



## Medium Chain Acyl CoA Dehydrogenase Deficiency (MCAD)

### 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."

### 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 MCAD are healthy, and will not have MCAD. 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.

#### KEY POINTS:

- **You have just heard that your baby may have MCAD. 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 MCAD.**
- **If treated early, children with MCAD can have healthy growth and development.**

Connecticut Department of Public Health

Connecticut Newborn Screening Program • 860.920.6628

Connecticut Newborn Diagnosis and Treatment Network • 860.837.7870

Adapted, with permission, from the Minnesota Department of Public Health

## What is MCAD?

Medium-chain acyl-CoA dehydrogenase deficiency (MCAD) is a condition in which the body is not able to break down certain fats. It is considered a fatty acid oxidation condition because people affected with MCAD 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?

Although these conditions cannot be cured, they can be treated. If further testing finds that your baby has MCAD, he or she may need to be on a special diet. A specialist may recommend medication, supplements, or frequent feedings. This will help to prevent health problems. If treated early, children with MCAD 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. The specialist and/or dietician will talk to you about the best plan for your baby.

## What are the signs and symptoms of MCAD?

If untreated, MCAD can cause significant medical problems. However, if the condition is found and treated early, individuals with MCAD can often lead healthy lives. The signs and symptoms can be very different from one baby to another. Some common early signs 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 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](http://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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