

Carnitine Palmitoyl Transferase Deficiency – Type 1A (CPT1)

What is CPT1?

Carnitine palmitoyl transferase I deficiency (CPT1) 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 CPT1 are unable to change some of the fats they eat and store in their body into the energy the body needs to function. This can cause too many unused fatty acids to build up in the body. Detecting the condition early and beginning treatment can often prevent the severe outcomes of CPT1.

What causes CPT1?

Enzymes help start chemical reactions in the body. CPT1 happens when an enzyme called “carnitine palmitoyl transferase” is missing or not working. This enzyme breaks down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.

What Symptoms or Problems Occur with CPT1?

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

Children with CPT1 usually start showing symptoms between the ages of 8 – 18 months. CPT1 may cause periods of illness called Metabolic Crises, or low blood sugar. Some of the first signs of a metabolic crisis are:

- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a metabolic crisis is not treated, a child with CPT1 can develop:

- breathing problems and seizures
- coma, sometimes leading to death

Many metabolic crises can lead to brain damage and learning problems. They can also have liver, heart or kidney problems. Some children with CPT1 never have any problems.

What is the Treatment for CPT1?

The following treatments are often used for children with CPT1:

1. Do not go a long time without food – Babies and young children with CPT1 should eat often to avoid low blood sugar or a metabolic crisis. They should not go without food for more than 4 to 6 hours. Some babies may need to eat more often.

2. Diet – Sometimes your child will need to eat a diet low in fat (lean meat and low-fat dairy foods) and high in carbohydrates (such as bread, noodles, fruits, vegetables). Your dietitian will make any needed diet changes.

3. Medium Chain Triglyceride (MCT) oil – Your doctor may prescribe MCT oil. This special oil has medium chain fatty acids that can be used for energy.

Things to Remember

In addition to the list of Symptoms or Problems, always call your doctor when your child has any of the following:

- vomiting
- diarrhea
- an infection
- a fever

Children with CPT1 need to eat extra starchy food (such as bread, cereal, and rice) and drink more fluids during any illness even if they don't want to eat. When they become sick, they often need to be treated in the hospital to prevent serious health problems. After 5 years of age the metabolic crises happen less often.