Biotinidase Deficiency

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 biotinidase deficiency. 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 are healthy, and will not have biotinidase deficiency.**
- **If treated early, children with biotinidase deficiency 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

A positive newborn screen for biotinidase deficiency can, in some cases, be indicative of exposure to heat. Most babies who have follow up testing for biotinidase deficiency are healthy, and will not have biotinidase deficiency. 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 biotinidase deficiency?

Babies with biotinidase deficiency cannot use the vitamin biotin. Biotin is found in many foods and is important for growth and development. An enzyme (a type of chemical in our body) called biotinidase helps separate biotin from the food we eat so that our body can then use it. In babies with biotinidase deficiency, the enzyme doesn't work well so the baby's body doesn't get enough biotin. Treatment is very effective for these babies, and can prevent any symptoms from occurring.

What does this mean?

Although this condition cannot be cured, it can be treated. If further testing shows that your baby has biotinidase deficiency, he or she will have to take a daily dose of biotin. This will prevent health problems such as hair loss, skin rash, and even seizures. If treated early, children with this condition 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. You will want to have these follow up tests done as soon as possible. In some cases, you may be asked to visit a specialist and/or dietician. The specialist and/or dietician may recommend a biotin supplement.

What are the signs and symptoms of biotinidase deficiency?

Babies that have biotinidase deficiency usually have no symptoms at birth. Within a few weeks or months after birth, symptoms may develop if the baby is not treated. The signs and symptoms can be very different from one baby to another. Some common early signs include: low muscle tone (floppiness), hair loss, or a rash. Once treatment begins, most of the symptoms will disappear. If treatment is started before symptoms appear, they usually won't develop any symptoms.

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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