ORGANIC ACIDURIAS

What are Organic Acidurias?

Organic Acidurias are a group of conditions due to a deficiency in an enzyme or chemical scissors that breaks down proteins in the body. If not treated early, children with this condition may present with vomiting, irritability, drowsiness, rapid breathing and coma.

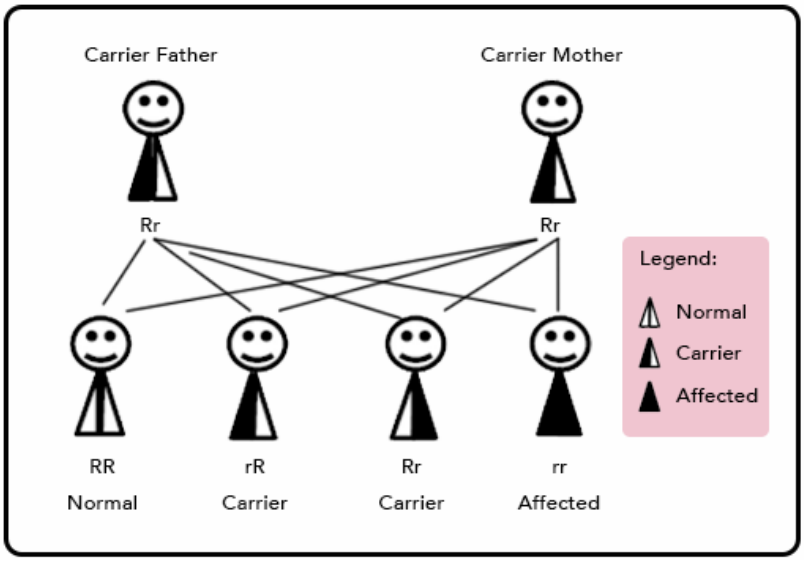
The name of the condition is derived from the substance that builds-up due to the deficiency of the said enzyme. An organic aciduria can be one of the following:

Glutaric aciduria type 1 - due to deficiency of the enzyme glutaryl-CoA dehydrogenase
3MCC deficiency - due to a deficiency of the enzyme 3-methylcrotonyl-CoA carboxylase
Propionic aciduria – due to a deficiency of the enzyme propionyl-CoA carboxylase
Methylmalonic aciduria– due to a defiency of the enzyme methymalonyl-CoA mutase
Isovaleric aciduria – due to a deficiency of the enzyme isovaleryl-CoA dehydrogenase

Other organic acid disorders include beta-ketothiolase deficiency and multiple carboxylase deficiency.

These group of disorders can be detected through newborn screening. Early detection can prevent the complications of these conditions.

What causes Organic Acidurias?



To efficiently use the food we eat, our body breaks it down to smaller units. Due to a lack of an enzyme or chemical scissors, children with this condition cannot effectively breakdown protein which causes the accumulation of toxic substances in the body.

The organic acidurias are an inherited condition. The gene for the different enzymes 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 organic acidurias have one working and one non-working gene coding for a particular enzyme needed in the

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breakdown of protein. 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 an organic acidemia. 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 ORGANIC ACIDURIAS?

Untreated children with this condition may present with vomiting, irritability, drowsiness, rapid breathing and coma. They may become seriously ill and it may cause death.

WHAT IS THE TREATMENT OF ORGANIC ACIDURIAS?

The main treatment of organic acidurias is through control of the diet. Children are placed on a low protein diet. They are also given carnitine, which is a medication that aids in the excretion of the toxic substances. Children with isovaleric aciduria are also given glycine which works similarly to carnitine. In some cases of methylmalonic aciduria, giving vitamin B12 is beneficial to the child.

Please consult with your attending physician prior to starting treatment.

WHAT SHOULD I DO WHEN MY BABY IS UNWELL?

Children with organic acidurias 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.

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