

Positive Result:

Blood Spot Screen Result Notification



Elevated C14:1 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C14:1 acylcarnitine.

What does this mean?

High levels of C14:1 acylcarnitines can indicate that your child has very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. A positive result does not mean your baby has VLCAD deficiency, but more testing is needed to know for sure.

What happens next?

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

What is very long-chain acyl-Co A dehydrogenase (VLCAD) deficiency?

VLCAD deficiency is part of a group of disorders called fatty acid oxidation disorders. With VLCAD, 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?

VLCAD deficiency is a lifelong condition. If untreated, it can cause:

- Lack of energy
- Muscle weakness (hypotonia)
- Poor appetite
- Vomiting
- Diarrhea
- Low blood sugar (hypoglycemia)

Children with VLCAD deficiency can benefit from prompt and careful treatment.

What treatment options are available?

Although VLCAD deficiency cannot be cured, it can be treated. Children with VLCAD deficiency are treated with a high-carbohydrate, low-fat diet and avoidance of fasting. Certain medications may be prescribed to help break down the fats. If treated before symptoms develop, children can have healthy growth and development.

Children with VLCAD deficiency should see their regular doctor and a doctor who specializes in VLCAD deficiency.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org