

## **Methyl malonic acidemia (methylmalonic aciduria)**

### **What is the Methyl malonic acidemia?**

Our body breaks down food specially proteins to smaller substances to be used efficiently for our body's function. For this, our body makes use of enzymes to cut up proteins into building blocks called amino acid, which further enter into energy cycle. Methylmalonic acidemia is genetic disorder in which the body is unable to break down the certain amino acids like methionine, threonine, isoleucine and valine and fats, which in turn leads to a buildup of a toxic level of methylmalonic acid in the blood. However, not always grouped together with the inherited versions, a severe nutritional deficiency of vitamin B12 can also result in pattern with identical symptoms and treatments as the genetic methylmalonic acidemias.

### **What are the signs and symptoms of Methyl malonic acidemia?**

Children usually present in the first few days of life with lethargy, poor oral intake, recurrent vomiting, dehydration, failure to thrive, respiratory distress, weak muscle tone (hypotonia), developmental regression, hepatomegaly and coma.

### **How do you diagnose Methyl malonic acidemia?**

Determination of organic acids in urine, amino acids in blood (plasma, drawn after 3-4 hours fasting), urine and CSF, acyl carnitine profile in blood (dried blood or plasma) and total plasma homocysteine which is essential in order to differentiate the various types of MMA. Additional testing may be undertaken to identify the specific enzyme deficiency, including enzyme analysis and molecular genetic testing.

### **What is the treatment of Methyl malonic acidemia?**

There is no specific treatment for methylmalonic acidemia. Treatment is focused on managing the symptoms. Options include:

- Aggressive treatment of decompensation events
- Special protein managed diet
- Vitamin B12 supplementation
- Medications such as carnitine
- Avoidance of stressors (such as fasting or illness) that can lead to a decompensation event
- Liver and kidney transplant in some cases.

### **What should I do when my baby is sick?**

When children with Methyl malonic acidemia are sick, they may present with drowsiness,

vomiting, seizures or convulsions and irritability. If not treated properly and immediately, it might lead to serious brain damage. Once these signs and symptoms are present, it is advised to bring your child to the hospital for management and alert your pediatrician or metabolic physician.