Galactosemia

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 galactosemia. Please understand that the newborn screening is just that--- a screening test. Further testing is required to confirm or rule out the diagnosis.**
- **Most babies who have out of range newborn screens do not actually have galactosemia, or have a very mild form that requires no treatment.**
- **If treated early, children with galactosemia 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 galactosemia are healthy, and will not be diagnosed with galactosemia. 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 galactosemia?

Galactosemia is a condition present at birth where the body is not able to use a milk sugar, called galactose. Galactose comes from food, including all breast milk, dairy products, and many baby formulas. The term "galactosemia" literally means too much galactose in the blood.

What does this mean?

Although these conditions cannot be cured, they can be treated. If further testing finds that your baby has galactosemia, he or she will have to stay on a galactose free diet throughout life. This can prevent serious health problems. If treated early, children with galactosemia can have healthy growth and development.

What happens next?

Your baby's doctor may ask for the newborn screen to be repeated or, 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. The specialist may want to switch your baby to soy-based formula since breastmilk and milk-based formulas contain galactose.

What are the signs and symptoms of galactosemia?

Signs and symptoms of galactosemia can be very different from one baby to another. Some babies do not show any symptoms. Other babies can have trouble feeding and gaining weight, lack energy or become irritable. If you become concerned about your baby's feeding or activity, please talk to your child's 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 answer questions and 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 professional medical advice, diagnosis or treatment.

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