

Positive Result:

Blood Spot Screen Result Notification



Elevated C5-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 C5-DC acylcarnitine.

What does this mean?

High levels of C5-DC acylcarnitine can indicate that your child has glutaric acidemia type I (GA-I). A positive result does not mean your baby has GA-I, 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 glutaric acidemia type I?

GA-I is part of a group of disorders called organic acid disorders. With GA-I, the body is unable to breakdown some proteins into energy the body needs to function. 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?

GA-I is a lifelong condition and is different for each child. Some children with GA-I have only a few health problems. In fact, some health problems are so mild that some mothers are found to have this disorder through their child's newborn screen.

Children with GA-I are usually healthy at birth, but they can have a large head (macrocephaly).

If untreated, GA-I can cause:

- Poor appetite
- Tiredness
- Difficulty moving
- Abnormal movements, such as twitches or spasms
- Muscle weakness (hypotonia)
- Rigid muscles
- Brain damage

Children with GA-I may benefit from prompt and careful treatment.

What treatment options are available?

Treatment for GA-I consists of a diet low in protein and medications during illness. Certain supplements may be prescribed. Even with treatment, however, long-term health problems can occur.

Children with GA-I 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