



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.
- 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 and urine carnitine analysis will reveal decreased free and total carnitine (C0) in plasma and overexcretion of carnitine in urine. The newborns 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:

(Click on the name to take you to the website. Complete URLs are listed in the Appendix)

[OMIM](#)

[Genetics Home Reference](#)

[STAR-G/HRSA](#)

Referral (local, state, regional and national):

[Testing](#)

[Clinical](#)

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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 healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. It may be prudent, however, to document in the patient's record the rationale for any significant deviation from these standards and guidelines.

APPENDIX: Resources with Full URL Addresses

Additional Information:

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

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

STAR-G/HRSA <http://www.newbornscreening.info/PRO/fattyaciddisorders/carnitine.html>

Referral (local, state, regional and national):

Testing <http://biochemgen.ucsd.edu/UCSDW3BG/Labchoose.asp>

Clinical

<http://www.genetests.org/servlet/access?id=8888891&key=RRqZcXXEUiAx9&fcn=y&fw=zLsf&filename=/clinicsearch/searchclinic.html>

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