

## Newborn Screening ACT Sheet

### [Decreased C0]

## Carnitine Uptake Defect (CUD; Primary Carnitine Deficiency)

**Differential Diagnosis:** Carnitine uptake defect (CUD); Maternal carnitine deficiency (primary or secondary); prematurity.

**Condition Description:** Carnitine Uptake Defect (CUD), a fatty acid oxidation (FAO) disorder, is caused by a defect in the carnitine transporter in the cell membrane. This leads to decreased free carnitine in cells and increased excretion of carnitine in urine. The resulting carnitine deficiency disrupts the transport of long-chain fatty acids into mitochondria, leading to decreased energy production, particularly in tissues with high energy needs (skeletal and heart muscle). FAO occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) after glycogen stores become depleted and energy production relies increasingly on fat metabolism. The presentation and age of onset of symptoms are variable.

#### **You Should Take the Following Actions:**

- Inform family of the newborn screening result.
- Ascertain clinical status (poor feeding, lethargy, tachypnea).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (tachycardia, hepatomegaly, hypotonia). If any of these signs are present or if the newborn is ill, transport to a hospital for further treatment in consultation with the metabolic specialist.
- Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
- Provide family with basic information about CUD and its management.
- Report final diagnostic outcome to newborn screening program.

**Diagnostic Evaluation:** **Plasma carnitines (free and total):** are decreased. **Urinary carnitine** excretion may be increased. **Molecular genetic testing** may confirm the diagnosis.

**Clinical Considerations:** Carnitine uptake defect has a variable presentation and age of onset. Characteristic manifestations include lethargy, hypotonia, hepatomegaly, and cardiac decompensation due to cardiomyopathy. Hypoketotic hypoglycemia is typical in acute episodes. These findings are rarely present in the neonatal period. Maternal carnitine deficiency (primary or secondary), other fatty acid oxidation defects, organic acidurias, and prematurity can cause low carnitine levels in a newborn.

#### **Local Referral Site:**

UAB Department of Genetics  
VHL108B 1670 University Blvd  
Birmingham, AL 35233  
Phone: 205-996-6983  
Fax: 205-975-6389

Alabama Newborn Screening Program  
1-866-928-6755

Dr. Dean recommends the following for abnormal carnitine values: **Alert level = carnitine less than 5.54**

If equal to or more than 34 weeks gestation:

1. **Send an acylcarnitine and plasma carnitine level (total/free carnitine).**
2. A carnitine level should be obtained on an infant's mother for an infant with an initial low level whose total and free carnitine normalizes as this may suggest a maternal carnitine deficiency.

If less than 34 weeks gestation or sick infant in the NICU no matter gestational age:

1. Carnitine supplementation as directed.
2. **Send an acylcarnitine and plasma carnitine level (total/free carnitine) after carnitine** supplementation is discontinued.
3. May need further evaluation if abnormal levels persist.

This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures.