

**Specialist Medical and Surgical
Services**

Clinical Genetics Service

Tier 2 Service Specification

September 2024

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1. Status

Approved to be used for mandatory nationwide description of services to be provided.

MANDATORY ☒ RECOMMENDED ☐

It is compulsory to use this Specification when purchasing services. No Districts should use a local service specification instead of this mandatory specification.

2. Review History

Review History	Date
First published on NSFL	June 2011
Updated: Revisions to the following sections: Maori Health, Access, Service Levels, Support Services, retired NS10014 purchase unit, Quality Requirements and Service Planning Information.	November 2016
Moved to Health NZ template. Updated links for PUDD and NSFL only. Amended DHB to become District/Region where appropriate. No other changes to content made.	September 2024
Consideration for next Service Specification Review	Within five years

Note: In September 2024 a small programme of work moved all Service Specifications to Health New Zealand branded templates. No amendments were made to the body text or content of the Service Specification, so references to DHB, Ministry of Health or other pre-2022 reforms vocabulary will still exist. A larger programme of work to review and revise all Service Specifications is planned for late 2024 to early 2025.

Note: Contact the NSF Team, Te Whatu Ora | Health New Zealand to discuss proposed amendments to the service specifications and guidance in developing new or updating and revising existing service specifications. NSF@tewhatuora.govt.nz

Nationwide Service Framework Library web site [here](#)

3. Introduction

This tier two service specification for the Clinical Genetics Service must be read in conjunction with the tier one Specialist Medical and Surgical Services and Services for Children and Young People service specifications as they contain generic details that are applicable to all service delivery.

3.1 Background

Since 1 July 2011, the Service's planning and funding decisions have been made at a national level because the Service requires a highly specialised workforce and has a relatively low volume patient group. This national approach supports the desired outcome for this Service to be sustainable, delivered consistently and that people are able to access it equitably.

The Service is provided by two district hosts with three regional service hubs and a regional outreach clinic programme to support equitable service access.

4. Service Definition

The Service is an integrated tertiary clinical and genetic testing service provided for families and individuals with, or at risk of, a range of inherited and sporadic congenital, genetic and chromosomal disorders. The Service will provide:

- direct care of referred patients including clinical assessment and genetic diagnosis
- advice on and/or management of genetic disorders and their consequences, including family follow up (such as anticipatory care, prenatal care and testing)
- advice and support to other health care professionals in the care of patients
- genetic risk estimation
- genetic counselling (including pre and post predictive testing)
- accessible information for families/other health professionals and patient support groups (written and spoken)
- information to individuals and families about sources of community support including support groups
- support to individuals and families (such as, in decision making about future pregnancies)
- a management plan for the secondary referrer which identifies appropriate onward referral to other clinical services for interventions arising from genetic disorders. The geneticist would be responsible for providing a management plan and may also work with the secondary referrer in the onward referrals
- education and training/teaching for health professionals and students within the health professions, across primary, secondary and tertiary care
- a family and whānau inclusive approach

- assistance to the DNA¹ storage service operations by gaining consent and collecting relevant samples
- a clinical interpretation and liaison support for laboratories that provide genetic analysis.

5. Service objectives

5.1 General

The key objective of the Service is to enhance and promote the health by enabling individuals and families and whānau to make informed health decisions.

This is achieved by having a comprehensive and integrated approach to the investigation, diagnosis, education and counselling for individuals and families and whānau and through increasing health professional knowledge and public awareness.

5.2 Māori Health

It is expected that there will be recognition of the genetic disorders with increased incidence in New Zealand Māori eg, hereditary diffuse gastric cancer, to raise awareness, increase identification and earlier treatment.

6. Service Users

All eligible individuals and their families and whānau referred with, or at risk of, a genetic disorder that meet the Service's entry criteria in 7.2 below.

7. Access

7.1 Referral Process

Referral into the Service is made by primary, secondary or tertiary health care professionals (HCPs). Family referrals may occur for family members of known at-risk individuals.

¹ DNA: Deoxyribonucleic Acid is a nucleic acid that contains the genetic instructions used in the development and functioning of all known living organisms.

7.2 Entry Criteria

- Newborns, infants and children with single gene disorders, chromosomal disorders, other known genetic syndromes, and 'syndromic' clinical presentations of uncertain cause.
- Complex metabolic disorders (in conjunction with the National Services Paediatric and Adult Metabolic Service).
- Pregnancy related abnormal fetal outcomes and other high risk pregnancies based on family history.
- Adults with, or at risk of, single gene disorders or other familial disorders.
- Adults with a strong family history of cancer or with a family cancer syndrome.
- Children and adults with an intellectual disability.

Access to the Service may occur with additional members added to a family file that remains open and accessible.

7.3 Exit Criteria

Exit for individuals is by agreement with the HCP at the clinic appointment or consultation and confirmed in writing to the individual patient and referrer. Patients that are discharged from the Service may be re-referred and provided follow-up assessment as required.

