Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)

What is MCAD Deficiency?

MCAD is a fatty acid oxidation defect due to deficiency in one of the enzymes (Medium-chain acyl-CoA dehydrogenase) involved in the breakdown of medium chain fatty acids (between 4 and 12 carbons long). Individuals who are missing this enzyme have an accumulation of "medium-chain" fatty acids which cause the symptoms of the disease.

What causes the disease?

Mutations in the *ACADM* gene are a potential cause of the Medium-chain acyl-CoA dehydrogenase (MCAD) 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 months of life such as poor feeding, vomiting, lethargy and hypoketotic hypoglycemia. These symptoms are frequently accompanied by seizures, Reye-like syndrome with hepatomegaly, elevated liver enzymes and hyperammonemia which could progress to a coma and eventually death, if not treated. Some infants suffered from sudden infant death syndrome.

How is the diagnosis confirmed?

The diagnosis of MCAD deficiency is established by measuring the acylcarnitine profile (Tandem MS) in the blood sample and confirmed by finding specific urine organic acids, enzyme testing and DNA molecular testing of the *ACADM* gene.

What is the treatment of the disease?

Frequent feedings ensure that a child with MCAD deficiency does not undergo any prolonged period of fasting. This is very effective in preventing metabolic crises and their sequelae. In an acute symptomatic episode, IV glucose should be given as soon as possible. Supplementation with carnitine and/or uncooked cornstarch as a source of glucose may also be considered. A Biochemical Geneticist should coordinate the treatment.