

Positive Result:

Blood Spot Screen Result Notification



Elevation of C4 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C4 acylcarnitine.

What does this mean?

High levels of C4 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 disorder with high levels of C4 acylcarnitine is short-chain acyl-CoA dehydrogenase (SCAD) deficiency. A positive result does not mean your baby has SCAD 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 of both mom and baby. Your baby will also be seen by a metabolic specialist.

What is short-chain-acyl-CoA dehydrogenase (SCAD) deficiency?

SCAD deficiency is part of a group of disorders called fatty acid oxidation disorders. With SCAD, 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 cause it?

SCAD deficiency is a lifelong condition. Most people with SCAD deficiency do not have any health problems. Some children develop health problems. Possible health problems include:

- Low blood sugar
- Sleepiness
- Vomiting
- Muscle weakness
- Poor growth and weight gain
- Seizures

Children with SCAD deficiency who are having health problems can benefit from prompt and careful treatment.

What treatment options are available?

Although SCAD deficiency cannot be cured, it can be treated. If treatment is recommended, children are treated with certain medications to help break down fat into energy and avoid fasting.

Some metabolic specialists want children with SCAD deficiency to have regular check-ups while other specialists do not. It is very important to follow the metabolic specialist's testing and follow-up recommendations.

Resources

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

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org