



Newborn Screening ACT Sheet

Decreased C0 and Other Acylcarnitines Carnitine Uptake Defect (CUD)

Differential Diagnosis: Carnitine uptake defect (CUD).

Condition Description: CUD is caused by a defect in the carnitine transporter that moves carnitine across the plasma membrane. Reduced carnitine limits acylcarnitine formation preventing transport of fatty acids into mitochondria, thereby limiting energy production. Tissues with high energy needs (skeletal and heart muscle) are particularly affected.

Medical Emergency: Take the Following IMMEDIATE Actions

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, lethargy, tachypnea).
- Consult with pediatric metabolic specialist. (See attached list.)
- Evaluate the newborn (tachycardia, hepatomegaly, reduced muscle tone).
- Initiate emergency treatment as indicated by metabolic specialist.
- Initiate timely confirmatory/diagnostic testing as recommended by specialist.
- Initial testing: (free and total) plasma carnitine and urine carnitine.
- Repeat newborn screen if the second screen has not been done.
- Educate family about signs, symptoms, and need for urgent treatment if infant becomes ill.
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma and urine carnitine analysis will reveal decreased free and total carnitine (C0) in plasma and overexcretion of carnitine in urine. The newborn's mother should be investigated, as well, because several cases of maternal CUD have been identified following an abnormal newborn screening result in their offspring. Transporter assays and OCTN2 gene sequencing establish the diagnosis.

Clinical Considerations: Carnitine transporter defect has a variable expression and variable age of onset. Characteristic manifestations include lethargy, hypotonia, hepatomegaly, and cardiac decompensation due to cardiomyopathy. Hypoglycemia is typical in acute episodes.

Additional Information:

OMIM

<http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=212140>

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=primarycarnitinedeficiency>

STAR-G/HRSA

<http://www.newbornscreening.info/Parents/fattyacid disorders/Carnitine.html> <http://www.newbornscreening.info/Pro/fattyacid disorders/Ctd.html>



Newborn Screening FACT Sheet

Carnitine Uptake Deficiency (CUD) or Carnitine Transport Deficiency (CTD)

What is CUD?

CUD is a kind of fatty acid oxidation disorder. People with CUD have problems breaking down fat into energy for the body.

What Causes CUD?

Enzymes help start chemical reactions in the body. CUD happens when an enzyme called “carnitine transporter” is either missing or not working right. This enzyme helps carry something called carnitine into our cells. Carnitine helps the body make energy from fats in food. It also helps us to use the fat that is already stored in the body.

What Symptoms or Problems Occur with CUD?

[Symptoms are something out of the ordinary that a parent notices.]

Babies with CUD first show signs of it between birth and age three. CUD can cause periods of illness called Metabolic Crises. Some of the first symptoms of a Metabolic Crisis are:

- too much sleepiness
- behavior changes (such as crying for no reason)
- irritable mood
- poor appetite

If a Metabolic Crisis is not treated, a child with CUD can develop:

- breathing problems
- swelling of the brain
- seizures
- coma, sometimes leading to death

Repeated Metabolic Crises can cause brain damage. This can cause learning problems or mental retardation. CUD can also cause an enlarged heart and weak muscles.

What is the Treatment for CUD?

The following treatments are often used for children with CUD:

1. L-carnitine – The doctor usually prescribes the life-long use of L-carnitine. This is safe and natural and helps the body make energy. It

also helps the body get rid of harmful wastes. L-carnitine can get rid of heart problems and muscle weakness in children with CUD.

2. Do not go a long time without food – Babies and young children with CUD should eat more often to avoid problems. They should not go without food for more than 4 to 6 hours. Some babies may need to eat even more often than this. It is important that babies be fed during the night. They need to be woken to eat if they do not wake up on their own.

3. Diet – Sometimes a low-fat, high carbohydrate diet (such as fruits, vegetables, bread, noodles) is used in addition to L-carnitine. Any changes in the diet should be made by a dietitian. Dietitians know what are the right foods to eat. Ask your doctor whether your child needs to have any changes in his or her diet.

Things to Remember

Always call your doctor when your baby has any of the following:

- poor appetite
- low energy or too much sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- continued muscle pain or weakness

Babies with CUD need to eat extra starchy food (such as rice, cereal, bread) and drink more fluids when they're sick - even if they don't feel hungry – or they could have a Metabolic Crisis. If they won't or can't eat, they may need to be treated in the hospital to prevent serious health problems.