Newborn Screening ACT Sheet
[Elevated C8 with Lesser Elevations of C6 and C10 Acylcarnitine] Medium-chain Acyl-CoA Dehydrogenase (MCAD) Deficiency

Differential Diagnosis:

Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency.

Condition Description:

MCAD deficiency is a fatty acid oxidation (FAO) disorder. FAO occurs during prolonged fasting and/ or periods of increased energy demands (fever, stress) when energy production relies increasingly on fat metabolism. In an 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). If signs are present or infant is ill, initiate emergency treatment with IV glucose. 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 analysis will show elevated octanoylcarnitine (C8). Urine acylglycine will show elevated hexanoylglycine. Diagnosis is confirmed by mutation analysis of the MCAD gene.

Clinical Considerations:

MCAD deficiency is usually asymptomatic in the newborn although it can present acutely in the neonate with hypoglycemia, metabolic acidosis, hyperammonemia, and hepatomegaly. MCAD deficiency is associated with high mortality unless treated promptly; milder variants exist. Hallmark features include vomiting, lethargy, and hypoketotic hypoglycemia. It is a significant cause of sudden death.

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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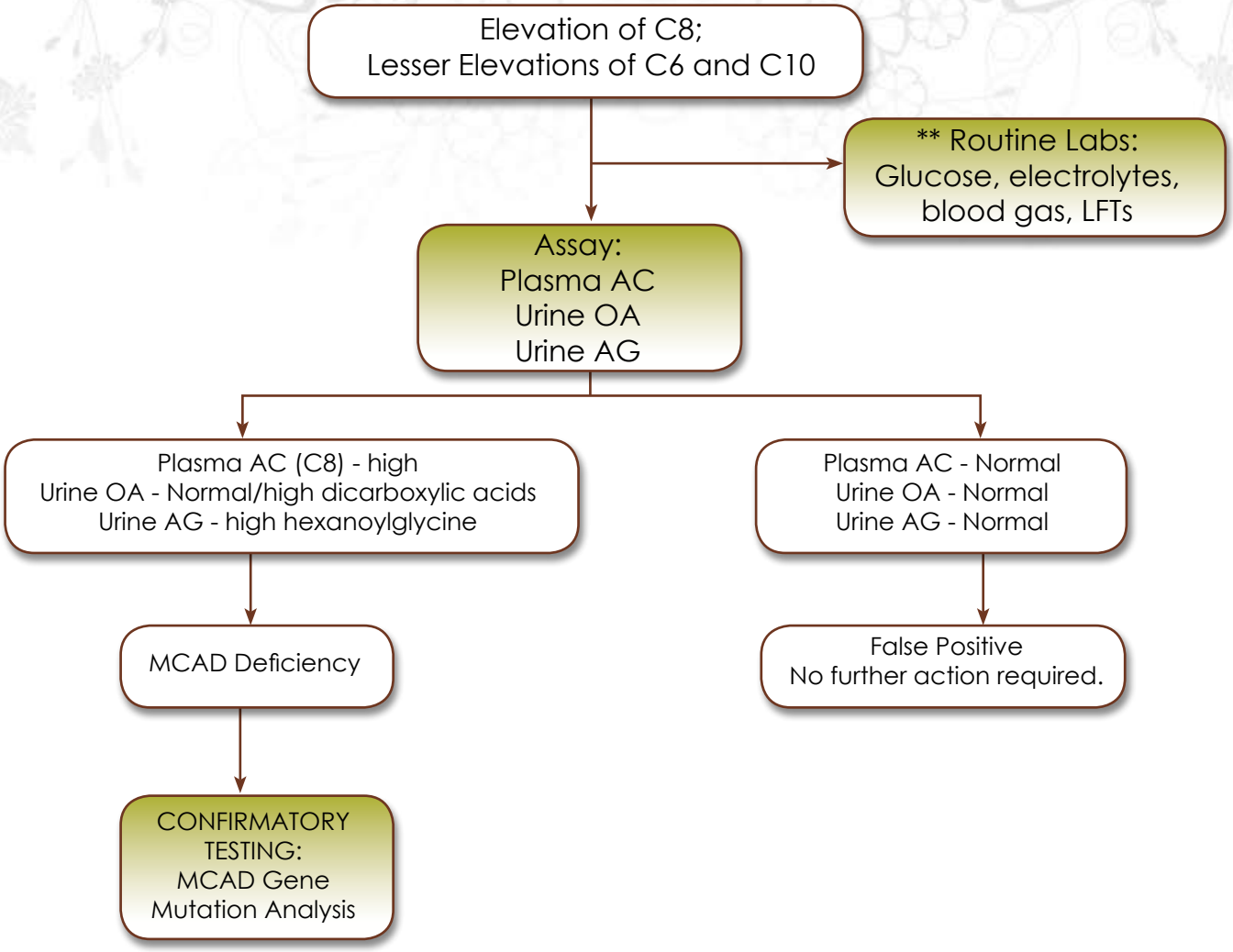
C8 Elevated & Lesser Elevations of C6 and C10



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

Abbreviations / Key:
LFTs = liver function tests
MCAD = Medium-chain acyl-CoA dehydrogenase
AC = acylcarnitine
OA = organic acid
AG = acylglycine
‡ = When the positive predictive value of screening is sufficiently high and the risk to the newborn is high, some initiate diagnostic studies that are locally available at the same time 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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