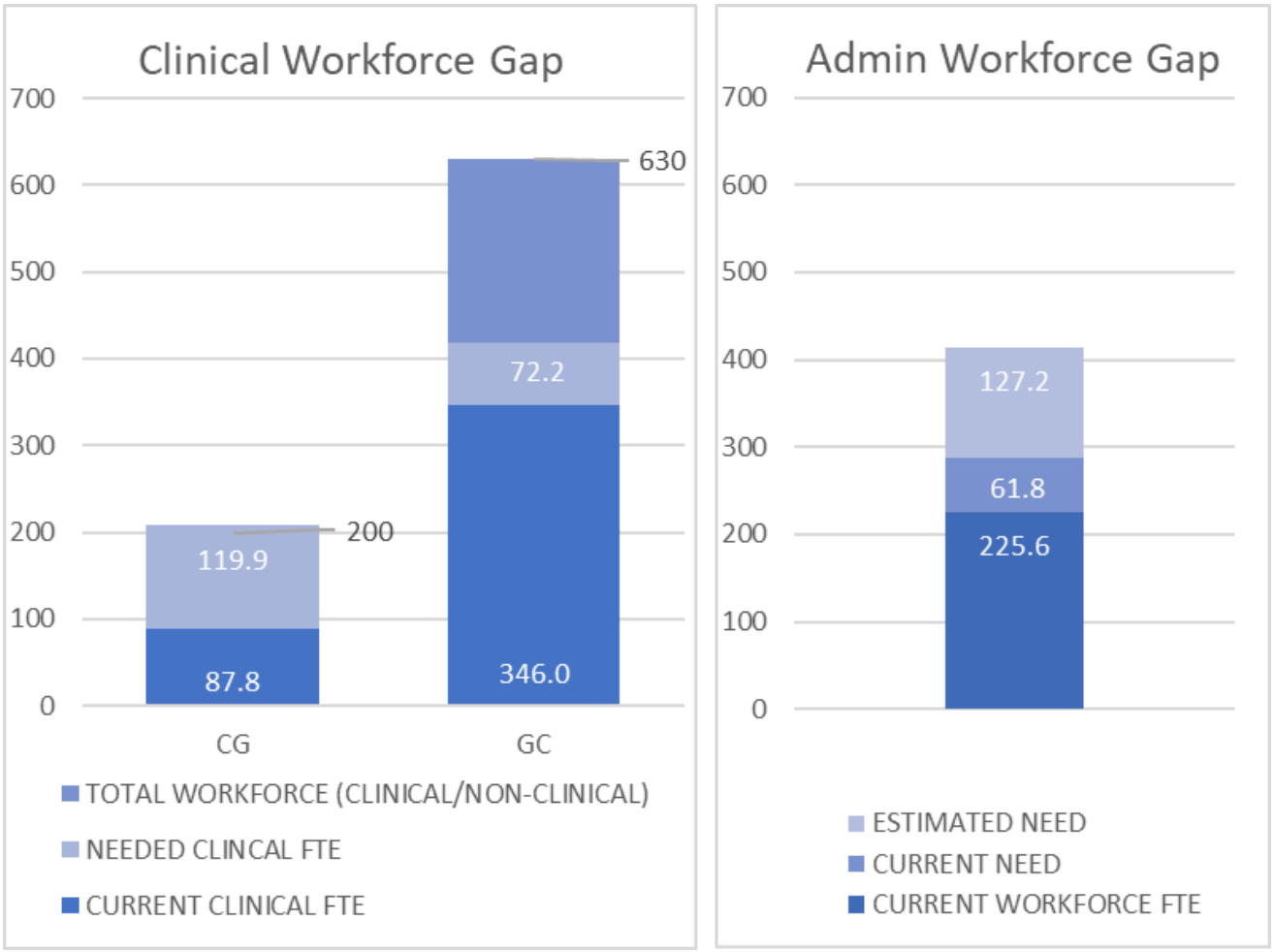
In considering the data above, it is important to recognise that it represents an ideal situation where all patients referred are provided a clinical service. The current reality is that increasingly stringent triage and eligibility criteria are being applied to referrals to manage waitlists. In addition, many services are seeing more patients than our estimates of clinical capacity would suggest. Differences in service resourcing (like administrative support) could be one explanation, or reduced time on other activities that, although non-clinical, are an important part of service delivery (like professional development, research, supervision of students and trainees, etc.). In addition, 84% of genetic counsellors and 93% of clinical geneticists report working extra hours above their regularly contracted hours[66].

