POMPE DISEASE: QUICK FACTS

Understanding Pompe Disease at a Glance

amda
Acid Maltase Deficiency Association

WHAT IS POMPE DISEASE?

Pompe disease is a rare, inherited metabolic and neuromuscular disorder caused by an insufficient amount of the lysosomal enzyme, acid alpha-glucosidase (GAA). It is caused by a defect in the GAA gene.

The enzyme breaks down glycogen inside lysosomes; when GAA activity is deficient, glycogen accumulates in cells — particularly in skeletal, respiratory, and cardiac muscles. This causes progressive muscle damage and weakness.

Pompe disease is also known as acid maltase deficiency and glycogen storage disease type II (GSD II).

Historically, the prevalence of Pompe disease was estimated at approxiately 1 in 40,000 births. However, with the addition of Pompe disease to newborn screening (NBS) programs across the nation, a 2024 study found that the prevalence was approximately 1 in 18,711 births

COMMON SIGNS AND SYMPTOMS

Pompe disease affects each person differently. Symptoms may include

- Progressive muscle weakness
- Difficulty walking, climbing stairs, or rising from a chair
- Frequent falls
- Difficulty breathing, particularly when lying down or sleeping
- Morning headaches or daytime fatigue related to breathing problems
- Difficulty chewing or swallowing
- Feeding difficulties and poor muscle tone in infants
- Enlarged heart or other cardiac involvement, particularly in infants

CLASSIFICATIONS

Though the disease exists on a spectrum, there are two main classifications:

Infantile-onset Pompe disease (IOPD)
Symptoms begin during infancy. The classic infantile form is characterized by severe muscle weakness, low muscle tone (hypotonia), cardiomyopathy/enlarged heart, and rapid progression. Without effective treatment, classic infantile disease can lead to cardiorespiratory failure and death during the first year of life.

Late-onset Pompe disease (LOPD)
Symptoms may begin in childhood, adolescence, or adulthood. Skeletal and respiratory muscle weakness are typically predominant, while significant cardiac involvement is less common than in classic infantile disease. Progression varies considerably from person to person.

HOW IS POMPE DISEASE DIAGNOSED?

Diagnostic tests include

- Measurement of GAA enzyme activity
- Genetic testing of the GAA gene
- Testing to evaluate muscle, respiratory, and cardiac function

Pompe disease is also included in newborn screening programs in the United States, although screening practices vary internationally.

HOW IS POMPE DISEASE INHERITED?

Pompe disease is inherited in an autosomal recessive manner.

When both parents carry a disease-causing variant in the GAA gene, each pregnancy has:

- 25% chance that the child will have Pompe disease
- 50% chance that the child will be a carrier
- 25% chance that the child will neither have Pompe disease nor be a carrier

CAN POMPE DISEASE BE TREATED?

Yes. Pompe disease is treatable.

Disease-specific treatments are available, including enzyme replacement therapy (ERT). The therapy provides the GAA enzyme that people with Pompe disease do not produce in sufficient amounts.

People with Pompe disease may also benefit from specialized care involving respiratory, cardiac, physical therapy, occupational therapy, nutrition, speech/swallowing, and other services.

Early diagnosis and appropriate treatment can make an important difference.

LEARN MORE ABOUT POMPE DISEASE

— SCAN HERE —

YOU ARE NOT ALONE

The Acid Maltase Deficiency Association (AMDA) provides education, support, advocacy, and information for individuals and families affected by Pompe disease.