Isolated Elevation of C5-DC

Routine Labs:
Glucose, electrolytes,
blood gas

Assay:
Plasma AC
Urine OA
Urine AC

Plasma AC - C5-DC/normal
Urine OA - GA1 profile/normal
Urine AC - C5-DC

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

Glutaryl-CoA dehydrogenase
(GCDH) assay,
fibroblasts

GCDH activity
low

GCDH activity
normal

Glutaryl-CoA dehydrogenase
(GCDH) assay,
GCDH Gene Analysis

Glutaric Acidemia Type 1
(GA1)

Optional Confirmatory Testing:
False positive
No further action
required.

Abbreviations/Key
DC = dicarboxylic
AC = acylcarnitine
OA = organic acid
GA = glutaric acid
GCDH = glutaryl-CoA dehydrogenase

‡ = When the positive predictive value of screening is sufficiently high and the risk to the baby is high, some initiate diagnostic studies at the same time as the confirmation of screening result is done.

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

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’s record the rationale for any significant deviation from these standards and guidelines.