C16-OH Elevated +/- C18 and Other Long-Chain Acylcarnitine species

Elevated C16-OH +/- C18:1-OH and Other long-chain acylcarnitines

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

Assay:
Plasma AC
Urine OA

Plasma AC – LCHAD/TFP profile
Urine OA – N/LCHAD/TFP profile

LCHAD/TFP deficiency

Optional Confirmatory Testing:
LCHAD/TFP enzyme assays; TFP gene(s) analysis

Plasma AC – Normal
Urine OA - Normal

Fatty acid oxidation in vitro assay (fibroblasts/lymphoblasts)

FAO – LCHAD/TFP profile
FAO - Normal

LCHAD/TFP deficiency
False positive. No further action required.

Abbreviations/Key:
AC = acylcarnitine
CPK = creatine phosphokinase
FAO = fatty acid oxidation
LCHAD = Long-chain 3-hydroxyacyl-CoA dehydrogenase
LFTs = liver function tests
OA = organic acid
TFP = Trifunctional protein

‡ - When the positive predictive value of screening is sufficiently high and the risk to the baby is high, some initiate diagnostic studies that are locally available at the same 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.

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