



Newborn Screening ACT Sheet

Elevated C5-DC Acylcarnitine Glutaryl-CoA Dehydrogenase Deficiency

Differential Diagnosis: Glutaric aciduria (GA-I)

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.

You Should Take the Following Actions

- **Contact family IMMEDIATELY to inform them of the newborn screening result.**
- **Consult with pediatric metabolic specialist. (See attached list.)**
- **Evaluate the newborn for macrocephaly and muscle hypotonia; initiate confirmatory/diagnostic testing as recommended by metabolic specialist.**
- **Initial testing: Plasma acylcarnitine profile, urine organic acids.**
- **Repeat newborn screen if the second screen has not been done.**
- **Refer to metabolic specialist to be seen as soon as possible – not any 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 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 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:

New England Metabolic Consortium

<http://www.childrenshospital.org/newenglandconsortium/NBS/descriptions/GAI.html>

Gene Tests/Gene Clinics

<http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=NexQvDbtfnPSK&gry=&fcn=y&fw=UOcs&filena>
[me=/profiles/oa-overview/index.html](http://www.genetests.org/servlet/access?db=geneclinics&site=gt&id=8888891&key=NexQvDbtfnPSK&gry=&fcn=y&fw=Ume=/profiles/oa-overview/index.html)

Genetics Home Reference

<http://ghr.nlm.nih.gov/condition=glutaricacidemia?typei>

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<http://www.newbornscreening.info/Pro/organicaciddisorders/GA1.html>
<http://www.newbornscreening.info/Parents/organicaciddisorders/GA1.html>



Newborn Screening FACT Sheet

Glutaric Acidemia, Type 1 (GA1)

What is GA1?

GA1 is a type of organic acid disorder. People with GA1 can't break down the amino acids lysine and tryptophan from the foods they eat.

What Causes GA1?

Enzymes help start chemical reactions in the body. GA1 happens when an enzyme called "glutaryl-CoA dehydrogenase" is missing or not working. This enzyme breaks down glutaric acid. Glutaric acid is made when the body breaks down lysine, hydroxylysine and tryptophan. When a child with GA1 eats food with these amino acids, glutaric acid builds up in the blood. All foods with protein contain these amino acids.

What Symptoms or Problems Occur with GA1?

[Symptoms are something out of the ordinary that a parent notices.]

Newborns with GA1 are usually healthy, but many are born with a large head. Other symptoms usually start between 2 months and 4 years of age.

GA1 causes periods of severe illness called Metabolic Crises. Some early signs of a Metabolic Crisis are:

- poor appetite
- too much sleepiness or lack of energy
- irritable mood
- feeling jittery
- nausea
- vomiting
- low muscle tone (floppy muscles and joints)
- weak muscles

If untreated, more symptoms can follow:

- tics or muscle spasms
- rigid muscles
- jerking movements of the arms and legs
- poor coordination and balance
- high levels of acids in the blood
- seizures
- brain swelling, or blood in the brain
- coma, sometimes leading to death

What is the Treatment for GA1?

Treatments for babies and children with GA1 are:

1. Medication – The doctor may prescribe riboflavin for your child. This is a vitamin that helps the body use protein. It helps remove glutaric acid from the blood. The doctor might also prescribe L-carnitine. This is safe and natural and helps the body make energy. Don't use any medicine without checking with your doctor.

2. Food plan, including medical foods and formula–

Most children need to eat foods low in lysine and tryptophan. The diet often includes special medical foods and formulas. The following foods should not be eaten at all or limited:

- milk, cheese and other milk products
- meat and poultry
- fish
- eggs
- dried beans and peas
- nuts and peanut butter

3. Blood tests – Regular blood tests will measure your child's amino acid levels.

Things to Remember

Minor illness such as a cold or the flu can cause a Metabolic Crisis in babies and children with GA1. Call your doctor right away when your child has any of the following:

- loss of appetite
- low energy or too much sleepiness
- vomiting
- fever
- infection or illness
- behavior or personality changes (such as crying for no reason)

Sick children often don't feel hungry. If they can't eat or show signs of a Metabolic Crisis, they may need to be treated in the hospital.

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