

Positive Result:

Blood Spot Screen Result Notification



Elevated C3-DC Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C3-DC acylcarnitine.

What does this mean?

High levels of C3-DC acylcarnitine can indicate that your child has malonic acidemia. A positive result does not mean your baby has malonic acidemia, 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 malonic acidemia?

Malonic acidemia is part of a group of disorders called organic acid disorders. With malonic acidemia, the body is unable to change some fats into energy the body needs to function. When the body cannot break down these fats, it can cause harmful toxins to build up and cause health problems.

What health problems can it cause?

Malonic acidemia is a lifelong condition. If untreated, it can cause:

- Poor feeding
- Sleepiness
- Vomiting
- Developmental delay
- Muscle weakness (hypotonia)
- Enlarged heart (cardiomyopathy)
- Seizures

Children with malonic acidemia may benefit from prompt and careful treatment.

What treatment options are available?

Although malonic acidemia cannot be cured, treatment may be beneficial. Because malonic acidemia is so rare, not much is known about the effectiveness of treatment. Treatment can include a diet low in fat.

Children with malonic acidemia 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