



Genetic Services of Western Australia

General Information Your Genetic Consultation

- **Biochemical tests** involve testing for certain substances in the body (such as proteins) for evidence of hereditary conditions and birth defects.

Genetic consultation

A genetic consultation is a confidential appointment which will:

- provide you with information about genetic conditions which run in families
- assess your risk of developing an inherited condition based on your medical and family history
- assess whether genetic testing is possible;
- recommend surveillance and risk management options
- provide you with information on early detection and lifestyle strategies for good health.

Your doctor or specialist can refer you to Genetic Services of WA or you can contact us directly. If we receive a referral, a genetic counsellor will contact you by phone to ask about you and your family's medical history. The genetic counsellor needs this information before they can assess your risk.

You may be asked to send in medical reports and other relevant information to assist the counsellor in preparing for your appointment. Please bring photographs of family members to the appointment if you think they may help the counsellor or geneticist identify hereditary conditions.

At the consultation the genetic counsellor will discuss any of your concerns and answer your questions. Your medical and family history will be reviewed and the likelihood that an inherited condition or disorder is present in your family will be discussed. Possible choices for treatment, early detection and prevention will also be discussed. Sometimes it will be necessary to gather more information and/or arrange tests.

Are interpreters available?

Yes, please contact us before your appointment if English is not your first language and you want to have an interpreter during the appointment.

What do I do now?

If you would like more information or to arrange an appointment please contact:

Your doctor
or

Genetic Services of Western Australia
King Edward Memorial Hospital
374 Bagot Road
SUBIACO WA 6008
Phone: (08) 9340 1525
Fax: (08) 9340 1678
Email: gswa@health.wa.gov.au

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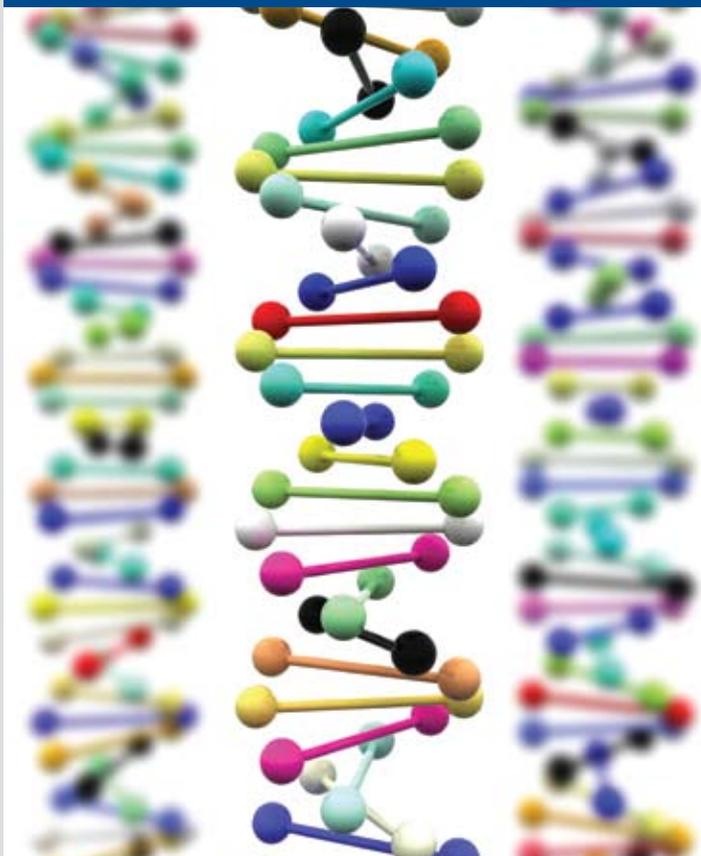
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Who are Genetic Services of WA?

Genetic Services of WA offers a range of services to people with a hereditary (passed down through families) condition and those people concerned about their risk of developing a hereditary condition. These services are provided by a range of staff, including genetic counsellors, clinical geneticists and laboratory staff.

What are hereditary (genetic) conditions?

Many genetic conditions and disorders run in families. These conditions and disorders may also be called hereditary because they are passed down in the genes from parents to their children. Hereditary conditions result from inheriting an altered (changed) gene that increases your risk of developing the condition or disorder.

How common are hereditary (genetic) conditions and birth defects?

Approximately 6 out of 10 of individuals in their lifetime will be affected by a condition which has some hereditary component. Genetic conditions can range from mild to very severe. Hereditary conditions and birth defects occur in 3–5% of all live births and a third of paediatric admissions to hospitals.

Why should I contact Genetic Services of WA?

The following list describes the most common reasons for seeking help from Genetic Services of WA:

- If you have been diagnosed with a genetic condition.
- If you or your partner is concerned about a condition in your family being passed on to your children.
- If you, your partner or a family member has had a child with a birth defect or hereditary (genetic) condition.
- If you are a woman in her mid-30s or older who is planning to have a child.
- If you and your partner are related (e.g. cousins) and are planning to have a child.
- If you have been exposed to a potential teratogen (or medication during pregnancy) which may cause a birth defect.
- If you have a history of stillbirth or if you have had more than three miscarriages.
- For detection of carrier status for a gene mutation, such as cystic fibrosis, Fragile X.
- If a fetal abnormality is unexpectedly detected during pregnancy, following routine prenatal tests such as ultrasound or increased risk first trimester or second trimester screen.
- If you have two or more close relatives who have had bowel, breast or ovarian cancer when they were younger than 50 years of age.

What services are available?

1. Early diagnosis and treatment

Hereditary conditions such as cystic fibrosis, muscular dystrophy and some neurological conditions can be detected early in some high-risk families. The diagnosis of a predisposition (tendency to develop) to some cancers, before the symptoms appear, can provide the opportunity for preventative measures or early treatment.

A range of screening and diagnostic tests can be carried out during pregnancy to detect conditions such as: Down syndrome, neural tube defects, muscular dystrophy, cystic fibrosis, haemophilia and many other genetic conditions. Screening is a test offered to all people, regardless of previous family history.

Screening tests include:

- *First Trimester Screening (Nuchal Translucency and maternal blood test)* combines results from a blood test and an ultrasound to provide information on the risk of having a baby with Down syndrome. This test can detect some other abnormalities and may also tell you if you have a multiple pregnancy (e.g. twins).
- *Maternal Serum Screening (MSS)* is a blood test that provides information on the risk of having a baby with a chromosome abnormality like Down syndrome or a neural tube defect like spina bifida.

- The newborn screening test (also called the Guthrie test) checks for signs of serious disorders when your baby is between 48 and 72 hours old. Disorders that can be detected include: phenylketonuria (PKU), hypothyroidism, galactosaemia and cystic fibrosis.

Diagnostic tests identify genetic conditions. These tests include:

- *Chorionic villus sampling:* when a small sample of placenta is taken for testing in a laboratory.
- *Ultrasound scan:* provides an image of the baby in the womb.
- *Amniocentesis:* when a small sample of amniotic fluid is taken for testing in a laboratory.

2. Familial Cancer Program

This program is run by Genetic Services of WA and provides a comprehensive service to individuals and families with a significant family history of breast, ovarian, bowel and other related familial cancers. A brochure is available on request.

3. Laboratory services

These tests include:

- *DNA (deoxyribonucleic acid)* tests examine an individual's DNA for gene changes.
- *Cytogenetic tests* examine an individual's chromosomes for abnormalities such as the extra number 21 chromosomes that results in Down syndrome (Trisomy 21). Chromosomes are the packages that contain our genetic material.