TEXAS Department of State Health Services

Newborn Screening ACT Sheet

Elevated C16 and/or C18:1 Acylcarnitine

Carnitine Palmitoyltransferase 2 (CPTII) Deficiency

Differential Diagnosis: Carnitine palmitoyl transferase type 2 (CPTII) deficiency, carnitine/acylcarnitine translocase (CACT) deficiency;

Condition Description: In both the translocase and CPTII deficiencies, the acylcarnitines cannot be transported into the mitochondria for fatty acid oxidation. Thus, the need for generation of energy from fatty acids during fasting or increased demand (fever, stress) cannot be met. In addition, the neonatal form of CPTII deficiency is associated with multiple congenital anomalies.

Conditions associated with this analyte have been identified by the Society of Inherited Metabolic Disorders (SIMD) as critical, and require immediate action.

MEDICAL EMERGENCY - Take the Following IMMEDIATE Actions:

- **Contact family to inform them of the newborn screening result and ascertain clinical status (lethargy, seizures).**
- **Evaluate infant (hepatomegaly, cardiac insufficiency; history of sudden unexpected death in a sibling; dysmorphic facies).**
- **Immediate telephone consultation with pediatric metabolic specialist to determine appropriate follow-up (See attached list).**
- **Emergency treatment if symptomatic and/or hypoglycemia is present.**
- **Initial testing: plasma carnitine, plasma acylcarnitine, and urine organic acids.**
- **Repeat newborn screen if second screen has not been done.**
- **Educate family about signs, symptoms and need for urgent treatment of hypoglycemia.**
- **Report findings to newborn screening program.**

Diagnostic Evaluation: Plasma acylcarnitine analysis reveals increased C16 and/or C18:1. Urine organic acid analysis reveals increased lactic acid and dicarboxylic acids.

Clinical Considerations: In the neonatal form of CPTII deficiency, the neonate is profoundly ill with marked hypoglycemia, metabolic acidosis, cardiac arrhythmias, and facial dysmorphism. Only rarely will these infants survive. In the later-onset muscular form of CPTII deficiency, the neonate is asymptomatic but muscle disease develops in the adolescent or adult years. Translocase deficiency presents similarly to the neonatal form of CPTII deficiency.

Additional Information:

American College of Medical Genetics and Genomics
https://www.acmg.net/StaticContent/ACT/C16_and-or_C18-1.pdf

Genetics Home Reference
http://ghr.nlm.nih.gov/condition/carnitine-palmitoyltransferase-ii-deficiency

STAR G FELSI
http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT2%20update%202011.pdf