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.

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.

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

ACMG
American College of Medical Genetics and Genomics
Translating Genes Into Health®

LOCAL RESOURCES: Insert State newborn screening program web site links

State Resource site *(insert state newborn screening program website information)*

Name	
URL	
Comments	

Local Resource Site *(insert local and regional newborn screening website information)*

Name	
URL	
Comments	

APPENDIX: Resources with Full URL Addresses

Additional Information:

Gene Reviews
http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi?book=gene&part=oa-overview

Genetics Home Reference
http://ghr.nlm.nih.gov/condition=glutaricacidemiatypei

Referral (local, state, regional and national):

Testing
http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/22682?db=genetests&country=United%20States

Clinical Services
http://www.ncbi.nlm.nih.gov/sites/GeneTests/clinic?db=GeneTests

Find Genetic Services
http://www.acmg.net/GIS/Disclaimer.aspx

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