

Positive Result:

Blood Spot Screen Result Notification



Elevation of C5 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C5 acylcarnitine.

What does this mean?

High levels of C5 acylcarnitine 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 common metabolic disorders with high levels of C5 acylcarnitine are isovaleric acidemia (IVA) and short/branched chain acyl-CoA dehydrogenase (SBCAD) deficiency. A positive result does not mean your baby has IVA, SBCAD deficiency, or a different metabolic disorder, 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 IVA and SBCAD deficiency?

IVA and SBCAD deficiency are part of a group of disorders called organic acid disorders. With IVA and SBCAD deficiency, the body is unable to break down certain proteins from food. When the body cannot break down these proteins, it can cause harmful toxins to build up and cause health problems.

What health problems can it cause?

Some children with IVA have few health problems, while others have very serious complications. If untreated, it can cause:

- Poor feeding
- Sleepiness
- Vomiting
- Seizures

Very few children with SBCAD deficiency develop health problems. Possible health problems include:

- Low blood sugar
- Sleepiness
- Poor feeding

Most people with SBCAD deficiency are of Hmong descent and have no health problems.

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

What treatment options are available?

Although IVA and SBCAD deficiency cannot be cured, they can be treated. Treatment for IVA and SBCAD deficiency (if needed) consists of a diet low in protein.

Children with IVA and SBCAD 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