NORTH CAROLINA NEWBORN SCREENING PROGRAM

Pompe Disease Fact Sheet for Parents (infantile-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 **infantile-onset Pompe disease (IOPD).** 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 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:

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

Babies with IOPD can have an enlarged heart, weak muscles, and feeding problems. Symptoms can become life-threatening if treatment is not started as soon as possible.

What happens next?

At your first genetics appointment, the doctor will take a sample of your baby's blood and urine to do more tests 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.

How is IOPD treated?

IOPD can be treated with enzyme replacement therapy (ERT). This medicine is given once per week or once every other week through an IV. If ERT is needed, you will need to return to your genetics provider as soon as possible.

Your baby may also need other types of treatment to stay healthy. This may include physical therapy or feeding therapy. Your baby may also need to see some specialty doctors. The baby's genetics team will make any referrals to these doctors that your baby needs.

Where do I go for more information?

Use your phone's camera to scan the QR codes.

Duke Pediatric Genetics

Acid Maltase Deficiency Association

Baby's First Test

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

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http://slph.ncpublichealth.com
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