Amino Acid Disorder (OTC/CPS)

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 an amino acid disorder. 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 amino acid disorders are healthy, and will not be diagnosed.**
- **If treated early, children with amino acid disorders can have healthy growth and**

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 amino acid disorders 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 are OTC and CPS?

Ornithine Transcarbamylase Deficiency (OTC) and Carbamoyl Phosphate Synthetase 1 Deficiency (CPS) are part of a group of disorders called amino acid disorders. Our body breaks down proteins in food into their basic building blocks (amino acids). During this process, a waste called ammonia is produced. When the body's process for removing waste is not working, dangerous amounts of ammonia begin to build up in the blood. If left untreated, this can result in developmental delays, or medical problems.

What does this mean?

Although OTC and CPS cannot be cured, they can be treated. If further testing finds that your baby has one of these amino acid disorders, he or she may have to be on a special low-protein diet. Certain medications may be prescribed to help lower the ammonia levels in the blood. This will prevent health problems such as developmental delays, learning disabilities or tight muscles. If treated early, children with amino acid disorders 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 and/or dietician.

What are the signs and symptoms of OTC and CPS?

Signs and symptoms of OTC and CPS 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, get cold easily and more. 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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