Genetics Uncoded:

Facts about

Usher Syndrome Type 3

What Your Test Results Mean

Carriers typically show no symptoms of Usher syndrome type 3; however, carriers are at an increased risk of having a child with Usher syndrome type 3. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Usher Syndrome Type 3 Explained

Usher syndrome type 3 is an inherited disorder characterized postlingual progressive hearing loss and onset of progressive vision loss in the second decade. Individuals with Usher syndrome type 3 are not able to produce the protein clarin 1, a protein required for nerve cell communication in the inner ear and retina. This protein is essential for proper hearing and vision. Deficient levels of clarin 1 cause the vision and hearing problems characteristic of the disease. The disease does not affect intelligence or lifespan of individuals with Usher syndrome type 3. Individuals with Usher syndrome type 3 may also experience difficulties with balance due to inner ear problems.

Treatment is mostly supportive. Optimizing communication is important. Because hearing loss is postlingual, speech is maintained; however, some individuals opt to learn sign language while others opt for hearing aids or cochlear implantation. Routine eye exams are recommended.

How the Genetics Work

The clinical features of Usher syndrome type 3 can be explained by pathogenic variants in the *CLRN1* gene. In general, individuals have two copies of the *CLRN1* gene. Carriers of Usher syndrome type 3 have a single variant in one copy of the *CLRN1* gene while individuals with Usher syndrome type 3 have variants in both copies of their genes, one inherited from each parent.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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