Family Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C16 and/or Elevated C18:1 Acylcarnitines

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C16 and/or C18:1 acylcarnitines.

What does this mean?

High levels of C16 and/or C18:1 acylcarnitines can indicate that your child has a metabolic disorder. If your baby has a metabolic disorder, more testing is needed to find out which metabolic disorder it is. The most likely are carnitine palmitoyl transferase type 2 (CPT-II) deficiency or CACT deficiency. A positive result does not mean your baby has CPT-II or CACT deficiency, but more testing is needed to know for sure.

What happens next?

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

What is CPT-II and CACT deficiency?

CPT-II and CACT deficiency are part of a group of disorders called fatty acid oxidation disorders. With CPT-II and CACT 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-II and CACT deficiency are lifelong conditions. If untreated, they can cause:

- Muscle weakness (hypotonia)
- Low blood sugar (hypoglycemia)
- Enlarged liver (hepatomegaly)
- Breathing problems
- Heart problems
- Seizures

Children with these disorders can benefit from prompt and careful treatment.

What treatment options are available?

Although CPT-II and CACT deficiency cannot be cured, they can be treated. Treatment consists of a high-carbohydrate, low-fat diet and avoidance of fasting. Certain supplements may be prescribed. Early treatment can be life-saving. Even with treatment, however, long-term health problems can occur.

Children with CPT-II and CACT 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

DEPARTMENT OF HEALTH

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155
Phone (800) 664-7772, Fax (651) 215-6285 ***interpreters available***

REV 12/2018