

Genetic Counselling

What is Genetic Counselling?

Genetic counselling is a service that is offered to people who have an inherited condition, or people who have, or may have family members with an inherited condition. This could be someone diagnosed with a rare genetic disorder and their families, expectant parents undergoing prenatal testing, or adults looking to learn more about their risk of inheritance.

Genetic counsellors aim to provide a full understanding of inheritance patterns, the risk of inheriting the condition, the different tests available, and the test results and implications. The service is available before testing takes place, during testing, and after you receive the results.

What can I expect?

Each person who attends genetic counselling has their own reasons for doing so, and it is important that the genetic counsellor understands your reasons. They will spend time to talk to you and explore your experience of the condition, understand your family, and gain an insight into any worries and concerns you may have.

Your Family Tree

The genetic counsellor will look at the prevalence of the condition and you will work together to draw out a family tree. This helps the counsellor to look at your family and the medical history, to assess the risk of having a genetic condition. It will also help you to make sense of your family history and understand the different patterns of inheritance. This video helps to explain autosomal recessive inheritance, one of the most common modes of inheritance in inherited metabolic disorders

<https://vimeo.com/album/5216273/video/273660858>

Consent Forms

The Genetic Counsellor will need you to sign a consent form to allow them to perform a genetic test. Prior to you signing they will ensure you have a full understanding of any tests and will provide information about the results. The consent form also gives permission to share the results with any relevant medical professionals such as your GP and your specialist, and family members who may also need a test, as well as permission to keep the sample on record in case you need it for the future. It is important to understand that there can be some uncertainties in genetic testing and your Genetic counsellor will explain this. They will also give a realistic time frame for when your results will be ready and plan in the best way for you to receive these results, usually a face-to-face appointment is the best approach as it gives your Genetic Counsellor the opportunity to support you and discuss your feelings.

Results

Your Genetic Counsellor will share your test results with you and fully explain the impact these may have on you and your family. They will help you to make sense of how you may be feeling and discuss your worries and concerns. They will also help to identify any next steps in relation to support and management of the condition and/or follow ups. They will also discuss the impact on other family members. There are different options of how to contact other family members, your Genetic

Counsellor may invite you to bring them in clinic with you. However, in most cases, you will be given a letter which can be passed on to your relatives which they can take to their GP for referral.

Your Genetic Counsellor will always ensure you have accurate and helpful information, this could include explanations as to why your baby and any future children may have an increased risk of having a condition, how tests are carried out, and any risks. They will also signpost you to information sources, or support groups so you can do your own research about living with the condition and will help you to discuss any options available to you, particularly important if your result means you are considering whether to continue with a pregnancy.

Regardless of your reason for accessing this service or the outcome of any tests, your Genetic Counsellor will always be understanding, non-judgemental, and supportive and will help you to navigate information, options, and empower you to make any decisions that are right for you.

How do I access Genetic Counselling?

You will usually be referred to a genetic counselling service by your GP. However, Genetic Counsellors are also based in certain clinics, especially in Inherited Metabolic Disorders/rare genetic disease clinics, where they will generally be a part of the multidisciplinary team involved in the care and management of yours or your child's condition.