

Positive Result:

Blood Spot Screen Result Notification



Elevated C0 Acylcarnitine (Free Carnitine)

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of free carnitine.

What does this mean?

High levels of free carnitine can indicate that your child has carnitine palmitoyl transferase type I (CPT-I) deficiency. A positive result does not mean your baby has CPT-I deficiency, but more testing is needed to know for sure.

What happens next?

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

What is carnitine palmitoyl transferase type I (CPT-I) deficiency?

CPT-I deficiency is part of a group of disorders called fatty acid oxidation disorders. With CPT-I deficiency, the body is unable to change some fats into energy the body 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?

CPT-I deficiency is a lifelong condition. If untreated, it can cause:

- Lack of energy
- Irritable mood
- Poor appetite
- Vomiting
- Diarrhea
- Low blood sugar (hypoglycemia)
- Seizures
- Coma, sometimes leading to death

Children with CPT-I deficiency can benefit from prompt and careful treatment.

What treatment options are available?

Although CPT-I deficiency cannot be cured, it can be treated. Children with CPT-I deficiency are treated with a high-carbohydrate, low-fat diet and avoidance of fasting. Certain supplements may be prescribed. If treated before symptoms develop, children can have healthy growth and development.

Children with CPT-I deficiency should see their regular doctor and a doctor who specializes in metabolic disorders.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org