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### Important points

- Genetics services includes clinical genetics (genetic counselling), laboratory and education services.
- Genetic counselling provides
  - Information
  - Supportive counselling regarding the diagnosis and risk for a genetic condition in the family
  - Diagnostic, carrier, predictive and presymptomatic genetic testing where appropriate
  - Management of conditions in some cases
- The health professional team providing genetic counselling may consist of clinical geneticists or other medical specialists, genetic counsellors and social workers
- Genetic counselling is provided as part of a comprehensive genetics service whose elements include clinical, laboratory and education
- The availability of genetics services varies throughout Australia and New Zealand

Genetic counselling is provided by a team of health professionals who work together to provide an individual or family with current information and supportive counselling (advice or guidance) regarding problems in growth, development and health that may have a genetic basis. This can assist families and individuals to understand and adjust to the diagnosis of a genetic condition.

### What happens in genetic counselling?

#### *The consultation*

During the consultation:

- A family health history is collected to provide information about the health of family members
- A diagnosis of a genetic condition may be made or confirmed in a pregnancy, after birth, in childhood or later in life. The diagnosis may be made on the basis of clinical features, biochemical tests or genetic tests (see Genetics Fact Sheet 21). This diagnosis may mean that other family members are at risk. On the other hand, a family member may be reassured to find that he/she does not have, or is unlikely to develop, the particular condition

Where there is a genetic condition in a family, the genetic counselling team may:

- Estimate the risks that other family members, or future children, will be affected by the condition. Often, however, a person is reassured following genetic counselling to find out that a condition is unlikely to recur in their family
- Discuss the impact and possible effects on the individual and their family in a supportive atmosphere. Management strategies can be developed and referral provided to appropriate community resources, including support groups. Both verbal and written information about the condition and its impact is provided to assist people in dealing with some of the issues that may arise from the diagnosis of a genetic condition
- Discuss if appropriate prenatal testing and other reproductive options to ensure that any decision is made on an informed

basis. Many genetic conditions can be diagnosed before birth (see Genetics Fact Sheets 17 & 18)

- If a genetic condition is identified by prenatal diagnosis, genetic counselling is the means by which current information and support is provided so that an informed decision can be made regarding the continuation of the pregnancy
- Where there has been exposure to a potential *teratogen* (chemical, drugs, medications, radiation or other environmental agents which can cause birth defects), genetic counselling provides an opportunity to obtain current information and support and discuss strategies and options
- Discuss and arrange appropriate genetic testing, including carrier, predictive and presymptomatic testing, where available (see Genetics Fact Sheet 21)

#### *Follow up*

After the initial consultation an opportunity may be provided to go over the information and offer on-going support as families and individuals learn about the condition. It is very common for people to think of many questions after the genetic counselling session, and new questions also arise as a condition develops.

Follow-up is provided in further consultations, if geographically possible, or by telephone.

A letter summarising the consultation(s) is also provided.

### Who provides genetic counselling?

Genetic counselling is provided by a multi-disciplinary team of professionals that may include:

- **Clinical geneticists and other specialist medical practitioners** with expertise in the genetics of their field of medicine eg oncologists (cancer genetics) and neurologists (eg Huntington disease and Alzheimer disease)
- **Genetic counsellors** are graduate health professionals with specialist training and certified by the Human Genetics Society of Australasia (HGSA) to provide genetic counselling.

Associate Genetic Counsellors are graduate health professionals in the process of completing the requirements of the HGSA to become a certified genetic counsellor. Genetic Counsellors and Associate Genetic Counsellors provide genetic counselling as part of a multi-disciplinary team. Some work in 'outreach' and are linked to a major genetics unit

- **Social workers** with a special interest in genetics and particular genetic conditions, work closely with clinical geneticists, genetic counsellors and support groups

### When should genetic counselling be sought?

There are a number of reasons for which genetic counselling is appropriate. These include:

- When there is a condition that runs in a family and individuals are concerned that they or their children will develop the condition
- Where a previous child is affected by a serious problem in growth, development or health
- Where one or more family members (blood relatives not related by marriage) have unusual features, or a serious health problem
- Where a woman is in her mid 30s or older and is either planning a pregnancy or is already pregnant
- When a couple are blood relatives (see Genetics Fact Sheet 16)

- Where an individual or their partner has some concerns about a condition in themselves or their family being passed on to their children
- When a fetal abnormality is detected during pregnancy
- When there is concern about exposure to some environmental agent such as drugs, medications, chemicals or radiation that might cause birth defects

### How can genetic counselling be accessed?

The availability of genetics services will vary throughout Australian States and Territories and New Zealand. Sometimes the service is based in specialist paediatric or obstetric departments of a hospital. Any individual or family with any of the concerns listed above can obtain genetic counselling at these centres.

It is preferable that referral to these genetic counselling services is through a general practitioner, a medical specialist or other health care professional.

**Other Genetics Fact Sheets referred to in this Fact Sheet: 16, 17, 18, 21**

#### Information in this Fact Sheet is sourced from:

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