

Understanding the Role of a Geneticist and Genetic Counsellor

This guide aims to give families a clear understanding of the role geneticists and genetic counsellors play in their journey with Usher syndrome, empowering them to seek the appropriate support when needed.

Families navigating a diagnosis of Usher syndrome will likely encounter geneticists and genetic counsellors. While the geneticist focuses on the medical and scientific aspects of the diagnosis, the genetic counsellor complements this by providing psychosocial support, education, and guidance, helping families process complex medical information and make informed decisions. Genetic counsellors may also assist with family planning decisions, such as the likelihood of future children being affected by Usher syndrome as well as options for prenatal testing.



While the geneticist's primary role is medical, a genetic counsellor focuses on education and support.

Geneticist: A geneticist is a medical doctor who specialises in the study of genes, genetic variations, and heredity in living organisms. Geneticists focus on understanding how genes influence health and disease, playing a key role in diagnosing and treating genetic conditions. The geneticist is responsible for ordering the appropriate genetic tests based on the patient's clinical symptoms and family history. Once the genetic tests are complete, the geneticist interprets the results to confirm a diagnosis. They determine the specific genetic mutation responsible for the condition and how it aligns with the clinical symptoms.

Genetic Counsellor: A genetic counsellor is a healthcare professional with specialist knowledge in human genetics, counselling, and health communication. They provide critical information and support to individuals and families dealing with genetic conditions. Genetic counsellors typically work as part of a multidisciplinary team that includes a geneticist. The genetic counsellor meets with the family to explain the geneticist's findings in a way that is easy to understand. They discuss the inheritance pattern, risk factors for other family members, and the long-term implications of the condition.

Key Responsibilities of a Geneticist:

Geneticists are responsible for diagnosing genetic disorders by evaluating symptoms and ordering tests.

Key Responsibilities of a Genetic Counsellor:

Genetic counsellors assess the implications of the genetic diagnosis for family members. They offer support to help individuals and families navigate the emotional aspects of the diagnosis. They can provide information about the likelihood of other family members being affected or being carriers of the same genetic mutation. This helps in making informed decisions about family planning and potential testing for relatives.

When Should You See a Geneticist or Genetic Counsellor?

You may want to consider seeing a geneticist or genetic counsellor for a variety of reasons, including:

Clinical Diagnosis: Genetic testing can confirm a clinical diagnosis by identifying specific gene mutations associated with a particular condition. This allows for a better understanding of the condition, helping healthcare providers predict the progression of the disease based on the genetic type. While a geneticist is responsible for ordering genetic tests and analysing the results, genetic counselling helps patients and their families understand the results of the genetic tests. The results can be complex, and a genetic counsellor explains the findings in a clear and accessible way.

Reproductive Genetic Counselling: Families with a history of genetic disorders, or those who have a child with a genetic diagnosis may seek genetic counselling before or during a subsequent pregnancy. This includes preconception counselling to understand the risk of passing on genetic disorders to their future children, prenatal testing during pregnancy, or pre-implantation genetic testing during IVF to select embryos without the known genetic condition. This helps in making informed reproductive choices.

Intervention Therapies/Research: Genetic counselling can help identify potential candidates for clinical trials or therapies, where cutting-edge interventions, such as cell or gene therapies, may be available. These treatments could be in experimental stages but offer hope for managing genetic conditions. Some studies on Usher syndrome will be gene-specific so it's important to get the genetic diagnosis confirmed by genetic testing if that is something a patient is interested in pursuing.

How to Find a Geneticist or Genetic Counsellor:

Families can ask their child's GP, paediatrician, or specialist (such as an ophthalmologist or audiologist) for a referral to a geneticist or a genetic counsellor.