

## BC Newborn Screening Program

## Information Sheet

### VLCAD 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 VLCAD 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. VLCAD (Very Long Chain Acyl-Co-A dehydrogenase) is an enzyme responsible for breaking down carbon chains that are between 12 and 18 carbons 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?

VLCAD deficiency is a rare condition; its incidence is estimated between 1/50,000 and 1/120,000 worldwide.

#### What causes the disease?

Mutations in the gene for VLCAD enzyme results in a deficient amount of enzyme or a defective enzyme.

#### What are the clinical features of the disease?

Although children with VLCAD deficiency may appear 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, vomiting or diarrhea, lethargy, and seizures. This can progress quickly to coma and death. Some children may also have significant heart problems and muscle weakness. In most cases, the first episode usually occurs in the first few months of life. There are also forms of VLCAD deficiency that will present later in childhood or even in adulthood.

#### How is the diagnosis confirmed?

The diagnosis of VLCAD deficiency is confirmed by measuring the levels and ratios of certain long-chain acylcarnitines by Tandem Mass Spectrometry (MS/MS).

A specific urine organic acid profile, enzyme testing, or mutation analysis of the VLCAD 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 VLCAD 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 VLCAD deficiency?

VLCAD deficiency is inherited as an autosomal recessive disease. Parents of a child with VLCAD 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 VLCAD 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 VLCAD deficiency have a 2/3 chance of being carriers. VLCAD carriers are healthy and do not have symptoms of the disease.

### Resources

<http://www.newbornscreening.info/Parents/fattyacid disorders/VLCADD.html>

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

<http://www.geneclinics.org>

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