Extrapolating on the 2017 HGSA and Australian Genomics Professional Status Survey of Genetic Counsellors and Clinical Geneticists[67] the current size of the potential clinical genetic healthcare professional workforce (including those trained but not practicing) is 200 clinical geneticists and 630 genetic counsellors. However, not all of those qualified are currently in clinical practice, and many work part-time (less than 1.0 FTE). A significant proportion of clinical staff time (17% for genetic counsellors and 10% for clinical geneticists) is also spent on administrative activities.[68] To make the most effective use of a limited clinical workforce, it is important to ensure that clinical staff have adequate support to enable them to practice at top of scope.

On average, clinical genetic services employ 0.52 FTE administrative support staff per clinical FTE.[69] This includes data management and genetic assistant support, which have been identified as critical roles in supporting clinical staff. To account for this, an estimated 0.66 FTE of administrative support is needed per FTE of clinical staff, specifically data management and genetic assistant support. This leaves an estimated workforce gap of 61.8 additional FTE of administrative support to meet the estimated current need across Australia and New Zealand. Improving administrative support, would increase the clinical capacity of clinical staff and genetic services.

[65] World Health Organization. (2023). Workload indicators of staffing need. WHO Press.
[66] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.
[67] Nisselle A, Macciocca I, Mackenzie F, Dunlop K, Metcalfe S & Gaff C, for the Australian Genomics Workforce & Education Working Group. (2018). Professional Status Survey of Genetic Counsellors and Clinical Geneticists.
[68] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.
[69] 2022 HGSA and Australian Genomics census of Australasian Professional Genetics and Genomics Services.

There are an estimated 87.8 FTE clinical geneticists and 346 FTE genetic counsellors in clinical practice in Australasia. A workforce gap of 119.9 FTE clinical geneticists and 72.2 FTE genetic counsellors is estimated across Australia and New Zealand. Clinical geneticists are the smallest workforce, largest workforce gap, and currently have the most limited workforce pipeline.



This is compounded by two thirds (61%) of clinical geneticists planning to retire in the next 10 years (compared to 11% of genetic counsellors).[70] It is likely that the service delivery data does not accurately reflect how many referrals could be appropriately managed by genetic counsellors, as referrals may be allocated to a clinical geneticist due to non-clinical factors such as clinic scheduling, availability, or billing.

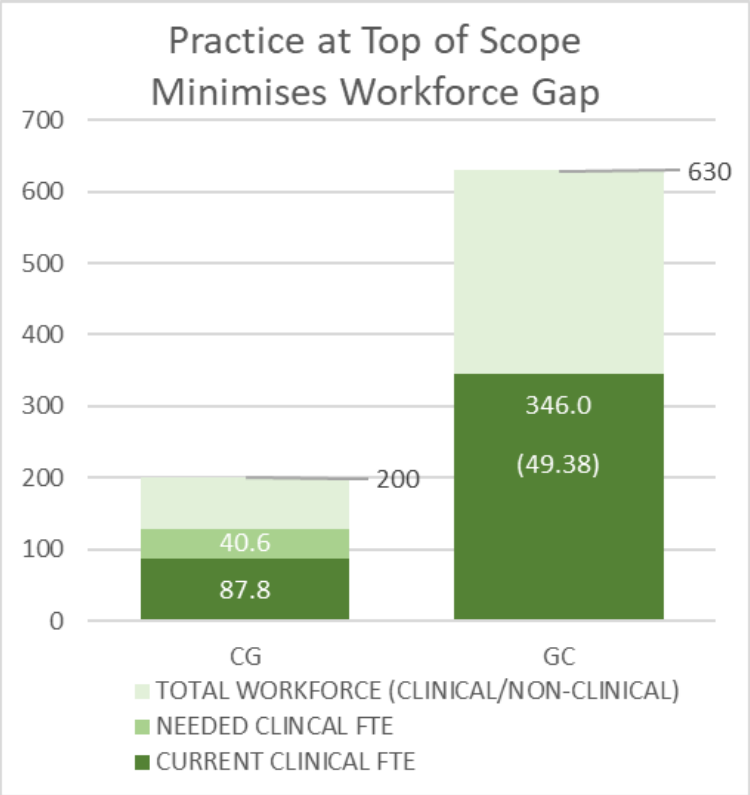
Recommendation:

