Galactosemia

What is newborn screening?

Newborn screening is a blood test done soon after birth to check for conditions that do not have symptoms 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 a screening test---it determines whether a baby has a risk of having a condition. If a baby's levels are "out of range," this cues the doctor to order additional testing. This additional testing (called diagnostic testing) is used to learn if, in fact, the condition is present. If galactosemia is diagnosed, then your healthcare team can further talk about treatment, if any is needed.

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

There are 4 typical outcomes from the follow up (diagnostic) testing. Most babies who have follow-up testing for galactosemia are healthy, and will not be diagnosed with galactosemia. In Connecticut, only about 1 out of 70 (1.4%) babies who have out of range screens for galactosemia end up receiving a true diagnosis of galactosemia. About 22 out of 70 (31%) of babies are found to be "carriers" of the galactosemia gene (carriers do not have the disorder). Finally, about 12 out of 70 (17%) of babies have a very mild form that requires no treatment. However, just because most babies with an out of range screening results do not end up with a true diagnosis, please do not think the follow up diagnostic testing is not important. Galactosemia CAN be a serious disorder if not treated. Therefore, it is important to follow your doctor's advice & get your baby tested quickly so that final results can be acted on, if needed.

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What happens next?

Your baby's doctor may ask for the newborn screen to be repeated with another heel stick, or they might need your baby to have a blood test drawn at a blood draw station. This follow up testing is important to know if treatment is needed. This testing can sometimes take 3-4 weeks to get results. 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 while we wait for the diagnostic results. **Unless told otherwise, you can continue to breastfeed and/or formula feed as you have been.**

What is galactosemia?

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

What would a diagnosis of galactosemia mean for my child?

Although galactosemia cannot be cured, it can be treated with diet changes. People with galactosemia will have to stay on a galactose free diet throughout their life. This can prevent serious health problems. If treated early, children with galactosemia can have healthy growth and development.

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 (are very sleepy) 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.

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

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