

Positive Result:

Blood Spot Screen Result Notification



Elevated C8 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C8 acylcarnitine.

What does this mean?

High levels of C8 acylcarnitine can indicate that your child has medium-chain acyl-CoA dehydrogenase (MCAD) deficiency. A positive result does not mean your baby has MCAD deficiency, but more testing is needed to know for sure.

What happens next?

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

What is medium-chain acyl-CoA dehydrogenase (MCAD) deficiency?

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

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

- Vomiting
- Lack of energy (lethargy)
- Low blood sugar (hypoglycemia)
- Seizures
- Breathing difficulties
- Liver problems
- Brain damage
- Coma
- Sudden death

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

What treatment options are available?

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