Genetics Uncoded:

# Facts about

# Glycogen Storage Disease Type V

## What Your Test Results Mean

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

## Glycogen Storage Disease Type V Explained

Glycogen storage disease type V is an inherited metabolic muscle disorder in which muscle cells cannot break down glycogen to release usable carbohydrates. A deficient level of the enzyme myophosphorylase causes individuals with glycogen storage disease type V to suffer from exercise intolerance. During the first few minutes of exercise, they will experience fatigue, muscle pain, and cramps. Excessive exercise can lead to kidney failure. These symptoms can appear during childhood, but often do not occur until adulthood. There is no treatment for glycogen storage disease type V except to restrict the frequency and intensity of exercise.

## How the Genetics Work

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

**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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