Family Fact Sheet

Positive Result:

Blood Spot Screen Result Notification

Minnesota Newborn Screening Program

Elevated C4-DC + C5-OH Acylcarnitines

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C4-DC + C5-OH acylcarnitines.

What does this mean?

High levels of these acylcarnitines 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 likely is 3-methylcrotonyl-CoA carboxylase (3-MCC) deficiency. A positive result does not mean your baby has 3-MCC deficiency, or a different metabolic disorder, but more testing is needed to know for sure.

A baby may screen positive because of elevations of C5-OH from their mother who may have 3-MCC deficiency but has no symptoms. Again, more testing (including testing the mother) is needed.

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 3-MCC deficiency?

3-MCC deficiency is part of a group of disorders called organic acid disorders. With 3-MCC 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?

Health problems for people with 3-MCC deficiency can range from mild to life-threatening. In fact, some are so mild that some mothers are found to have this disorder through their child's newborn screen.

However, if untreated, 3-MCC can cause:

- Poor feeding
- Sleepiness
- Vomiting and diarrhea
- Muscle weakness (hypotonia)
- Seizures

Children with 3-MCC deficiency can benefit from prompt and careful treatment.

What treatment options are available?

Although 3-MCC deficiency cannot be cured, it can be treated. Treatment for 3-MCC deficiency (if needed) consists of a diet low in protein.

Children with 3-MCC 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

DEPARTMENT OF HEALTH

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155
Phone (800) 664-7772, Fax (651) 215-6285 ***interpreters available***

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