TEXAS Health and Human Services
Texas Department of State Health Services
Jennifer A. Shuford, M.D., M.P.H.
Commissioner

Newborn Screening FACT Sheet

Pompe Disease

What is Pompe Disease?
Pompe is a metabolic muscle disorder that interferes with processing and storage of complex sugars called glycogen. The build-up of glycogen in muscle cells causes the muscle to break down.
Three types of Pompe disease have been described (see below), which differ in severity and age of onset with effects on muscle, heart, and lung function. Pompe disease occurs in ~ 1 in 40,000 births.

What Causes Pompe Disease?
Pompe Disease is a lysosomal disease caused by changes in the acid alpha-glucosidase (*GAA*) gene. It is an autosomal recessive inheritance, meaning both parents are genetic carriers of the disease but don't have the disease itself.
Reduced GAA enzyme activity causes glycogen to build up in the tissues of major organs including muscle.

Types of Pompe Disease and their symptoms:
The classic form of infantile-onset Pompe Disease (IOPD) presents within a few months of life with signs and symptoms such as poor tone, heart defects, poor weight gain, and respiratory problems. If untreated, this form of Pompe Disease often leads to death from heart failure in the first year of life.

The non-classic form of IOPD appears by age one. It is characterized by delayed motor skills (such as rolling over and sitting) and progressive muscle weakness. The heart may be abnormally large (cardiomegaly) but affected individuals usually do not experience heart failure.

The muscle weakness in this disorder leads to serious breathing problems and most children with non-classic infantile-onset Pompe Disease live only into early childhood.

Late onset Pompe Disease (LOPD) describes onset after twelve months of age, though symptoms may not present until later in childhood, adolescence, or adulthood.
Typically, symptoms include progressive muscle weakness and respiratory insufficiency. Cardiac disease is less commonly reported.

What to Expect with Pompe Disease care:
Enzyme Replacement Therapy (ERT) is available for all forms and should be started under guidance of a specialist.

Alglucosidase alfa has been used for ERT for more than 15 years. It improves breathing without a ventilator as well as heart and skeletal muscle function in patients.

Avalglucosidase alfa, which has similar efficacy to alglucosidase alfa, has been approved as a second option for ERT.

ERT should be initiated as soon as possible for patients with the infantile form after evaluating cross-reactive immunogenic material (CRIM) status and determining if immune therapy is required. For late onset forms, therapy should be initiated at the first signs of muscle weakness.

Regardless of the type of Pompe Disease, most children will need additional services including physical therapy, respiratory therapy, and dietary management.

Things to Remember

There is no cure for Pompe Disease, but medication and therapy can help manage some symptoms and potentially slow the course of the disease.

Resources:

Medline Plus
https://medlineplus.gov/genetics/condition/pompe-disease/

Muscular Dystrophy Association
https://www.mda.org/sites/default/files/2021/09/MDA-Pompe-disease-Fact-Sheet-2021.pdf

Health Resources & Services Administration
https://newbornscreening.hrsa.gov/conditions/pompe-disease

Navigate Life Texas
https://www.navigatelifetexas.org/en/diagnosis-healthcare/children-with-multiple-disabilities-rare-conditions-undiagnosed

TEXAS Health and Human Services | Texas Department of State Health Services

Where do I go for more information?
Use your phone's camera to scan the QR code.

Next steps after a positive Newborn Screening for Pompe Disease

Your baby had a positive Newborn screen result, which is a blood test performed on every baby that identifies those at risk for rare disorders. Your baby's results show absent or reduced acid alpha- glucosidase (GAA) enzyme activity. When there is not enough GAA enzyme activity, glycogen builds up in the body. This causes certain organs and muscle to not work properly. This means there is a chance that your baby has a condition called Pompe Disease.

More testing is in progress, but additional confirmatory testing and an exam are needed as soon as possible.

What Happens

- It is important to follow instructions provided to you to have the laboratory tests drawn. These results will help confirm or rule out the diagnosis.
- Your baby's doctor will help arrange a visit to a specialist clinic that is familiar with Pompe Disease. The specialist will check your baby's health and discuss the test results in more detail at the visit. The team may also include a genetic counselor because this is a genetic disease. It is important to keep this appointment. A quick diagnosis and early treatment may lessen the impact of the disease.

What is Pompe Disease?

Pompe Disease is a rare inherited disorder caused by variants in a gene called acid alpha- glucosidase (GAA). This gene controls how much GAA enzyme the body can produce. People with Pompe are missing this enzyme or have less enzyme activity. When there is not enough GAA enzyme activity a type of carbohydrate called glycogen builds up in the body. This causes certain organs and muscle to not work properly. There are different types of Pompe Disease that range in severity and age when problems begin. The specialists you meet with will help figure out if your child has Pompe Disease, or if your child is not affected.

What are possible complications of Pompe Disease?

Symptoms and age of onset vary from person to person depending on the type of Pompe Disease. In infants, problems can include low muscle tone, an enlarged or weakened heart, poor feeding, and growth, and breathing problems. Late-onset symptoms include muscle weakness and breathing problems.

What is the treatment?

Babies with Pompe Disease are treated with Enzyme Replacement Therapy (ERT) and other supportive care which can include occupational and physical therapy to help muscle strength. If there are heart problems, a cardiologist can help manage those as well.

Where can I get more information?

In addition to speaking with your baby's doctors, here are some starting resources for information about Pompe Disease:

- Genetics Home Reference: https://ghr.nlm.nih.gov/condition/pompe-disease
- Baby's First Test-Newborn Screening: http://www.babysfirsttest.org/newborn-screening/conditions/pompe
- Association for Glycogen Storage Disease (AGSD) https://www.agsdus.org/