Carnitine Palmitoyltransferase Deficiency

Carnitine palmitoyltransferase (CPT) deficiency is a rare condition that causes muscle weakness and other symptoms. It happens because of a problem with one of two enzymes, CPT1 or CPT2.

CPT enzymes help get fatty acids into cells to use for energy. They are very important for certain cells, such as muscle cells. With CPT deficiency, cells that need fatty acids for energy may not work well. This leads to symptoms such as muscle weakness and heart or liver problems.

Types of CPT deficiency

There are 2 main types of CPT deficiency:

- **CPT1 deficiency.** This type is from a problem with the CPT1 enzyme. The CPT1 enzyme helps get fatty acids inside your cells. This form of CPT deficiency is very rare.
- **CPT2 deficiency.** This type is from a problem with the CPT2 enzyme. The CPT2 enzyme helps make sure your body can use fatty acids once they are inside your cells. There are several forms of CPT2 deficiency. The severe neonatal form shows up in the first few days of life. The infantile form affects children usually before age 1. The one that affects adults is called the myopathic form.

What causes CPT deficiency?

CPT deficiency is caused by an abnormal gene. A problem with the gene might cause the CPT1 or CPT2 enzymes to be missing or not work normally. You may have CPT deficiency if it runs in your family. In most cases, a person needs to inherit an abnormal gene from both parents.

Symptoms of CPT deficiency

CPT1 deficiency can cause:

- Symptoms of low blood sugar (hypoglycemia)
- Liver problems, such as an enlarged liver
- Nervous system damage from liver problems
- Coma and sudden death
- Heart failure

CPT2 deficiency can affect each person differently, depending on how well the CPT2 protein is working. Symptoms may be mild to severe. For periods of time, people with CPT deficiency may not have any symptoms. Very severe forms can affect newborns or babies. These are more rare.

The myopathic form of CPT2 deficiency causes milder symptoms. It doesn't cause heart or liver problems. People with this form usually have a normal lifespan. Symptoms may start any time up to age 60. Symptoms can appear with skipping meals, a lot of exercise, or illness. Symptoms may include:

- Symptoms of low blood sugar (hypoglycemia)
- Temporary muscle pain
- Muscle breakdown
- Muscle weakness

- Dark urine

Diagnosing CPT deficiency

You may be diagnosed by a neurologist or geneticist. You'll be asked about your symptoms and medical history. The doctor may also ask about your family's medical history. You'll be given a physical exam that may include a nervous system exam. Tests may also be done, such as:

- **Blood tests.** These are done to check for signs of CPT deficiency in the blood. And they check for enzymes in the blood that can show liver disease.
- **Exercise tests.** These help find the type of metabolic problem.
- **Genetic test.** This kind of test can confirm a CPT deficiency.
- **Heart tests.** Tests such as an echocardiogram can show if the heart is affected.
- **Urine test.** This test looks for a protein called ketones. It also looks for another substance in the urine to check for muscle breakdown.

Treatment for CPT deficiency

A change in diet is the main treatment for CPT deficiency. Your diet should be high in carbohydrates and low in fats and protein. It's also important to eat often. This can prevent low blood sugar. It can also prevent the body from using fats for energy. Also, don't do things that may trigger symptoms. These may include skipping meals, being exposed to cold, stress, and getting a lot of exercise. Eating high-carbohydrate meals before exercise may help. If you are told to fast for a blood test or procedure, ask your healthcare provider about getting a dextrose infusion during the fasting period so you won't become weak.

Possible complications of CPT deficiency

In some cases, the myopathic form of CPT2 deficiency can cause kidney failure. This happens when muscle breaks down during exercise. This creates a chemical that can damage the kidneys. You may need IV (intravenous) fluids or dialysis to prevent or treat this.

Heart weakness (cardiomyopathy) is a possible complication of some forms of CPT deficiency. A weakened heart may not be able to pump blood as well. This can lead to symptoms such as swelling and shortness of breath. Untreated heart weakness may lead to death early in life.

Liver problems are also a possible complication. Liver problems can lead to low blood sugar (hypoglycemia). This is most often triggered by an infection. Severe low blood sugar can cause brain damage. This can lead to coma and death.

The liver and heart are not affected in the myopathic form.

Preventing CPT deficiency

If the condition runs in your family, you may choose to see a genetic specialist before you have a baby. Genetic testing can be done to find out your risk of passing the disease to your child. If you do have the abnormal gene for the condition, you have choices. If you use in vitro fertilization, the embryos can be tested for the disease. Amniocentesis can also be used to test for the condition in early pregnancy.