

Secondary Carnitine Deficiency



Carnitine is a nutrient that helps the body's cells work normally. Secondary carnitine deficiency is when there isn't enough carnitine in the blood. This can cause muscle weakness. It can also cause heart or liver problems. Secondary carnitine deficiency can be caused by a number of health problems.

How carnitine works in the body

You get carnitine through some of the foods you eat. It helps get fatty acids into cells to use for energy. Carnitine is vital for certain cells, such as muscle cells. With less carnitine, cells that need fatty acids for energy may not work well.

What causes secondary carnitine deficiency?

A variety of health problems can lower the levels of carnitine in the blood. They may do this by increasing the amount sent out in urine. Or they may cause the body to absorb less from food. The health problems that can cause it include:

- Certain metabolic disorders
- Certain medicines, such as valproate
- Digestive diseases that don't allow your body to absorb nutrients
- Kidney disease, especially with dialysis
- Liver disease
- Malnutrition
- Mitochondrial disease

Symptoms of secondary carnitine deficiency

Symptoms can occur a bit differently in each person. You may have no symptoms, or your symptoms may be mild to severe. Some symptoms may appear if you skip meals, get a lot of exercise, or have an illness. Symptoms can include:

- Decreased or floppy muscle tone
- Fatigue
- Irritability
- Muscle weakness
- Shortness of breath
- Swelling (edema)

Diagnosing secondary carnitine deficiency

The process to diagnose carnitine deficiency starts with a medical history and a physical exam. The healthcare provider will ask about your symptoms. They may also ask about your family's medical history. The physical exam may include a neurological exam. Tests may also be done, such as:

- **Blood tests.** These are done to check the levels of carnitine in the blood. They also check for creatine kinase, which shows muscle damage. And they check for enzymes in the blood that can show liver disease.
- **Exercise tests.** These help identify the type of metabolic problem.
- **Heart tests.** Tests, such as echocardiography, can show if your heart is affected.
- **Urine test.** This test looks for a protein called ketones.

Treatment for secondary carnitine deficiency

The main treatment for carnitine deficiency is L-carnitine supplements. This is the form of carnitine that your body can use. It is available in liquid and pill form. L-carnitine increases the amount of carnitine in your blood and inside cells. This can help prevent many of the symptoms of the disease. You may need to take L-carnitine for only a limited time. You may need repeated blood tests to check your carnitine levels. Other dietary changes may be helpful. These include eating more carbohydrates and less fat.

It's also important to not do things that may set off symptoms. These include skipping meals, being exposed to cold, and getting a lot of exercise.

What happens if you don't get treated?

Carnitine deficiency has 2 possible complications:

- **Heart weakness.** Heart problems may be the first symptom of carnitine 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. Heart problems respond well to treatment with L-carnitine. Your healthcare provider may want to check your heart for signs of weakness over time.
- **Liver problems.** Liver problems may also be the first symptoms of a carnitine deficiency. The liver may enlarge and not function as well as it should. Liver problems can lead to episodes of low blood sugar (hypoglycemia). Infections are often the trigger for these episodes. Severe hypoglycemia can lead to coma and death if not treated right away with a sugar called dextrose. Liver problems may not respond well to treatment with L-carnitine. If you know you are prone to carnitine deficiency, consider asking your healthcare provider about keeping fasting periods before testing as short as possible (or if you might even have an IV infusion of dextrose during that time.)

When to call your healthcare provider

Call your healthcare provider right away if any of the following occur:

- Shortness of breath
- Swelling
- Symptoms of low blood sugar
- Fatigue
- Decrease in muscle tone

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