C14:1 Elevated +/- Other Long-Chain Acylcarnitines

Elevation of C14:1 +/- other long-chain AC

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

Plasma AC, quantitative

Plasma AC - VLCAD profile

Plasma AC - Normal

VLCAD Deficiency

VLCAD gene sequencing

VLCAD Deficiency

VLCAD gene sequencing

Fatty Acid Oxidation Probe Assay, Fibroblast Culture

FAO – C12<C14<C16

FAO – C10<C12>C14

FAO - Normal

Severe VLCAD deficiency

Mild VLCAD deficiency

Normal

Abbreviations:
AC = acylcarnitine
CPK = creatine phosphokinase
FAO = Fatty acid oxidation
LFT = liver function test
VLCAD = very long-chain acyl-CoA dehydrogenase

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

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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