That adequate administrative support of at least 0.66 FTE administrative staff per FTE of clinical staff be prioritised to enable clinical staff to practice at top of scope and minimise the current workforce gaps.

Consideration of the capabilities of genetic counsellors to make the most effective use of their clinical skills and scope of practice could in turn alleviate pressure on the smaller clinical geneticist workforce, helping to bridge the workforce gaps.

[70] 2022 HGSA and Australian Genomics census of Australasian Professional Genetic Workforce.

Adequate administrative support has the potential to address the genetic counsellor workforce gap. The available genetic counsellor FTE would then have the potential to offset the clinical geneticist workforce gap, resulting in a reduced clinical geneticist workforce gap of 40.6 FTE across Australia and New Zealand.



7.5. Consent

Informed consent is essential to the appropriate delivery of clinical genetic services. Services should be aware of organisational, national and state legal requirements that apply in their jurisdiction. A nationally consistent consent form is used in New Zealand, which is routinely reviewed, and there is an ongoing initiative to adopt and endorse standard wording for consent in Australia.

Examples of genomic consent forms include the Australian Genomics National clinical consent forms and the NSW Ministry of Health for the Australian Health Minister's Advisory Council (AHMAC) Project Reference Group on Health Genomics National Model of Consent for Clinical Genomic Testing. Both have supporting patient resources. While these may provide a valuable resource and framework for consistency, services should ensure appropriate approval of consent forms at a local level. A limitation of existing models is that they do not address Data Sovereignty.

7.6. Testing

Clinical genetics services must be able to facilitate genetic testing. A close working relationship between the services and the laboratories providing testing facilitates appropriate test requests, supports result interpretation, and allows for clinical prioritisation of requests. Clinical genetics units are generally situated in, or closely affiliated with, an institution which provides a comprehensive range of these services.

There are some evidence-based consensus guidelines for genetic testing (for example the eviQ Cancer Genetics guidelines for genetic testing), and some Medicare Benefits Schedule (MBS) item numbers for genetic tests for patients meeting specific criteria. However, eligibility may be specific to the individual service, their budget, and funding model (see FUNDING below). When public funding is not available, consideration should be given to facilitating access to and interpretation of self-funded options.

There are a variety of genetic tests that may be offered to clients. The choice of which test is the most clinically appropriate should be determined as part of the genetic assessment process. The client/family should be informed about the nature of the sample required, appropriateness of the test, the information the test is seeking, the limitations of the test, and the possible results and their implications. It is important that clients have an estimated timeframe for the result and arrangements for providing the results should be agreed to by the client/family beforehand. Clients and families should be made aware of data and sample storage, use and governance.

7.6.1.Laboratory services

In order to provide comprehensive service to clients, clinical genetics units may work with laboratories in the public or private sectors. Samples for testing in a clinical diagnostic setting should be referred to accredited laboratories. Turnaround times for results can be variable and need to be included along with waitlists when considering adequate and appropriate timeframes for the delivery of clinical care.

Guidelines for the accreditation of public and private laboratories are not addressed in this document. In Australia, laboratory services are accredited by the National Association of Testing Authorities (NATA) and in New Zealand by International Accreditation New Zealand (IANZ), both under the National Pathology Accreditation Advisory Council (NPAAC) Requirements.

