
Trifunctional Protein (TFP) 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 TFP deficiency?

What causes this disease?

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. Trifunctional Protein (TFP) Deficiency occurs when a group of enzymes which function together, called “trifunctional protein”, is either missing or not working properly. The job of TFP is to help break down “long-chained” fats, along with the LCHAD (Long Chain 3-Hydroxyacyl-CoA Dehydrogenase) enzyme. Individuals who are missing TFP have an accumulation of long-chained fatty acids and are unable to make ketone bodies for energy.

What is its incidence?

TFP deficiency is a rare disease; its incidence is unknown.

What are the clinical features of the disease?

Although babies with TFP 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 irritability, hypotonia, lethargy, hypoketotic hypoglycemia, vomiting or diarrhea, and seizures. This can progress quickly to coma and death. These children may also have developmental delays, poor weight

gain, cardiomyopathy, breathing problems, muscle weakness, and liver problems. Periods of heavy exercise can also trigger episodes. The presentation of TFP deficiency is variable and there may be individuals with the disorder who have a milder course and or/an age of onset in childhood or adulthood.

How is the diagnosis confirmed?

The diagnosis of TFP 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 may also assist in confirming the diagnosis. Diagnostic testing is arranged by specialists at your regional treatment centre.

What is the treatment of the disease?

Frequent feedings ensure that a child with TFP deficiency does not undergo any prolonged period of fasting. A special diet low in long-chain fats may be prescribed. Supplementation with carnitine may also be considered. Treatment is coordinated by specialists at your regional treatment centre.

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 TFP deficiency?

TFP deficiency is inherited as an autosomal recessive disease. Parents of a child with TFP 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

TFP 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 TFP deficiency have a 2/3 chance of being carriers. TFP carriers are healthy and do not have symptoms of the disease.

Resources

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

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