A VISUAL GUIDE TO UNDERSTANDING POMPE DISEASE

WHAT PEOPLE LIVING WITH POMPE DISEASE—AND THEIR FAMILIES—NEED TO KNOW

Pompe disease is a debilitating genetic disorder that is inherited from both parents in an autosomal recessive fashion. Depending on where you live, Pompe disease affects between 1 in 33,000 to 1 in 300,000 individuals. It can affect people of all ethnicities.

People who have Pompe disease have difficulty making a specific lysosomal enzyme. The purpose of a lysosomal enzyme is to help break down waste materials in a cell. In Pompe disease, the affected enzyme is acid α-glucosidase, also known as GAA. Normally, GAA breaks down a complex carbohydrate called glycogen and converts it into a simple sugar.

People with Pompe disease either don't have any GAA or not enough GAA. This causes the glycogen to build up in body tissues such as skeletal muscles, liver, and heart. This buildup remains in the body's cells, especially the muscles, which is what causes the symptoms of Pompe disease. It can be classified as a lysosomal storage disorder, neuromuscular disease, acid maltase deficiency, or glycogen storage disorder.

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

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

This guide is meant to help you understand a complicated disease. How parents pass down Pompe disease?

Acid α-glucosidase (GAA) is an enzyme that breaks down complex carbohydrates into simple sugars. Cell is the basic building block of all living things. DNA is the 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 is a special type of protein that speeds up a reaction that takes place within a cell. Lysosome is a specialized fluid-filled sac found in the cells that contains enzymes. Lysosomal storage disorder is a group of over 50 diseases resulting from the accumulation of waste products in lysosomes. Mutation is a permanent error in the DNA code. Neuromuscular disorder is a group of disorders that affect the nerves that control voluntary muscles.

What do these words mean? Want to learn more?

International Pompe Association
worldpompe.org
United Pompe Foundation
unitedpompe.com
Acid Maltase Deficiency Association
amda-pompe.org
Muscular Dystrophy Association
mda.org
The Association for Glycogen Storage Disease UK
www.agsd.org.uk
Australian Pompe’s Association
australianpompe.com
Canadian Association of Pompe
www.pompecanada.com
Selbsthilfegruppe Glykogenose Deutschland e.V.
glykogenose.de
Vereniging Spierziekten Nederland (VSN)
www.vsn.nl
The National Organization for Rare Disorders
rarediseases.org
EURORDIS
eurordis.org

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How does Pompe disease affect families?

Pompe disease is inherited through a person’s genes. People affected by Pompe disease 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. 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. 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.

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. It can affect people of all ethnicities. People who have Pompe disease have difficulty making a specific lysosomal enzyme. The purpose of a lysosomal enzyme is to help break down waste materials in a cell.

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

People with Pompe disease either don’t have any GAA or not enough GAA. This causes the glycogen to build up in body tissues such as skeletal muscles, liver, and heart. Normally, GAA breaks down a complex carbohydrate called glycogen and converts it into a simple sugar.

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

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The Association for Glycogen Storage Disease UK
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Australian Pompe’s Association
australianpompe.com

Canadian Association of Pompe
www.pompecanada.com

Selbsthilfegruppe Glykogenose Deutschland e.V.
glykogenose.de

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The National Organization for Rare Disorders
rarediseases.org

EURORDIS
eurordis.org

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What causes Pompe disease?

Everyone has information coded into their cells called DNA—which is inherited from one’s parents.

Sometimes, mutations occur in the DNA code that makes up a particular gene.

Certain DNA mutations make very little or no GAA.

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

What are the symptoms of Pompe disease?

Symptoms can vary based on how well the GAA enzyme is functioning.

INFANTILE SYMPTOMS

NERVOUS SYSTEM

• Developmental delay
• Motor delay

LUNGS

• Difficulty breathing
• Respiratory infections

SKELETON AND MUSCLE

• Rapid muscle weakness
• Diminished muscle tone
• Floppy muscles
• Large protruding tongue
• Head lag

GASTROINTESTINAL

• Feeding difficulties
• Poor appetite
• Difficulty swallowing
• Failure to thrive
• Enlarged liver

LUNGS

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

GASTROINTESTINAL

• Difficulty chewing and swallowing
• Poor weight gain
• Chronic constipation

LUNGS

• Difficulty breathing
• Respiratory infections

SKELETON AND MUSCLE

• Muscle weakness, especially limbs and lower body
• Back pain
• Inability to physically exercise
• Difficulty walking
• Difficulty climbing stairs
• Gait abnormalities
• Joint tightening
• Winging of shoulder blades
• Reduced spine movement
• Abnormal curvature of the spine
• Motor delay
• Loss of bladder or bowel control

LATE-ONSET SYMPTOMS

NERVOUS SYSTEM

• Developmental delay
• Motor delay

LUNGS

• Difficulty breathing
• Respiratory infections

SKELETON AND MUSCLE

• Rapid muscle weakness
• Diminished muscle tone
• Floppy muscles
• Large protruding tongue
• Head lag

GASTROINTESTINAL

• Feeding difficulties
• Poor appetite
• Difficulty swallowing
• Failure to thrive
• Enlarged liver

How is Pompe disease treated?

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

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

It has been shown to slow the progression of disease and may improve a person’s outcome.

However, there are unmet needs with current ERT, such as people building up a serious resistance to treatment.

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. CRIM-negative infants are more likely to develop an immunity to ERT and may not respond as well as CRIM-positive infants.

Different types of ERT and other treatments are being researched and tested to provide an alternative for those living with Pompe disease.

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

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A doctor suspects Pompe disease—what happens next?

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

There is glycogen buildup

GAA activity is present

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

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.
- **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 waste products in lysosomes.
- **Mutation**: A permanent error in the DNA code.
- **Neuromuscular disorder**: A group of disorders that affect the nerves that control voluntary muscles.

Want to learn more?

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