

# Glycogen Storage Disease Type III

## What Your Results Mean

Test results indicate that you are a carrier of glycogen storage disease type III. Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of glycogen storage disease type III 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 glycogen storage disease type III, each of your children has a 1 in 4 (25%) chance to have the condition.

## Glycogen Storage Disease Type III Explained

### What is Glycogen Storage Disease Type III?

Glycogen storage disease type III 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.



## Prognosis

Prognosis is typically mild. Individuals with glycogen storage disease type III are at an increased risk for infant fatalities due to seizures caused by low blood sugar. With proper management, life expectancy is not impacted.

## Treatment

Glycogen storage disease type III has no cure, but high-protein diets and physical therapy may alleviate symptoms.



### Resources

#### Association for Glycogen Storage Disease

<https://www.agsdus.org/type-ii.php>

#### Genetics Home Reference

<https://ghr.nlm.nih.gov/condition/glycogen-storage-disease-type-iii>

#### National Society of Genetic Counselors

<https://www.nsgc.org/>