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

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APPENDIX: Resources with Full URL Addresses

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/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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