TEXAS Department of State Health Services

Newborn Screening FACT Sheet

Elevated C16 and/or C18:1 Acylcarnitine

Carnitine Palmitoyltransferase 2 (CPTII) Deficiency

What is CPTII?
Carnitine palmitoyltransferase type II deficiency (CPTII) 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 CPTII are unable to change some of the fats they eat into energy the body needs to function. This can cause too many unused fatty acids to build up in the body. The health outcomes and treatment of CPTII depend on the age of the onset. Detecting the condition early and beginning treatment can often prevent many of the severe health outcomes of CPTII.

What causes CPTII?
When we eat food, enzymes help break it down. Some enzymes help break down fats into their building blocks, called fatty acids. Other enzymes break down these fatty acids. In CPTII, the enzyme carnitine palmitoyltransferase II is not working correctly. This enzyme's job is to prepare fatty acid to be broken down inside the mitochondria. Mitochondria are the energy making factories of cells. Carnitine helps bring fatty acids into the mitochondria. The fatty acids cannot enter unless they are attached to carnitine. Once inside, carnitine palmitoyltransferase II removes carnitine so the fatty acid can be broken down for energy. Individuals with CPTII cannot remove carnitine from the fatty acids in the mitochondria, so they cannot break down fatty acids for energy. Fatty acids are important sources of energy for the heart, especially when sugars are low, such as between meals.

What Symptoms or Problems Occur with CPTII?
[Symptoms are something out of the ordinary that a parent notices]

There are three main forms of CPTII which differ in their age of onset: an extremely severe form in newborns (neonatal), a severe form in babies and young children, and a mild form in adults. Most individuals with CPTII have the "classic" adult form. Classic CPTII has different signs than CPTII in babies or children.

Signs of CPTII in babies include:

- sleeping longer or more often
- weak muscle tone (known as hypotonia)
- behavior changes (such as crying for no reason)
- irritability
- poor appetite
- fever / diarrhea / vomiting
- low blood sugar (known as hypoglycemia)
- trouble breathing/Seizures
- developmental delays

What is the Treatment for CPTII?
The following treatments are often used for children with CPT II

1. Dietary Treatment - Your baby may need to be on a restricted diet in order to avoid certain foods that his or her body cannot break down. A dietician or nutritionist can help you plan a healthy diet for your child. Eating often can also help your baby avoid many of the signs mentioned in the symptoms section.

2. Supplements and Medications - Medium Chain Triglyceride (MCT) oil supplements are a common treatment for CPTII.

Your Doctor might also prescribe L-carnitine supplements. L-carnitine is a substance naturally made by the body, but your baby might not make enough of it.

3. Lifestyle Changes – Your baby may need to avoid the cold because cold weather can trigger some signs of CPTII, such as muscle weakness (see Symptoms or Problems).

Things to Remember
Always call your doctor when your child has any of the Symptoms or Problems mentioned.

Babies and children with CPTII need to eat extra starchy food and drink more fluids during any illness even if they may not feel hungry or they could have a Metabolic Crisis. Children who are sick often don't want to eat. If they won't or can't eat, they may need to be treated in the hospital to prevent a metabolic crisis. Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.