

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

Differential Diagnosis: Glutaric aciduria (GA-1). Maternal expression of disorder possible.

Condition Description: GA-1 is caused by a defect of glutaryl-CoA dehydrogenase which limits the metabolism of glutaryl-CoA to crotonyl-CoA, resulting in increased glutaric acid or its metabolites that are toxic.

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### ***YOU SHOULD TAKE THE FOLLOWING 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.
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Diagnostic Evaluation: Urine organic acid analysis should be ordered promptly, and will be diagnostic if it shows increased 3-hydroxyglutaric acid with or without increased glutaric acid. If urine organic acids don't confirm the diagnosis, the metabolic specialist will consider analyzing glutarylcarnitine 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.

### Additional Information:

[Gene Reviews](#)

[Genetics Home Reference](#)

### Referral (local, state, regional and national):

[Testing](#)

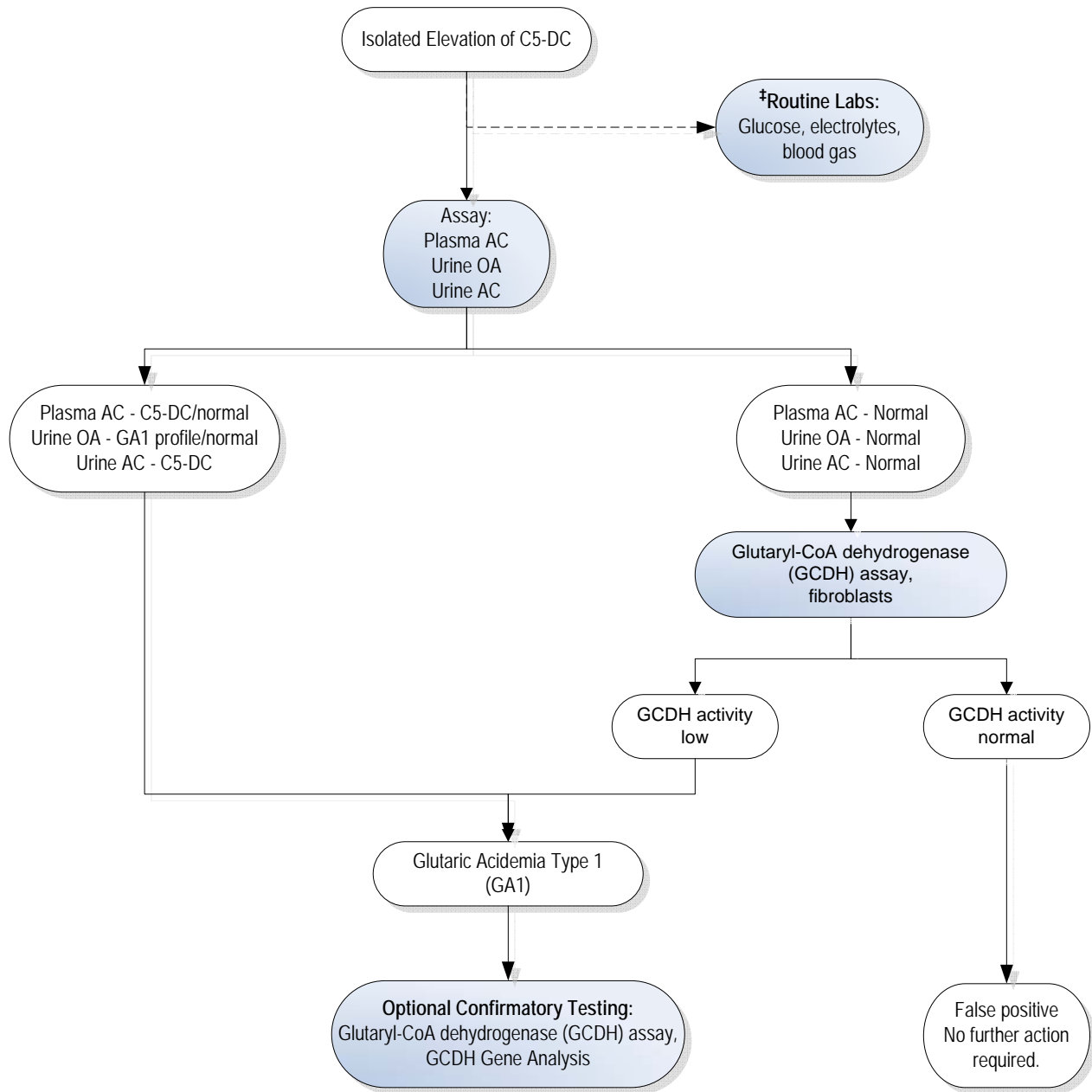
[Clinical Services](#)

[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.



## C5-DC Elevated (Isolated)



### 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.

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