

Positive Result:

Blood Spot Screen Result Notification



Elevated C4 and Elevated C5 Acylcarnitines

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of both C4 and C5 acylcarnitines.

What does this mean?

High levels of C4 and C5 acylcarnitines can indicate that your child has glutaric acidemia type II (GA-2). A positive result does not mean your baby has GA-2, 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 glutaric acidemia type II (GA-2)?

GA-2 is part of a group of disorders called fatty acid oxidation disorders. GA-2 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?

GA-2 is a lifelong condition and is different for each child. Some babies have severe symptoms that are present at birth and are life-threatening. Others have a less severe form that begins in childhood or adulthood.

If untreated, it can cause:

- Lack of energy
- Poor feeding
- Vomiting
- Low blood sugar (hypoglycemia)
- Muscle weakness (hypotonia)
- Trouble breathing
- A "sweaty feet" smell
- Seizures

Children with GA-2 can benefit from prompt and careful treatment.

What treatment options are available?

Although GA-2 cannot be cured, it can be treated. Treatment consists of a high-carbohydrate, low-fat diet and avoidance of fasting. Certain medications and supplements may be prescribed. Early treatment provides the most benefit to children. Even with treatment, however, long-term health problems can occur.

Children with GA-2 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