TEXAS Department of State Health Services

Newborn Screening ACT Sheet

Elevated C0/C16+C18

Carnitine Palmitoyl Transferase 1 Deficiency (CPT1)

Differential Diagnosis: Carnitine palmitoyl transferase 1 deficiency (CPT1).

Condition Description: This disorder is caused by a deficiency of the enzyme CPT1, preventing the fatty acid carnitine-acylcarnitine linkage required to transport fatty acids into the mitochondria. This results in accumulation of free carnitine (C0) and prevents the fatty acid oxidation response necessary to generate energy during fasting and increased energy needs (fever, stress).

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).**
- **Immediate telephone consultation with metabolic specialist (See attached list).**
- **Evaluate the newborn (lethargy, hepatomegaly, seizures).**
- **Initiate emergency treatment as indicated by metabolic specialist.**
- **Initiate timely confirmatory/diagnostic testing as recommended by specialist.**
- **Initial testing: plasma carnitine, plasma acylcarnitine analysis.**
- **Repeat newborn screen if second screen has not been done.**
- **Educate family about signs, symptoms and need for urgent treatment of hypoglycemia (lethargy, seizures).**
- **Report findings to newborn screening program.**

Diagnostic Evaluation: Plasma acylcarnitine showing elevated free carnitine C0 with low or normal long-chain acylcarnitines. CPT1 enzyme assays and CPT1A gene sequencing establish the diagnosis.

Clinical Considerations: CPT1 can have a variable presentation. Critical hypoketotic hypoglycemia is a common presenting feature. Newborns may appear asymptomatic, but can progress to fasting hypoketotic hypoglycemia, lethargy, hepatomegaly, and seizures, usually precipitated by fasting or acute illness.

Additional Information:

American College of Medical Genetics and Genomics
https://www.acmg.net/StaticContent/ACT/C0_C16-C18.pdf

Genetics Home Reference
http://ghr.nlm.nih.gov/gene=cpt1a

STAR G FELSI
http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT1.html