



# Genetic counselling

Information for Doctors

As with any pathology test, a patient having a genetic test needs to understand the purpose and potential consequences of having the test. In addition, genetic tests raise specific issues, either because the test is new or because the potential consequences of the test can involve a patient's relatives.

Genetic counsellors are accredited allied health professionals who can assist doctors and patients prior to a genetic test being performed, or after the genetic test has been reported.

Sonic Genetics maintains a list of genetic counselling services nationally (available on our website). We also provide free genetic counselling in a number of specific situations for patients or couples who have been tested by Sonic Genetics, through one of the Sonic Healthcare pathology laboratories.

This leaflet provides doctors with an outline of the purpose and process of genetic counselling.

Genetic counselling involves the provision of technical genetic information and emotional support in a way which helps a person (or couple) make informed decisions about what they want to do with that information.



**The provision of complex information to an uninformed person can take time, starting from basic concepts and building up to the specifics of the person's situation. There must also be opportunities for questions at each step.**

A genetic counselling session will usually cover the following technical information:

- The disorder of concern; many of these disorders are rare
- The genetic basis of the disorder, and the relevance of any test results
- The clinical significance of the result for the patient and (if relevant) their partner and genetic relatives; this discussion could include implications for current and future children
- Reproductive options which may be relevant, including the potential use of donor gametes, pre-implantation genetic diagnosis, prenatal diagnosis (by CVS), or simply accepting the risk that has not been clarified.

A genetic counsellor would also address the following psychosocial issues:

- Some people are shocked by the diagnosis or possibility of a genetic disorder; this possibility is usually unexpected
- The information and implications for both the person and their relatives may be emotionally challenging
- A person's initial beliefs and understanding may not align with the technical information
- A person and their partner or other relatives may have different beliefs, or respond differently to the same technical information
- The business of 'considering reproductive options' is itself confronting and typically very different from a couple's desire to simply 'have a baby' by natural means
- Each reproductive option carries its own practical, financial, emotional and ethical consequences.

These technical and psychosocial factors combine to create a complex situation in which people may need to make decisions which could have ramifications for both them and their families for years to come. A genetic counsellor seeks to inform and support a person in making a decision that must ultimately be theirs, and theirs alone.

## The genetic counselling session

### Family history-taking

A three-generation family history of both partners will be taken and used to develop a genetic pedigree. Please advise your patients that they should find out as much information as possible about their family history before the session with a genetic counsellor. The taking of the family history may identify genetic risks for other relevant disorders which would warrant discussion and/or investigation.

### The disorder

Information about the disorder identified (or potentially identified) through genetic testing, its potential physical and/or developmental and/or intellectual impact, its general age of onset and available resources will be provided. The amount of information given will be adapted to the interest and needs of the couple. The variability of genetic disorders means that it may be difficult to predict how severe the disorder in a specific person will be or the age at which it is likely to occur.

### The genetic basis of the disorder and the risks of children or other relatives being affected

The explanation of genes, chromosomes and inheritance will be adapted to the couple's interest and needs. This provides the basis for understanding the risk of a child or other genetic relatives being affected.

### Decision-making

The combination of complex technical information, a serious disorder (which is typically rare), emotional sequelae and implications for relatives is confronting. Many people will need time to consider and reflect in making any decisions that need to be made, and will feel completely overwhelmed.

The situation is even more complex when considering a reproductive issue, as a couple may not have the same views on disability, the severity of a disorder, and appropriate reproductive strategies.

Clarifying and resolving these issues will take time and may involve a number of sessions with the genetic counsellor. The goal of the process is to ensure that a person has the information, support and guidance to allow them to make decisions and choices with confidence.

### Referring for genetic counselling

Genetic counselling services are provided through public sector services and private providers (please refer to [www.sonicgenetics.com.au/doctors/resources](http://www.sonicgenetics.com.au/doctors/resources)). Sonic Genetics does not provide these services, and patients and doctors may contact these services directly.

Sonic Genetics provides free genetic counselling in certain situations for patients who have one of our tests. Details are provided with eligible test requests and reports, and conditions apply.