Genetics Uncoded:

Facts about

FKTN-Related Walker-Warburg Syndrome

What Your Test Results Mean

Carriers typically show no symptoms of *FKTN*-related Walker-Warburg syndrome (*FKTN*-related WWS); however, carriers are at an increased risk of having a child with *FKTN*-related WWS. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

FKTN-Related Walker-Warburg Syndrome Explained

Walker-Warburg syndrome is the most severe congenital muscular dystrophy. *FKTN*-related WWS is characterized by brain malformations and eye abnormalities, hypotonia, muscle weakness, developmental delay, and occasional seizures. Individuals with *FKTN*-related WWS are not able to properly produce the protein fukutin. Without fukutin, another protein called alpha-dystroglycan, essential for the development of muscle fibers and neural cells, cannot function properly, leading to the severe symptoms of the disease. *FKTN*-related WWS is typically lethal within the first few months of life.

Treatment of individuals with *FKTN*-related WWS typically includes supportive care. Most individuals do not live beyond the age of three.

How the Genetics Work

There are several genes known to cause Walker-Warburg syndrome. The clinical features of *FKTN*-related WWS can be explained by pathogenic variants the *FKTN* gene. In general, individuals have two copies of the *FKTN* gene. Carriers of *FKTN*-related WWS have a single variant in one copy of the *FKTN* gene while individuals with *FKTN*-related WWS have variants in both copies of their genes, one inherited from each parent.

Questions? Contact us at **1-855-776-9436** to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.

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