Elevation of C8; Lesser Elevations of C6 and C10

†Routine Labs: Glucose, electrolytes, blood gas, LFTs

Assay: Plasma AC
Urine OA
Urine AG

Plasma AC (C8) - high
Urine OA - Normal/high dicarboxylic acids
Urine AG - high hexanoylglycine

MCAD Deficiency

Plasma AC - Normal
Urine OA - Normal
Urine AG - Normal

False Positive
No further action required.

CONFIRMATORY TESTING:
MCAD Gene Sequencing

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.

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.