Services making use of overseas laboratories should ensure that they meet comparable local accreditation requirements (for example Clinical Laboratory Improvement Amendments (CLIA) in the USA). They should also be aware of data and sample storage, use and governance, processes for result review and reclassification, and be able to communicate this with clients and families.

7.6.1.1. Research laboratories

In some cases, a research laboratory may be the only available option or may be in the best interests of a client. It is common practice to confirm research results in a clinically accredited laboratory. All human genetic research requires approval from a Human Research Ethics Committee(s).

The NHMRC's National Statement on Ethical Conduct in Human Research[71] provides guidelines for genetic testing performed as part of a research study. In New Zealand, the *Te Nohonga Kaitiaki*[72] guidelines apply to the planning and execution of genomic research.

Some research samples are collected on the understanding that no results will be given to individual participants, and this must be made clear to participants in advance of the consent process. Where individual results are to be disclosed to participants, research groups would benefit from the involvement of a genetic counsellor, preferably before consent to the research is obtained, but especially in association with result disclosure.

7.6.2.Types of test

National Pathology Accreditation Advisory Council (NPAAC)[73] defines Level 2 and Level 3 DNA tests as those that have the potential to lead to complex clinical issues, for which specialised knowledge is needed for the DNA test to be requested, and for which professional genetic counselling and written informed consent should precede and accompany the test. The term 'professional genetic counselling' distinguishes genetic counselling provided by professional genetic counsellors from that provided by other professions (NHMRC, 2010)[74].

NPAAC also defines as Level 2 tests:

- where guidelines developed by the National Health and Medical Research Council or a national medical specialty college recommend pre-test genetic counselling and written consent
- a clinically affected child being tested for a disorder that typically presents in adulthood (see 7.5.2.4. Predictive and Presymptomatic testing, below

7.6.2.1. Diagnostic test

A diagnostic test is usually ordered by a medical provider to make or confirm a suspected diagnosis, or to exclude a differential diagnosis. An increasing number of diagnostic genetic tests are being ordered by non-genetics health professionals through mainstreaming, which will still require professional genetic counselling in the event of a positive or complex result.

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[71] National Health and Medical Research Council, Australian Research Council and Universities Australia. (2023). National Statement on Ethical Conduct in Human Research.

[72] Hudson M, Thompson A, Wilcox P, Mika J, Battershill C, Stott M, Brooks RT, Warbrick L. (2021). Te Nohonga Kaitiaki Guidelines for Genomic Research on Taonga Species. Te Kotahi Research Institute. University of Waikato, Hamilton New Zealand. Genomics Aotearoa. Te Kotahi Research Institute. 1-84.

[73] Australian Commission on Safety and Quality in Health Care. (2022). Requirements for medical testing for human genetic variation. 3rd ed.

[74] National Health and Medical Research Council, Australian Research Council and Universities Australia. (2010). Medical Genetic Testing: information for health professionals.

7.6.2.2. Carrier testing

A carrier test is primarily used to determine if an adult, with or without symptoms, has a genetic variation (also called a mutation), which increases the chance that their children will have the disorder in question. Minors would generally only have carrier testing performed when the resulting information will be used to help with their health management in the immediate future. However, there are some well-established screening programs (see 7.6.2.3 Screening tests, below) that perform carrier testing in minors (for example the Wolper Jewish Hospital Community Genetics Program High School Genetics Screening Program).

The HGSA publishes a position statement on Genetic Carrier Testing for Recessive Conditions and endorses the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) policy on Genetic Carrier Screening.

7.6.2.3. Screening tests

Screening tests are usually tests performed on a specific population, which provide a personalised risk assessment, for example blood-spot screening in newborns, or prenatal screening in pregnant people. When performed prenatally, screening tests, such as ultrasound +/- biochemical analysis may reveal an increased risk of fetal anomaly.

Genetic screening may also be available to specific 'high risk' groups in order to clarify individual status. For example, carrier and cancer predisposition screening for founder variants offered to people of Jewish ancestry.

