

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. Fatty acid oxidation occurs mainly 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.

YOU SHOULD TAKE THE FOLLOWING 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, transport infant to hospital for emergency treatment that would include IV glucose and any further treatment in consultation with the metabolic specialist.
- If infant is normal initiate timely confirmatory/diagnostic testing, as recommended by specialist.
- Educate family about need for infant to avoid fasting and the need for immediate medical attention if the infant even becomes mildly ill (poor feeding, vomiting, or lethargy).
- Report findings to newborn screening program.

Diagnostic Evaluation: Plasma acylcarnitine analysis will show a characteristic pattern consistent with MCADD. Urine organic acid analysis may also show an abnormal profile. Diagnosis may be 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. Untreated MCAD deficiency is a significant cause of sudden death.

Additional Information:

[Emergency Treatment Protocol \(New England Consortium of Metabolic Programs\)](#)
[Genetics Home Reference](#)

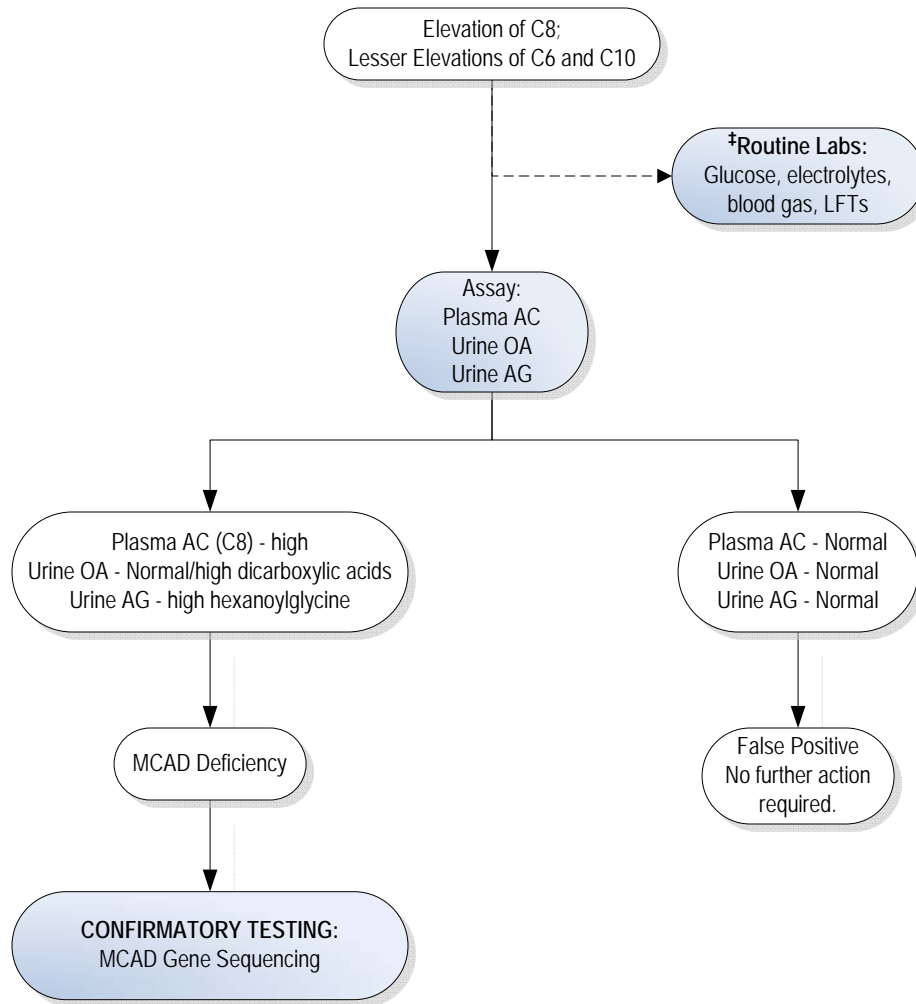
Referral (local, state, regional and national):

[Testing](#)
[Clinical Services](#)
[Find Genetic Services](#)

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.



C8 Elevated + Lesser Elevations of C6 and C10



Abbreviations/Key

AC = acylcarnitine

AG = acylglycine

LFTs = liver function tests

MCAD = Medium-chain acyl-CoA dehydrogenase

OA = organic acid

† = 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.

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

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