

3-Hydroxy-Hexadecanoyl Disorders - (Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) and Trifunctional Protein Deficiency (TFP))

Fatty acid oxidation disorders

What is it?

Long Chain Hydroxyacyl-CoA Dehydrogenase Deficiency (also known as LCHAD) and Trifunctional Protein deficiency (TFP) are inherited fatty acid oxidation disorders. Patients with fatty acid oxidation disorders, like LCHAD and TFP, cannot breakdown fats to energy. Once the body uses up its primary source of energy (glucose, or blood sugar), the body begins to fail because it cannot then make energy from fats. Therefore, people with LCHAD and TFP must eat on a very regular basis and should not go long without food.

What are the symptoms?

A person with LCHAD or TFP can appear normal at birth. Symptoms are variable and range from recurrent episodes of hypoglycemia and muscle weakness, to heart, liver, or eye problems. These symptoms can progress very quickly to coma, cardiac arrest, brain damage, or even death in children who are not eating well. In addition, mothers of babies with LCHAD or TFP can have problems during their pregnancy such as HELLP syndrome or acute fatty liver of pregnancy. Many symptoms of LCHAD and TFP can be prevented by immediate treatment and lifelong management. People with LCHAD or TFP typically receive follow-up care by a team of professionals that are experienced in treating people with metabolic disorders.

Inheritance and frequency

LCHAD and TFP are inherited in an autosomal recessive manner. This means that for a person to be affected with LCHAD or TFP, he or she must have inherited two non-working copies of the gene responsible for causing LCHAD and TFP. Usually, both parents of a person affected with an autosomal recessive disorder are unaffected because they are carriers. This means that they have one working copy of the gene, and one non-working copy of the gene. When both parents are carriers, there is a 1 in 4 (or 25%) chance that both parents will pass on the non working copies of their gene, causing the baby to have LCHAD or TFP. Typically, there is no family history of LCHAD or TFP in an affected person. LCHAD and TFP are rare fatty acid oxidation disorders; the total number of people affected with LCHAD and TFP is not known.

How is it detected?

LCHAD and TFP may be detected through newborn screening. A recognizable pattern of elevated chemicals alerts the laboratory that a baby may be affected. Confirmation of newborn screening results is required to make a firm diagnosis. This is usually done by a physician that specializes in metabolic conditions, or a primary care physician.

How is it treated?

LCHAD and TFP are treated by eating frequently and avoiding fasting. Babies may be given a special formula, and sometimes medication, as recommended by a genetic metabolic medical professional.

DISCLAIMER: This information is not intended to replace the advice of a genetic metabolic medical professional.

For more information:

Genetics Home Reference

Website: <http://www.ghr.nlm.nih.gov>

Save Babies Through Screening Foundation

4 Manor View Circle

Malvern, PA 19355-1622

Toll Free Phone: 1-888-454-3383

Fax: (610) 993-0545

Email: email@savebabies.org

Website: <http://www.savebabies.org>

FOD (Fatty Oxidation Disorder) Family Support Group

1559 New Garden Rd, 2E

Greensboro, NC 27410

Phone: (336) 547-8682 [8am - 8pm EST every day]

Fax: (336) 292-0536 [email/call ahead between 8am and 8pm before faxing]

Email: deb@fodsupport.org

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

United Mitochondrial Disease Foundation

8085 Saltsburg Road, Suite 201

Pittsburgh, PA 15239

Phone: (412) 793-8077

FAX: (412) 793-6477

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