

Genetic services in Victoria

Genetic services can help people, who are affected by or who are at risk of inherited conditions or birth defects, to make informed decisions about their health care. Services provided include genetic diagnosis, screening and testing, counselling and support.

Genetic services also provide education for professionals and undertake research to develop new genetic tests and improve knowledge about genetic conditions and their causes.

Knowledge about genetics is growing

Our understanding of genetics is expanding rapidly. This is, in part, due to the Human Genome Project, which has mapped (and aims to analyse) every human gene. It is anticipated that nearly all major genes responsible for susceptibility to disease, and some genes which offer protection, will be identified in the near future.

As a result, the number of genetic tests is expected to grow and genetic testing could include a wide range of uses. The emphasis of genetic services will shift from genetic screening and testing for rare single gene disorders (such as Huntington's disease and fragile X syndrome) and chromosome disorders (such as Down syndrome) to knowledge about genes for a range of common and complex disorders that will inform public health and management of individuals and families.

Genetic services in Victoria

Genetics services funded by the Department of Human Services are:

- The Victorian Clinical Genetics Service (VCGS), located at the Murdoch Children's Research Institute (MCRI)
- The Victorian Cancer Cytogenetics Service (VCCS), located at St Vincent's Hospital
- Thalassaemia Services, located at Monash Medical Centre, Clayton
- The Victorian Family Cancer Genetic Services, located at Royal Melbourne Hospital, Peter MacCallum Cancer Institute (PMCI), VCGS and Monash Medical Centre (MMC).

Types of services provided

Clinical genetic services currently available in Victoria include:

- Clinical diagnostic services for individuals with birth defects and genetic disorders of childhood.
- Diagnostic, predictive and counselling services for individuals at risk of, or affected by, adult onset genetic disorders. These services may be provided in joint clinics with a specialist.
- Information and counselling for people considering pre-implantation genetic diagnosis or prenatal diagnosis, or following the diagnosis of fetal abnormalities.
- Counselling of individuals, couples and families affected by (or perceived to be at risk of) genetic disorders or birth defects, and referral to appropriate community resources, including support groups.
- Identification of affected individuals and carriers of genetic disorders in extended family members, as appropriate.
- Involvement in the long-term management of individuals with rare complex genetic disorders, such as inborn errors of metabolism.
- Training for genetic counsellors and genetic fellows.
- Genetic education for professionals.

- Telephone counselling service for individuals, doctors and other health professionals concerning aspects of genetic disorders and teratogenesis (deformities present from birth).

Genetic screening and testing services

Screening refers to a test that is offered to all people, regardless of previous family history. Carrier testing is a type of screening and is generally used for those known to be at risk. Screening and testing services available in Victoria include:

- **Maternal serum screening (MSS)** – MSS is offered to all pregnant women for the detection of Down syndrome and neural tube defects.
- **Newborn screening** – all newborn babies are screened for phenylketonuria (PKU), hypothyroidism, cystic fibrosis and another 20 metabolic disorders.
- **Carrier screening for cystic fibrosis (CF)** – screening for CF carriers is currently available on a fee-for-service basis.
- **Thalassaemia screening** – couples in antenatal care are offered screening for haemoglobinopathies.

Genetic diagnostic laboratory services

Laboratory services include a range of genetic techniques to diagnose and assist in managing particular genetic conditions. Genetic diagnostic laboratory services currently available in Victoria include:

- **Newborn screening** – every newborn baby is screened for phenylketonuria (PKU), hypothyroidism, cystic fibrosis and 20 metabolic conditions.
- **DNA diagnostic tests** – DNA from individuals is examined to look for mutations in genes that may indicate a specific disorder. These tests include tests for bowel cancer genes. Breast cancer gene testing is performed at Royal Melbourne Hospital and Peter MacCallum Cancer Institute laboratories.
- **Metabolic diagnostic tests** – these are performed to diagnose or monitor the treatment of inborn errors of metabolism.
- **Cytogenetic diagnostic tests** – these are used to identify changes in the number or structure of chromosomes and may help to diagnose a disorder in pregnancy or any time after birth.
- **Thalassaemia diagnostic test** – this is a blood test to determine if a person is a carrier.

Support groups

The Genetic Support Network of Victoria (GSNV) aims to ensure that all people with a genetic disorder in Victoria have appropriate and accurate information and support to enable them to manage the challenges to their health and wellbeing. The GSNV facilitates an exchange of information, resources and assistance in order to support a number of existing genetic support groups and aid the development of new groups.

The Thalassaemia Society encourages and facilitates the formation and development of support groups for people with Thalassaemia and related haemoglobin conditions and for their families. They also provide an advocacy service for people with haemoglobin conditions, designed to increase awareness in the general community and, on request, to act as a spokesperson on behalf of individuals and families.

Where to get help

- Your doctor
- Genetic Health Services Victoria and VCGS Pathology Tel. (03) 8341 6201
- Genetic Support Network of Victoria Tel. (03) 8341 6315
- Thalassaemia Society Tel. (03) 9888 2211

Things to remember

- Genetic services in Victoria cover diagnosis, screening and testing, counselling, education, clinical research, and the ongoing management of individuals and families with particular birth defects and genetic disorders.
- The Genetic Support Network of Victoria (GSNV) supports many groups that provide a point of contact for parents and people with the same genetic condition.

This page has been produced in consultation with, and approved by:

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