The HGSA endorse the Royal Australian and New Zealand College of Obstetricians and Gynaecologists (RANZCOG) policy on Genetic Carrier Screening and the Position Statement from the International Society for Prenatal Diagnosis on the use of non-invasive prenatal testing for the detection of fetal chromosomal conditions in singleton pregnancies.

NPAAC defines expanded carrier and couple reproductive risk screening, for conditions for which there is no specific treatment as Level 2 tests.

7.6.2.4. Presymptomatic and Predictive Testing

Predictive tests are performed on an individual for a variant that has been identified in one of their blood relatives. Predictive tests are presymptomatic if the individual has no symptoms at the time of testing.

Predictive or pre-symptomatic DNA testing in an unaffected person for conditions for which there is no simple treatment would usually be defined as Level 2 tests by NPAAC, as specific consent and counselling issues are associated. NPAAC includes testing an apparently unaffected child or foetus for a mutation already defined in the family as a Level 2 test. The HGSA publishes a position statement on Predictive and Pre-symptomatic Genetic Testing in Adults and Children.

7.6.2.5. Whole genome/exome/transcriptome analysis

NPAAC defines whole genome/exome/transcriptome analysis and large gene panels for complex conditions affecting multiple systems as Level 3 tests due to the complexity of the analysis and probability of incidental/secondary/unsolicited findings (including family relationships). The laboratory is required to maintain documentation of specific consent requirements.

7.7. Documentation

Local health service and legislative requirements for record keeping should be followed. Health professionals must comply with national and/or state privacy legislation and only disclose confidential information as permitted/required by law. With the client's consent, copies of relevant correspondence and reports should be sent to their current healthcare provider(s) including the referring provider and their primary care general practitioner.

The HGSA has published a position statement on the Use of Human Genetic and Genomics Information in Healthcare Settings.

Recommendation:
In addition to mandated health recordkeeping requirements, clinical genetic services should account for:

- the value of information for future generations
- the importance of individual information and family information as a whole for blood relatives

8. SERVICE DATA

The HGSA is often asked to advocate for and provide information about clinical genetic services in Australia and New Zealand. The data needed to help do this is now being captured by the biennial workforce surveys administered by the HGSA Workforce Surveys Steering Committee. Most services already report data to hospitals, health regions, and government health departments.

Services should be able to collect and report on the following service metrics, which are not exhaustive. The most common queries are about wait lists and wait times, and the "service gap" between the number of referrals received and appointments delivered. Service gaps may be underestimated with the implementation of referral criteria.

Capacity constraints and waitlist management have meant the implementation of referral and eligibility criteria for access to many public hospital clinical genetic services. The application of referral criteria has implications for the ability to measure the true demand for genetic counselling and clinical genetic services and the reduced access for patients who would have historically been able to access an appointment in a public hospital clinical genetic service.

8.1. SERVICE METRICS

There are a number of service metrics that can be valuable in estimating workload and workforce demands at the service, state, regional, and national level. These metrics are based on information that has been requested from the HGSA by health services, states, and federal government to inform workforce measurement and planning, and which informed the development of the HGSA and Australian Genomics census of the Australasian Professional Genetic Workforce and Professional Genetics and Genomics Services. These metrics are outlined on the following page.

Where available, time on task data is needed for workforce estimations like the World Health Organisation Workforce Indicators of Staffing Need (WISN). Services should consider how they can collect and report this data.

Health service activities	The time necessary for a well-trained, skilled and motivated worker to perform clinical activities to professional standards in the local circumstances (mins per patient/activity OR rate of working i.e. activities per day/week/month): • types of clinical activities (e.g. appointments, follow up, phone, letters) • Include preparation and follow-up (e.g. notes, documentation)
Support activities	X hours per working day OR as X% of working time: • Performed by all members of the staff type • e.g. Recording and reporting, meetings, CPD
Additional activities	X hours per working day OR as X% of working time: • Performed only by certain (not all) members of the staff type. • e.g. supervision of students/other staff, management

