What is Pompe disease?

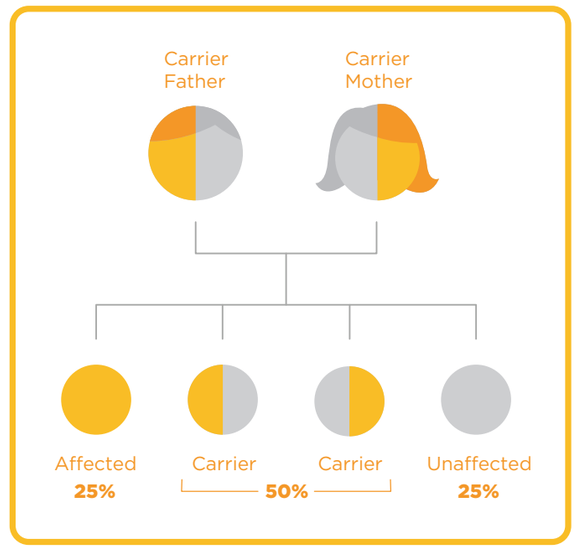
This guide is meant to help you understand a complicated disease

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

How does Pompe disease affect families?

How parents pass down Pompe disease

- 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*
- 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

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?

These additional resources will help you to understand Pompe disease

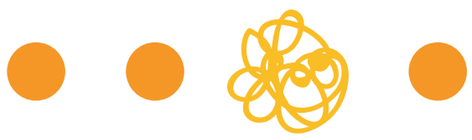
- **International Pompe Association**
  worldpompe.org
- **United Pompe Foundation**
  unitedpompe.com
- **Acid Maltase Deficiency Association**
  amda-pompe.org
- **Muscular Dystrophy Association**
  mda.org
- **The Association for Glycogen Storage Disease UK**
  www.agsd.org.uk
- **Australian Pompe's 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

A VISUAL GUIDE TO UNDERSTANDING POMPE DISEASE

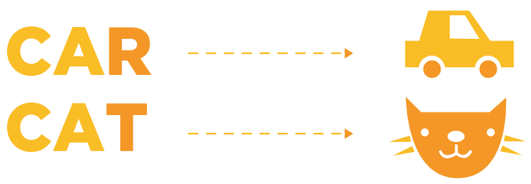
WHAT PEOPLE LIVING WITH POMPE DISEASE— AND THEIR FAMILIES— NEED TO KNOW

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



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

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

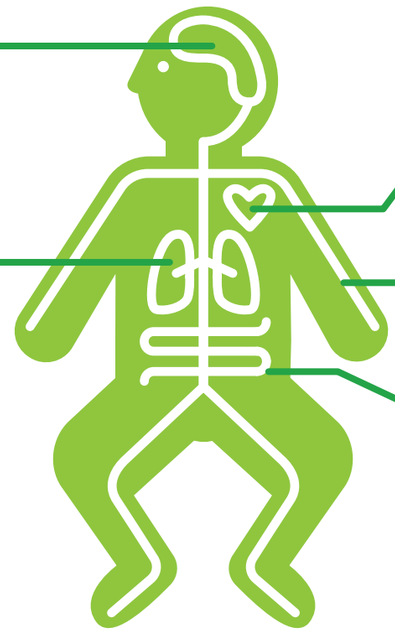
What are the symptoms of Pompe disease?

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



HEART
- Enlarged heart
- Abnormal heartbeat

SKELETON AND MUSCLE
- Rapid muscle weakness
- Diminished muscle tone
- Floppy muscles
- Large protruding tongue
- Head lag

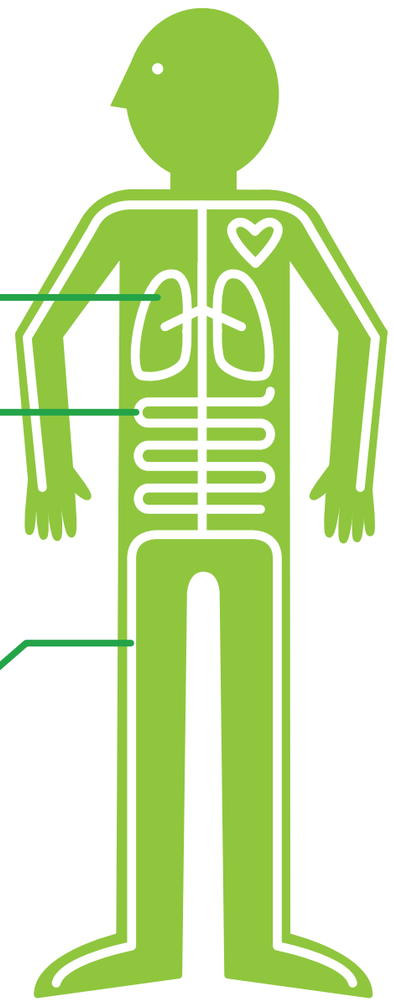
GASTROINTESTINAL
- Feeding difficulties
- Poor appetite
- Difficulty swallowing
- Failure to thrive
- Enlarged liver

LATE-ONSET SYMPTOMS

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

GASTROINTESTINAL
- Difficulty chewing and swallowing
- Poor weight gain
- Chronic constipation

SKELETON AND MUSCLE
- Muscle weakness, especially limb 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



How is Pompe disease treated?

ERT

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

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

A doctor suspects Pompe disease—what happens next?

YOU GET A BLOOD TEST TO MEASURE THE ACTIVITY IN YOUR ***GAA ENZYME*** **OR** YOU GET A MUSCLE BIOPSY TO MEASURE GLYCOGEN BUILDUP

GAA ACTIVITY IS ***ABSENT OR REDUCED*** / THERE ***IS GLYCOGEN BUILDUP***

GAA ACTIVITY IS ***PRESENT*** / THERE IS ***NO GLYCOGEN BUILDUP***

YOU DO NOT HAVE POMPE DISEASE

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

(+) IT ***DETECTS*** A COMMON MUTATION

YOU HAVE POMPE DISEASE

(–) IT ***DOES NOT DETECT*** A COMMON MUTATION, BUT THE DOCTOR STILL SUSPECTS POMPE DISEASE

YOU GET A ***GAA GENE SEQUENCING TEST***

(+) IT ***DETECTS*** A MUTATION THAT CAUSES POMPE DISEASE

(–) IT ***DOES NOT DETECT*** A MUTATION THAT CAUSES POMPE DISEASE