



HUMAN GENETICS SOCIETY OF AUSTRALASIA

ARBN. 076 130 937 (Incorporated Under the Associations Incorporation Act)

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Title:	HGSA Glossary of Terms
Document Number:	2024GL02
Publication Date:	February 2024
Location:	https://hgsa.org.au/Web/Consumer-resources/Policies-Position-Statements.aspx
Replaces:	N/A
Last Review Date:	N/A
Next Review Date:	February 2025 Annual review to ensure currency with other HGSA documents

This document is published online, please access the most current version at the location above.

HGSA Glossary of Terms

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Activity based funding (ABF)

A funding model for public health services in Australia based on the direct clinical services they provide to patients.

Advanced Trainee in Clinical Genetics

A medical practitioner with Royal Australasian College of Physicians (RACP) Basic Training as a physician or paediatrician (or equivalent as determined by RACP) who has is undertaking advanced training in Clinical Genetics for physicians through the RACP Advanced Training Committee (ATC) in Clinical Genetics¹.

Advanced Training Committee (ATC) in Clinical Genetics

Supervises advanced training in clinical genetics through the Royal Australasian College of Physicians (RACP).

Australian Health Practitioner Regulation Agency (AHPRA)

The national organisation responsible for implementing the National Registration and Accreditation Scheme (NRAS) across Australia in partnership with National Boards to ensure that Australia's registered health practitioners are suitably trained, qualified and safe to practise.

Australasian

Pertaining to Australia and Aotearoa New Zealand.

Australasian Association of Clinical Geneticists (AACG)

A special interest group (SIG) of the Human Genetics Society of Australasia (HGSA) that connects, trains and represents medical specialists and trainees in clinical genetics, and provides comprehensive expert knowledge and advice on medical genetics in Australia and New Zealand.

Australasian Society of Genetic Counsellors (ASGC)

A special interest group (SIG) of the Human Genetics Society of Australasia (HGSA) that represents members of the HGSA who provide genetic counselling or work in related areas. The majority of members are either training or certified as genetic counsellors.

Associate Genetic Counsellor

A job title used to describe a genetic counsellor with a recognised tertiary qualification in genetic counselling who has not yet completed FHGSA Certification.

¹ Royal Australian College of Physicians (RACP). 2023. [Advanced Training in Clinical Genetics Handbook](#).

Block funding

A budget-based funding model used for some public health services in New Zealand, and in Australia where Activity Based Funding (ABF) may not be appropriate.

Board of Censors for Genetic Counsellors (BOCGC)

The HGSA Board of Censors for Genetic Counselling is responsible for the professional regulation of genetic counsellors as a self-regulating health profession in Australia and New Zealand. It ensures that individual genetic counsellors have relevant professional skills and knowledge to provide safe and effective delivery of services within their scope of practice.

Carrier test

Primarily used to determine if an adult, with or without symptoms, has a genetic variation, which increases the chance that their children will have the disorder in question.

Clinical genetic service

A multidisciplinary team that provides healthcare for people and families who are affected by, or are at risk of developing, a genetic health condition. The majority of clinical genetic services are based in the public health setting, with a growing number of private services. Services may provide a general genetic service and/or have several subspecialty interests and conduct clinics or research in these areas. Clinics may have a local, regional, state-wide, or national referral base. Roles, responsibilities, clinic structures, and other features can vary significantly. Although supported by a range of healthcare and other professionals, core clinical genetic services are provided by genetic healthcare providers supported by administrative staff.

Clinical geneticist

A medical practitioner with Royal Australasian College of Physicians (RACP) Basic Training as a physician or paediatrician (or equivalent as determined by RACP) who has completed advanced training in Clinical Genetics for physicians through the RACP Advanced Training Committee (ATC) in Clinical Genetics². Applicants must be recommended by the 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 HGSA grants the title 'Fellow of the HGSA' (FHGSA) in Clinical Genetics to those who have achieved FRACP (Clinical Genetics).

Continuing Professional Development (CPD)

"A range of learning activities through which health professionals maintain and develop throughout their career to ensure that they retain their capacity to practise safely, effectively and legally within their evolving Scope of Practice". (Allied Health Professions project, 2002, UK).

