

Glutaric Acidemia, type 1 (GA1)

(glue-ter-ic acid-ee-me-ah)

What is a positive newborn screen?

Newborn screening is done on tiny samples of blood taken from your baby's heel 24 to 48 hours after birth. Newborn screening tests for rare, hidden disorders that may affect your baby's health and development. The newborn screen suggests your baby might have a disorder called GA1 (gee-A-1).

A positive newborn screen does not mean your baby has GA1, but it does mean your baby needs more testing to know for sure.

Your baby's doctor will help arrange for more testing by specialists in disorders like GA1.

What is glutaric acidemia?

GA1 affects an enzyme needed to break down proteins from the food we eat, so they can be used for energy and growth. In GA1, the enzyme used to break down proteins is missing or not working properly.

A person with GA1 doesn't have enough enzyme to break down protein containing lysine and tryptophan, which causes a substance called glutaric acid to build up in the body. Glutaric acid is harmful to the body and causes health problems if it builds up.

GA1 is a disorder that is passed on, or inherited, from a child's mother and father. Because GA1 is a genetic disease, family members are at risk of having GA1 too, even if no one in the family has had it before.

What problems can glutaric acidemia cause?

GA1 is different for each child. Some children with GA1 have only a few health problems, while other children may have very serious complications. Babies with GA1 are usually healthy at birth, but many are born with a large head.

If GA1 is not treated, a child might develop:

- Sleepiness
- Feeding problems
- Vomiting
- Muscle weakness
- Bleeding into the brain or eyes
- Cerebral palsy
- Seizures
- Coma

It is very important to follow the doctor's instructions for testing and treatment.

What is the treatment for glutaric acidemia?

GA1 can be treated. The treatment is life-long. Treatment for children with GA1 can include:

- Medications to help the body use protein and remove glutaric acid
- Medications to help the body use energy
- Diet low in lysine and tryptophan - a dietician will help you set up the best diet for your child

Children with GA1 should see their regular doctor, a doctor who specializes in GA1, and a dietician.

Children with GA1 can benefit from prompt and careful treatment.

Resources for parents

Genetics Home Reference

<http://ghr.nlm.nih.gov>

Organic Acidemia Foundation

www.oaanews.org

MN Children & Youth with Special Health Needs

(651) 201-3650 or (800) 728-5420

MN Newborn Screening Program

www.health.state.mn.us/newbornscreening

Newborn Screening Program, 601 Robert St. N., St. Paul, MN 55155, Phone (800) 664-7772, Fax (651) 201-5471