



## GA-1 (Glutaric Acidemia, Type 1)

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### What is it?

GA-1 stands for glutaric acidemia, type 1. It is one type of organic acid disorder. People with GA-1 have problems breaking down the amino acids lysine and tryptophan from the food they eat.

### What causes it?

In order for the body to use protein from the food we eat, it is broken down into smaller parts called amino acids. Special enzymes then make changes to the amino acids so the body can use them.

GA-1 occurs when an enzyme, called glutaryl-CoA dehydrogenase is either missing or not working properly. This enzyme's job is to break down a substance called glutaric acid. Glutaric acid is made when the amino acids lysine, hydroxylysine and tryptophan are processed by the body. Whenever a child with GA-1 eats food containing lysine or tryptophan, glutaric acid and other harmful substances build up in the blood. Lysine and tryptophan are found in all foods that contain protein.

### If GA-1 is not treated, what problems occur?

Babies with GA-1 are usually healthy at birth, although many are born with a larger-than-average head size. Other symptoms usually start between 2 months and 4 years of age.

GA-1 causes episodes of severe illness called metabolic crises. Some of the first symptoms of a metabolic crisis are:

- 1) Poor appetite.
- 2) Extreme sleepiness or lack of energy.
- 3) Irritability.

- 4) Jitteriness.
- 5) Nausea.
- 6) Vomiting.
- 7) Low muscle tone (floppy muscles and joints).
- 8) Muscle weakness.

If untreated, other symptoms then follow:

- 1) Tics or spasms of the muscles
- 2) Rigid muscle contractions called spasticity
- 3) Involuntary jerking movements of the arms and legs, called dystonia
- 4) Poor coordination and balance problems
- 5) Increased levels of acidic substances in the blood, called metabolic acidosis
- 6) Seizures
- 7) Swelling of the brain or blood in the brain
- 8) Coma, sometimes leading to death.

Episodes of metabolic crisis can be triggered by:

- 1) Illness or infection.
- 2) Fever.

Other effects of GA-1 that can happen even without a metabolic crisis are:

- 1) Poor growth.
- 2) Enlarged liver.
- 3) Low muscle tone.
- 4) Progressive spasticity.
- 5) Dystonia, an involuntary movement disorder.
- 6) Repeated episodes of fever.
- 7) Excessive sweating.
- 9) Delays in walking and other motor skills.
- 10) Learning delays and mental retardation.
- 11) Speech problems.
- 12) Brain damage.

Some people have very mild or no symptoms and are found to be affected only after a brother or sister is diagnosed.

### **What is the treatment for GA-1?**

Your baby's primary doctor will work with a metabolic doctor and a dietician to care for your child.

Prompt treatment is needed to prevent a metabolic crisis. You need to start treatment as soon as you know your child has GA-1. Certain treatments may be advised for some children but not others. Treatment usually is needed throughout life.

The following are treatments often recommended for babies and children with GA-1:

#### ***Medication***

Riboflavin is a vitamin that helps the body use protein. It may also help remove glutaric acid from the blood. Your doctor may recommend that your child take riboflavin supplements by mouth.

Some children may be helped by L-carnitine. This is a safe and natural substance that helps body cells make energy. It also helps the body get rid of harmful wastes. Your doctor will decide whether or not your child needs L-carnitine supplements. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor.

Do not use any medication without checking with your metabolic doctor.

Children with symptoms of a metabolic crisis need medical treatment right away. They often need to be treated in the hospital. During a metabolic crisis, children may be given fluids, glucose, insulin, carnitine and other medications by IV to help get rid of harmful substances in the blood. Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

#### ***Food Plan, Including Medical Foods and Formula***

Most children need to eat a diet made up of foods low in lysine and tryptophan. Special medical foods and a special formula are usually part of the diet. Your dietician will create a food plan that has the right amount of protein, nutrients and energy for your child.

#### ***Low-Protein Diet***

Foods that will need to be avoided or strictly limited include:

- 1) Milk, cheese and other dairy products.
- 2) Meat and poultry.
- 3) Fish.
- 4) Eggs.
- 5) Dried beans and legumes.
- 6) Nuts and peanut butter.

Many vegetables and fruits have only small amounts of lysine and tryptophan and can be eaten in carefully measured amounts.

Do not remove all protein from the diet. Your child still needs a certain amount of protein for normal growth and development. Any changes in the diet should be made under the guidance of a dietician.

#### ***Medical Foods and Formula***

There are medical foods such as special low-protein flours, pastas and rice that are made especially for people with organic acid disorders.

A special medical formula that contains the right level of amino acids and nutrients for your child may be recommended. Your metabolic doctor and dietician will tell you whether your child should be on this formula and how much to use.

Your child's exact food plan will depend on many things such as his or her age, weight, general health and blood test results. Your dietician will fine-tune your child's diet over time.

The long-term benefits of the special diet and medical foods are not yet known. However, it is important to follow the food plan as long as your doctor advises.

### ***Regular Blood Tests***

Your child will have regular blood tests to measure his or her amino acid levels. Urine tests also may be done. Your child's diet and medication may need to be adjusted based on blood and urine test results.

### ***Call Your Doctor at the Start of Any Illness***

For some babies and children, even minor illness can lead to a metabolic crisis. In order to prevent problems, call your doctor right away when your child has any of the following:

- 1) Loss of appetite
- 2) Low energy or extreme sleepiness
- 3) Vomiting
- 4) Infection or illness
- 5) Fever
- 6) Behavior or personality changes

Children with GA-1 need to eat more carbohydrates and drink more fluids when they are ill – even if they're not hungry – or they could have a metabolic crisis. Also, they need to avoid eating protein during any illness.

