

Carnitine Uptake Defect (CUD)

(metabolic condition: fatty acid oxidation disorder) *Information for Health Professionals*

Also known as:

- carnitine transport defect
- renal carnitine transport defect
- systemic carnitine deficiency

What are fatty acid oxidation disorders?

Fatty acid oxidation disorders (FAOD) are a group of inherited conditions in which fatty acids cannot be broken down to provide the body with energy. Fatty acids are a key source of energy during fasting or stress, such as during an illness. An inability to metabolize fatty acids leads to an accumulation of toxic metabolites in the body along with cellular energy deficiency which in combination can cause serious health problems.

What is CUD?

Carnitine is required to transport fatty acids into the mitochondria of the cell where breakdown occurs to produce energy. In CUD, carnitine that is present in the diet or synthesized by the liver cannot be transported into cells. This results in a deficiency of carnitine inside the cells leading to an inability to use fatty acids for energy production.

Maternal CUD is a condition that occurs when an infant is born to a woman with untreated CUD. In these situations, infants will have low carnitine at birth but are unaffected and do not require treatment.

What causes CUD?

CUD is caused by pathogenic variants in the carnitine transporter gene resulting in very low carnitine levels.

How common is CUD?

CUD is a rare condition with an estimated incidence of 1 in 50,000 infants born in the United States.

What are the clinical features of CUD?

Although infants with CUD may appear normal at birth, during a period of fasting (such as during a common illness), a child who was previously healthy in the first few months of life may present with lethargy, hypoketotic hypoglycemia and hypotonia. They may also present with cardiomyopathy, muscle weakness and an enlarged liver. They are at risk for encephalopathy, coma and sudden death.

What is the screening test for CUD?

A decreased concentration of carnitine is detected on the newborn blood spot screen. Newborn blood spot screening will not detect all infants with CUD. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of CUD is confirmed by the finding of very low total and free carnitine in a blood sample together with high levels of carnitine excreted in urine. Further testing may include enzyme analysis in a skin biopsy and/or molecular genetic analysis of the carnitine transporter gene. Blood and urine carnitine levels in mothers of infants with low carnitine can distinguish between maternal CUD and CUD in the infant. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is CUD treated?

CUD is treated by lifelong supplementation with carnitine and avoidance of fasting in order to avoid metabolic crisis. Prompt treatment of a metabolic crisis with intravenous fluids and glucose is necessary. Recurrent metabolic crisis may result in neurological damage or death.

Is CUD inherited?

CUD is inherited as an autosomal recessive disorder. Parents of a child with CUD are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. CUD carriers are healthy. Genetic counselling is available to families with CUD.

Additional resources are available through:

Clinical & Metabolic Genetics Program (Edmonton)

8-53 Medical Sciences Building
8440 – 112 St. NW
Edmonton, AB T6G 2H7
Phone: 780-407-7333
Fax: 780-407-6845

Emergency consultations:

Phone 780-407-8822 and ask for the specialist on call for metabolic diseases.

Clinical & Metabolic Genetics Program (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7587
Fax: 403-955-3091

Emergency consultations:

Phone 403-955-7211 and ask for the specialist on call for metabolic diseases.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening

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