Very long-chain acyl-CoA dehydrogenase deficiency (VLCAD)

What is VLCAD Deficiency?

VLCAD is one of the fatty acid oxidation defects due to a deficiency of one of the enzymes involved in breaking down of long chain fatty acids (between 12 and 18 carbons long). Individuals missing this enzyme have an accumulation of "very long-chain" fatty acids which cause the symptoms of the disease.

What causes the disease?

Mutations in the *ACADVL* gene are a potential cause of the of Very long-chain acyl-CoA dehydrogenase (VLCAD) 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 such as poor feeding, vomiting, lethargy and hypoketotic hypoglycemia. These symptoms are frequently accompanied by seizures, cardiomyopathy and muscle weakness which could progress to a coma and eventually death, if not treated. There are also forms of VLCAD deficiency that present in later years reaching adulthood.

How is the diagnosis confirmed?

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

What is the treatment for the disease?

Frequent feeding ensures 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. In addition, a special diet low in long-chain fats may be prescribed. The treatment should be coordinated by a Biochemical Genetics Specialist and a Metabolic Genetics dietitian.