SERVICE METRICS	
Patient Demographics	• Age • Sex • Post code • Country of birth • Indigenous status and self-identified ethnicity
Staffing	• Headcount • FTE • Contract type (permanent/temporary/casual) • Employer sponsored visas • Seniority levels (headcount and FTE) • Funding for roles (e.g. temporary grant funded vs permanent health department funded)
Vacancies	• FTE and seniority level of advertised positions for each type of staff member • Contract type (permanent/temporary/casual) • Vacancies (unfilled/filled/filled by an alternatively qualified candidate to what was advertised)
Triage	• Definitions of clinical triage categories • Recommended appointment timeframe for each triage category

Referrals	• Number of referrals received (inpatient/outpatient, adult/paediatric where possible)
	• Number of appointments delivered
	  ○ New/review appointments (to be able to determine service gap between number of referrals received and number of appointments delivered)
	  ○ Genetic counsellor/Clinical Geneticist/co-consult
	  ○ mode of delivery (face to face vs telehealth phone/online)
	• Patient specific advice by letter/email only
	  ○ in NZ classified as “non-contact” advice
	  ○ classified as clinical activity under ABF in Australia

Waitlists	• Date of receipt of referral to appointment date • Actual wait time for each triage category • Wait time for a first appointment • (by each type of clinical staff)

Testing	• Number of tests requested
	• Types of tests requested with some broad categories e.g.[75]:
	  ○ Variant specific tests (segregation/cascade)
	  ○ Single gene tests
	  ○ Multiple gene panels
	  ○ Whole exome/genome sequencing
	  ○ Chromosomal tests (microarray/karyotype)
	• Turn-around time for test results (for each triage category)

[75] Fennell, A. P., Hunter, M. F., & Corboy, G. P. (2020). The changing face of clinical genetics service delivery in the era of genomics: a framework for monitoring service delivery and data from a comprehensive metropolitan general genetics service. *Genetics in medicine: official journal of the American College of Medical Genetics*, *22*(1), 210–218. https://doi.org/10.1038/s41436-019-0602-2

9. FUNDING

9.1. Activity Based Funding

Activity based funding (ABF) is a way of funding hospitals based on the direct clinical services they provide to patients. The Independent Health and Aged Care Pricing Authority (IHACPA) sets price and cost benchmarks to assist the Commonwealth Government in delivering activity based funding (ABF) and block funding for public hospital services.

In the IHACPA Tier 2 Non-Admitted Services Definitions Manual Version 8.0 (2023-24) Genetics is classified as a 20 series - Medical consultation:20.08 Genetics, as usually provided by a clinical geneticist, that explicitly excludes genetic counselling services provided by a genetic counsellor (40.66).

Tier 2 Version 8.0 included a new class for genetic counsellors as a 40 series - Allied health and/or clinical nurse specialist intervention: 40.66 Genetic counselling. This defines the service as the provision of genetic counselling provided in a specialist clinic/unit led by a health practitioner who is not a physician or surgeon. It does not address the specialist qualifications of appropriately trained and certified genetic counsellors. It also explicitly excludes services provided in a specialist genetic clinic (20.08).

Despite the HGSA engaging with IHACPA, there remain inconsistencies across jurisdictions in interpretation and application of ABF to funding of genetic services. It is not transparent how or whether the IHPA costing for Genetics 20.08 applies to activity by genetic counsellors working in genetic services, and the classifications are applied inconsistently across jurisdictions. Anecdotally, the General Medicine code 40.53 has been applied in some jurisdictions. It is unclear how the classification applies to specialised genetic clinics or genetic services provided in other clinics. For example, the classification of familial cancer clinics in some jurisdictions as 20.42 Medical oncology rather than 20.08 Genetics is weighted differently, which leads to inconsistencies and inequities in funding genetic services.

Recommendation:

Clear guidance and up-to-date costing studies from IHACPA are needed to support national consistency in the use of ABF codes and equity in genetic service delivery.