² Royal Australian College of Physicians (RACP). 2023. [Advanced Training in Clinical Genetics Handbook](#).

Diagnostic test

Usually ordered by a medical provider to make or confirm a suspected diagnosis, or to exclude a differential diagnosis.

Fellow of the HGSA in Clinical Genetics (FHGSA, Clinical Genetics)

The HGSA grants the title ‘Fellow of the HGSA’ in Clinical Genetics to those who have achieved FRACP (Clinical Genetics) after completing advanced training in Clinical Genetics for physicians through the Royal Australasian College of Physicians (RACP) Advanced Training Committee (ATC) in Clinical Genetics.³

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 geneticist⁴, but who have additional specialised training in clinical genetics. However, this **does not** entitle them to statutory specialist registration in clinical genetics.

Fellow of the HGSA in Genetic Counselling (FHGSA, Genetic Counselling)

The HGSA grants the title ‘Fellow of the HGSA’ in Genetic Counselling to those who have achieved certification after assessment by the HGSA Board of Censors for Genetic Counselling. This replaces the term “Part 2 Certification”.

Genetic/genomic

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.⁵ These terms may be used interchangeably.

Genetic assistant

An allied health or clinical assistant who completes tasks delegated by a qualified genetic healthcare professional to support the delivery of genetic services. 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.

³ Royal Australian College of Physicians (RACP). 2023. [Advanced Training in Clinical Genetics Handbook](#).

⁴ Medical Board of Australia. (2018). [Medical List of specialties, fields and related titles Registration Standard](#).

⁵ Commonwealth of Australia Department of Health. National Health Genomics Policy Framework. 2017.

Genetic associate

A job title previously used in New Zealand interchangeably with the title genetic counsellor.

Genetic counselling⁶

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

Genetic counsellor⁷

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 Human Genetics Society of Australasia (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 HGSA administers the training, certification, and regulation of genetic counsellors in Australia and New Zealand. There is currently no legislative or statutory protection in Australia or New Zealand for the title of genetic counsellor.

Genetic counsellors have completed a tertiary qualification in genetic counselling from a program accredited or recognised by the HGSA and can apply to become a Member of the HGSA in Genetic Counselling (MHGSA, Genetic Counselling). Genetic counsellors trained, certified, or working

⁶ 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>

⁷ Australasian Society of Genetic Counsellors. (2023). [Definition of Genetic Counsellor](#).

overseas can also apply for MHGSA. MHGSA acknowledges the minimum qualification requirements to practice as a genetic counsellor.

MHGSA genetic counsellors practicing in Australia or New Zealand can undertake clinical certification through the HGSA Board of Censors for Genetic Counselling (BOC). 'Fellow of the HGSA' (FHGSA) in Genetic Counselling is granted by the HGSA Council to those who complete certification.

Genetic health professionals

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

Genetic health professionals may work in clinical care and/or other areas (research, academia, education, policy, industry and pathology).

Genetic healthcare providers

Medical providers/specialists, nurses, and allied health care providers with expertise and/or training in genetics) employed/embedded in a genetic service to support the activities of a clinical genetic service.

Human Genetics Society of Australasia (HGSA)

The peak professional body for those working in the field of human genetics in Australia and New Zealand.

Mainstreaming

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.

Medical Council of New Zealand (MCNZ)

The national body that registers New Zealand doctors as competent and fit to practise under the Health Practitioners Competence Assurance Act 2003.

Member of the HGSA in Genetic Counselling (MHGSA, Genetic Counselling)

The HGSA grants MHGSA to genetic counsellors who have completed a tertiary qualification in genetic counselling from a program accredited or recognised by the HGSA. Genetic counsellors trained, certified, or working overseas can also apply for MHGSA. MHGSA acknowledges the minimum qualification requirements to practice as a genetic counsellor. This replaces the term "Part 1 Certification)

Medicare

Medicare is a Commonwealth health insurance scheme-based funding model that pays a benefit through the Medicare Benefits Schedule (MBS) to subsidise the cost of healthcare not otherwise funded by the government. For more information, see the [Australian Government Department of Health and Aged Care collection “Medicare billing in public hospitals”](#)

Metropolitan

Major urban areas with 100,000 or more residents.⁸

National Alliance of Self Regulating Health Professions (NASRHP)

The national peak body that sets benchmarked standards for self-regulating allied health professions in Australia.

