POMGNT1-Related Disorders

What Your Results Mean

Test results indicate that you are a carrier of a *POMGNT1*-related disorder. Carriers typically show no symptoms. You and your partner or donor would both have to be carriers of a *POMGNT1*-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 or donor are carriers for the *POMGNT1*-related disorder, each of your children has a 1 in 4 (25%) chance to have the condition.

POMGNT1-Related Disorders Explained

What are *POMGNT1*-Related Disorders?

Limb-Girdle Muscular Dystrophy

Limb-girdle muscular dystrophy is a term for a group of diseases that cause weakness and wasting of the muscles in the arms and legs. The muscles most affected are those closest to the body (proximal muscles), specifically the muscles of the shoulders, upper arms, pelvic area, and thighs. The severity, age of onset, and features of limb-girdle muscle dystrophy vary among the many subtypes of this condition and may be inconsistent even within the same family. Signs and symptoms may first appear at any age and generally worsen with time, although in some cases they remain mild. Intelligence is generally unaffected with limb-girdle muscular dystrophy; however, developmental delay and intellectual disability have been reported in rare forms of the disorder.

Muscular Dystrophy-Dystroglycanopathy Congenital with Brain and Eye Anomalies A3 (MDDGA3)

An autosomal recessive disorder characterized by congenital muscular dystrophy, eye abnormalities, a brain malformation known as cobblestone lissencephaly, and underdevelopment of the cerebellar and pontine brain structures. Affected individuals typically present with eye abnormalities, congenital glaucoma, mental retardation, fluid around the brain (hydrocephalus), generalized muscle weakness and myoclonic jerks. Included diseases are the more severe Walker-Warburg syndrome and the slightly less severe muscle-eye-brain disease.

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Muscular Dystrophy-Dystroglycanopathy Congenital with Mental Retardation B3 (MDDGB3)

MDDGB3 is an autosomal recessive disorder characterized by congenital muscular dystrophy associated with intellectual disabilities and mild structural brain abnormalities. Other features include white matter changes, cerebellar cysts, pontine underdevelopment, and eye abnormalities.

Retinitis Pigmentosa 76 (RP76)

Retinitis pigmentosa is an autosomal recessive condition characterized by progressive degeneration of the retina, the light sensitive membrane that coats the inside of the eyes. Individuals typically have night vision blindness and loss of mid peripheral visual field. As their condition progresses, they lose their far peripheral visual field and eventually central vision as well.

Prognosis

Prognosis depends upon the type of *POMGNT1*-related disorder. Some conditions, such as Walker-Warburg syndrome, have a poor prognosis as life expectancy is typically three years. However, retinitis pigmentosa should not affect life expectancy as it only affects vision.

Treatment

There is no cure for *POMGNT1*-related disorders, therefore treatment is symptomatic. Medical management can require the coordinated efforts of a team of specialists including pediatricians, geneticists, orthopedic surgeons, neurologists, eye specialists, and other health care professionals.

Resources

Genetics Home Reference
https://ghr.nlm.nih.gov/gene/POMGNT1#conditions
https://ghr.nlm.nih.gov/condition/limb-girdle-muscular-dystrophy#:~:text=Description,%2C%20pelvic%20area%2C%20and%20thighs.
National Organization for Rare Disorders
https://rarediseases.org/rare-diseases/walker-warburg-syndrome/
Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/156/muscle-eye-brain-disease#:~:text=Muscle%20eye%20brain%20disease%20(MEB,%2C%20glaucoma%20%2C%20and%20brain%20abnormalities.
National Society of Genetic Counselors
https://www.nsgc.org/

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