The HGSA continues to advocate for the ancillary costs of providing genetic services, including significant preparation time and case complexity, test costs (that are often borne by the genetic service budget) and operational costs including essential administrative staff (who do not directly generate ABF qualified service events) to be adequately accounted for in the combined pricing for geneticists and genetic counsellors. Other clinical activities that are an essential part of Clinical Genetic Services have historically not been included as ABF qualified service events, like consultation to provide advice to other health professionals. There remains inconsistency in the application of ABF to “non-contact” clinical service events like advice provided to patients by letter or electronic correspondence only, and multidisciplinary case review meetings.

A number of genetic services provide supported mainstreaming programmes for non-genetics health professionals. This is expanding as genetic and genomic information becomes increasingly accessible and important for treatment and management beyond familial risk. However, management from specialist genetic/familial cancer services can be resource intense, requiring provision of significant training and support. The impact on individual genetic services reliant on patient facing interactions for Activity Based Funding has not been published, and it is not clear whether they will have capacity to provide the same level of support for the long-term.

9.2. Medicare (Australia)

Although the Medical Services Advisory Committee (MSAC) recognises that "genetic counselling represents best practice for patients or family members undergoing genetic testing for a heritable disease" there is no mechanism for this to be funded separately using the Medicare Benefits Schedule. The MSAC Guidelines outline that "genetic counselling cannot be funded separately via the MBS. Consequently, it is suggested that genetic counselling be captured within an appropriate consultation item claimed by the professional responsible for the care of the patient (usually the professional requesting the test)".[76]

There are no Medicare Benefits Schedule (MBS) item numbers for genetic counselling provided by a genetic counsellor. Consultations specifically for the provision of genetic counselling by medical practitioners do not have existing MBS items. However, existing MBS items may apply to genetic counselling performed by a medical provider (whether a clinical geneticist or non-genetics physician in the mainstream setting).

A Medicare benefit cannot be claimed if the service has already been paid for through another mechanism or arrangement with the Australian Government or a state or territory government. Working arrangements in a public hospital can be complex, and services should ensure that the same service is not paid for twice through both public hospital and MBS funding. For more information, see the Australian Government Department of Health and Aged Care collection "Medicare billing in public hospitals".

9.3. Block funding

Block funding is a traditional model of funding for hospital services based on a fixed amount.

Although historically the majority of genetic services in Australia have been block funded, most are now funded or transitioning to ABF.

The New Zealand national service is effectively block funded. Activity levels are measured and tracked via Purchase Unit (PU) codes under Te Pae Tata, the New Zealand Health Plan[77], but funding is not altered based on these.

[76] Australian Government Department of Health and Aged Care. (May 2021). Guidelines for preparing assessments for the Medical Services Advisory Committee

[77] Manatū Hauora Ministry of Health. (2023). Purchase unit codes.

Recommendation:

Funding models should account for:

- Genetic test, pathology, and other necessary investigations.
- Obligations to provide services across regional or state boundaries.
- Medical and socioeconomic cost reduction and the flow-on financial benefits of genetic services to a range of hospital, health, community and social services.
- Funds for genetic counselling and clinical genetics trainees, as well as trainees from other health disciplines and medical specialties.
- Resources to teach genetic counselling, medical and other undergraduate health students.
- Sufficient funding to support adequate supervision, continuing education, and professional development
- Capacity to undertake clinical, biomedical, psychosocial and service-related research and evidence-based service improvement.

10. REFERENCES

See Footnotes.

10.1. Archival HGSA documents

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10.2. Comparators and Reference Frameworks

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Zellerino, B. C., Milligan, S. A., Gray, J. R., Williams, M. S., & Brooks, R. (2009). Identification and prioritization of quality indicators in clinical genetics: an international survey. *American journal of medical genetics. Part C, Seminars in medical genetics*, *151C* (3), 179–190. https://doi.org/10.1002/ajmg.c.30220

11. GLOSSARY OF TERMS

Available online at HGSA Policies and Position Statements.

12. VERSION HISTORY

Date	Summary of changes
19 March 2024	7.4.1 Service Capacity Correction from “new patient referrals” to “available clinical hours” per year. Annual capacity for new patient referrals added.