SLC35A3-Related Disorders

What Your Results Mean

Test results indicate that you are a carrier of *SLC35A3*-related disorders. Carriers are not expected to show symptoms. You and your partner would both have to be carriers of an *SLC35A3*-related disorder 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 *SLC35A3*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

SLC35A3-Related Disorders Explained

What is an *SLC35A3*-Related Disorder?

An *SLC35A3*-related disorder is an inherited condition that is characterized by arthrogryposis (immobility of the joints due to muscle fibrosis), intellectual disabilities, and seizures. In addition, individuals may have small head size, poor muscle tone, and a diagnosis of autism.

Prognosis

Prognosis is currently unknown, as it has only been noted in a small subset of individuals.

Treatment

Treatment is supportive and symptomatic, as there currently is no cure. Anti-seizure medications help with seizures and physical and occupational therapy can be beneficial for affected individuals.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/condition/SLC35A3
National Society of Genetic Counselors
https://www.nsgc.org/

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