TEXAS Health and Human Services | Texas Department of State Health Services

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: blood sugar, plasma (free and total) carnitine levels, and maternal plasma (free and total) carnitine levels;***
- **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; and**
- **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

Online Mendelian Inheritance in Man (OMIM) – Entry #212140
U.S. National Library of Medicine, Medline Plus – Arginase deficiency
STAR G FELSI – Carnitine Transporter Deficiency
STAR G FELSI – CTD

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