Understanding 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 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 is caused by an abnormal gene.

How does carnitine work in the body?

You get carnitine through some of the foods you eat. It helps get fatty acids into the energy factories of cells (mitochondria) 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 primary carnitine deficiency?

It's caused by an abnormal gene. The gene causes a problem with a protein that carries carnitine into cells from the blood. In some cases, this only leads to low carnitine levels in muscle. It is also called carnitine uptake defect.

What are the symptoms of primary carnitine deficiency?

Children tend to show symptoms in the first few years of life. Symptoms can occur a bit differently in each person. Your child may have no symptoms, or they may be mild to severe. Symptoms can include:

- Decreased or floppy muscle tone
- Enlarged liver
- Muscle weakness
- Fatigue
- Irritability or confusion
- Delayed movement (motor development)
- Poor feeding
- Episodes of very low blood glucose triggered by routine viral illnesses
- Swelling (edema)
- Shortness of breath
- Weak or enlarged heart

How is primary carnitine deficiency diagnosed?

Your baby may be diagnosed through standard newborn screening tests.

The process to diagnose carnitine deficiency starts with a health history and a physical exam. The healthcare provider will ask about your child's symptoms. They may also ask about your family's health 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.
- **Urine test.** This test looks for a protein called ketones.
- **Genetic test.** This kind of test can confirm primary carnitine deficiency.
- **Heart tests.** Tests such as echocardiography can show if the heart is affected.

How is primary carnitine deficiency treated?

If your baby or child has been diagnosed with primary carnitine deficiency, the healthcare provider will prescribe carnitine to be taken by mouth each day. Your child should not have any long periods of fasting or skipping meals. And they should be watched closely if they become ill for any reason. In some cases, IV (intravenous) sugar may be needed. Your child will need yearly check-ins with a cardiologist to screen for the development of cardiomyopathy (weakness of the heart muscles).