

## Newborn Screening ACT Sheet

# [Elevated C5-DC Acylcarnitine] Glutaric Acidemia Type I (GA I)

**Differential Diagnosis:** None.

**Condition Description:** Glutaric acidemia Type I (GA I) is caused by deficiency of glutaryl-CoA dehydrogenase, an enzyme in the lysine degradation pathway, leading to elevated glutaric acid, glutarylcarnitine (C5-DC) and other toxic metabolites. Neonatal presentation is rare, but affected patients are at high risk of neurodegenerative crises triggered by common childhood illnesses.

### **You Should Take the Following IMMEDIATE Actions:**

- Inform family of the newborn screening result
  - Ascertain clinical status (poor feeding, macrocephaly, hypotonia).
  - Consult with pediatric metabolic specialist.
  - Evaluate the newborn (poor feeding, macrocephaly, hypotonia).
  - Initiate confirmatory/diagnostic testing and management, as recommended by the specialist.
  - Provide family with basic information about GA-I and its management.
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- Report findings to newborn screening program.

**Diagnostic Evaluation:** Urine organic acids: Glutaric and 3-hydroxyglutaric acids are typically elevated in GA I but may be only mildly elevated or have normal levels in some affected individuals. Plasma acylcarnitines will demonstrate elevated glutarylcarnitine (C5-DC). Urine glutarylcarnitine (C5-DC): may help identify patients with GA I who have low glutaric acid excretion by urine organic acid analysis. Molecular genetic testing can confirm the diagnosis.

**Clinical Considerations:** Neonates with GA I typically do not have acute symptoms but can be macrocephalic, most often presenting later in infancy. This disorder typically manifests as an acute encephalopathic crisis triggered by an intercurrent infectious illness, fasting, or other stressors. Rarely children may present with a chronic cerebral palsy presentation without an apparent acute metabolic crisis. Presymptomatic initiation of treatment can prevent the onset of irreversible neurologic symptoms. Treatment includes a diet low in lysine, carnitine supplementation, and emergency treatment during intercurrent illness with the goal of averting catabolism. Untreated late onset GA I (> 6 years of age) may manifest with other nonspecific neurologic abnormalities including headaches, vertigo, dementia and ataxia. Even with treatment all patients with GA I may be at increased risk for additional manifestations (e.g. renal disease). An infant with GA I presenting with macrocephaly and a subdural hematoma may phenotypically mimic shaken baby syndrome.

### **Additional Information:**

[How to Communicate Newborn Screening Results](#)

[Gene Reviews](#)

[Medline Plus](#)

[Condition Information for Families- HRSA Newborn Screening Clearinghouse](#)

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

[Find a Genetics Clinic Directory](#)

[Genetic Testing Registry](#)

## State and Other Resources

### State Newborn Screening Program

Metabolic (Bloodspot) Screening, Montana's Newborn Screening Programs

406-444-0984, [dphhs.mt.gov/ecfsd/cshs/newbornscreeningprograms/](https://dphhs.mt.gov/ecfsd/cshs/newbornscreeningprograms/)

Short-term Follow-up Coordinator - Crystal Fortune, MT Laboratory Services Bureau

406-444-0930 or 800-821-7284 [cfortune@mt.gov](mailto:cfortune@mt.gov)

### Genetics/Metabolics Consultants

Shodair Children's Hospital Medical Genetics Staff will contact you with recommendations.

Metabolic Clinic: 406-444-1099; Clinics held across Montana: 406-444-1016

[shodair.org/about-shodair/contact/](https://shodair.org/about-shodair/contact/)

**If emergency consultation with a metabolic specialist is necessary**, contact One Call (24/7) at Children's Hospital Colorado (800-525-4871) and ask to speak to the metabolic specialist on call.

### Information for Clinicians and Families

Montana Medical Home Portal (see Newborn Disorders and Parents & Families sections)

[mt.medicalhomeportal.org/newborn/glutaric-acidemia-type-1](https://mt.medicalhomeportal.org/newborn/glutaric-acidemia-type-1)

### Parent/Family Support

Organic Acidemia Association

[www.oaaneews.org/ga-i.html](https://www.oaaneews.org/ga-i.html)

## National Resources (with web addresses)

### Additional Information

How to Communicate Newborn Screening Results

[www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf](https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/Resources/achdnc-communication-guide-newborn.pdf)

Gene Reviews

[www.ncbi.nlm.nih.gov/books/NBK546575/](https://www.ncbi.nlm.nih.gov/books/NBK546575/)

Medline Plus

[medlineplus.gov/genetics/condition/glutaric-acidemia-type-i/](https://medlineplus.gov/genetics/condition/glutaric-acidemia-type-i/)

Condition Information for Families-HRSA Newborn Screening Clearinghouse

[newbornscreening.hrsa.gov/conditions/glutaric-acidemia-type-i](https://newbornscreening.hrsa.gov/conditions/glutaric-acidemia-type-i)

Genetic and Rare Diseases Information Center

[rarediseases.info.nih.gov/diseases/10321/3-methylglutaconyl-coa-hydratase-deficiency-auh-defect](https://rarediseases.info.nih.gov/diseases/10321/3-methylglutaconyl-coa-hydratase-deficiency-auh-defect)

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

Find a Genetics Clinic Directory

[clinics.acmg.net](https://clinics.acmg.net)

Genetic Testing Registry

[www.ncbi.nlm.nih.gov/gtr/](https://www.ncbi.nlm.nih.gov/gtr/)