Pompe Disease

What is newborn screening?

Newborn screening is a blood test to check for conditions that can be hidden at birth. Soon after birth, a nurse takes a few drops of blood from your baby's heel. With newborn screening many conditions can be found and treated early.

Your baby's newborn screen flagged for a condition called Pompe disease. Please remember that just because the newborn screen flagged for Pompe disease, it does not mean that your child has a diagnosis of Pompe disease.

KEY POINTS:

- **You have just heard that your baby's newborn screen was out of range. Please understand that the newborn screening is just that: a screening test. Further testing is needed before a diagnosis can be made.**
- **When Pompe is detected early and treatment is started, many babies with the condition are able to live longer lives with improved growth and development.**

What does my baby's newborn screen show?

The newborn screen found low levels of an enzyme called "acid alpha-glucosidase" (called GAA for short). When GAA is low, it means that your baby could have Pompe disease. Not all babies with an out-of-range screening result will go on to get a diagnosis of Pompe disease. However, since some babies will be diagnosed with Pompe disease, it is important to have the follow-up testing done as soon as possible, so that treatment can be started if needed. Your baby should follow up with a neurologist, a doctor who specializes in Pompe disease. The neurologist can discuss your baby's results in more detail.

What is Pompe disease?

Pompe disease happens when children are missing all or some GAA. The GAA enzyme is needed to break down glycogen (a large sugar) into glucose (the form of sugar that the body uses for energy). When glycogen is not broken down properly, it builds up in the body and can cause health problems.

There are different forms of Pompe disease, such as infantile-onset and late-onset. The type of Pompe disease a person is diagnosed with depends on several factors such as the age when problems begin and what organs

are affected. If Pompe is diagnosed, medical specialists can help figure out which type a person has. The newborn screen cannot determine the type of Pompe disease that a baby would have.

What health problems can it cause?

Pompe disease is different for each child. Pompe is a lifelong condition that can result in serious health problems. If untreated, it can cause:

- Muscle weakness
- Enlarged liver
- Heart problems
- Breathing problems

What treatment options are available?

Although Pompe disease cannot be cured, some of the symptoms can be treated. Possible treatments include:

- Enzyme replacement therapy (ERT).
- Supportive therapies like physical therapy (to help with muscle weakness) and respiratory therapy (to help with lung and heart health).

Children with Pompe disease will need to be followed by a team of healthcare workers. They should see their regular doctor (pediatrician) and healthcare providers who specialize in Pompe disease. If a diagnosis is confirmed, your healthcare team will discuss the symptoms and how to monitor and treat your child in more detail.

What if I still have questions?

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Screening Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org as a resource.

SCAN HERE for Information about *NEWBORN SCREENING* including TIPS for preparing for blood draws and collecting urine samples on newborns, and more

This fact sheet contains general information and is for information purposes only. Every child is different and some of this information may not apply to your child specifically. This sheet does not replace medical advice, diagnosis or treatment from your child's healthcare provider.

Connecticut Department of Public Health
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