KANSAS DEPARTMENT OF HEALTH AND ENVIRONMENT

NEWBORN SCREENING ACT SHEET

SCREEN FOR: DECREASED C0 AND OTHER ACLYCARNITINES

CONDITION: CARNITINE UPTAKE DEFECT (CUD)

DIFFERENTIAL DIAGNOSIS: Carnitine uptake defect (CUD).

METABOLIC 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 the mitochondria, thereby limiting energy production. Tissues with high energy needs (skeletal and heart muscle) are particularly affected.

MEDICAL EMERGENCY - ACTION TO BE TAKEN IMMEDIATELY:

- 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.

CONFIRMATION OF DIAGNOSIS: 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 EXPECTATIONS: 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.

REPORTING: Report diagnostic result to family and Kansas NBS program.

SPECIALISTS:

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