



## Pompe disease

Pompe disease is an inherited disorder caused by the buildup of a complex sugar called glycogen in the body's cells. The accumulation of glycogen in certain organs and tissues, especially muscles, impairs their ability to function normally.

Researchers have described three types of Pompe disease, which differ in severity and the age at which they appear. These types are known as classic infantile-onset, non-classic infantile-onset, and late-onset.

The classic form of infantile-onset Pompe disease begins within a few months of birth. Infants with this disorder typically experience muscle weakness (myopathy), poor muscle tone (hypotonia), an enlarged liver (hepatomegaly), and heart defects. Affected infants may also fail to gain weight and grow at the expected rate (failure to thrive) and have breathing problems. If untreated, this form of Pompe disease leads to death from heart failure in the first year of life.

The non-classic form of infantile-onset Pompe disease usually appears by age 1. It is characterized by delayed motor skills (such as rolling over and sitting) and progressive muscle weakness. The heart may be abnormally large (cardiomegaly), but affected individuals usually do not experience heart failure. The muscle weakness in this disorder leads to serious breathing problems, and most children with non-classic infantile-onset Pompe disease live only into early childhood.

The late-onset type of Pompe disease may not become apparent until later in childhood, adolescence, or adulthood. Late-onset Pompe disease is usually milder than the infantile-onset forms of this disorder and is less likely to involve the heart. Most individuals with late-onset Pompe disease experience progressive muscle weakness, especially in the legs and the trunk, including the muscles that control breathing. As the disorder progresses, breathing problems can lead to respiratory failure.

### Frequency

Pompe disease affects about 1 in 40,000 people in the United States. The incidence of this disorder varies among different ethnic groups.

### Causes

Mutations in the *GAA* gene cause Pompe disease. The *GAA* gene provides instructions for producing an enzyme called acid alpha-glucosidase (also known as acid maltase). This enzyme is active in lysosomes, which are structures that serve as recycling centers within cells. The enzyme normally breaks down glycogen into a simpler sugar called glucose, which is the main energy source for most cells.

Mutations in the *GAA* gene prevent acid alpha-glucosidase from breaking down glycogen effectively, which allows this sugar to build up to toxic levels in lysosomes. This buildup damages organs and tissues throughout the body, particularly the muscles, leading to the progressive signs and symptoms of Pompe disease.

### **Inheritance Pattern**

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

### **Other Names for This Condition**

- acid maltase deficiency
- acid maltase deficiency disease
- alpha-1,4-glucosidase deficiency
- AMD
- deficiency of alpha-glucosidase
- GAA deficiency
- glycogen storage disease type II
- glycogenosis Type II
- GSD II
- GSD2
- Pompe's disease

### **Diagnosis & Management**

#### Genetic Testing Information

- What is genetic testing?  
[/primer/testing/genetictesting](#)
- Genetic Testing Registry: Glycogen storage disease type II, infantile  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0751173/>
- Genetic Testing Registry: Glycogen storage disease, type II  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0017921/>

#### Research Studies from ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22Glycogen+Storage+Disease+Type+II%22+OR+%22Pompe+disease%22>

### Other Diagnosis and Management Resources

- Baby's First Test  
<https://www.babysfirsttest.org/newborn-screening/conditions/pompe>
- GeneReview: Pompe Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK1261>
- National Organization for Rare Disorders (NORD) Physician Guide  
<https://rarediseases.org/physician-guide/pompe-disease/>

### **Additional Information & Resources**

#### Health Information from MedlinePlus

- Health Topic: Carbohydrate Metabolism Disorders  
<https://medlineplus.gov/carbohydratemetabolismdisorders.html>

#### Genetic and Rare Diseases Information Center

- Glycogen storage disease type 2  
<https://rarediseases.info.nih.gov/diseases/5714/glycogen-storage-disease-type-2>

#### Additional NIH Resources

- National Institute of Neurological Disorders and Stroke: Myopathy Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Myopathy-Information-Page>
- National Institute of Neurological Disorders and Stroke: Pompe Disease Information Page  
<https://www.ninds.nih.gov/Disorders/All-Disorders/Pompe-Disease-Information-Page>

#### Educational Resources

- MalaCards: neurological manifestations of pompe disease  
[https://www.malacards.org/card/neurological\\_manifestations\\_of\\_pompe\\_disease](https://www.malacards.org/card/neurological_manifestations_of_pompe_disease)
- Merck Manual Consumer Version: Overview of Hereditary Metabolic Disorders  
<https://www.merckmanuals.com/home/children-s-health-issues/hereditary-metabolic-disorders/overview-of-hereditary-metabolic-disorders>
- Orphanet: Glycogen storage disease due to acid maltase deficiency  
[https://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=365](https://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=365)

#### Patient Support and Advocacy Resources

- Acid Maltase Deficiency Association  
<http://www.amda-pompe.org/>
- Association for Glycogen Storage Disease  
<https://www.agsdus.org/type-ii.php>

- Association for Glycogen Storage Disease UK  
<http://www.agsd.org.uk/>
- International Pompe Association  
<https://worldpompe.org/>
- Metabolic Support UK  
<https://www.metabolicsupportuk.org/>
- Muscular Dystrophy Association  
<https://www.mda.org/disease/metabolic-myopathies>
- National Organization for Rare Disorders (NORD)  
<https://rarediseases.org/rare-diseases/pompe-disease/>
- Resource list from the University of Kansas Medical Center  
<http://www.kumc.edu/gec/support/glycogen.html>

#### Clinical Information from GeneReviews

- Pompe Disease  
<https://www.ncbi.nlm.nih.gov/books/NBK1261>

#### Scientific Articles on PubMed

- PubMed  
[https://www.ncbi.nlm.nih.gov/pubmed?term=%28Glycogen+Storage+Disease+Type+II%5BMAJR%5D%29+AND+%28%28glycogen+storage+disease+type+II%5BTIAB%5D%29+OR+%28pompe\\*%5BTIAB%5D%29+OR+%28acid+maltase+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D](https://www.ncbi.nlm.nih.gov/pubmed?term=%28Glycogen+Storage+Disease+Type+II%5BMAJR%5D%29+AND+%28%28glycogen+storage+disease+type+II%5BTIAB%5D%29+OR+%28pompe*%5BTIAB%5D%29+OR+%28acid+maltase+deficiency%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D)

#### Catalog of Genes and Diseases from OMIM

- GLYCOGEN STORAGE DISEASE II  
<http://omim.org/entry/232300>

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