Genetic Counsellors are health professionals with training in genetics and counselling. They provide information and psychosocial support to individuals or families who have concerns about a genetic condition in their family.

People often seek genetic counselling to help understand the risk for them and other family members of having a child affected by a genetic condition. Couples are then able to make informed decisions about future childbearing (please see ‘What are my family planning options’ below). In addition, genetic counsellors aim to help individuals, couples and families understand and adapt to the medical and psychological implications of the diagnosis of a genetic condition in their family.

It should be stressed that genetic counsellors will not tell, or even advise, a couple as to whether or not they should have children, or make any other decisions for that matter, such as whether to have a genetic test and who to tell the results to. However, genetic counsellors are skilled at helping people to reach decisions which are appropriate for the individual and their family.

A first genetic counselling appointment will usually take between 1-2 hours. Your genetic counsellor will take a record of your family and personal medical history. They will ask questions about your relatives such as names, dates of birth and death, causes of death and general health. The results of any genetic tests already taken will be discussed as well as any further testing that might be possible.

Sometimes it may not be possible to get a genetic diagnosis or a particular genetic test might not be available. In this case the counsellor will provide more general information based on the family history. If a diagnosis isn’t made it may be appropriate to visit the clinic at a later point in time as the field of genetics is advancing at a fast pace.

The counsellor will provide information on the condition, how it may affect family members and how it is inherited in your family. Any relevant support resources and research information will also be passed on. They will provide support and counselling to promote informed choices which may include information on family planning options (see below).

You may like to bring a family member or friend along to your genetic counselling appointment for additional support. After the initial consultation an opportunity may be provided to go over the information and offer on-going support.
Is the counselling confidential?
Yes. Genetic Counsellors will not talk about your information with anyone, even other members of your family. This ensures the privacy of both you and your family members. The information you discuss with your genetic counsellor will be sent out to you in a letter after your appointment, which can be used to help you inform family members if you wish. Your information may be given to another medical practitioner in the course of your management (if relevant) only after consent is given to do so.

How can I prepare for my appointment to the genetic counselling service?
Before attending a genetic counselling session, it may be helpful to find out as much information as you can about the medical history of both sides of your family as this is the type of information discussed at your appointment.

It’s a good idea to write down any questions you think of before going to a session to make sure they are answered.

What are my family planning options?
For couples that find out that they are at risk of passing on a genetic condition, one option, of course, is to accept the risk and have children. A genetic counsellor can help you to understand the risk so your decision is an informed one. Other couples may consider adoption.

Another option is to do IVF using an egg donor if it is the woman who is the carrier or sperm donor if it is the man who is the carrier of the condition. A donor embryo may also be an option. If having a biological child is important to a couple there are other options detailed below.

Prenatal testing
Prenatal testing involves testing the fetus in early pregnancy to find out if it is affected by the genetic condition. If the test comes back positive the couple then have the option to terminate the pregnancy or continue armed with the knowledge that will help them plan and prepare for the future.

Prenatal tests are only available for some neuromuscular disorders and ideally genetic counselling should be sought well before becoming pregnant. A certain amount of information needs to be known about the particular condition running in your family and its genetic cause for the test to be done. Your genetic counsellor will be able to provide information on whether it will be possible to do the test in your case and give advice on next steps.

There are two different methods of prenatal testing:

- Chorionic Villus Sampling (CVS) is usually done around weeks 11 to 12 of pregnancy and involves using fine needle to take a small sample of the placenta. One woman in one hundred (1%) will have a miscarriage as a result of this test.
• Amniocentesis is usually done around weeks 15 to 18 of pregnancy and involves inserting a fine needle to collect a sample of the amniotic fluid that surrounds the baby. Amniocentesis has a one in two hundred risk of causing a miscarriage.

