A VISUAL GUIDE TO UNDERSTANDING POMPE DISEASE

INFORMATION FOR PEOPLE LIVING WITH POMPE DISEASE—AND THEIR FAMILIES
What is Pompe disease?

For additional information, talk to your healthcare provider.

This guide is meant to help you understand a complicated disease.

Pompe disease is a debilitating genetic disorder that is inherited from both parents in an autosomal recessive fashion.1

It can be classified as a lysosomal storage disorder, neuromuscular disease, acid maltase deficiency, or glycogen storage disorder.2

There are 2 main types of Pompe disease: infantile and late-onset. Classic infantile appears within a few months of birth, nonclassic infantile appears in the first year. Late-onset appears later, in childhood or adulthood.3

Usually, the earlier symptoms appear, the more severe the symptoms can be.4

Sometimes it’s difficult for doctors to diagnose Pompe disease, since several symptoms can be confused with other neuromuscular disorders.5

How does Pompe disease affect families?

How parents pass down Pompe disease

- Pompe disease is inherited through a person’s genes. People with the mutated gene either have the disease or are carriers of the disease. Carriers have 1 copy of the mutated gene and 1 normal copy of the gene. They typically do not show symptoms. Those who have the disease have 2 copies of the mutated gene. If both parents are carriers, there is a 25% chance that each child could have Pompe disease.6

- If 1 parent has Pompe disease and the other parent is unaffected, then none of the children will develop Pompe disease, but all will be carriers.7

- If 1 parent has the disease and the other parent is a carrier, there is a 50% chance that each child could be a carrier and a 50% chance that each child could develop Pompe disease.8

What should I know about Pompe disease?

Depending on where you live, Pompe disease affects between 1 in 33,000 to 1 in 300,000 individuals.9

- People who have Pompe disease have difficulty making a specific lysosomal enzyme.10

- In Pompe disease, the affected enzyme is acid α-glucosidase, also known as GAA.11

- Normally, GAA breaks down a complex carbohydrate called glycogen and converts it into a simple sugar.12

- People with Pompe disease either don’t have any GAA or don’t have enough GAA. This causes the glycogen to build up in body tissues such as skeletal muscles, smooth muscle, and heart muscle.13

- This buildup remains in the body’s cells, especially the muscles, which is what causes the symptoms of Pompe disease.14
What causes Pompe disease?

Everyone has information coded into his or her cells called DNA—which is inherited from his or her parents.

Sometimes, mutations occur in the DNA code that makes up a particular gene. Think of it like spelling. One wrong letter can completely change the meaning of a word.

Certain DNA mutations lead to making very little or no GAA enzyme.

People with certain types of mutations may make some GAA, but not enough. Plus, it doesn’t always function correctly.

What are the symptoms of Pompe disease?

Symptoms can vary based on the level of deficiency and how well the GAA enzyme is functioning.

INFANTILE SYMPTOMS

NERVOUS SYSTEM
• Developmental delay
• Motor delay

LUNGS
• Difficulty breathing
• Respiratory infections

GASTROINTESTINAL
• Difficulty chewing and swallowing
• Poor weight gain
• Chronic constipation
• Loss of bladder or bowel control

SKELETON AND MUSCLE
• Muscle weakness, especially limb and lower body
• Back pain
• Inability to physically exercise
• Difficulty walking
• Difficulty climbing stairs
• Joint tightening
• Winging of shoulder blades
• Reduced spine movement
• Abnormal curvature of the spine
• Motor delay

LATE-ONSET SYMPTOMS

NERVOUS SYSTEM
• Motor delay

LUNGS
• Lung infections
• Respiratory weakness
• Difficulty breathing
• Sleep apnea
• Sleepiness
• Morning headache

GASTROINTESTINAL
• Difficulty chewing and swallowing
• Poor weight gain
• Chronic constipation
• Loss of bladder or bowel control

SKELETON AND MUSCLE
• Muscle weakness, especially limb and lower body
• Back pain
• Inability to physically exercise
• Difficulty walking
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How is Pompe disease treated?

Currently, enzyme replacement therapy (ERT) is the only FDA-approved treatment for Pompe disease.

ERT works by replacing the nonfunctioning or missing GAA with functioning GAA.

An infant's cross-reactive immunological material (CRIM) status can help determine his or her response to treatment. CRIM-positive infants make some GAA, while CRIM-negative infants make no GAA.

There are other investigational therapies, such as gene therapy, being researched for Pompe disease. These investigational products are not FDA-approved for the treatment of Pompe disease, and their safety and efficacy for use in Pompe patients has yet to be established.

To learn more about ongoing research trials, search for Pompe disease on either clinicaltrials.gov or clinicaltrialsregister.eu, or talk to your healthcare provider.

A doctor suspects Pompe disease—here’s an example of what can happen

YOU GET A BLOOD TEST TO MEASURE THE ACTIVITY IN YOUR GAA ENZYME

YOU GET A MUSCLE BIOPSY TO MEASURE GLYCOGEN BUILDUP

YOU GET A DNA TEST TO FIND COMMON MUTATIONS ASSOCIATED WITH POMPE DISEASE

YOU GET A GAA GENE SEQUENCING TEST

GAA ACTIVITY IS ABSENT OR REDUCED

GAA ACTIVITY IS PRESENT

THERE IS GLYCOGEN BUILDUP

THERE IS NO GLYCOGEN BUILDUP

IT DETECTS A COMMON MUTATION

IT DOES NOT DETECT A COMMON MUTATION, BUT THE DOCTOR STILL SUSPECTS POMPE DISEASE

IT DETECTS A MUTATION THAT CAUSES POMPE DISEASE

IT DOES NOT DETECT A MUTATION THAT CAUSES POMPE DISEASE

YOU DO NOT HAVE POMPE DISEASE

YOU HAVE POMPE DISEASE

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What do these words mean?

**Acid α-glucosidase (GAA)**
An enzyme that breaks down complex carbohydrates into simple sugars.

**Cell**
Basic building block of all living things.

**Deoxyribonucleic acid (DNA)**
Basic unit that allows for the transmission of genetic information from one generation to the next and contains instructions, or code, for making proteins and enzymes.

**Enzyme**
A special type of protein that speeds up a reaction that takes place within a cell.

**Lysosome**
A specialized fluid-filled sac, found in the cells, that contains enzymes.

**Lysosomal storage disorder**
A group of over 50 diseases resulting from the accumulation of the deficient enzyme’s substrate in lysosomes.

**Mutation**
A permanent error in the DNA code.

**Neuromuscular disorder**
A group of disorders that affect the nerves that control muscles.

References