

Positive Result:

Blood Spot Screen Result Notification



Elevated C4-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 C4-OH acylcarnitine.

What does this mean?

High levels of C4-OH acylcarnitine can indicate that your child has medium/short-chain hydroxyacyl-CoA dehydrogenase (M/SCHAD) deficiency. A positive result does not mean your baby has M/SCHAD deficiency, but more testing is needed to know for sure.

What happens next?

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

What is medium/short-chain hydroxyacyl-CoA dehydrogenase (M/SCHAD) deficiency?

M/SCHAD deficiency is part of a group of disorders called fatty acid oxidation disorders. With M/SCHAD, 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?

M/SCHAD deficiency is a lifelong condition. If untreated, it can cause:

- Sleepiness
- Vomiting
- Low blood sugar
- Seizures
- Heart and breathing problems
- Coma, sometimes leading to death

Children with M/SCHAD deficiency can benefit from prompt and careful treatment.

What treatment options are available?

Although M/SCHAD deficiency cannot be cured, it can be treated. Children with M/SCHAD 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 M/SCHAD deficiency should see their regular doctor and a doctor who specializes in M/SCHAD 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