Genetics Uncoded:

Facts about

Usher Syndrome Type 1F

What Your Test Results Mean

Carriers typically show no symptoms of Usher syndrome type 1F; however, carriers are at an increased risk of having a child with Usher syndrome type 1F. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Usher Syndrome Type 1F Explained

Usher syndrome type 1F is an inherited disorder characterized by congenital sensorineural hearing loss and progressive vision impairment presenting in adolescence. Individuals with Usher syndrome type 1F are not able to produce the protein protocadherin 15, a protein essential for proper development of the cells of the inner ear and photoreceptors in the eye. This protein is essential for proper hearing and vision. Deficient levels of protocadherin 15 cause the vision and hearing problems that are characteristic of the disease. In addition to vision and hearing problems, individuals with Usher syndrome type 1F may have problems with balance and typically have delayed motor milestones as a result. The disease does not affect intelligence or lifespan; however, many individuals do not develop speech and benefit from learning other forms of communication such as sign language.

Treatment is mostly supportive. Optimizing communication at an early age is important. While hearing aids are not effective for individuals with Usher syndrome type 1F, some families opt for cochlear implantation. Routine eye exams are recommended.

How the Genetics Work

Usher syndrome type 1F represents approximately 7-12% of Usher type 1. The clinical features of Usher syndrome type 1F can be explained by pathogenic variants in the *PCDH15* gene. In general, individuals have two copies of the *PCDH15* gene. Carriers of Usher syndrome type 1F have a single variant in one copy of the *PCDH15* gene while individuals with Usher syndrome type 1F 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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