FKTN-Related Disorders (Including Walker-Warburg Syndrome)

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of *FKTN*-related disorders (including Walker-Warburg syndrome). Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of *FKTN*-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 or donor are carriers for *FKTN*-related disorders, each of your children has a 1 in 4 (25%) chance to have the condition.

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What is Walker-Warburg Syndrome?

Walker-Warburg syndrome is a genetic condition that affects the development of the muscles, brain, and eyes. It belongs to a group of congenital muscular dystrophies and is considered the most severe of them. Signs and symptoms first appear at birth. Walker-Warburg syndrome is characterized by brain malformations, eye abnormalities, hypotonia, muscle weakness, developmental delay, and occasional seizures.

What is an *FKTN*-Related Disorder?

Other conditions caused by a change in the FKTN gene include Fukuyama congenital muscular dystrophy and limb girdle muscular dystrophy type 2M. Individuals with Fukuyama congenital muscular dystrophy first show signs and symptoms during infancy. These include muscle weakness, brain abnormalities, glaucoma, cataracts, and nearsightedness. Individuals additionally have a moderate to severe intellectual disability and may not be able to walk or stand. However, it is important to note that these signs and symptoms are less severe than in Walker-Warburg syndrome. Limb girdle muscular dystrophy type 2M is the mildest of FKTN-related disorders. Age of onset varies for limb girdle muscular dystrophy type 2M. Symptoms include muscle weakness typically occurring in the shoulders, thighs, upper arms, and pelvic area of individuals. Some individuals may have dilated cardiomyopathy. The eyes and brain are not affected in this milder form.

Prognosis

Prognosis depends on the severity of the diagnosis. Individuals diagnosed with Walker-Warburg syndrome typically do not live beyond the age of three. Individuals with Fukuyama congenital muscular dystrophy typically live into their teens or early adulthood. Life expectancy is typically unaffected by limb girdle muscular dystrophy type 2M.

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Treatment

Treatment of individuals with Walker-Warburg syndrome and *FKTN*-related disorders is supportive as there is no cure. Supportive treatment may include medication for seizures and physical and occupational therapy. Some individuals benefit from shunting for fluid buildup around the brain (hydrocephalus). Placement of a feeding tube may be necessary. Treatment for limb girdle muscular dystrophy type 2M may include physical therapy, occupational therapy, and monitoring of the heart.

Resources

Cure Congenital Muscular Dystrophy
https://www.curecmd.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/walker-warburg-syndrome/
National Society of Genetic Counselors
https://www.nsgc.org/

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