Methylmalonic Acidemia (MMA)

What is Methylmalonic Acidemia?

Methylmalonic Acidemia is an organic acid disorders due to a deficiency in one of the enzymes involved in catabolism of methylmalonyl Co-A. This enzyme is called methylmalonyl Co-A mutase. The deficiency leads to the accumulation of methylmalonic acid and harmful substances in the blood.

What causes the disease?

Mutations in the *MUT* gene are a potential cause of the methylmalonyl Co-A mutase enzyme deficiency.

What are the clinical features of the disease?

The affected babies are usually normal at birth, however, they present symptoms in their first few weeks of life such as acute metabolic crises triggered by URTI or other catabolic state characterized by poor feeding, vomiting, lethargy that progress to coma and eventually death if untreated. It is frequently accompanied by metabolic acidosis with high anion gap, ketonuria, hypoglycemia, hyperammonemia. There is later onset form with variable presentations.

How is the diagnosis confirmed?

The diagnosis of MMA is confirmed by finding specific urine organic acids and plasma acylcarnitine profiles, in addition to plasma aminoacids. DNA molecular testing of the *MUT* gene as well as enzymatic studies may also aid in confirming the diagnosis.

What is the treatment of the disease?

A low protein diet, special metabolic formula, and vitamin B12 injections are often recommended to children with MMA. Children should not be kept without food for extended periods of time. Supplementation with carnitine and antibiotics may also be considered. Treatment can prevent metabolic crises and their sequelae. In an acute symptomatic episode, IV D10% and fluids can be given, along with other medications that can help the body excrete harmful substances and decrease the level of acid in the blood. A Biochemical genetics specialist and a Metabolic Genetics dietitian should coordinate the treatment.