Treatment for Primary Carnitine Deficiency (Child)

Carnitine is a nutrient that helps the body's cells work normally. Primary carnitine deficiency is when not enough carnitine can get into the energy factories (mitochondria) of the cells in the body. This can cause muscle weakness. It can also cause heart or liver problems. Primary carnitine deficiency is a rare health problem a child is born with. It's caused by an abnormal gene.

Types of treatment

The main treatment for carnitine deficiency is L-carnitine supplements. This is the form of carnitine that the body can use. It's available in liquid and pill form. L-carnitine increases the amount of carnitine in the blood and inside cells. This can help prevent many of the symptoms of the disease. Your child will likely need to take L-carnitine for the rest of their life.

It's also important for your child to avoid things that may trigger symptoms. These include skipping meals, being exposed to cold, and getting a lot of exercise.

What happens if your child is not treated?

Carnitine deficiency has two 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. Untreated heart weakness may lead to death early in life. But heart problems respond well to treatment with L-carnitine. Your child's healthcare provider may want to check your child's heart for signs of weakness over time.
- **Liver problems.** Liver problems may also be the first symptoms of a carnitine deficiency. They often affect children in the first few years of life. The liver may get larger and not function as well as it should. This may cause problems such as poor feeding and irritability. 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.

Preventing primary carnitine 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 (IVF), the embryos can be tested for the disease. Amniocentesis can also be used to test for the condition in early pregnancy. Carnitine deficiency is also included in some newborn screening panels.

When to call your child's healthcare provider

Call your child's healthcare provider right way if your child has any of these:

- Shortness of breath
- Irregular heart rate
- Swelling
- Symptoms of low blood sugar
- Extreme tiredness (fatigue)

- Decrease in muscle tone