7.4 Time

A clinical triage process will determine level of priority. Triage will occur within a maximum of five working days from referral.

- *Urgent referral:* clinical response/contact within two working days
- *Priority referral:* assessment² within six weeks of referral.
- *Routine referral:* assessment within four months of referral.

8. Service Components

8.1 Processes

The Service components provided include the following.

- **Triage** – All referrals received are triaged, according to the Human Genetics Society of Australasia (HGSA) standards. Triage is by urgency, identifies the preferred clinic location, and clinical staff level required.
- **Assessment** – This component includes clinical history, confirmation of family history, examination, clinical diagnosis risk assessment and appropriate diagnostic investigations.

² Assessment includes provision of non-contact or telephone assessments

- **Specialist non-contact assessment** – An assessment of patient records and any relevant diagnostic test results provided by a referrer (without the patient present). A written plan of care is developed for the patient and provision of that plan and other necessary advice is discussed with, and sent to the referring clinician, the patient and their general practitioner.
- **Genetic counselling** – Includes pedigree analysis, risk assessment and investigations, and provision of surveillance and management guidelines for genetic disorders.
- **Genetic investigations** – Includes genetic testing of affected individuals (proband) and, as appropriate, at risk unaffected individuals, as determined by genetic services clinical staff.
- **Consultative and advisory services** – Includes genetics advice to other specialists and primary health care practitioners to help them to assess, diagnose and manage individuals and families and whānau.
- **Onward referral to clinical services** – Currently interventions arising out of the assessment and genetic counselling components are managed by referral to the appropriate clinical service. This may change in the future.

8.2 Pacific Health

The Service will consider the cultural requirements of Pacific patients and their families, support their access to the Service and facilitate consent procedures.

8.3 Health for other Ethnic Groups

This will include specific screening and genetic counselling for ethnic groups with recognised risk, for example Ashkenazi Jewish patients and families for cancer or metabolic diseases.

8.4 Settings

The Service is mainly provided as an outpatient service. Individuals and their families and whānau will be seen at the closest regional service base (hub) or outreach clinic that can see them within the priority timeframe. Telephone consultation with the patients may be necessary and appropriate in some circumstances.

The Service may be provided to neonates and other individuals (including pregnant patients and cancer patients) on an in-patient basis as and where appropriate.

8.5 Service Levels

The Service will be provided at a tertiary hospital level, Monday to Friday.

8.6 Support Services

The following support services are required to be provided as an integral part of the Service but are funded as part of another service.

- Laboratory sServices (cytomolecular, biochemistry, metabolic).
- Radiology services (especially for skeletal dysplasia).

8.7 Key Inputs

The major input to the Service is the workforce which includes clinical geneticists and genetics counsellors.

8.7.1 Clinical Geneticists

Clinical geneticists are physicians who have undergone specialty training in genetics after general professional training in medicine or paediatrics. Their vocational requirements are covered under the Health Practitioners Competency Assurance Act. The HGSA Guideline 2010GD02: Role of the Clinical Geneticist (September 2010) describes in more detail the role of the clinical geneticist.

8.7.2 Genetic Counselling

Genetic counselling is the process by which patients and relatives at risk of a disorder that may be hereditary are advised of the consequences of the disorder, the probability of developing or transmitting it and ways in which this may be prevented, avoided or ameliorated.

Genetic counsellors are tertiary-trained health professionals with specialist training in genetics and counselling, certified by the HGSA in genetic counselling³.

9. Service Linkages

Generic service linkages are described in the tier one Specialist Medical and Surgical Services service specification.

The following service linkages are critical to the provision of the Service.

Critically Linked Providers	Nature of Linkage	Accountabilities associated with linkages
Lead Maternity Carers (LMCs)	LMCs are a referral source and have ongoing involvement with individuals and family/whānau.	To provide education, information or expert opinion particularly in relation to reproductive genetics. Referral guidelines recommend consultation with an obstetrician.
Maternal Fetal Medicine (MFM) and High Risk Pregnancy Clinics	MFM is a referral source and has ongoing involvement with individuals and family/whānau.	To provide education, information or expert opinion particularly in relation to reproductive genetics (on referral).
Perinatal Pathology	Liaise and work with pathologist.	To assist with diagnosis of genetic disorders and inform need for family/whānau genetic counselling.

³ Genetic Associates in New Zealand must have HGSA certification in Genetic Counselling (or equivalent) and ongoing participation in Maintenance of Proficiency, HGSA.

