

Glycogen Storage Disease Type IV/ Adult Polyglucosan Body Disease

What Your Results Mean

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

Glycogen Storage Disease Type V Explained

What is Glycogen Storage Disease Type V?

Glycogen storage disease type V, also known as adult polyglucosan body disease, is an inherited metabolic muscle disorder in which muscle cells cannot break down glycogen to release usable carbohydrates. As a result, the muscles become easily fatigued. During the first few minutes of exercise, affected individuals experience fatigue, muscle pain, and cramps. The symptoms typically stop with rest, and individuals are able to resume exercising (known as the “second wind” phenomenon). Excessive exercise can cause muscles to breakdown and release a chemical called myoglobin into the body. Myoglobin makes the urine red or brown and is toxic to the kidneys, causing life-threatening kidney failure in some patients. These symptoms can appear during childhood but often do not start until adulthood.



Prognosis

Prognosis is generally favorable, and lifespan is not affected.

Treatment

There is no cure for glycogen storage disease type V. Restricting the frequency and intensity of exercise is the mainstay of treatment.



Resources

Association for Glycogen Storage Disease

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

Genetics Home Reference

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

National Society of Genetic Counselors

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