

PARENT FACT SHEET

DISORDER

Glutaric acidemia, type 1 (GA-1)

CAUSE

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 a 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 NOT TREATED

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 two months and four years of age. GA-1 causes episodes of severe illness called metabolic crises. A metabolic crisis can cause brain damage and problems with involuntary movements of the muscles and tendons (this is called spasticity).

TREATMENT OPTIONS

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is needed throughout life.

- Your child’s dietician will develop a food plan made up of foods low in lysine and tryptophan. A special formula will need to be given to your child as an infant. Special medical foods will also be part of this food plan.
- No changes to your child’s diet should occur without permission and supervision of the dietician.
- Riboflavin is a vitamin that helps the body use protein. It may also help remove glutaric acid from the blood. Your child’s metabolic specialist will prescribe the proper dose and make any changes when necessary.
- Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps the body make energy. It also helps the body get rid of harmful wastes. Your metabolic specialist will prescribe this medication, if necessary.
- Your child will have regular blood tests to measure their amino acid levels. Urine tests may also need to be done. The results of these tests may result in medication or diet changes.
- Children with GA-1 need to eat more carbohydrates and drink more fluids when they are ill – even when they are not hungry. They also should avoid all protein when they are ill.
- Contact your child’s doctor immediately at the start of any illness. Children with GA-1 may need to be treated in a hospital to prevent serious health problems.

IF TREATED

With prompt and lifelong treatment, children with GA-1 can often live healthy lives with typical growth and learning.

For more information go to the following website: <http://www.newbornscreening.info>