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.