



Newborn Screening ACT Sheet [Elevated C14:1 +/- other long-chain acylcarnitines] Very Long-Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

Differential Diagnosis:

Very long-chain acyl-CoA dehydrogenase (VLCAD) def.

Condition Description:

VLCAD deficiency is a fatty acid oxidation (FAO) disorders. Fatty acid oxidation occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In a FAO disorder, fatty acids and potentially toxic derivatives accumulate because of a deficiency in one of the mitochondrial FAO enzymes.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Contact family to inform them of the newborn screening result and ascertain clinical status (poor feeding, vomiting, lethargy).
- Consult with pediatric metabolic specialist.
- Evaluate the newborn (poor feeding, lethargy, hypotonia, hepatomegaly, arrhythmia, evidence of cardiac decompensation). If signs are present or infant is ill, initiate emergency treatment with IV glucose and oxygen. Transport to hospital for further treatment in consultation with metabolic specialist. If infant is normal initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Educate family about need for infant to avoid fasting. Even if mildly ill, immediate treatment with IV glucose is needed.
- Report findings to newborn screening program

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:**Diagnostic Evaluation:**

Plasma acylcarnitine profile may show increased C14:1 acylcarnitine (and lesser elevations of other long chain acylcarnitines). Diagnosis is confirmed in consultation with the metabolic specialist by mutation analysis of the VLCAD gene and additional biochemical genetic tests.

Clinical Expectations:

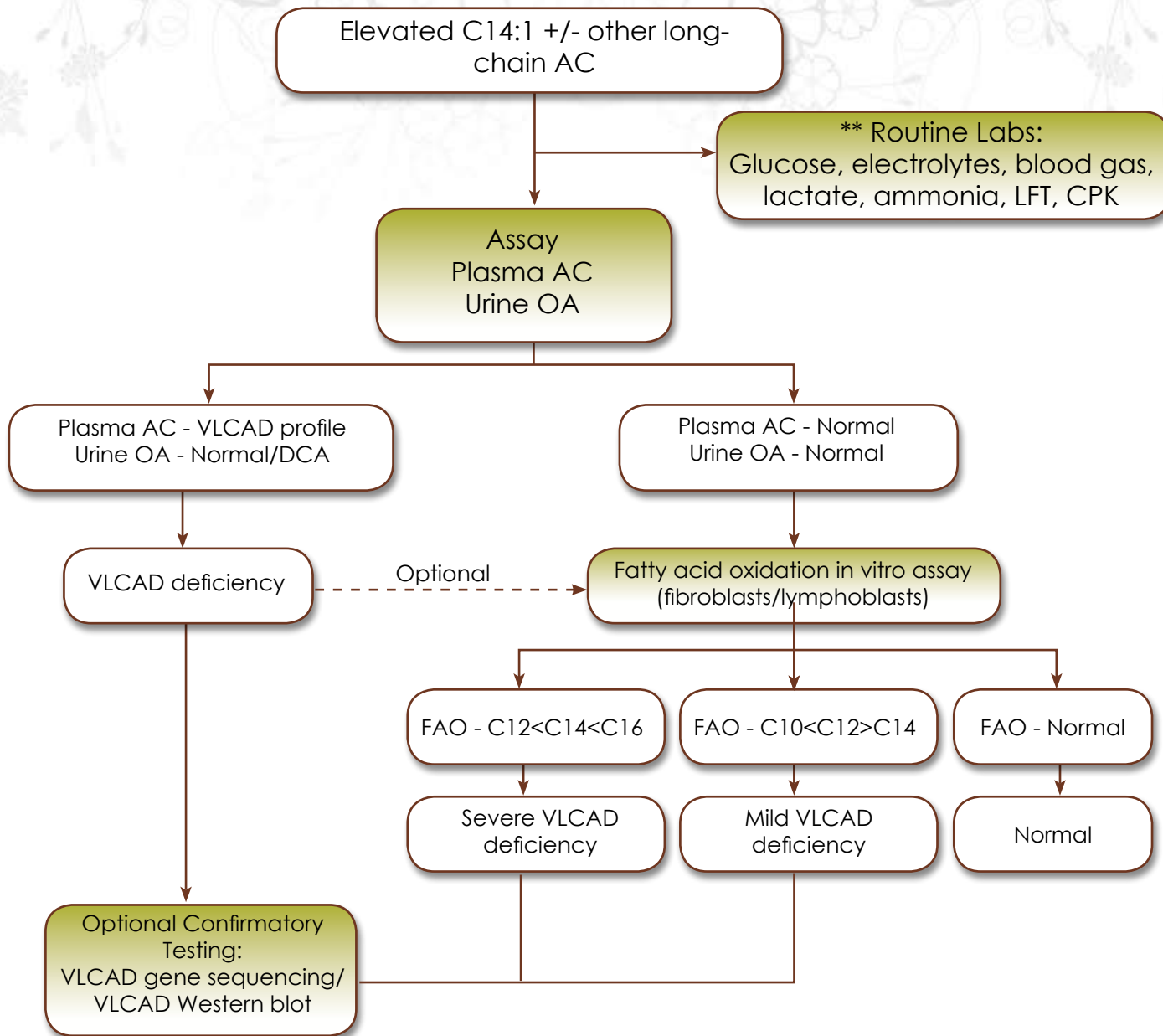
VLCAD deficiency may present acutely in the neonate and is associated with high mortality unless treated promptly; milder variants exist. Features of severe VLCAD deficiency in infancy include hepatomegaly, cardiomyopathy and arrhythmias, lethargy, hypoketotic hypoglycemia, and failure to thrive. Treatment is available.

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





C14:1 Elevated +/- Other long Chain AC



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

Abbreviations / Key:

LFT = liver function test

AC = acylcarnitine

VLCAD = very long-chain acyl-CoA dehydrogenase

FAO = Fatty acid oxidation

CPK = creatine phosphokinase

OA = organic acid

DCA = dicarboxylic acid

** = 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 time as the confirmation of the screening result is done.

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