Newborn Screening ACT Sheet
[Elevated C5-DC Acylcarnitine]
Glutaryl-CoA Dehydrogenase Deficiency

Differential Diagnosis:
Glutaric aciduria (GA-1)

Condition Description:
GA-I is caused by a defect of glutaryl-CoA dehydrogenase which limits the metabolism of glutaryl-CoA to crotonyl-CoA, resulting in increased glutaric acid (toxic) and its metabolites.

MEDICAL EMERGENCY: TAKE THE FOLLOWING IMMEDIATE ACTIONS:

- Contact family IMMEDIATELY to inform them of the newborn screening result.
- Consult with pediatric metabolic specialist.
- Evaluate the newborn for macrocephaly and muscle hypotonia, initiate confirmatory/ diagnostic testing as recommended by metabolic specialist.
- Refer to metabolic specialist to be seen as soon as possible but not later than three weeks.
- Educate family about diagnostic possibilities, complexity of diagnostic work-up and the possibility of neurodegenerative crisis with an intercurrent infectious illness.
- IMMEDIATE treatment with IV glucose is needed for intercurrent infectious illness.
- Report findings to newborn screening program.

(See flowchart for information concerning the specific actions)

You Should Take the Following Actions:

Diagnostic Evaluation:
Urine organic acid analysis will reveal elevated glutaric acid and 3-hydroxyglutaric acid, should be ordered promptly and is often diagnostic. If urine organic acids don't confirm the diagnosis, the metabolic specialist will consider analyzing lutarylcarnitine in urine and 3-hydroxyglutaric acid in blood and CSF, enzyme assay in fibroblasts, and molecular analysis of the GCDH gene.

Clinical Considerations:
The neonate with glutaric acidemia type I is usually macrocephalic but otherwise asymptomatic. Later signs include metabolic ketoacidosis, failure to thrive, and sudden onset of dystonia and athetosis due to irreversible striatal damage. With appropriate treatment, 60-70% of patients will not suffer neurodegenerative disease.

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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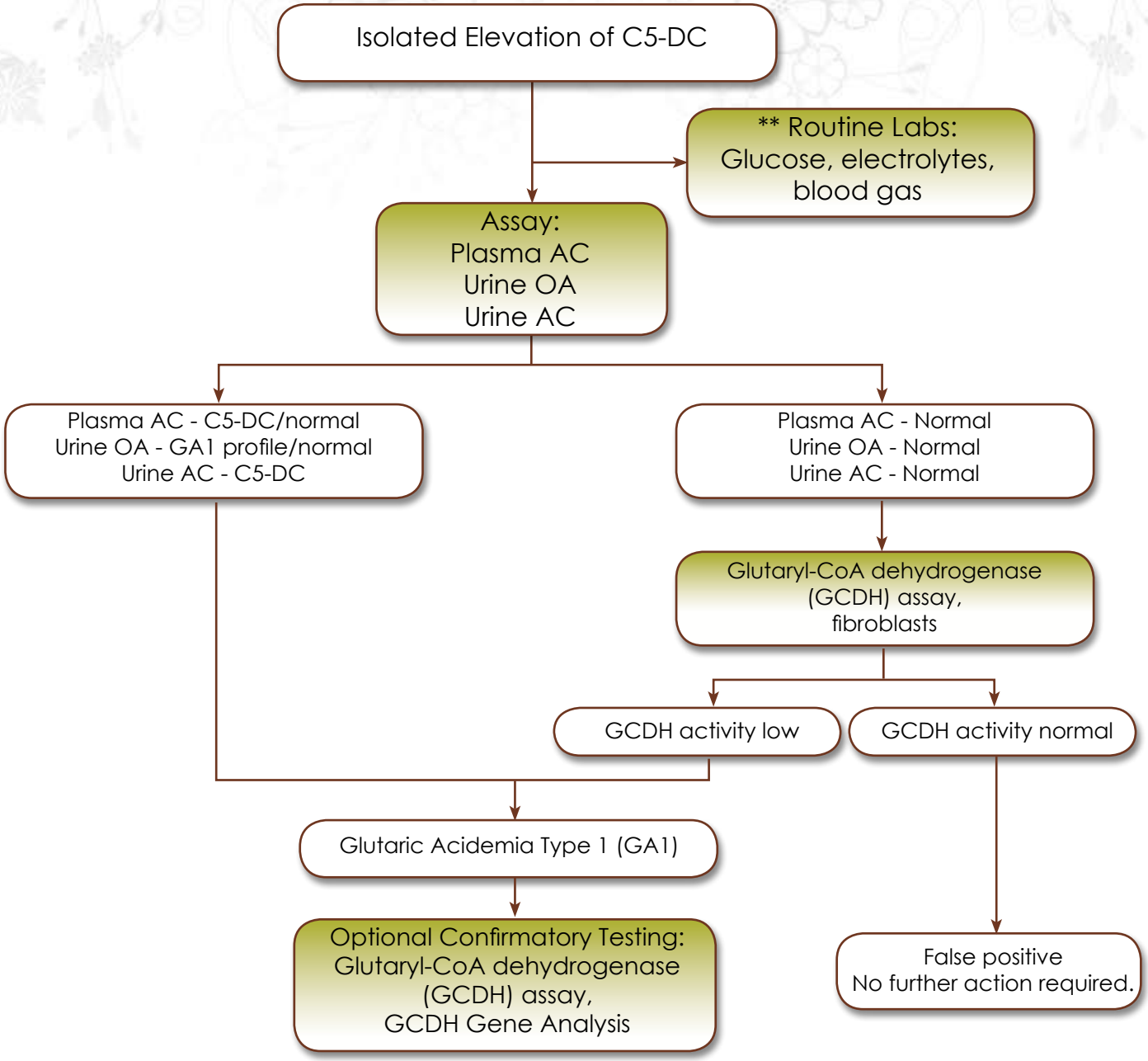
C5-DC Elevated (Isolated)



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

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.

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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