Children who are sick often don't want to eat. If they can't eat, or if they show signs of a metabolic crisis, they may need to be treated in the hospital.

### **What happens when GA-1 is treated?**

With prompt and lifelong treatment, children with GA-1 often can live healthy lives with typical growth and learning. Early treatment can help prevent episodes of metabolic crisis and the resulting health effects.

Even with treatment, some children continue to have episodes of metabolic crisis. This can lead to brain damage and long-term problems with involuntary movements and spasticity. After age 6, metabolic crises are less common.

### **What causes the glutaryl-CoA dehydrogenase enzyme to be absent or not working correctly?**

Genes tell the body to make various enzymes. People with GA-1 have a pair of genes that do

not work correctly. Because of the changes in this pair of genes, the glutaryl-CoA dehydrogenase enzyme either does not work properly or is not made at all.

### **Is GA-1 inherited?**

GA-1 is inherited and affects both boys and girls equally.

Everyone has a pair of genes that make the glutaryl-CoA dehydrogenase enzyme. In children with GA-1, neither of these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with GA-1 rarely have the condition themselves. Instead, each parent has a single nonworking gene for GA-1. They are called carriers. Carriers do not have GA-1 because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have GA-1. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

### **Can other members of the family have GA-1 or be carriers?**

#### ***Having GA-1***

The brothers and sisters of a baby with GA-1 have a chance of being affected, even if they haven't had symptoms. Finding out whether other children in the family have GA-1 is important because early treatment may prevent serious health problems. Talk with your metabolic doctor or genetic counselor about testing your other children.

#### ***GA-1 Carriers***

Brothers and sisters who do not have GA-1 still have a chance to be carriers like their parents. Except in special cases, carrier testing should be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent chance to be a carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with GA-1.

When both parents are carriers, newborn screening results are not sufficient to rule out GA-1 in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

### **Can other family members be tested?**

#### ***Diagnostic Testing***

Brothers and sisters of a child with GA-1 can be tested using blood, urine or skin samples.

#### ***Carrier Testing***

If both gene changes have been found in your child, other family members can have DNA testing to see if they are carriers.

### **How many people have GA-1?**

About one in every 40,000 white babies in the United States is born with GA-1.

### **Does GA-1 happen more frequently in a certain ethnic group?**

GA-1 occurs in people from all parts of the world. It is more common in people of Amish background in the United States, the Ojibway Indians in Canada, and people of Swedish ancestry.

### **Does GA-1 go by any other names?**

GA-1 is sometimes also called:

- 1) Glutaric aciduria type 1.
- 2) Glutaryl-CoA dehydrogenase deficiency.
- 3) Dicarboxylic aminoaciduria.
- 4) Glutarate-aspartate transport defect.

### **Where can I find more information?**

Organic Acidemia Association

[www.oaanews.org](http://www.oaanews.org)

Children Living with Inherited Metabolic Diseases (CLIMB)

[www.climb.org.uk](http://www.climb.org.uk)

Save Babies Through Screening Foundation

[www.savebabies.org](http://www.savebabies.org)

Genetic Alliance

[www.geneticalliance.org](http://www.geneticalliance.org)

National Coalition for PKU and Allied Disorders

[www.pku-allieddisorders.org](http://www.pku-allieddisorders.org)

International Organization of Glutaric Acidemia

[www.glutaricacidemia.org](http://www.glutaricacidemia.org)

### **Children's Special Health Services (CSHS)**

State Capitol Judicial Wing

600 E. Boulevard Ave., Department 301

Bismarck, ND 58505-0269

Toll Free: 800.755.2714

701.328.2436

Relay TDD: 701.328.3975

CSHS website: [www.ndhealth.gov/CSHS](http://www.ndhealth.gov/CSHS)

North Dakota Department of Health website:

[www.ndhealth.gov](http://www.ndhealth.gov)

Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

### ***Family Resources***

Family to Family Network

Center for Rural Health

University of North Dakota

School of Medicine and Health

P.O. Box 9037

Grand Forks, ND 58202-9037

Toll Free: 888.434.7436

701.777.2359

Fax: 701.777.2353

Email: [NDF2F@medicine.nodak.edu](mailto:NDF2F@medicine.nodak.edu)

[www.medicine.nodak.edu/crh](http://www.medicine.nodak.edu/crh)

Pathfinder Services of ND  
Pathfinder Family Center  
1600 2<sup>nd</sup> Ave. SW, Ste. 19  
Minot, ND 58701  
Toll Free: 800.245.5840  
701.837.7500  
Relay TDD: 701.837.7501  
Email: [ndpath01@ndak.net](mailto:ndpath01@ndak.net)  
[www.pathfinder.minot.com](http://www.pathfinder.minot.com)



North Dakota Department of Health  
Newborn Screening Program  
600 E. Boulevard Ave., Dept. 301  
Bismarck, ND 58505-0200  
800.472.2286 or 701.328.2493  
[www.ndhealth.gov/familyhealth](http://www.ndhealth.gov/familyhealth)

Family Voices of North Dakota, Inc.  
P.O. Box 163  
Edgeley, ND 58433  
Toll Free: 888.522.9654  
701.493.2634  
Fax: 701.493.2635  
[www.geocities.com/ndfv](http://www.geocities.com/ndfv)

\*\*This fact sheet has general information.  
Every child is different and some of these facts  
may not apply to your child specifically.  
Certain treatments may be recommended for  
some children but not others. All children  
should be followed by a metabolic doctor in  
addition to their primary care provider.

### **Acknowledgement**

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