



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

The three reported children with DE-RED had symptoms starting as an infant. All three had a small head (microcephaly), brain abnormalities, muscle weakness (hypotonia), and poor feeding and/or difficulties growing and gaining weight (failure to thrive). Other symptoms were found in some, but not all, of the three diagnosed children. These include seizures, vomiting, movement differences (such as significant muscle stiffness, difficulties walking, and/or difficulties with coordination), life threatening response to an infection (sepsis), and eye/vision changes. Children with DE-RED may have a shortened lifespan.

#### What treatment options are available?

Because there have only been three children reported with DE-RED, the medical community is still learning about this condition and its treatment. The suggested treatment consists of a special diet, medications, and avoiding fasting. Children with DE-RED should see their regular doctor and a metabolic specialist.

### Resources



Scan here for more information about newborn screening in Minnesota or visit our website:  
[www.health.state.mn.us/people/newbornscreening](http://www.health.state.mn.us/people/newbornscreening)

#### MedlinePlus:

[www.medlineplus.gov/genetics](http://www.medlineplus.gov/genetics)

#### Save Babies Through Screening Foundation:

[www.savebabies.org](http://www.savebabies.org)

#### Newborn Screening Info Center:

[www.newbornscreening.hrsa.gov](http://www.newbornscreening.hrsa.gov)