

Positive Result:

Blood Spot Screen Result Notification



Absent/Reduced C0 Acylcarnitine (Free Carnitine)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has no or low levels of free carnitine.

What does this mean?

Absent or low levels of free carnitine can indicate that your child may have carnitine uptake defect (CUD). A positive result does not mean your baby has CUD, but more testing is needed to know for sure.

Some babies may have low free carnitine on newborn screening because the mother has low carnitine levels. These babies do not have CUD and their carnitine levels should become normal over time. More testing for both mom and baby is needed to determine if this is the case.

What happens next?

Your baby's doctor or a metabolic specialist familiar with CUD will help arrange for more testing. Your baby will also be seen by a metabolic specialist.

What is carnitine uptake defect (CUD)?

Carnitine uptake defect is part of a group of disorders called fatty acid oxidation disorders. CUD prevents the body from using fat from food for energy it needs to function. Using stored fat for energy is especially important between meals when the body is not getting new energy from food. During periods without food (fasting) or illness, health problems can begin.

What health problems can it cause?

Health problems for people with carnitine uptake defect can range from mild to life-threatening. In fact, some are so mild that some mothers are found to have this disorder through their child's newborn screen.

However, if untreated, CUD can cause:

- Lack of energy
- Vomiting
- Low blood sugar (hypoglycemia)
- Muscle weakness
- Heart and liver problems
- Seizures
- Coma
- Sudden death

Children with CUD may benefit from prompt and careful treatment.

What treatment options are available?

Although CUD cannot be cured, it can be treated. Children with CUD are treated with a high-carbohydrate, low-fat diet and avoidance of fasting. Carnitine supplementation may be prescribed. If treated early, children can have healthy growth and development.

Children with CUD should see their regular doctor and a doctor who specializes in this condition.

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org