FATTY ACID OXIDATION DISORDERS:

GLUTARIC ACIDURIA TYPE II [GA2]

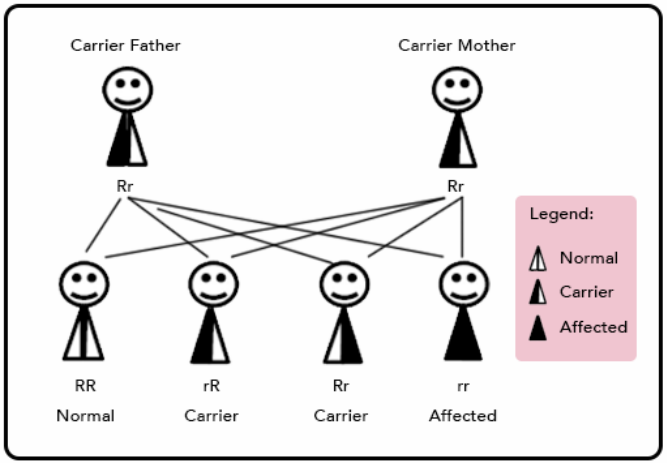
What is GA2?

Glutaric Aciduria Type II is due to a deficiency in the electron transfer flavoprotein and/or the ETF-ubiquinone oxidoreductase enzyme or chemical scissors that breaks down fat and proteins in the body. The deficiency of one or both of the enzymes may cause Glutaric Aciduria type II. Children with this condition cannot properly breakdown fat and some amino acids (or the building blocks of protein) in their body or from the food they eat.

Some children born with this condition may not display any signs or symptoms until later in life but some may present at infancy with nausea, vomiting, weakness and low blood sugar which may lead to seizures, coma and death. This disorder can be detected through newborn screening. Early detection can prevent the complications of this condition such as low blood sugar, increased sleepiness, and muscle weakness among others.

What causes GA2?

To efficiently use the food we eat, our body breaks fats and proteins down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively utilize fat or some amino acids (or the building blocks of protein) from their body or from the food they eat.



Glutaric Aciduria Type II is an inherited condition. The gene for the 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 II have one working and one non-working gene coding for a particular enzyme needed in the breakdown of fat and some proteins. 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 II. 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 GA2 Deficiency?

Children born with this condition appear normal at birth but if treatment is not given early they can present with low blood sugar which can lead to seizures, coma and death. Muscle weakness may be among the complications too.

GLUTARIC ACIDURIA TYPE II [GA2]

WHAT IS THE TREATMENT OF GA2?

The main treatment of GA2 is through control of the diet. Children with this condition should aim for a diet high in carbohydrates and low in protein and fat. If the child is well or does not have any illness, he/she should eat regular meals and avoid fasting more than 6 hours.

Riboflavin, a vitamin, has been found to be helpful. Carnitine, a supplemental medication essential for muscle energy production may also be of benefit for some patients.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL OR HAS AN ILLNESS (LIKE RESPIRATORY OR GASTROINTESTINAL INFECTION)?

Children with GA2 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 and severe infection). Your child may present with lethargy, seizures or convulsions, 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 GA2 patient's breakdown of protein