ORGANIC ACIDURIAS

What are Organic Acidurias?

Organic acidurias are an important class of inherited metabolic disorders arising due to defect in intermediary metabolic pathways of carbohydrate, amino acids and fatty acid oxidation. It leads to accumulation of organic acids in tissues and their subsequent excretion in urine.
The name of the condition in most instances is derived from the substance that builds-up due to the deficiency of the said enzyme. For example:

- Propionic aciduria due to a deficiency of the enzyme propionyl-CoA carboxylase
- Methylmalonic aciduria due to a deficiency of the enzyme methymalonyl-CoA mutase
- Isovaleric aciduria due to a deficiency of the enzyme isovaleryl-CoA dehydrogenase

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.
They are inherited condition and the gene for enzymes is contained in the genetic material that is inherited from the 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. 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 acidemias.

What are the signs and symptoms of Organic Acidurias?

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

How do you diagnose Organic Acidurias?

They are usually diagnosed in infancy by urinary excretion of abnormal amounts or types of organic acids. The diagnosis is usually made by detecting an abnormal pattern of organic acids in a urine sample by gas chromatography-mass spectrometry. Confirmatory diagnosis may require enzyme assays and molecular analysis.

What is the treatment of Organic Acidurias?

The main treatment of organic acidurias is through control of the diet and synthetic amino acid based formulas. Secondary carnitine deficiency is common and hence carnitine is also given in some cases. Treatment of the acute manifestations may also include dialysis, correction of fluid and electrolyte imbalances, correction of acidosis as well as maintenance of cerebral function with adequate perfusion, oxygen and glucose as the case demands. Even when the treatment is not available, the identification of organic acidurias is important for the genetic counseling and for making the pre-natal diagnosis possible in a future gestation.

What should I do when my baby is unwell or has an illness (like respiratory or gastrointestinal infection)?

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. If not treated properly and immediately, it might lead to serious brain damage and death. In case of any suspicion, please bring your child to the hospital for management and alert your pediatrician or metabolic physician.