

Positive Result:

Blood Spot Screen Result Notification



Elevated C16-OH Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C16-OH acylcarnitine.

What does this mean?

High levels of C16-OH can indicate that your child has long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency or trifunctional protein (TFP) deficiency. A positive result does not mean your baby has LCHAD or TFP deficiency, but more testing is needed to know for sure.

What happens next?

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

What is LCHAD and TFP deficiencies?

LCHAD and TFP deficiencies are part of a group of disorders called fatty acid oxidation disorders. With LCHAD and TFP, 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?

LCHAD and TFP deficiencies are lifelong conditions. If untreated, they can cause:

- Lack of energy
- Feeding difficulties
- Low blood sugar (hypoglycemia)
- Muscle weakness (hypotonia)
- Liver problems

Children with LCHAD and TFP deficiencies may benefit from prompt and careful treatment.

What treatment options are available?

Although LCHAD and TFP deficiencies cannot be cured, they can be treated. Children with these disorders are treated with a high-carbohydrate, low-fat diet and avoidance of fasting. Certain supplements may be prescribed. Early treatment provides the most benefit to children. Even with treatment, however, long-term health problems can occur.

Children with LCHAD and TFP deficiencies should see their regular doctor and a doctor who specializes in these 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