

Usher Syndrome Type IIA/ *USH2A*-Related Disorders

What Your Results Mean

Test results indicate that you are a carrier of Usher syndrome type IIA/*USH2A*-related disorders. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of Usher syndrome type IIA/*USH2A*-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 IIA/*USH2A*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

Usher Syndrome Type IIA/*USH2A*-Related Disorders Explained

What is Usher Syndrome Type IIA/*USH2A*-Related Disorders?

Usher syndrome type IIA/*USH2A*-related disorders is a group of inherited disorders characterized by mild-to-severe hearing loss and vision loss that worsen over time. Individuals with this type of Usher syndrome typically have hearing loss at birth that can get more severe over time. Vision loss begins in adolescence or adulthood and progressively worsens over time.

Prognosis

Individuals have 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 is important. Some individuals may benefit from hearing aids or cochlear implantation and speech training to normalize language, while others opt to learn sign language. 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/>