

Positive Result:

Blood Spot Screen Result Notification



Elevated C10:2 Acylcarnitine

What was found on the newborn screen?

The newborn screen that was collected at birth found that your baby has high levels of C10:2 acylcarnitine.

What does this mean?

High levels of C10:2 acylcarnitines can indicate that your child has 2, 4--Dienoyl-CoA reductase deficiency (DE-RED). A positive result does not mean your baby has DE-RED, 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 2,4-Dienoyl-CoA Reductase Deficiency (DE-RED)?

DE-RED is part of a group of disorders called fatty acid oxidation disorders. With DE-RED, 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?

Because only two children have ever been reported to have DE-RED, it is very difficult to know how the disorder affects children.

The two children shared the following symptoms:

- Life threatening, whole body response to an infection (sepsis)
- Small head (microcephaly)
- Muscle weakness (hypotonia)
- Poor feeding
- Shortened lifespan

What treatment options are available?

Because there have only been two patients reported with DE-RED, the medical community is still learning about this condition and the effectiveness of treatment. The suggested treatment for children with DE-RED consists of a special diet, medications, and avoidance of fasting.

Children with DE-RED should see their regular doctor and a metabolic specialist.

Resources

Genetics Home Reference:
<http://ghr.nlm.nih.gov>

Save Babies Through Screening Foundation:
www.savebabies.org

Baby's First Test:
www.babysfirsttest.org