Newborn Screening ACT Sheet
[Elevated C16-OH and Other Long Chain Acylcarnitines]
Long-chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)

Differential Diagnosis:
- Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency;
- Trifunctional protein (TFP) deficiency.

Condition Description:
LCHAD and TFP deficiencies are fatty acid oxidation (FAO) disorders. FAO occurs during prolonged fasting and/or periods of increased energy demands (fever, stress) after glycogen stores become depleted and energy production relies increasingly on fat metabolism. Fatty acids and potentially toxic derivatives accumulate in FAO disorders which are caused by deficiency in one of the enzymes involved in FAO.

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 infant (hepatomegaly, cardiac insufficiency; history of sudden unexpected death in a sibling; maternal liver disease during pregnancy; hypoglycemia). If signs are present or infant is ill, initiate emergency treatment in consultation with metabolic specialist.
- Educate family about signs and symptoms of hypoglycemia and metabolic acidosis.
- Report findings to newborn screening program.

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

Confirmation of Diagnosis:
Hypoglycemia, elevated liver transaminases, bilirubin, lactate, ammonia, and creatine phosphokinase (CPK) are suggestive of LCHAD and TFP deficiencies. Plasma acylcarnitine and urine organic acid analysis are first-line tests to determine if the appropriate LCAHD/TFP profiles are present. Differentiation between both disorders requires further biochemical and molecular genetic testing in cultured fibroblasts derived from a skin biopsy.

Clinical Considerations:

LCHAD and TFP deficiencies typically present acutely and are associated with high mortality unless treated promptly; milder variants exist. Hallmark features include hepatomegaly, cardiomyopathy, lethargy, hypoketotic hypoglycemia, elevated liver transaminases, lactic acidosis, and failure to thrive.

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

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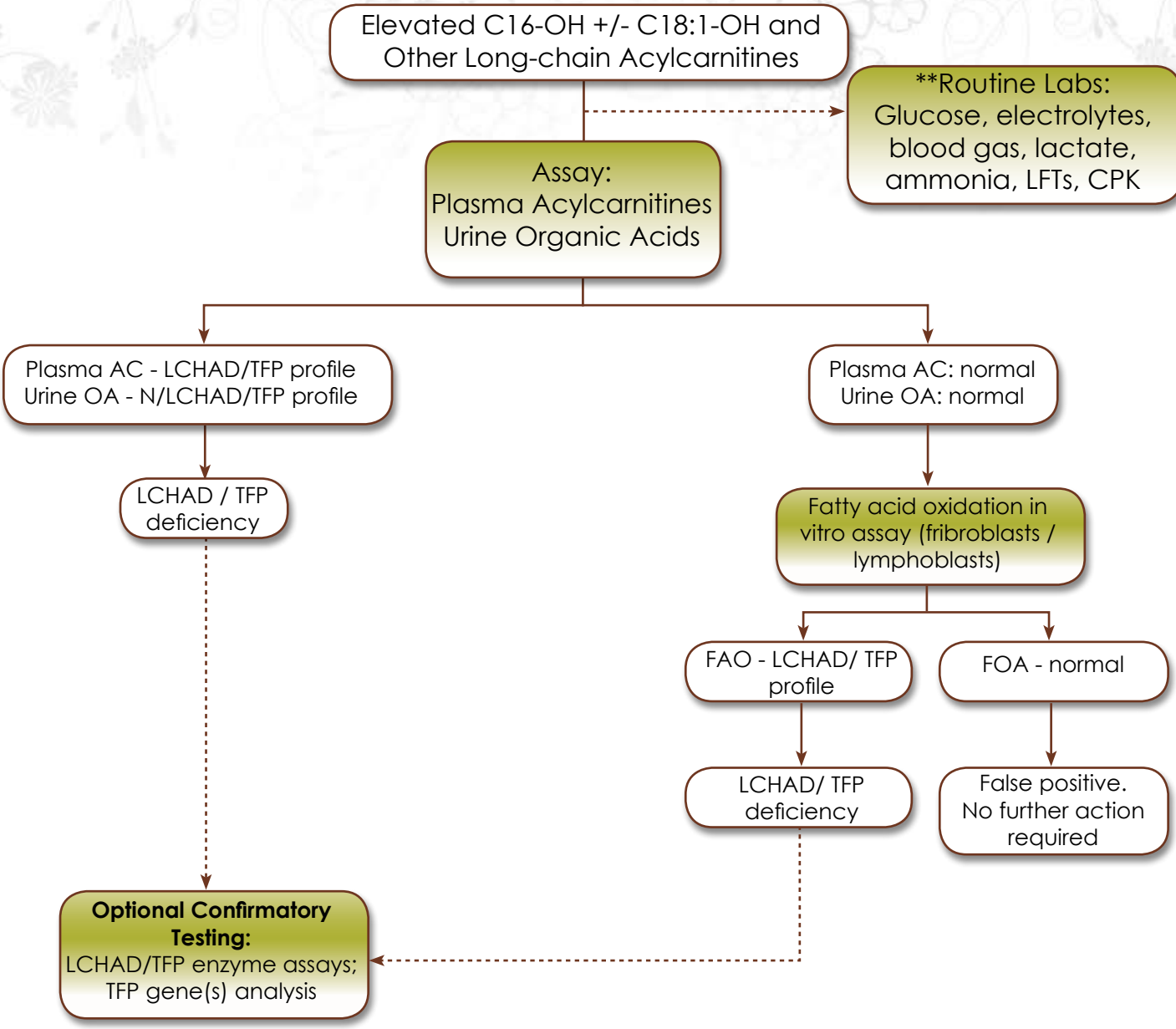
C16-OH Elecated +/- C18:1-OH and Other Long-Chain Acylcarnitines



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

Abbreviations / Key:
LCHAD = Long-chain 3-hydroxyacyl-CoA dehydrogenase
LFTs = liver function tests
AC = acylcarnitine
FAO = fatty acid oxidation
TFP = Trifunctional protein
CPK = creatine phsophokinase
OA = organic 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 as confirmation of the screening result is done.

Disclaimer: These standards and guidelines are designed primarily as an educational resource for physicians to help them provide quality clinical services. Adherence to these standards and guidelines does not necessarily ensure a successful medical outcome. These standards and guidelines 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. 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. It may be prudent, however, to document in the patient record the rationale for any significant deviation from these standards and guidelines.

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