

Newborn Screening ACT Sheet [Elevated C16 and/or C18:1 Acylcarnitine] Carnitine Palmitoyltransferase 2 (CPT2) Deficiency

Differential Diagnosis: Carnitine palmitoyltransferase (CPT2) deficiency Carnitine/acylcarnitine translocase (CACT) deficiency;

Condition Description: In both the translocase and CPT2 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 CPT2 deficiency is associated with multiple congenital anomalies.

YOU SHOULD TAKE THE FOLLOWING ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Evaluate infant (hepatomegaly, cardiac insufficiency; history of sudden unexpected death in a sibling; dysmorphic facies).
- Consult/refer to a metabolic specialist to determine appropriate follow-up.
- Emergency treatment if symptomatic and/or hypoglycemia is present.
- 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 CPT2 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 CPT2 deficiency, the neonate is asymptomatic but muscle disease develops in the adolescent or adult years. Translocase deficiency presents similarly to the neonatal form of CPT2 deficiency.

Additional Information:

[Gene Tests](#)

[Genetics Home Reference](#)

[CPT2](#)

[CACT](#)

Referral (local, state, regional and national):

[Testing](#)

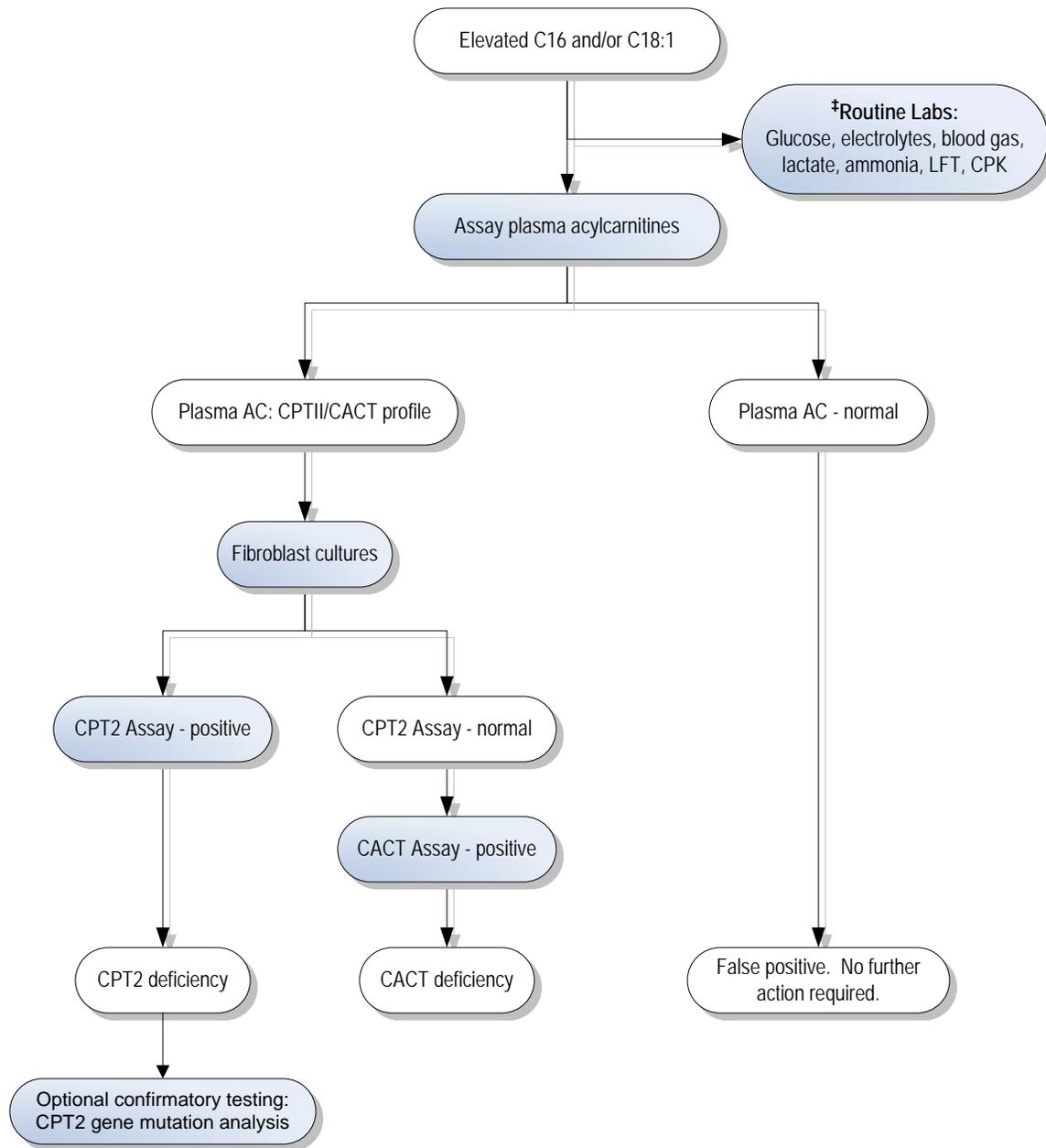
[Clinical Services](#)

[Find Genetic Services](#)

Disclaimer: This guideline is designed primarily as an educational resource for clinicians to help them provide quality medical care. It should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. Adherence to this guideline does not necessarily ensure a successful medical outcome. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this guideline. Clinicians also are advised to take notice of the date this guideline was adopted, and to consider other medical and scientific information that become available after that date.



C16 and/or C18:1 Elevated



Abbreviations/Key:

AC = acylcarnitine

CACT = carnitine - acylcarnitine translocase

CPK = creatine phosphokinase

CPT2 = carnitine palmitoyltransferase 2

LFT = liver function tests

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

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

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