

Carnitine Uptake Deficiency (CUD) General Overview

Q. What is CUD?

A. CUD is a treatable disorder that affects the way the body transports fats. If left untreated, CUD can cause life-threatening illness.

Q. What happens to fats in a child with CUD?

A. In a child with CUD, fats cannot be transported to the site in the body's cells where they can be used to produce energy. The enzyme responsible for CUD does not transport carnitine properly. Carnitine is needed to break down fats, and if the enzyme does not work well, fats build up in the body.

Q. What are the effects of having CUD if it is not treated?

A. Untreated CUD can lead to extremely low blood sugar levels, breathing problems, coma and possible death. The long-term effects of untreated CUD may include heart or liver problems, muscle weakness, brain damage and developmental delay.

Q. What is the treatment for CUD?

A. Treatment for CUD involves avoiding fasting and having meals that are high in carbohydrates and low in fats (a special formula may be required). Infants should have at least one nighttime feeding, or a late-night snack, to reduce the time they go without eating. Treatment should begin shortly after birth. Some doctors may prescribe dietary supplements, such as L-carnitine. Emergency care must be taken if a person with CUD becomes ill and has difficulty keeping food down. This is usually treated in the hospital with an IV. People with CUD require specialized treatment through a clinic with experience in treating this disorder.

Q. Why would a child have CUD?

A. CUD is an inherited disorder. It results when a baby receives a double-dose of a non-working CUD gene (one from each parent). For more information about this, contact your health care provider or a genetic counselor.

Q. What are the chances that a child will be born with CUD?

A. The prevalence of CUD is approximately 1:175,000. However, parents who have a child with CUD each carry one copy of the non-working gene. That means, with each pregnancy, there is a one in four chance of the child having CUD.

For more information about CUD, please see the Disorders section or our website: www.doh.wa.gov/nbs.



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