



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 by 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 **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.