FKRP-Related Disorders (Including Walker-Warburg Syndrome)

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of *FKRP*-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 *FKRP*-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 *FKRP*-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 *FKRP*-Related Disorder?

Other conditions caused by a change in the *FKRP* gene include limb girdle muscular dystrophy type 2I (LGMD2I), congenital muscular dystrophy type 1C (MDC1C), and rarely, variants in the *FKRP* gene are associated with muscle-eye-brain disease. Age of onset of symptoms for LGMD2I is variable as individuals may present with symptoms anytime between age one and 40. The condition is characterized by weakness of the hip and shoulder girdle, difficulty walking, spinal abnormalities including scoliosis, lordosis and kyphosis, and cardiomyopathy. Individuals may also experience difficulty breathing. The severity varies between affected individuals with some never requiring a wheelchair.

MDC1C causes muscle weakness, brain abnormalities, and intellectual disability but usually does not affect the eyes. Muscle-eye-brain disease causes muscle weakness, eye problems, and intellectual disability. The signs and symptoms of muscle-eye-brain disease are less severe than those of Walker-Warburg syndrome.

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Revised: 7/2020

Prognosis

Prognosis depends on the severity of the diagnosis. Individuals diagnosed with Walker-Warburg syndrome typically do not live beyond the age of three. Lifespan for individuals with limb girdle muscular dystrophy type 2I is unknown, though many types of limb girdle muscular dystrophy are associated with a shortened lifespan. Prognosis for congenital muscular dystrophy type 1C is typically poor as these individuals typically have a shortened lifespan. Prognosis for muscle-eye-brain disease is fair as individuals can survive for more than 70 years, though quality of life may be affected. Supportive care enhances the quality of life and the life expectancy.

Treatment

Treatment of individuals with Walker-Warburg syndrome and *FKRP*-related disorders is supportive as there is no cure. Supportive treatment for Walker-Warburg syndrome 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 2I may include physical therapy, occupational therapy, and monitoring of the heart. Individuals with congenital muscular dystrophy type 1C may also benefit from physical, occupational, and speech therapy. Additionally, they may require the use of various devices (e.g., canes, braces, walkers, wheelchairs) to assist with walking and mobility, surgery to correct skeletal abnormalities such as scoliosis, and regular monitoring of the heart and the respiratory system for the development of such associated complications.

Resources
Cure Congenital Muscular Dystrophy
https://www.curecmd.org/
National Organization for Rare Disorders (NORD)
https://rarediseases.org/rare-diseases/walker-warburg-syndrome/
https://rarediseases.org/rare-diseases/congenital-muscular-dystrophy/
Genetic and Rare Diseases Information Center
https://rarediseases.info.nih.gov/diseases/12533/limb-girdle-muscular-dystrophy-type-2i
National Society of Genetic Counselors
https://www.nsgc.org/

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