Tyrosinemia

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.

KEY POINTS:

- **You have just heard that your baby may have tyrosinemia. 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 for tyrosinemia are healthy, and will not be diagnosed.**
- **If treated early, children with tyrosinemia 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 tyrosinemia are healthy, and will not be diagnosed. In fact, about 10% of babies have transient tyrosinemia, meaning it lasts only a short time. This is likely due to vitamin C deficiency or an immature liver. No treatment is necessary for transient tyrosinemia. 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.

Connecticut Department of Public Health
Connecticut Newborn Screening Program • 860.920.6628
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Adapted, with permission, from the Minnesota Department of Public Health

What is tyrosinemia?

Tyrosinemia is part of a group of disorders called amino acid disorders. With tyrosinemia, the body is unable to break down protein from the food we eat containing tyrosine. This causes tyrosine to build up in the blood. There are three types of tyrosinemia, each with slightly different symptoms and treatments.

What does this mean?

Although tyrosinemia cannot be cured, it can be treated. If further testing finds that your baby has tyrosinemia, he or she may have to be on a special low protein diet. Certain medications may be prescribed to help lower the tyrosine levels in the blood. This will prevent health problems with the kidney and liver, skin and eyes, poor weight gain, and more. If treated early, children with tyrosinemia 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.

What are the signs and symptoms of tyrosinemia?

Signs and symptoms of tyrosinemia 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 have diarrhea or vomiting. 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.