

Elevated C10:2 2,4-Dienoyl-Coenzyme A Reductase Deficiency (DE RED)

What is DE RED?

2,4 Dienoyl-CoA reductase deficiency (DE RED) is a condition in which the body is unable to break down certain fats. It is considered a fatty acid oxidation condition because people affected with DE RED are unable to change some of the fats they eat into the energy the body needs to function. DE RED can cause weak muscle tone and can lead to the presence of too much acid in the blood. The effectiveness of treatment is currently unknown.

What Causes DE RED?

When we eat food, enzymes help break it down. Some enzymes break down fats into their building blocks, called fatty acids. Other enzymes break down these fatty acids. The enzyme 2, 4 Dienoyl-CoA reductase helps break down unsaturated fatty acids. In DE RED, the body may either not make enough or make non-working 2, 4 dienoyl-CoA reductase. When this happens, the body cannot fully break down unsaturated fatty acids. This causes harmful substances to build up in the body.

What Symptoms or Problems Occur with DE RED?

[Symptoms are something out of the ordinary that a parent notices.]

DE RED is extremely rare. A baby with this condition had signs including:

- small body and head size
- short torso, arms, and fingers
- weak muscle tone (known as hypotonia)
- poor appetite
- vomiting
- irritability
- delayed weight gain

What is the Treatment for DE RED?

Dietary Treatments - A baby with DE RED was treated with a low-lysine formula. Lysine is a building block of proteins. The baby had dangerously high levels of this substance in her blood.

Medications and Supplements - This baby was also treated with L-carnitine supplements. L-carnitine is a natural substance made by the body, but babies with DE RED might not make enough of it. These supplements help the body break down fats for energy and get rid of harmful substances that may build up. Your baby's doctor needs to write a prescription for L-carnitine supplements.

Things to Remember

DE RED is extremely rare.