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

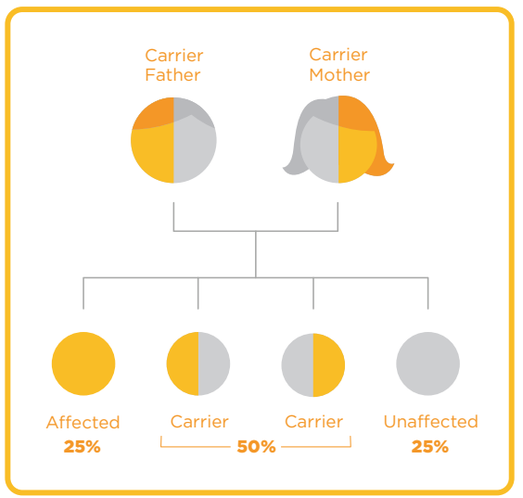
This guide is meant to help you understand a complicated disease

- Pompe disease is a debilitating ***genetic disorder*** that is inherited from both parents in an autosomal recessive fashion[1,2]
- It can be classified as a ***lysosomal storage disorder***, ***neuromuscular disease***, ***acid maltase deficiency***, or ***glycogen storage disorder***[1,3]
- 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[2,3]
- 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[3]

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[5]
- 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[6]
- 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[7]



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[1]

It can affect people of all ethnicities[8]

People who have Pompe disease have difficulty making a specific lysosomal enzyme[9]

The purpose of a lysosomal enzyme is to help break down specific nutrient substrates in a cell[8,10]

In Pompe disease, the affected enzyme is acid α-glucosidase, also known as *GAA*[1]

Normally, *GAA* breaks down a complex carbohydrate called glycogen and converts it into a simple sugar[8]

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[3,11,12]

This buildup remains in the body's cells, especially the muscles, which is what causes the symptoms of Pompe disease[1]

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[13]

CAR

CAT

Think of it like spelling. One wrong letter can completely change the meaning of a word

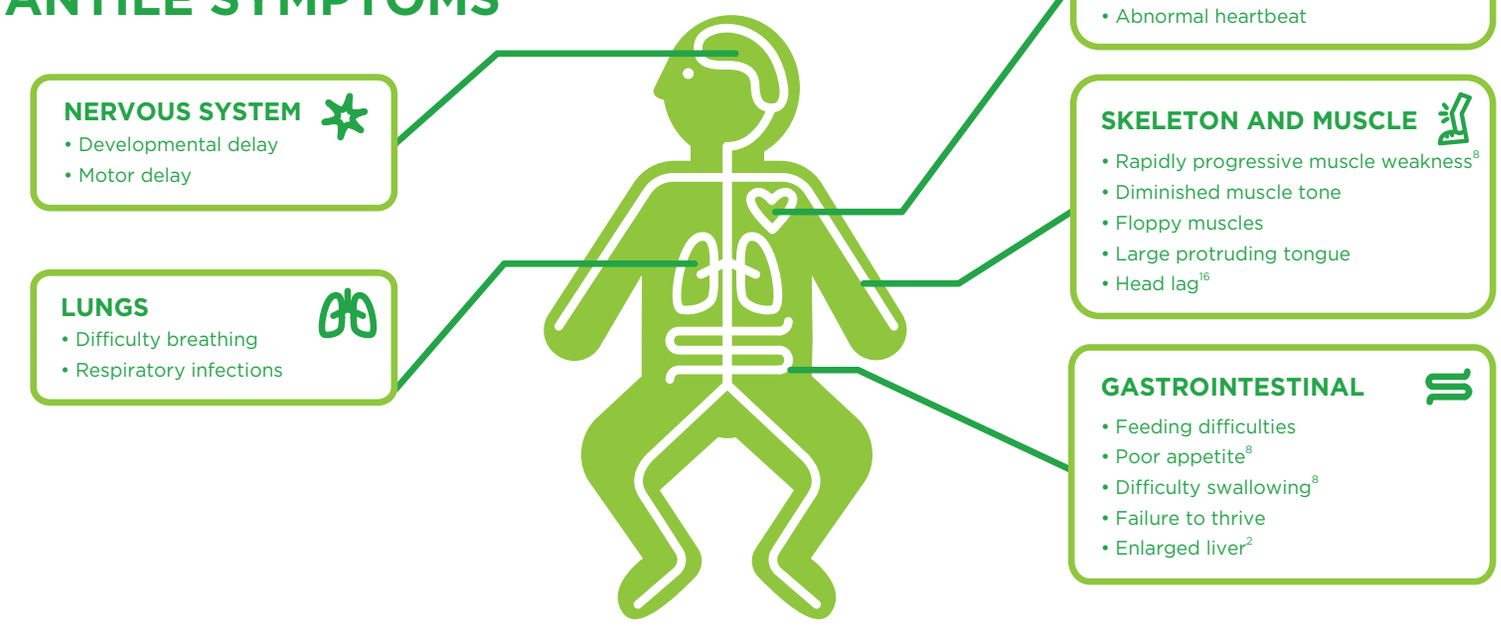
Certain DNA mutations lead to making very little or no *GAA*[11]

