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My46 Trait Profile

Pompe disease

Other Names: Glycogen storage disease type II, Acid maltase deficiency, acid alpha-glucosidase deficiency, GAA deficiency

Pompe disease is a genetic condition characterized by the body's inability to break down a large sugar called glycogen. It is caused by mutations in the GAA gene that makes the protein called acid alpha-glucosidase.

Characteristics of Pompe disease

Pompe disease is a progressive genetic condition caused by a lack of an enzyme called acid alpha-glucosidase (GAA). GAA is an enzyme necessary to break down glycogen (a large sugar) into simpler sugars called glucose. Without enough GAA enzyme, the glycogen gets stored in many tissues, particularly in the heart and muscles. As this the amount of storage progresses, disease symptoms become debilitating and life threatening. Disease severity and onset of symptoms are highly variable. Early or infantile-onset Pompe disease is the classic form of the condition and symptoms are usually apparent by the first few months of life. This form is characterized by cardiomegaly/cardiomyopathy (enlarged heart), hepatomegaly (enlarged liver), severe hypotonia (low muscle tone), and death due to failure of the heart and lungs within the first year of life. In late or adult-onset Pompe disease, signs or symptoms often do not appear until later in childhood, adolescence or adulthood. The typical characteristics of adult-onset Pompe disease are progressive muscle weakness especially in the legs and trunk, as well as respiratory weakness. There is usually no heart involvement in adult-onset Pompe disease.

Diagnosis/Testing

A diagnosis of Pompe disease can be made by measuring the GAA enzyme activity from a blood sample. A person with Pompe disease will have deficient (low) enzyme activity. The diagnosis can be confirmed by genetic testing for a change or mutation in a gene called GAA. This gene is responsible for making the GAA enzyme. Mutations in the GAA gene prevent the enzyme from working properly, and the result is a build up of glycogen in various organs of the body.

Management/Surveillance

Enzyme replacement therapy (ERT) is a medical treatment available for both infantile and adult onset Pompe disease. ERT provides the GAA enzyme that is missing in individuals with Pompe disease through regular intravenous (IV) infusions. ERT is a lifelong treatment for Pompe disease and is not a cure.

Supportive care for Pompe disease focuses on managing symptoms. Physical therapy can be used to help with movement and maintain muscle strength. Respiratory support (e.g., CPAP or BiPAP) can be used to help with breathing difficulties. Regular care and follow up with cardiology, gastroenterology, pulmonology and other specialist as needed is recommended to manage symptoms.

Mode of inheritance

Pompe disease is inherited in an autosomal recessive pattern. This means that an individual has to inherit two GAA mutations (i.e., one from each parent) to be affected with Pompe disease. If both parents are carriers of a GAA mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with Pompe disease.

Risk to family members

Parents of a child with Pompe disease are carriers of Pompe disease. If a sibling of a child with Pompe disease is unaffected, he/she has a 2 in 3 (or 66%) chance of being a carrier of Pompe disease.

Special considerations

Due to disease related heart problems, individuals with Pompe disease are at an increased for anesthesia complications. It is important to select a cardiac anesthesiologist who has experience with enlarged hearts and that the surgery or procedure be performed at an experienced hospital with an ICU in the event serious complications arise.

Resources

International Pompe Association
http://www.worldpompe.org/
Pompe Community, Genzyme sponsored Patient Information
http://www.pompe.com
Genetics Home Reference: Pompe disease
http://ghr.nlm.nih.gov/condition/pompe-disease

References

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The National Organization for Rare Disorders"Physician's Guide to Pompe Disease (Glyocogen Storage Disease, Type II; Acid Maltase Deficiency"

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