Glutaric Acidemia, Type II

What is newborn screening?

Newborn screening is a blood test to check for conditions that might be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel soon after birth. This blood sample is required for all newborn babies.

Newborn screening is not the same as diagnostic testing. A diagnostic test can tell with more certainty whether or not a child has a condition. On the other hand, a screening test simply indicates that a child may have a condition. The purpose of a screening test is to find babies that should have diagnostic testing. When a child with an out-of-range newborn screening result has a follow-up test result within the normal range, it is sometimes called a "false positive."

KEY POINTS:

- **You have just heard that your baby may have glutaric acidemia, type II. Please understand that the newborn screening is just that---a screening test. Further testing is required to confirm or rule out the diagnosis.**
- **Many babies who have out of range newborn screens are healthy, and will not be diagnosed.**
- **If treated early, children with glutaric acidemia, type II can have healthy growth and development.**

What if my baby needs more testing?

If you are told that your baby needs follow-up testing, it does not necessarily mean that your baby is at risk. An out of range result may occur because:

- The sample was too small
- The sample was collected too early
- The sample was collected too close to a feeding
- The baby was born too early or had a low birth weight

Most babies who have follow up testing for glutaric acidemia, type II are healthy, and will not be diagnosed. However, out of range screening results CAN indicate a disorder, so it is important to follow your doctor's advice & get your baby tested quickly so that final results can be confirmed.

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What is glutaric acidemia, type 2?

Glutaric acidemia, type II (GA-2) is a very rare condition in which the body is not able to break down certain fats and proteins. Babies affected with GA-2 are unable to convert some of the fats and proteins they eat into energy the body needs to function. This causes harmful substances to build up in the body.

What does this mean?

Although GA-2 cannot be cured, it can be treated. If further testing finds that your baby has GA-2, he or she will have to stay on a special diet throughout life. Some children may benefit from medication or supplements. This will help to prevent health issues such as weak muscle tone and heart problems. If treated early, children with GA-2 can have healthy growth and development.

What happens next?

Your baby's doctor may ask for your baby to have more testing. This follow up testing is important to know if treatment is needed. In some cases, you may be asked to visit a healthcare specialist and/or dietician. The specialist will talk to you about the best plan for your baby.

What are the signs and symptoms of GA-2?

Some babies may not show any symptoms of GA-2. Other babies might have trouble feeding or gaining weight, or sleeping longer or more often. Additional signs of GA-2 may include: fever, trouble breathing, behavior changes or vomiting. If you become concerned about your baby's growth, feeding or activity, please talk to your pediatrician.

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 Diagnosis and Treatment 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 an accurate and informative resource.

This fact sheet was written for information purposes only. It should not replace medical advice, diagnosis or treatment.

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