National Pathology Accreditation Advisory Council (NPAAC)

The Australian body responsible for developing and maintaining standards for the accreditation of pathology laboratories by the National Association of Testing Authorities (NATA) in Australia and International Accreditation New Zealand (IANZ) in New Zealand.

Outreach

Genetic services attached to and supported by a major genetics service that provide services to other metropolitan, regional, rural, or remote areas. 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. Some outreach services are provided via telehealth/telemedicine.

Practice

“[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,

⁸ Australian Government Department of Health (2019) [Modified Monash Model \(MMM\)](#) and [Stats NZ \(2017\) urban rural classification](#)

*research, advisory, regulatory or policy development roles, for example. This work can be of a paid or formal volunteer nature on a full or part-time basis.*⁹

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 associated with specific consent and counselling issues requiring professional genetic counselling.¹⁰

Royal Australasian College of Physicians (RACP)

The professional body that connects, trains and represents medical specialists and trainee specialists across Australia and Aotearoa New Zealand.

Regional¹¹

Large urban centres (and surrounding areas) with ~50,000-100,000 residents

Registration

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.

The HGSA publishes an online register of HGSA registered genetic counsellors who are engaged in the regulatory process, which includes active participation in or completion of certification and continuing professional development (CPD). At present, registration through the HGSA is not mandatory for genetic counsellors. There is no statutory legislation to protect the title of genetic counsellor, or to protect the public from accessing services from people using this title without qualification, training, or certification as a genetic counsellor.

⁹ NASRHP; [Self Regulating Health Profession Peak Bodies Membership Standards](#); 2/12/2016

¹⁰ Australian Commission on Safety and Quality in Health Care. [Requirements for medical testing for human genetic variation](#). 3rd ed. Sydney: ACSQHC; 2022

¹¹ Australian Government Department of Health (2019) [Modified Monash Model \(MMM\)](#) and [Stats NZ \(2017\) urban rural classification](#)

Remote¹²

Areas servicing less than 1000 residents. Note none of New Zealand is classified as remote due to its geographical size.

Rural¹³

Large and medium rural towns (and surrounding areas) with ~1,000-50,000 residents

Screening test

Usually performed on a specific population, which provide a personalised risk assessment. For example blood-spot screening in newborns, or prenatal screening in pregnant individuals.

Self-regulation

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 statutory legislation.

Supervision

For genetic counsellors, supervision 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. 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.

Some genetic counsellors 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.

In addition to supervision, 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.

¹² Australian Government Department of Health (2019) [Modified Monash Model \(MMM\)](#) and [Stats NZ \(2017\) urban rural classification](#)

¹³ Australian Government Department of Health (2019) [Modified Monash Model \(MMM\)](#) and [Stats NZ \(2017\) urban rural classification](#)

Telehealth/telemedicine/“telegenetics”

The use of information and communication technologies to deliver health care when patients and care providers are not in the same physical location¹⁴.

Whole genome/exome/transcriptome analysis

Extensive genomic analysis for complex conditions affecting multiple systems. Usually associated with complex analysis and a probability of incidental/secondary/unsolicited findings (including family relationships) requiring professional genetic counselling and documentation of specific consent.¹⁵

Workload indicators of staffing need (WISN)

A method developed by the World Health Organisation (WHO) to assess both workforce needs and workload pressure to estimate staff requirements.¹⁶

¹⁴ National Telehealth Service. Te Whatu Ora, Health New Zealand.

<https://www.tewhatauora.govt.nz/our-health-system/digital-health/national-telehealth-service/>

¹⁵ Australian Commission on Safety and Quality in Health Care. [Requirements for medical testing for human genetic variation](#). 3rd ed. Sydney: ACSQHC; 2022

¹⁶ World Health Organization. (2023). Workload indicators of staffing need. WHO Press.