ORGANIC ACIDURIAS:

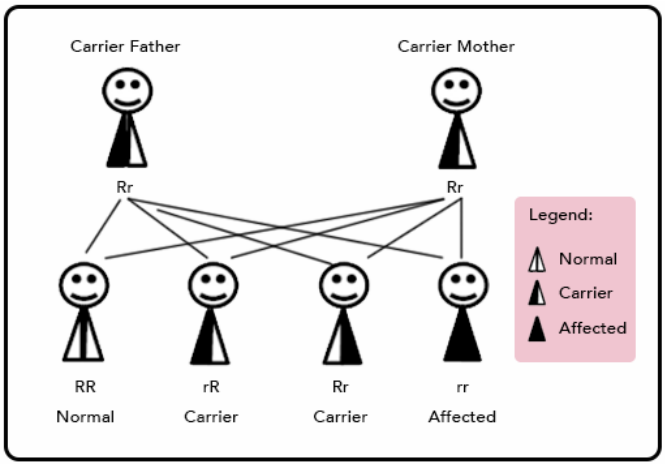
GLUTARIC ACIDURIA TYPE I [GA1]

What is GA1?

Glutaric Aciduria is due to a deficiency of an enzyme (or chemical scissors) called Glutaryl Co-A Dehydrogenase which is needed to break down the amino acids lysine and tryptophan. Amino acids are the building blocks of proteins. Children born with this condition may have an increased size of their head and may have convulsions, spasms of the muscles, involuntary movements of the arms or legs, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition.

What causes GA1?

To efficiently use the food we eat, our body breaks down the amino acids to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively break down the amino acids lysine and tryptophan. The accumulation of glutaric acid, a by-product of these amino acids, causes the signs and symptoms of this condition.



The gene for the glutaryl-CoA dehydrogenase enzyme is contained in the genetic material that we inherit from our parents. Because one part of the genetic material comes from the father and the other from the mother, the gene comes in pairs. In order to work correctly, at least one of the pairs should be working.

Parents of children with Glutaric Aciduria Type 1 have one working and one non-working gene coding for this enzyme. They do not manifest the disease but can pass them on to their children. They are known as **carriers**.

If the child inherits the non-working gene from both parents, he or she will have glutaric aciduria type 1. Thus, in each pregnancy, there is a 25% chance that the child will have the disorder, 50% chance of being a carrier and 25% chance of having two working genes.

What are the signs and symptoms of GA1?

Children born with this condition may have an increased size of their head and may have convulsions, spasms of the muscles, involuntary movements of the arms or legs, coma and death.

GLUTARIC ACIDURIA TYPE I [GA1]

WHAT IS THE TREATMENT OF GA1?

The main treatment of GA1 is through control of the diet. The child's diet should be low in tryptophan and lysine. Tryptophan and lysine are contained in protein rich foods. Children with glutaric aciduria are given a special milk formula without tryptophan or lysine. They are also given carnitine, a medication that helps the cells in the body to make energy.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL?

Children with GA1 may have a metabolic crisis which is a serious health condition caused by the build-up of toxic substances in the blood. A metabolic crisis occurs when a child is sick, has not eaten or drank well or during stressful events (such as surgery, severe infection and vaccination). Your child may present with lethargy, seizures or convulsions, increased involuntary movements, irritability and vomiting. If not treated properly and immediately, it might lead to serious brain damage and death. Once these signs and symptoms are present, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.

Normal breakdown of protein vs GA1 patient's breakdown of protein