NORTH CAROLINA NEWBORN SCREENING PROGRAM

Pompe Disease Fact Sheet for Parents (late-onset Pompe disease)

All babies born in North Carolina are screened at birth for certain medical conditions that can be treated to prevent serious illness. This is called newborn screening. Your baby's newborn screen was positive for **late-onset Pompe disease (LOPD)**. This is also called glycogen storage disease type II.

The signs of Pompe disease may not be obvious at birth. In some cases, Pompe disease can become serious very quickly if it is not treated. Your baby needs more testing as soon as possible to confirm the diagnosis.

What happens next?

At your first genetics appointment, the doctor will take a sample of your baby's blood and urine to confirm if your baby has Pompe disease. Someone will call you to share the test results. The person who calls will tell you what to do next.

For now, take your baby home and provide care as usual. Contact your baby's regular doctor (pediatrician) if you have any concerns about your baby's health.

What is Pompe disease?

Pompe disease is a rare but treatable condition that mainly affects the baby's muscles. Individuals with Pompe disease cannot break down a certain type of sugar, called glycogen, because they do not make enough of an enzyme called acid alpha-glucosidase (GAA). As a result, glycogen builds up in the body and causes health problems.

What are the symptoms of Pompe disease?

There are two types of Pompe disease:

- Late-onset Pompe disease (LOPD): Children with this type have some GAA enzyme. Symptoms may begin during the first year of life or later on in childhood or adolescence.
- Infantile-onset Pompe disease: Babies with this type have low levels or no GAA. Symptoms typically begin shortly after birth and are generally more severe than LOPD.

Symptoms of LOPD can include muscle weakness and delayed motor skills. Babies with LOPD may have trouble sitting up and walking and have milder symptoms than babies with the other type of Pompe disease. Babies with LOPD will not have an enlarged heart or weak muscles at birth like babies with infantile-onset Pompe disease.

How is LOPD treated?

LOPD can be treated with enzyme replacement therapy (ERT). This medicine is given once per week or once every other week through an IV.

Treatment for LOPD is not usually started right away. This is different than treatment for infantile-onset Pompe disease. Your baby will be seen by a genetics team who is familiar with Pompe disease. Your baby's genetics team will decide when it is time to start treatment. This can be different for each child with LOPD.

Your baby may also need other types of care to keep their muscles healthy, such as physical therapy. Your baby may also need to see some other specialty doctors. Your baby's genetics team will talk to you about this and make any referrals to these specialists that your baby needs.

Where do I go for more information?

Use your phone's camera to scan the QR code below.

Duke Pediatric Genetics

Acid Maltase Deficiency Association

Baby's First Test

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health
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http://slph.ncpublichealth.com
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