Genetics Uncoded:
FACTS ABOUT POMPE DISEASE

AKA | Acid maltase deficiency, Alpha-1,4-glucosidase deficiency, AMD, Alpha-glucosidase deficiency, Glycogenesis type II, Glycogen storage disease type II, GSD II

What Your Test Results Mean
Testing results indicate that you are a carrier of Pompe disease. Carriers typically show no symptoms of Pompe disease; however, carriers are at an increased risk of having a child with Pompe disease. Risk for the current or future pregnancies is dependent on your partner’s carrier status. Carrier testing of your partner is recommended in addition to consultation with a genetic counselor.

Pompe Disease Explained
Pompe disease is an inherited disorder characterized by muscle weakness and breathing problems. In this disorder, affected individuals 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, and experience a failure to thrive. 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 and less severe than the infantile-onset forms. It is characterized by progressive muscle weakness and breathing problems that may lead to respiratory failure. Enzyme replacement therapy is clinically available for individuals with this disorder. 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 the mutations in the GAA gene. In general, individuals have two copies of the GAA gene. Carriers of Pompe disease have a single mutation in one copy of the GAA gene, while individuals with Pompe disease have mutations 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%.

Recommended Next Steps
Carrier testing of your partner is recommended in addition to consultation with a genetic counselor for more detailed risk assessment.

Questions?
Contact us at (855) 776-9436 to set up an appointment to discuss your results in more detail with a NxGen MDx genetic counselor.