C4 and C5 +/- Other Acylcarnitines Elevated

Routine Labs: glucose, electrolytes, blood gas, ammonia

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

Plasma AC – GA2 Profile
Urine OA – GA2 Profile
Urine AG – GA2 Profile

Plasma AC – C4 +/- C5
Urine OA – EE/IVG
Urine AG – EE + IVG

Optional Confirmatory Testing:
ETHE1 Gene Sequencing

Optional Confirmatory Testing:
ETF/ETF-QO Gene Sequencing

Optional Confirmatory Testing:
ETF/ETF-QO Assay

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.