C16 and/or C18:1 Elevated

Elevated C16 and/or C18:1

‡Routine Labs:
Glucose, electrolytes, blood gas, lactate, ammonia, LFT, CPK

Assay plasma acylcarnitines

Plasma AC: CPTII/CACT profile

Plasma AC - normal

Fibroblast cultures

CPT2 Assay - positive

CPT2 Assay - normal

CACT Assay - positive

CPT2 deficiency

CACT deficiency

Optional confirmatory testing:
CPT2 gene mutation analysis

False positive. No further action required.

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.

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.

© American College of Medical Genetics, 2009

(Funded in part through MCHB/HRSA/HHS grant #U22MC03957)