Genetics Uncoded:

Facts about

Glycogen Storage Disease Type 3

What Your Test Results Mean

Carriers typically show no symptoms of glycogen storage disease type 3; however, carriers are at an increased risk of having a child with glycogen storage disease type 3. Because risk for offspring depends on both parents' carrier status, carrier testing regardless of sex is recommended.

Glycogen Storage Disease Type 3 Explained

Glycogen storage disease type 3 is an inherited metabolic muscle disorder caused by lack of the glycogen debrancher enzyme. The missing enzyme causes abnormal glycogen to build up in the muscles and liver. Symptoms include an enlarged liver, low blood sugar, and growth retardation. Later in life, muscle weakness and cardiomyopathy (heart muscle weakness) may develop. Glycogen storage disease type 3 has no cure, but high-protein diets and physical therapy may alleviate some symptoms. Individuals with glycogen storage disease type 3 are at an increased risk for infant fatalities due to seizures caused by low blood sugar, but most people with this disease live well into adulthood.

How the Genetics Work

Glycogen storage disease type 3 is an autosomal recessive disorder caused by pathogenic variants in the *AGL* gene. In general, individuals have two copies of the *AGL* gene. Carriers of Glycogen storage disease type 3 have a single variant in one copy of the *AGL* gene while individuals with Glycogen storage disease type 3 have variants in both copies of their *AGL* genes, one inherited from each parent. Risk for two carriers to have a child with the disorder is 25%.

Questions? Contact us at **1-866-661-7966** to set up an appointment to discuss your results in more detail with a Clarity Genetics genetic counselor.

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