

<b>Disease Name</b>	<b>Carnitine uptake deficiency</b>
<b>Alternate name(s)</b>	Systemic carnitine deficiency, Carnitine deficiency, Carnitine transporter deficiency
<b>Acronym</b>	SCD, CUD
<b>Disease Classification</b>	Fatty Acid Oxidation Disorder
<b>Variants</b>	N/A
<b>Variant name</b>	N/A
<b>Symptom onset</b>	Infancy or childhood with fasting hypoglycemia, weakness and/or cardiomyopathy.
<b>Symptoms</b>	Hypoketotic hypoglycemia, seizures, vomiting, lethargy progressing to coma. Chronic muscle weakness, cardiomyopathy, hepatomegaly.
<b>Natural history without treatment</b>	Non-progressive developmental delay due to hypoglycemia, cardiomyopathy and muscle weakness.
<b>Natural history with treatment</b>	Developmental delay, if present, is not reversed by treatment. Cardiomyopathy and muscle weakness can be reversed by treatment.
<b>Treatment</b>	Carnitine supplementation, no fasting.
<b>Physical phenotype</b>	Cardiomyopathy, muscle weakness.
<b>Inheritance</b>	Autosomal recessive
<b>General population incidence</b>	1/40,000
<b>Ethnic differences</b>	No
<b>Population</b>	N/A
<b>Ethnic incidence</b>	N/A
<b>Enzyme location</b>	Muscle, heart, kidney, leukocytes and fibroblasts
<b>Enzyme Function</b>	Transports carnitine into cells
<b>Missing Enzyme</b>	Carnitine transporter
<b>Metabolite changes</b>	Decreased free carnitine in plasma, increased carnitine in urine, decreased carnitine in muscle.
<b>Prenatal testing</b>	Protein analysis in cultured amniocytes, biochemical analyte testing. If a mutation in a proband is detected, DNA prenatal diagnosis via CVS or amniocytes is possible.
<b>MS/MS Profile</b>	Reduced concentrations of free carnitine and various acylcarnitine species.
<b>OMIM Link</b>	<a href="http://www.ncbi.nlm.nih.gov/omim/212140">http://www.ncbi.nlm.nih.gov/omim/212140</a>
<b>Genetests Link</b>	<a href="http://www.genetests.org">www.genetests.org</a>
<b>Support Group</b>	FOD Family Support Group <a href="http://www.fodsupport.org">http://www.fodsupport.org</a> Save Babies through Screening Foundation <a href="http://www.savebabies.org">http://www.savebabies.org</a> Genetic Alliance <a href="http://www.geneticalliance.org">http://www.geneticalliance.org</a>

## Newborn Screening ACT Sheet [Decreased C0 and other Acylcarnitines] Carnitine uptake Defect (CUD)

**Differential Diagnosis:** Carnitine uptake defect (CUD), maternal carnitine deficiency and prematurity.

**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 long-chain fatty acids into mitochondria, thereby limiting energy production. Tissues with high energy needs (skeletal and heart muscle) are particularly affected.

### **YOU SHOULD TAKE THE FOLLOWING ACTIONS:**

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, lethargy, tachypnea).
- Consult with pediatric metabolic specialist.
- 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.
- Educate family about signs, symptoms, and need for urgent treatment if infant becomes ill.
- Report findings to newborn screening program.

**Diagnostic Evaluation:** Plasma carnitine analysis will reveal decreased free and total carnitine (C0) in plasma in an affected infant. If the total and free carnitine are normal in the infant, it may suggest a maternal carnitine deficiency and plasma carnitine analysis in the mother is indicated. Transporter assays in fibroblasts and *SLC22A5* (OCTN2 carnitine transporter) gene sequencing establish the diagnosis. Prematurity should be considered in the differential 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. These are rarely present in the neonatal period.

### **Additional Information:**

[OMIM](#)  
[Genetics Home Reference](#)

### **Referral (local, state, regional and national):**

[Testing](#)  
[Clinical Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It 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. Adherence to this guideline does not necessarily ensure a successful medical outcome. 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 guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.

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