

Carnitine Uptake Defect (CUD) – Fatty Acid Oxidation Defect (FAOD)

What are Fatty Acid Oxidation Defects?

FAODs occur when fats (fatty acids) cannot be broken down in the body. Fats are an important source of energy for the body, especially during periods of fasting. As the carbon chains in fatty acids are broken down, energy is released and the products of this process are used to make ketone bodies, another source of energy.

What is Carnitine Uptake Defect?

Carnitine is used to transport long-chain fats into the mitochondria. It is produced in the liver and can also be found in the diet, mainly in meat. In Carnitine Uptake Defect, a deficiency of carnitine in the tissues results in a block in the process of fatty acid oxidation and an impaired ability to use fats to produce energy and ketone bodies.

What is its incidence?

CUD is a rare condition. Its incidence is unknown.

What are the clinical features of the disease?

Children with CUD may appear normal at birth. Typically, however, they may present with cardiomyopathy, muscle weakness, and hypotonia. Also, during a period of fasting (such as during a common illness), a child who was previously healthy may present with lethargy, hypoketotic hypoglycemia and an enlarged liver. They are at risk for encephalopathy, coma, and sudden death.

How is the diagnosis confirmed?

The diagnosis of CUD is confirmed by measuring total carnitine levels in blood; affected individual have greatly reduced levels. Conversely, urine analysis shows increased amounts of carnitine. Enzyme analysis using a skin biopsy may also help

to confirm the diagnosis. Diagnostic testing is arranged by specialists at your regional treatment centre.

What is the treatment of the disease?

Lifelong dietary supplementation with carnitine is very effective in preventing metabolic crises and their sequelae. If cardiomyopathy has developed in the untreated child, this can regress with carnitine supplementation. Frequent feedings also ensure that a child with a carnitine uptake defect does not undergo any prolonged period of fasting. Treatment is coordinated by specialists at your regional treatment centre.

What is the outcome of treatment?

If treatment is able to prevent metabolic crises, children with CUD have a good prognosis. However, repeated metabolic episodes can cause neurological damage over time.

Can a family have more than one child with Carnitine Uptake Defect?

CUD is an autosomal recessive disease. Parents of a child with CUD are assumed to be carriers for the disease and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with this condition. Prenatal testing for CUD can be done as early as 10-12 weeks of pregnancy. Genetic counselling to discuss the benefits of prenatal testing options in more detail is recommended.

Unaffected siblings of a child with CUD have a 2/3 chance of being carriers. Carriers for Carnitine Uptake Defect are healthy and do not have symptoms of the disease.

Resources

<http://www.fodsupport.org/>