



# Parent Information Sheet

## What is Pompe Disease?

Pompe disease (Pompe) is a **treatable** genetic condition affecting the body's ability to break down complex sugars from food. Pompe is classified as a lysosomal storage disorder. Lysosomal storage disorders are a group of rare inherited metabolic disorders that result from defects to the lysosome (an important part of each cell in the human body). Pompe can cause health issues such as breathing problems, heart problems, and a shortened lifespan if left untreated.

Pompe can be identified through newborn screening. Confirmatory testing is done using a blood sample from the baby, often directed by a metabolic specialist. A primary care physician or metabolic specialist may order additional tests, coordinate further care, and refer to genetic counseling. A metabolic specialist is also able to coordinate prompt treatment.

About one in  
**40,000**  
births in the US are  
affected by Pompe.

### Pompe is treatable.

Early diagnosis through newborn screening enables treatment to start early to stop disease progression and improve long term health outcomes.

## Symptoms

There are three types of Pompe (classic infantile-onset, non-classic infantile-onset, and late onset) and the severity and the age at which symptoms begin differs among them. Patients who develop symptoms early in life tend to be affected more severely and have a faster rate of disease progression compared to those who develop symptoms later in life.

Common symptoms of Pompe include:

- Delayed milestones
- Enlarged heart
- Feeding difficulties
- Muscle weakness
- Difficulty breathing
- Difficulty gaining weight

## Treatment

Although Pompe cannot be cured, it can be treated. Early intervention is important because treatments are most effective in improving children's quality of life when started within the first few weeks of life before symptoms start.

Treatment is typically done through a multi-disciplinary approach that can include Enzyme Replacement Therapy (ERT), physical therapy, occupational therapy, and sometimes dietary modifications. These treatment options can help prevent or reduce the symptoms of the disorder, leading to a better quality of life and longer life span.

**If left untreated, Pompe can cause severe breathing and heart complications, as well as death.**

### Act Quickly

Symptoms can worsen rapidly without treatment.

## Supports & Resources

### Additional Pompe Resources:

- <https://www.rarediseases.org/rare-diseases/pompe-disease/#affected>[www.curesma.org/](http://www.curesma.org/)
- <https://pompediseasenews.com/>
- <https://www.unitedpompe.com/>

### Additional Newborn Screening Information:

- <https://www.babysfirsttest.org/>
- <http://www.savebabies.org/>

### Kansas Department of Health and Environment Resources:

- (785) 296-1313: Special Healthcare Needs (SHCN) for care coordination\* and financial assistance\*\*

\*Care Coordination is available to minors diagnosed with a condition screened for by KS NBS

\*\*Financial assistance is based on meeting financial eligibility requirements

- (785) 291-3363: Kansas Newborn Screening for additional question

