New national genetic service launched

A new national genetic service has been established to improve access to genetics clinics and genetic testing in New Zealand. Genetic Health Service New Zealand (GHSNZ), launched in May 2012, provides genetic diagnosis, genetic counselling, assistance in managing genetic conditions and expert advice and education on genetic diseases.

GHSNZ encompasses the former Northern, Central and Southern Regional Genetics Services. The service is funded by the National Health Board, and provided by Auckland and Capital and Coast DHBs, and is part of the public health system. Genetic services are delivered from three main centres: Christchurch (South Island Hub), Wellington (Central Hub) and Auckland (Northern Hub), and outreach clinics throughout the country.

The Northern Hub covers the upper half of the North Island. It is based at Auckland Hospital and provides outreach clinics in Whangarei, Hamilton, Tauranga, Rotorua and Gisborne.

The Central Hub covers the lower half of the North Island and Nelson/Marlborough. It is based at Wellington Hospital and provides outreach clinics in New Plymouth, Whanganui, Hastings, Palmerston North, Porirua, Lower Hutt and Nelson-Marlborough.

The South Island Hub covers all of the South Island except Nelson/Marlborough. It is based at Christchurch Hospital and provides outreach clinics in Greymouth, Timaru, Dunedin, Queenstown and Invercargill.

Who provides the services?

GHSNZ clinical staff includes clinical geneticists and genetic associates.

Clinical Geneticists are doctors with specialist training in medical genetics. Patients are referred to them for diagnosis of a genetic condition.

Genetic Associates are health professionals with post-graduate qualifications in medical genetics and genetic counselling. Patients are likely to see them after their condition has been diagnosed.

What services are provided?

GHSNZ services include:

- Assistance in the diagnosis, clinical management of genetic disease and identification of preventable complications by early surveillance
- Advice about inheritance of genetic conditions, further information and genetic testing for those affected by or perceived to be at risk of genetic disorders in extended families
- Telephone enquiry service for doctors, midwives and other health professionals concerning genetic diseases
- Genetics education for professional and lay groups

Conditions frequently seen at genetic clinics in New Zealand

Patients most often seen in New Zealand genetics clinics include those with:

- Abnormal pre-natal scans or screening tests, e.g. amniocentesis with abnormal karyotype
- Dysmorphic features and developmental delay
- Neurological disorders with features suggestive of an inherited condition
- Fragile X syndrome
- Connective tissue disorders, e.g. Marfan syndrome, Ehlers-Danlos, osteogenesis imperfecta
- Familial cancer syndromes, e.g. hereditary breast and ovarian cancer, Lynch syndrome
- Metabolic disorders
- Inherited cardiac conditions, e.g. Long QT syndrome, hypertrophic cardiomyopathy
- Cystic fibrosis
- Muscular dystrophies (Duchenne, Becker, myotonic)
- Chromosome alterations
- Adult onset disorders of many systems, e.g. renal, ophthalmology, respiratory

Genetic Testing

Genetic testing should only be requested by GHSNZ clinical staff (or by a doctor in their specific scope of practice). Written consent is required by most reference laboratories. The majority of genetic testing is done overseas and is expensive.

All ages are covered, including:

Pre-conception/prenatal:

- Diagnostic, pre-conceptional, pre-natal or pre-symptomatic tests for a genetic condition and reproductive options including pre-implantation genetic diagnosis (PGD)
- Diagnostic assessment if there are concerns about foetal abnormalities during pregnancy
- Information and genetic counselling concerning prenatal diagnosis

Childhood:

- Diagnostic assessment of infants or children with dysmorphic features or developmental delay
- Genetic assessment for a child and their family/whānau with a likely genetic disorder
- Post-mortem review where a genetic disorder is suspected

Adulthood:

- Genetic risk assessment and testing for familial cancer syndromes
- Diagnostic assessment for adult-onset genetic disorders
- Explanation and further information about a genetic diagnosis
- Discussion of the implications of a family history of a genetic condition
- Recommendations for surveillance and management of genetic conditions

Patients seen through GHSNZ receive a detailed explanatory letter after their assessment, with copies sent to their General Practitioner and referring doctor (if different).

Who should be referred to the genetic service?

Referrals should be considered for a patient with an identified family history of a genetic disorder, or where a patient has been diagnosed with a genetic disorder or is at risk of a genetic disorder.

A common reason for referral to genetics services is for familial cancer syndromes. Approximately 5% of people with breast cancer and 2% of people with colon cancers have an inherited genetic susceptibility. There are more than 50 familial cancer syndromes, which increase susceptibility for specific cancers. Genetic testing is available for most familial cancer syndromes, but screening for a mutation usually has to start in an affected family member.

Referral is recommended for people with:

- A blood relative who is a known carrier of a familial cancer gene mutation, e.g. BRCA1, BRCA2, MLH1, MSH2, APC
- Ashkenazi Jewish ethnicity (increased occurrence of familial cancer syndromes)
- A personal or family history of breast cancer under age 40 years
- Male breast cancer
- High-grade serous ovarian cancer (any age)
- Colon cancer under age 40 years
- Multiple primary tumours (excluding lung, skin, cervix) under age 70 years, e.g. breast and ovarian, endometrial and colorectal cancer
- Rare tumour under age 45 years or any age if there is a close relative with a similar tumour, e.g. phaeochromocytoma, paraganglioma, sarcoma, glioblastoma, choroid plexus carcinoma, retinoblastoma
- Two or more first or second degree relatives on the same side of the family with colon or endometrial cancer with one diagnosed under age 60 years
- Two or more first or second degree relatives on the same side of the family with breast cancer under age 60 years and/or ovarian cancer at any age

How do you refer a patient to the genetic service?

Referrals to clinics can be arranged by faxing or posting a request to Auckland, Wellington or Christchurch centres. Detailed medical information is usually required.

Contact addresses:

Genetic Health Service NZ – Northern Hub
Auckland Hospital
Private Bag 92 024
Auckland Mail Centre
Auckland 1142
Ph: (09) 307 4949 Ext. 25870
Toll Free: 0800 476 123
Fax: (09) 307 4978
Email: GenSec@adhb.govt.nz

Genetic Health Service NZ – Central Hub
Wellington Hospital
Private Bag 7902
Wellington South
Ph: (04) 385 5310
Toll free: 0508 364 436
Fax: (04) 385 5822
Email: genetic.services@ccdhb.org.nz

Genetic Health Service NZ – South Island Hub
Christchurch Hospital
Private Bag 4710
Christchurch 8140
Ph: (03) 378 6574
Toll free: 0508 364 436
Fax: (03) 379 1343
Email: genetic.servicenz@cdhb.health.nz

For more information see:
www.geneticshealthservice.org.nz

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