

Carnitine Uptake Deficiency (CUD) (Car-ni-teen Up-tak De-fin-shen-see)

What is carnitine uptake deficiency?

Carnitine Uptake Deficiency (CUD) is a metabolic disorder. This means the body has a chemical imbalance. CUD is a condition that prevents the body from using fats for energy. People with this condition are missing an enzyme or the enzyme does not work right. CUD can look like another metabolic condition called Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD). CUD is inherited. This means it is present at birth.

What type of problems occurs with carnitine uptake deficiency?

A baby who has this condition may develop low blood sugar. The baby may develop a lack of energy (lethargy) and poor muscle tone. If left untreated, this condition can lead to heart failure, liver problems, coma, and death. Early diagnosis and treatment will help prevent these problems.

What is the chance my baby will have carnitine uptake deficiency?

This condition occurs in less than 1 in every 100,000 births. Babies born with this condition have a changed gene from each parent. A person who has one changed gene is called a carrier. A person who is a carrier does not have symptoms. If both parents are carriers, either parent can pass on the changed gene to their baby. If both parents pass on the changed gene, the baby will have the condition. If both parents are carriers, for each pregnancy:

- There is a 25% chance that the baby will be born with this condition. •
- There is a 50% chance that the baby will be a carrier for this condition.
- There is a 25% chance that the baby will not be born with this condition and will not be a carrier.

What is the treatment of carnitine uptake deficiency?

The treatment of this condition is to avoid long periods without food or drink. This means that your child should eat or drink nutritional supplements at least every 3 to 5 hours. A diet that has a lot of carbohydrates is recommended. Your baby's metabolic doctor will help you make sure that your baby gets the right diet and medical care.

Where in Virginia can my baby be taken care?

Please speak to your baby's pediatrician about obtaining a referral to a pediatric metabolic specialist in your area. If you want to know more about this condition, please contact Virginia Newborn Screening Services, Virginia Department of Health. The Web site is <http://www.vahealth.org/gns>.



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