



Carnitine Uptake 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."

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 carnitine uptake deficiency are healthy, and will not have carnitine uptake 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.

KEY POINTS:

- **You have just heard that your baby may have carnitine uptake 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 carnitine uptake deficiency.**
- **If treated early, children with carnitine uptake deficiency 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 carnitine uptake deficiency?

Carnitine uptake deficiency (CUD) affects the body's ability to recycle an important nutrient called carnitine. Carnitine helps the body make energy from fats and also helps the body use stored fat. A person with CUD may not be able to break down fat properly. CUD is different for each child. Some children with CUD do not have any health problems, while other children may have very serious complications.



What does this mean?

Although these conditions cannot be cured, they can be treated. If further testing finds that your baby has CUD, he or she may need to be on a special diet. Sometimes, a doctor will recommend feeding your baby more frequently. Certain medications and supplements may be prescribed. This can help to prevent health problems. If treated early, children with CUD 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 CUD?

Some babies with CUD never have serious signs or symptoms. Sometimes signs and symptoms may not happen until later in life. Signs and symptoms could include: sleepiness, not eating well or having trouble gaining weight, or behavior changes. 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 as an accurate and informative resource.

This fact sheet was written for information purposes only. It should not replace medical advice, diagnosis or treatment.