

## **Carnitine Deficiency**

**Last updated June 2010**

### **Definition**

One of a group of muscle diseases that interfere with the processing of fats for energy production

### **Cause**

Either a defect in a gene for a protein, such as the carnitine transporter OCTN2, that brings carnitine into the cell ("primary" carnitine deficiency); or a deficiency of carnitine secondary to other metabolic diseases ("secondary" carnitine deficiency)

### **Onset**

Infancy to early adulthood

### **Symptoms**

If confined to muscles, weakness in the hips, shoulders, and upper arms and legs; face and tongue muscles may also be weak; heart muscle weakness may occur; in more severe cases, in which other tissues are affected, also includes low blood sugar, fatigue, vomiting, abdominal pain, growth retardation, low weight, enlarged liver, episodes of brain function abnormalities

### **Progression**

Usually slowly progressive

### **Inheritance**

Primary carnitine deficiency usually autosomal recessive, meaning caused by the contribution of a defective gene from each parent; secondary carnitine deficiency inheritance depends on primary cause