



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).

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (lethargy, seizures).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (lethargy, hepatomegaly, seizures); 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 of hypoglycemia (lethargy, seizures).
- Report findings to newborn screening program.

(See [flowchart](#) for information concerning the specific actions)

You Should Take the Following Actions:

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:

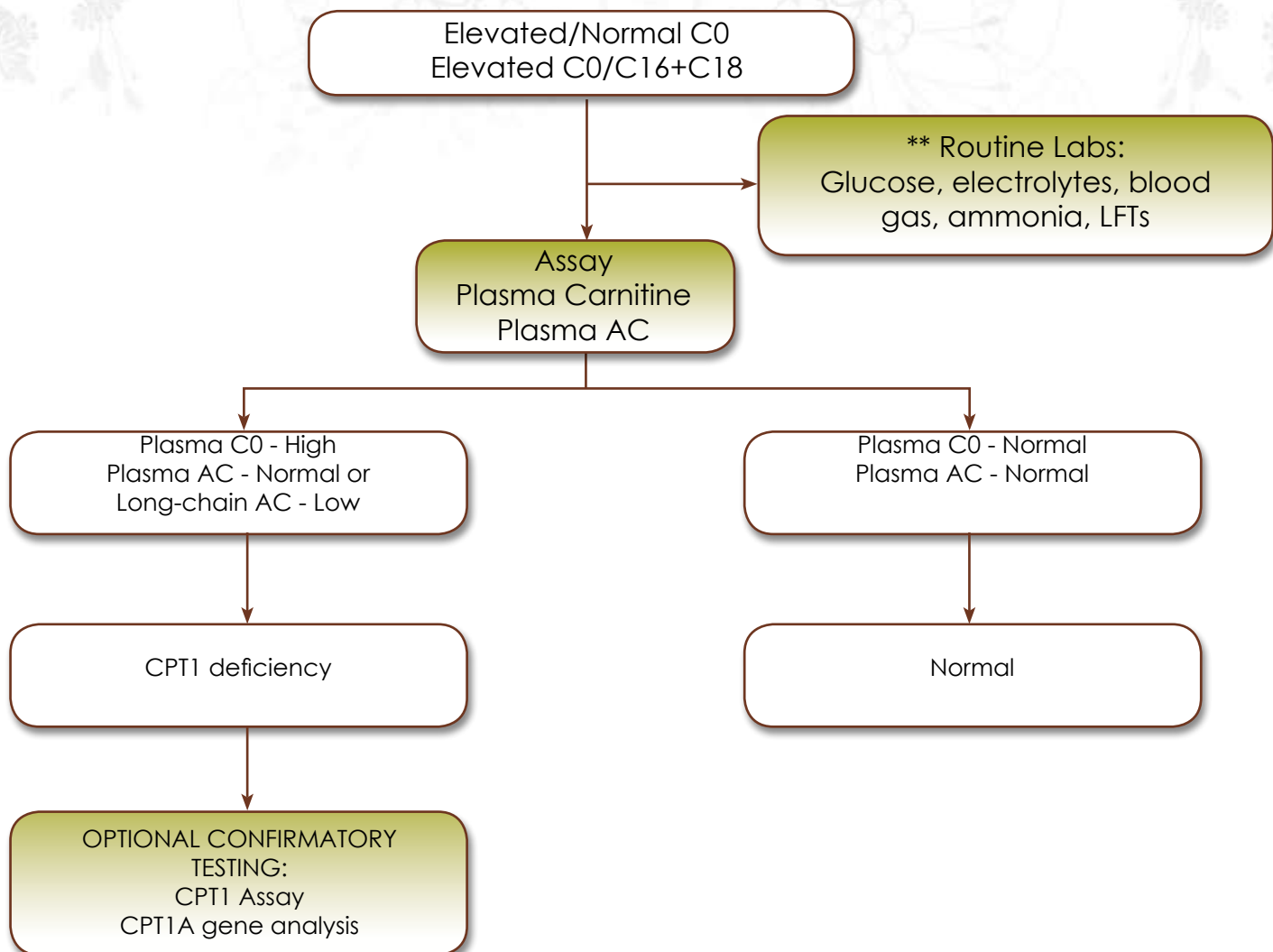
CPT1A can have a variable presentation. Critical hypoketotic hypoglycemia is a common presenting feature. Newborns may appear asymptomatic but can progress to fasting hypoketotic hypoglycemia, lethargy, hepatomegaly, and seizures, usually precipitated by fasting or acute illness.

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality medical services. In determining the propriety of any specific procedure or test, the healthcare provider should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen





C0 Elevated; C0/C16/C16+C18 Elevated



Actions are shown in shaded boxes; results are in the unshaded boxes.

Abbreviations / Key:

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AC = acylcarnitine

LFTs = liver function tests

CPT1 = carnitine palmitoyl transferase

** - When the positive predictive value of screening is sufficiently high and the risk to the infant is high, some initiate diagnostic studies that are locally available at the same time as confirmation of the screening result is done.

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