

# Parent Information

## Inconclusive Newborn Screening Test Result for POMPE DISEASE

(Pom-PAY)

### What is Newborn Screening?

After babies are born, five drops of blood are collected from the baby's heel to test for certain diseases. These conditions usually cannot be found simply by looking at or examining a baby.

### What is Pompe Disease?

Pompe disease is an inherited condition that affects the heart and muscles. It is a lysosomal storage disorder. People with Pompe disease have problems breaking down a sugar called glycogen. When, glycogen is not broken down, it builds up in the body and causes health problems. There are several types of Pompe disease: classic infantile-onset, non-classic infantile-onset, and late-onset. Pompe disease is different for each child and differs depending on the form of the disease. Newborn screening cannot tell the difference between these types. However, doctors will determine which type, if any, your child has.

### What does an inconclusive result mean?

An inconclusive test result means that your baby needs to follow up with a specialist to check for Pompe disease. Your baby's doctor will help arrange for a visit with doctors who have expertise in Pompe disease.

### What do I need to do now?

Your baby's doctor will tell you what is needed to follow up on your baby's screening results. If your baby does have Pompe disease, finding out quickly will allow treatment to be started promptly. It is also important to follow all of your baby's doctor's instructions for testing and for treatment.

### What problems can Pompe cause?

Babies with the classic infantile-onset of Pompe may begin to have heart problems, poor muscle tone, muscle weakness, feeding problems, and develop serious breathing problems if untreated. These health problems may begin right away or within a few months after birth.

Babies with the non-classic infantile-onset form of Pompe disease have delayed motor skills and progressive muscle weakness by one year of age. They have serious breathing problems and they may also have some heart problems.

Late-onset Pompe disease can develop later in childhood, adolescent years, or even in adulthood. Patients may have serious muscle weakness, and significant breathing problems.

### How is Pompe treated?

Although Pompe disease cannot be cured, some of the symptoms can be treated. Possible treatments include: physical therapy, respiratory therapy and other supportive therapies, as well as enzyme replacement therapy (ERT). Treatment is life-long.

Children with Pompe disease benefit from specialized treatment. These children should see their primary care doctor and doctors with expertise in Pompe disease.

### Resources for parents

[www.babysfirsttest.org](http://www.babysfirsttest.org)

[www.dhhs.ne.gov/nsp/](http://www.dhhs.ne.gov/nsp/)

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