**IVF and preimplantation genetic diagnosis (PGD)**

Recent technological advances have allowed couples that carry certain genetic conditions to have treatment at fertility (IVF) clinics to prevent passing on the condition. This typically involves couples undergoing standard in vitro fertilisation (IVF) during which eggs are fertilised by sperm outside the womb and then the resulting embryos are tested in a procedure known as preimplantation genetic diagnosis (PGD). The embryos are grown in the laboratory for several days and the embryo’s DNA is tested for the presence of the genetic change. Only embryos that do not carry the genetic change are transferred back into the woman’s womb. In Australia PGD must be paid for privately.

Success rates for having a child from an IVF cycle followed by PGD vary from IVF centre to centre but tend to follow standard IVF success rates. As for prenatal testing above, PGD is only available if the affected gene is known. A new diagnostic test needs to be developed for each couple and this can take up to six months.

A new PGD technique called karyomapping has become available in some fertility clinics. It uses a DNA finger-printing technique to identify which embryos have inherited the altered gene. Again, the affected gene needs to be known, but the test is much quicker to set up than previous PGD tests. Another advantage is that information about the whole genome is obtained which can allow chromosomal errors, which can cause the embryo to fail to implant or to miscarry, to be identified. As a result the embryos with the best chance of producing a healthy child can be selected, potentially improving the IVF success rate.

It is important to consider that all IVF procedures can be stressful and some couples require many rounds of treatment for success.

**How do I find a Genetic Counselling service near me?**

Most clinical genetics services require a referral from a health professional, so your GP, neurologist or other specialist is an important first port of call. Self-referral may be acceptable to some clinical genetics units; you can call them to check. The Human Genetic s Society of Australia has a list of genetic counselling services in Australia and New Zealand [https://www.hgsa.org.au/asgc/find-a-genetic-counsellor](https://www.hgsa.org.au/asgc/find-a-genetic-counsellor)

**Background information**

**What is a genetic condition?**

All of our inborn traits, from the colour of our eyes to our blood types, are determined by our genes - chemical bits of information that are the basic units of hereditary. Genes are carried on the rod-like structures known as chromosomes, which are found in every cell in our bodies (except red blood cells). We have more than 20,000 genes and certain changes (called mutations) in certain genes lead to the development of genetic conditions such as muscular dystrophy.
Genetic conditions are passed down through families. Which family members are at risk and how big that risk is differs for different conditions. This is due to what is called “patterns of inheritance”. For example, some conditions usually only affect males (e.g. Duchenne muscular dystrophy). Some genes are also described as being “recessive” or “dominant” which affects the risk of a condition being inherited.

Sometimes a genetic condition appears for the first time in a family with the genetic change not being present in either parent. This is due to a spontaneous genetic change in the egg or sperm or in the fertilized egg shortly after the egg and sperm cells unite during conception. In some conditions such as Duchenne muscular dystrophy and facioscapulohumeral muscular dystrophy it is thought that up to one third of cases may be due to spontaneous genetic changes.

It must be emphasized, however, that absence of a family history does not mean that the condition has resulted from a spontaneous mutation. It may be that the mutation has been in the family for a number of generations and has not shown up before, just by chance. For many conditions carrier testing is available to help to clarify this (see below).

**What is carrier testing?**

A carrier is a person who usually shows no symptoms of a condition but who carries the genetic change which causes it. Sometimes carriers can have very mild symptoms of the condition, as can be the case with carriers of the genetic change that causes Duchenne muscular dystrophy.

There is a risk that a 'carrier' will pass the faulty gene on to his or her children. The level of that risk depends on the inheritance pattern of the condition (this is described in the factsheet relating to the particular condition in which you are interested, or you can discuss it with your genetic counsellor).

Carrier tests are done by examining the DNA in a blood sample. They are most straightforward if the specific genetic change responsible for the disorder in a particular family is known. This is because the scientists in the laboratory will then know exactly where in the genetic code to look for the change. If the genetic change is not known other options may be possible depending on the condition and what is known about it.

**Further information**

- Factsheets on individual neuromuscular disorders, which include information on inheritance, can be found on the MDA website [http://www.mda.org.au](http://www.mda.org.au) ('Disorders' tab at the top of the page)