POMPE DISEASE

WHAT IS POMPE DISEASE?

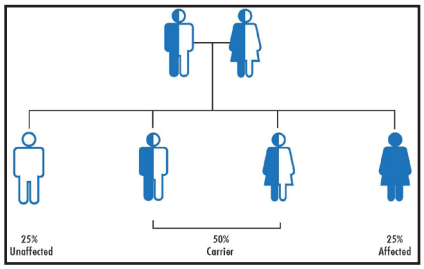
Pompe disease is classified as a lysosomal storage disorder (LSD). Lysosomes are cell organelles that digest and recycle various molecules. Disorders that result in the accumulation of molecules within the lysosomes, including Pompe disease, are identified as LSDs.

THE CAUSE

Pompe disease is caused by mutations in the GAA gene. Normal function of this gene produces an enzyme called acid alpha-glucosidase that is active in the lysosomes. The lack of this enzyme results in the inability of cells to break down glycogen into glucose. Accumulation of glycogen causes primary damage in muscle tissues.

INHERITANCE PATTERN

Pompe disease is an inherited autosomal recessive (AR) condition. In order to be affected, the individual must inherit one mutation in the gene from each parent. Each parent is referred to as a carrier and does not show symptoms of the condition.



When two carrier parents of an AR condition have children, there is a 25% chance of having an affected child with each pregnancy.

Pompe disease affects 1 in 40,000 births.

PATIENT CARE

DIAGNOSIS

1. Urine test to measure oligosaccharides.
2. Enzymatic testing to measure acid alpha-glucosidase activity levels in blood or skin cells.
3. DNA mutation analysis of the GAA gene.

MANAGEMENT

The management team should consist of cardiology, genetics, physical therapy, gastroenterology, neurology, and pulmonology.

TREATMENT

Enzyme replacement therapy is the approved treatment for all patients with Pompe disease. The treatment is called alglucoside alfa (Lumizyme) and is manufactured by Sanofi Genzyme.

Gene therapy for Pompe disease is being developed by Actus Therapeutics with ongoing research and clinical trials.

Clinical trials are underway for a new enzyme replacement therapy for Pompe disease by Amicus Therapeutics.

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WHAT ARE THE SYMPTOMS?

There are three types of Pompe disease, classified according to the age of onset of symptoms and severity of the disease. General symptoms include: myopathy, cardiomegaly, hypotonia, respiratory failure, hepatomegaly, and failure to thrive.

Classic infantile-onset Pompe begins within the first few months of life. Symptoms include myopathy, hypotonia, hepatomegaly, and cardiac defects. Infants have failure to thrive and will succumb to heart failure in the first year of life if untreated.

In Non-classic infantile-onset Pompe, symptoms become apparent around age one and include delayed motor skills and progressive muscle weakness. The heart may be enlarged, but patients do not usually have heart failure. This type can be accompanied by compromised breathing. Patients usually survive into early childhood.

Late-onset Pompe Disease may not show symptoms until adolescence or adulthood. Patients experience progressive muscle weakness in the trunk and limbs. Progressive disease can lead to respiratory failure.

RESOURCES

Gene Reviews: MPS1
ncbi.nlm.nih.gov/books/NBK1261/

Genetics Home Reference
ghr.nlm.nih.gov/condition/pompe-disease

NCBI Testing Registry
ncbi.nlm.nih.gov/gtr/conditions/C0751173/

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