NxGen MDx

Usher Syndrome Type IIIA

What Your Results Mean

Test results indicate that you are a carrier of Usher syndrome type IIIA. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Usher syndrome type IIIA for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner are carriers for Usher syndrome type IIIA, each of your children has a 1 in 4 (25%) chance to have the condition.

Usher Syndrome Type IIIA Explained

What Is Usher Syndrome Type IIIA?

Usher syndrome type IIIA is an inherited disorder characterized by severe-to-profound hearing loss and vision loss that worsens over time. Individuals with this type of Usher syndrome typically have normal hearing at birth and begin to experience hearing loss during late childhood or adolescence, after the development of speech, and the hearing loss becomes more severe over time. Vision loss begins in late childhood or adolescence and progressively worsens over time. Some individuals with Usher syndrome type IIIA may also experience difficulties with balance due to the development of inner ear abnormalities.

Prognosis

Individuals have severe hearing and vision impairment. The condition does not affect a person's life expectancy or intelligence.

Treatment

Treatment is mostly supportive; there is no cure. Optimizing communication is important. Because hearing loss is post lingual, 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.

Resources

Usher Syndrome Coalition
https://www.usher-syndrome.org/

National Institute on Deafness and Other Communication Disorders
https://www.nidcd.nih.gov/health/usher-syndrome

National Society of Genetic Counselors
https://www.nsgc.org/

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