Genetics Uncoded:

NxGen MDx

Facts about Pompe Disease

Pompe Disease Explained

Pompe disease 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 Pompe 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 Pompe disease is much milder form of the disorder. It is characterized by progressive muscle weakness and breathing problems that may lead to respiratory failure. Enzyme replacement therapy is clinically available and management is typically overseen by a group of metabolic specialists.

How the Genetics Work

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

What Your Test Results Mean

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

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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Revised 12/15/16