Medium Chain Acyl CoA Dehydrogenase Deficiency (MCADD)

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.

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 MCADD. Please understand that the newborn screening is just that---a screening test. Further testing is needed to confirm or rule out the diagnosis.**
- **Most babies who have out of range newborn screens are healthy, and will not have MCADD.**
- **If treated early, children with MCADD 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 MCADD are healthy, and will not have MCADD. 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 MCADD?

Medium-chain acyl-CoA dehydrogenase deficiency (MCADD) is a condition in which the body is not able to break down certain fats. It is a fatty acid oxidation condition because people with MCADD are unable to change some of the fats they eat into energy the body needs to function. Instead, too many unused fatty acids build up in the body.

What does this mean?

If further testing finds that your baby has MCADD, they will meet with a geneticist and dietician to discuss MCADD in more detail and to go over questions that you might have. A specialist may recommend medication, supplements, or frequent feedings. This will help to prevent health problems. Children with MCADD can have healthy growth and development.

What happens next?

Your baby's doctor may ask 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.

What are the signs and symptoms of MCADD?

If untreated, MCADD can cause medical problems. However, if the condition is found and treated early, individuals with MCADD can often lead healthy lives. The signs and symptoms can be very different from one baby to another. Some common early sings include: extreme sleepiness, poor appetite, vomiting, diarrhea, changes in behavior, or fever. If you become concerned about your baby's growth, 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 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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