Glycogen Storage Disease Type II (Pompe Disease)

NxGen MDx

What Your Results Mean

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

Glycogen Storage Disease Type II (Pompe Disease) Explained

What is Glycogen Storage Disease Type II (Pompe Disease)?

Glycogen storage disease type II is an inherited disorder characterized by muscle weakness and breathing problems. Individuals with this disorder cannot break down glycogen due to a shortage of the enzyme acid alpha-glucosidase. Glycogen then builds up to toxic levels in the lysosomes, damaging the body's organs and tissues.

There are three types of glycogen storage disease type II disease: classic infantile-onset, non-classic infantile-onset, and late-onset. The classic infantile-onset form is characterized by muscle weakness, poor muscle tone, hepatomegaly, and heart defects. Affected infants begin to have symptoms within the first few months of life. Non-classic infantile-onset is characterized by delayed motor skills and progressive muscle weakness. Affected individuals usually die early in childhood. Late-onset glycogen storage disease type II disease is a much milder form of the disorder. Symptoms typically begin in late childhood or adulthood, characterized by progressive muscle weakness and breathing problems that may lead to respiratory failure.

Prognosis

Prognosis is generally poor. Earlier onset of symptoms is typically associated with faster disease progression. Individuals with the classic infantile-onset form typically die within the first year of life, and the survival of those with the non-classic infantile-onset form is usually only early childhood. Those with the late-onset form of glycogen storage disease type II typically die in their twenties or thirties from respiratory failure.

Treatment

Enzyme replacement therapy is clinically available; however, it is not curative. Management is typically overseen by a group of metabolic specialists. Treatment is otherwise directed at managing an individual's specific symptoms.

Resources

Association for Glycogen Storage Disease
http://www.agsdus.org/type-ii.php
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/pompe-disease
National Society of Genetic Counselors
https://www.nsgc.org/

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