Usher Syndrome Type ID/ *CDH23*-Related Disorders

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of Usher syndrome type ID/*CDH23*-related disorders. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Usher syndrome type ID/*CDH23*-related disorders 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 ID/*CDH23*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

Usher Syndrome Type ID/*CDH23*-Related Disorders Explained

What is Usher Syndrome Type ID/*CDH23*-Related Disorders?

Usher syndrome type ID/*CDH23*-related disorders is a group of inherited disorders characterized by mild-to-severe hearing loss and vision loss that worsens over time. The hearing loss is usually present at birth, and vision loss begins in childhood and progressively worsens over time. The cause of hearing loss for these individuals is inner ear abnormalities. The inner ear is what also helps maintain the body's balance and orientation in space. Therefore, individuals with Usher syndrome type ID/CDH23-related disorders can also have problems with balance and delayed motor milestones as a result. Many individuals do not develop speech and benefit from learning alternative forms of communication, such as sign language.

Prognosis

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

Treatment

Treatment is mostly supportive, as there is no cure. Optimizing communication at an early age is important. Hearing aids are usually not effective, and some individuals opt for 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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