Critically Linked Providers	Nature of Linkage	Accountabilities associated with linkages
Secondary and tertiary hospital services (including but not limited to neurology, cardiology, ophthalmology, respiratory, paediatrics and fertility services).	Services complete their specialty specific investigations to assist with genetics diagnosis.	Work with relevant professionals regarding investigation requirements.
Oncology and Haematology	Refer and/or liaise.	Clinical genetics HCPs will liaise regarding current management options to ensure individuals and family/whānau well informed. Refer for management.
Breast, bowel and gynaecological surgical services	Refer and/or liaise.	Liaise with relevant HCPs regarding current management options to ensure individuals and family/whānau well informed. Refer for management
Neonatal and child health services	Inpatient assessment where timely assessment is required on neonatal intensive care units at hubs. Works closely with the secondary referrers.	Work with relevant HCPs regarding investigation requirements on referral.
New Zealand Familial Gastrointestinal Cancer Service	Refers to registry and uses information from registry.	To support family and whānau diagnosis and tracking.
New Zealand Cancer Registry	Refers to registry and uses information from registry.	To support family and whānau diagnosis and tracking.

Other linkages include, but are not limited to the following.

Other Linked Providers	Nature of Linkage	Accountabilities associated with linkages
Well Child / Tamariki Ora (WCTO) service providers	Liaise and work with relevant WCTO providers.	Work with other relevant HCPs whenever there are concerns relating to an individual.
Other hospital services	Refer to relevant hospital services as appropriate.	Work with other relevant HCPs whenever there are concerns relating to an individual. Referral on for management of conditions arising from genetic disorders.
Disease or disability based community groups relevant to inherited, genetic or chromosomal disorders.	Refer or liaise re individuals and their family as appropriate. Provide education, information or expert opinion.	Provide education, information or expert opinion. Work with other relevant HCPs and provide support whenever there are concerns relating to an individual.

10. Exclusions

The following exclusions apply.

- Non-health related genetic testing such as paternity testing.
- Genetic testing ordered by non-genetics staff (eg, primary, secondary and tertiary care providers).

11. Quality Requirements

The Service must comply with the quality requirements described in the Operational Policy Framework or, as applicable, Crown Funding Agreement Variations, contracts or service level agreements

The Service will ensure all laboratories that are accessed for genetic testing (cytogenetic, molecular, metabolic and biochemical) will comply with **International Accreditation New Zealand** or equivalent accreditation and should participate in recognised quality assurance programmes.

Clinical audit will align with the requirements of the HGSA Standards Framework⁴.

⁴ Clinical Genetics Services Standards Framework <http://www.hgsa.org.au/documents/item/1798>

12. Purchase Units

Purchase Unit Codes are defined in Health New Zealand's Nationwide Service Framework Purchase Unit Data Dictionary. The following codes apply to this Service.

PU Code	PU Description	PU Definition	Unit of Measure
NS10010	National Services Clinical Genetics 1st Attendance	First attendance to a Geneticist or Medical Officer at Registrar level or above or Nurse Practitioner for a specialist assessment within a clinical genetic service.	Attendance
NS10011	National Services Clinical Genetics Subsequent Attendance	Follow up attendances to a Geneticist or Medical Officer at Registrar level or above or Nurse Practitioner for a specialist assessment within a clinical genetic service.	Attendance
NS10012	National Services Clinical Genetics Counselling 1st Attendance	First Attendance for genetic counselling provided for a patient and their family and whanau by a Genetic Associate within a clinical genetic service.	Attendance
NS10013	National Services Clinical Genetics Counselling Subsequent Attendance	Follow-up Attendance to Genetic Counselling provided for a patient and their family and whanau by a Genetic Associate within a clinical genetic service.	Attendance
M00010	Medical non contact First Specialist Assessment - Any health specialty	A review is undertaken by a Registered Medical Practitioner of Registrar level or above, or a Registered Nurse Practitioner, of patient records and any diagnostic test results. The original referral should only be generated after a face to face contact by the referrer.	Written Plan of Care

These purchase units, used in conjunction with Mode of Delivery (refer to the Purchase Unit Data Dictionary), and Health Specialty Code – M87 Specialist Medical Genetics, will allow comprehensive reporting of activity within the service.

The Service must also comply with the requirements of national data collections where applicable eg, National Non Admitted Patient Collection (NNPAC).

Explanations of the units of measure (UOM) from the purchase unit table.

Unit of Measure	Unit of Measure Definition
Attendance	Number of attendances to a clinic/department/acute assessment unit or domiciliary.
Written Plan of Care	Written plan of care provided by the specialist to the referring GP

13. Reporting Requirements

13.1 Narrative report

A narrative report is to be written annually and provided to the Ministry of Health by 31 July of each year, for the previous financial year.

This report must identify issues and trends in the Service provision, for example, waiting times, service risks and service utilisation.

13.2 Service Planning Information

The Service will be cognisant of future workforce needs and participate in succession planning.

The HGSA Clinical Genetics Committee has developed a Standards Framework for Clinical Genetics Services in Australasia (2013 GL01, May 2013) to provide a foundation for current and future Clinical Genetics Services development, delivery and monitoring of outcomes and quality. The HGSA Standards Framework is incorporated into the quality requirements of this service specification in section 8 above.

14. Glossary

Not required

15. Appendices

Not required