People with certain types of mutations may make some *GAA*, but not enough. Plus, it doesn't always function correctly[4,11,14]

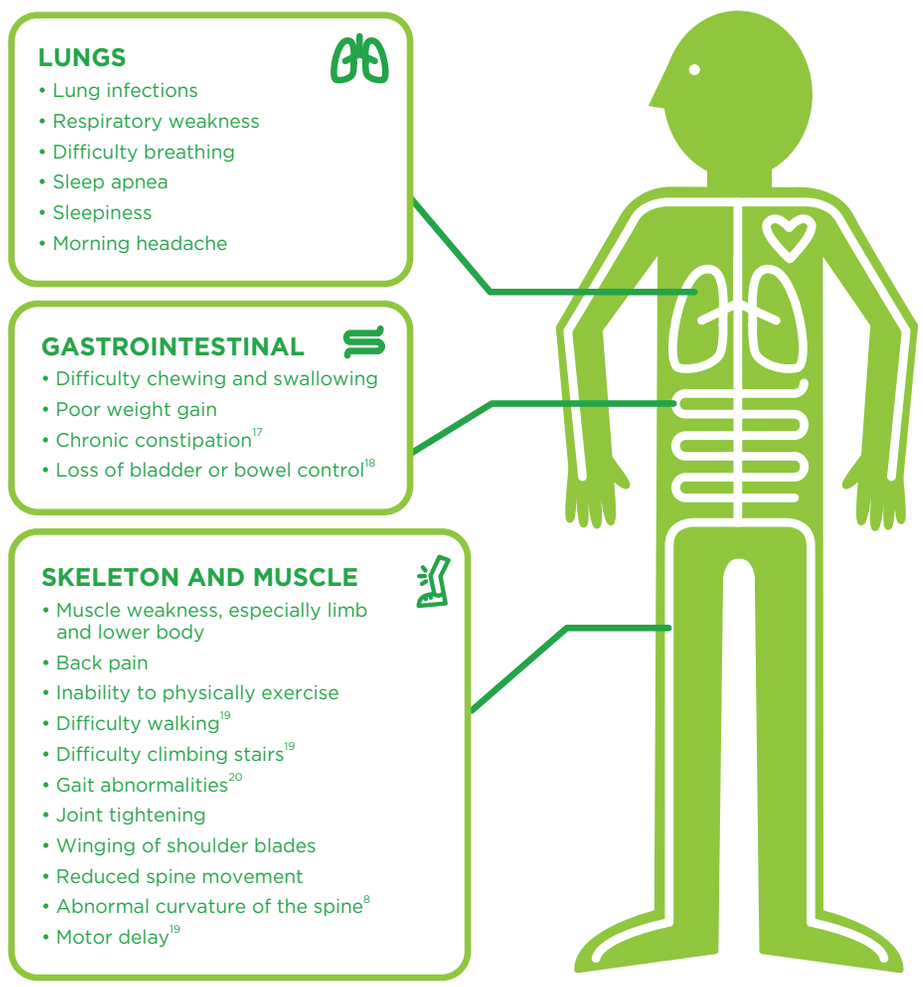
What are the symptoms of Pompe disease?

Symptoms can vary based on the level of deficiency and how well the *GAA* enzyme is functioning[4]

INFANTILE SYMPTOMS[3]



LATE-ONSET SYMPTOMS[3]



How is Pompe disease treated?

