

BC Newborn Screening Program

Information Sheet

LCHAD Deficiency – Fatty Acid Oxidation Defect (FAOD)

What are Fatty Acid Oxidation Defects?

FAODs occur when fats (fatty acids) cannot be broken down in the body. Fats are an important source of energy for the body, especially during periods of fasting. Fatty acids are transported into cells and then taken into the mitochondria to be broken down.

What is LCHAD deficiency?

Fatty acids are made up of carbon chains. As these carbon chains are broken down, energy is released and the products of this process are used to make ketone bodies, another source of energy. LCHAD (Long Chain 3-Hydroxyacyl-CoA Dehydrogenase) is an enzyme responsible for breaking down carbon chains that are between 12 and 18 carbon atoms long. Individuals who are missing this enzyme have an accumulation of these “long-chain” fatty acids and are unable to make ketone bodies for energy.

What is its incidence?

LCHAD deficiency is a rare disease; its incidence is estimated at 1/80,000 in BC.

What causes the disease?

Mutations in the gene for LCHAD enzyme results in a deficient amount of enzyme.

What are the clinical features of the disease?

Although children with LCHAD deficiency are normal at birth, during a period of fasting (such as during a common illness), a child who was previously healthy may present with hypoketotic hypoglycemia, hypotonia, cardiomyopathy, vomiting or

diarrhea, lethargy, and seizures. This can progress quickly to coma and death. These children may also have developmental delays, poor weight gain, and eye, nerve, and liver problems. The first episode usually occurs in infancy or early childhood.

How is the diagnosis confirmed?

The diagnosis of LCHAD deficiency can be made by finding elevated levels of medium to long chain acylcarnitines on Tandem Mass Spectrometry (MS/MS) analysis of a blood sample. A specific urine organic acid profile, enzyme analysis, or mutation analysis of the LCHAD gene may also assist in confirming the diagnosis. Diagnostic testing is arranged by specialists at BC Children’s Hospital.

What is the treatment of the disease?

Frequent feedings ensure that a child with LCHAD deficiency does not undergo any prolonged period of fasting. 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. Treatment is coordinated by specialists at BC Children’s Hospital.

What is the outcome of treatment?

Treatment can be effective in preventing metabolic crises and their sequelae.

Can a family have more than one child with LCHAD deficiency?

LCHAD deficiency is inherited as an autosomal recessive disease. Parents of a child with LCHAD deficiency are assumed to be carriers for the disease and have a 1 in 4 (25%) chance, in each pregnancy, of having another child with this condition. Prenatal testing for LCHAD deficiency can be done as early as 10-12 weeks of pregnancy. Genetic counselling to discuss the benefits of prenatal testing options in more detail is recommended.

Unaffected siblings of a child with LCHAD deficiency have a 2/3 chance of being carriers. LCHAD carriers are healthy and do not have symptoms of the disease.

Resources

<http://www.fodsupport.org/>

<http://www.geneclinics.org>

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