

## Glutaric Acidemia, type 1 (GA1)

### Action required

Contact metabolic specialist today.  
See infant today.

### Issues to discuss with metabolic specialist

- Laboratory evaluation of infant
  - § Should testing be performed by primary care or metabolic clinic
  - § Plasma acylcarnitine analysis
    - 0.1 mL frozen plasma in sodium heparin green top tube
  - § Plasma hydroxy fatty acids
    - 0.5 mL frozen plasma in sodium heparin green top tube
  - § Urine organic acids
    - 4.0 mL random urine, frozen

### False Positives

- Screening result can be impacted by certain drug therapies containing pivalic acid or with administration of TPN

### Review with family

Family has **not** been notified of result by MDH.

After discussion with metabolic specialist, contact family to coordinate clinic visit, lab work, and referral to metabolic clinic.

Prompt follow-up is critical.

### NICU issues

Newborn screens cannot be accurately interpreted after drug therapies containing pivalic acid or administration of TPN.

GA1 has been mistaken for child abuse.

### Clinical summary

Glutaric acidemia (GA1) is an autosomal recessive disorder that results from the defective activity of glutaryl-CoA dehydrogenase, an enzyme involved in breaking down lysine, tryptophan, and glutaric acid. Newborns are usually asymptomatic, but macrocephaly may be present at birth. If an infant is not screened and/or left untreated, symptoms may begin to appear in early infancy or childhood and can include metabolic ketoacidosis, failure to thrive, retinal hemorrhage, dystonia, cerebral palsy, and seizures.

Affected children require a lysine/tryptophan-free diet, life-long riboflavin and carnitine supplementation, and monitoring by both primary care and specialty providers.

**Incidence of GA1:** Rare. ~1:40,000; more prevalent in the Amish community: ~1:300

### Clinical expectations

If treated promptly, children with GA1 can live healthy lives with normal growth and development.

Appropriate treatment can prevent neurodegenerative disease. Metabolic crises can still occur even with treatment.

Affected children should be monitored for amino acid levels.

### Resources

**GeneTests:** [www.genetests.org](http://www.genetests.org)

**OMIM:** [www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM](http://www.ncbi.nlm.nih.gov/sites/entrez?db=OMIM)

**ACT Sheets:** [www.acmg.net/resources/policies/ACT/condition-analyte-links.htm](http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm)

**MN Newborn Screening Program:**  
[www.health.state.mn.us/newbornscreening](http://www.health.state.mn.us/newbornscreening)