ERT

Currently, enzyme replacement therapy (ERT) is the only FDA-approved treatment for Pompe disease[21]

ERT works by replacing the nonfunctioning or missing *GAA* with functioning *GAA*[21]

CRIM+

CRIM–

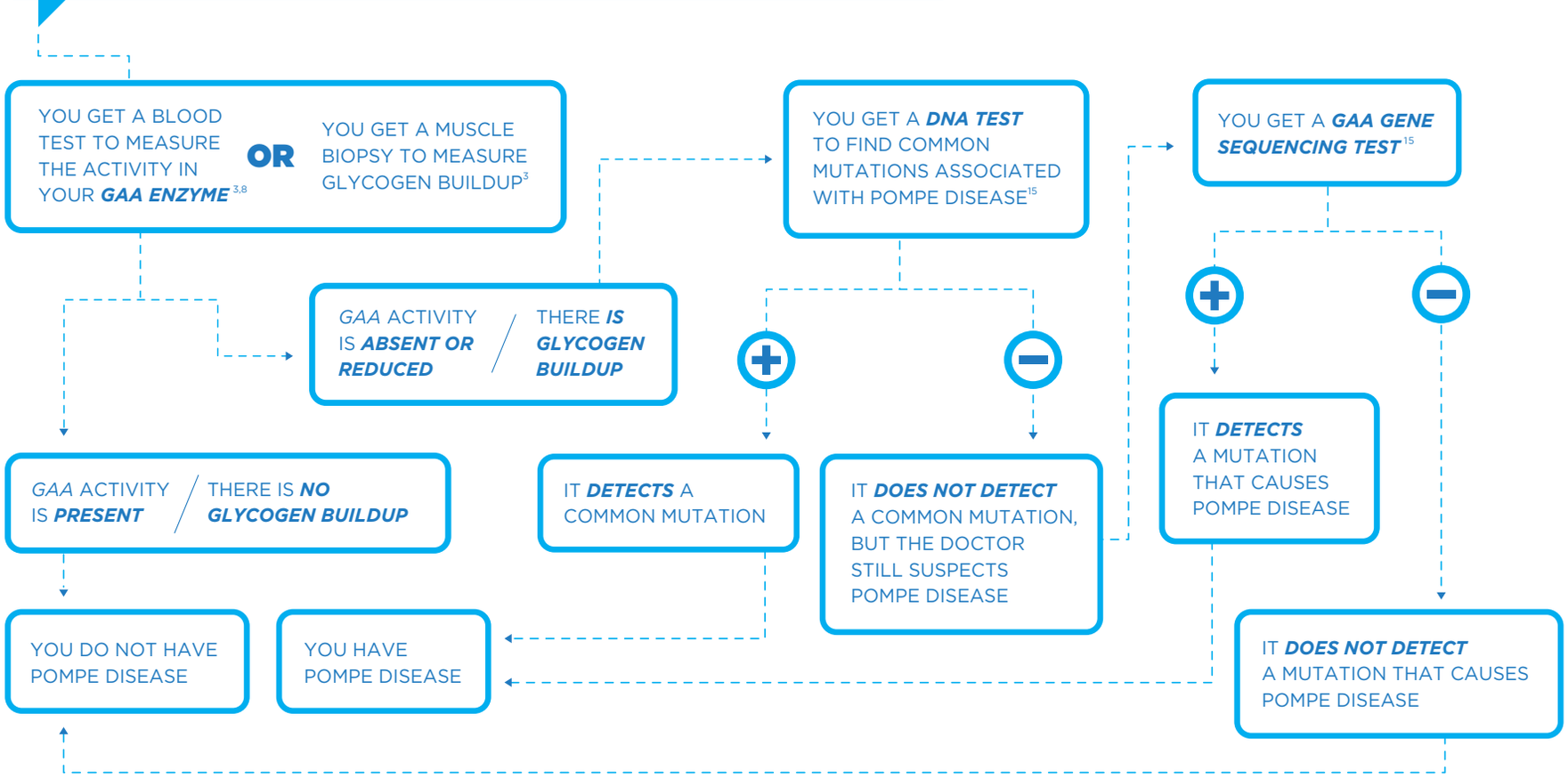
An infant's cross-reactive immunological material (CRIM) status can help determine his or her response to treatment. CRIM-positive infants make some *GAA*, while CRIM-negative infants make no *GAA*[22]

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[8]

pompe disease

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



What do these words mean?

Acid α-glucosidase (GAA)
An enzyme that breaks down complex carbohydrates into simple sugars[8]

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[23]

Mutation
A permanent error in the DNA code

Neuromuscular disorder
A group of disorders that affect the nerves that control muscles[24]

Other resources that may be helpful are below.

Also, talk to your healthcare provider.

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

Spierziekten Nederland
https://www.spierziekten.nl/

The National Organization for Rare Disorders
rarediseases.org

EURORDIS
eurordis.org

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