NUTRICIA Metabolic Nutrition *Fact Sheet*

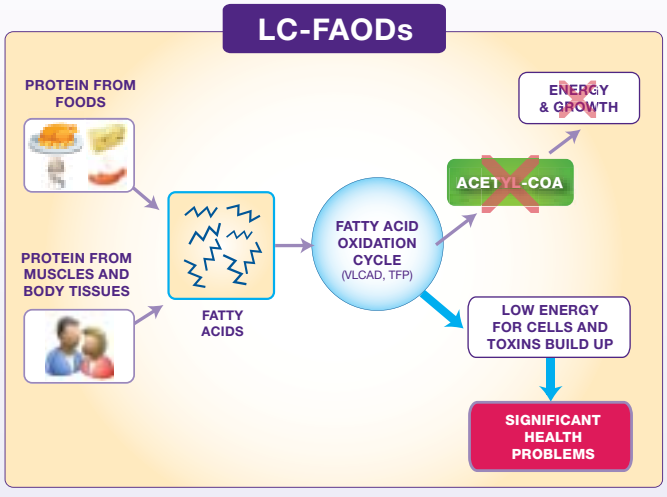
Long-Chain Fatty Acid Oxidation Disorders

An experienced healthcare professional should be consulted for the management of Long-Chain Fatty Acid Oxidation Disorders

What are Long-Chain Fatty Acid Oxidation Disorders?

Abbreviations & Definitions:	
Acetyl-CoA	A fatty acid; used by the cell to produce energy
EFA	Essential fatty acid
FAOD	Fatty acid oxidation disorder
HELLP Syndrome	Pregnancy complication involving high blood pressure and the liver
LCFA	Long-chain fatty acid
LC-FAOD	Long-chain fatty acid oxidation disorder
LCHAD	Long-chain 3-hydroxyacyl-CoA dehydrogenase; an enzyme
MCT	Medium chain triglyceride
TFP	Trifunctional protein; three enzymes that work together
VLCAD	Very long-chain acyl-CoA dehydrogenase; an enzyme

Long-chain fatty acid oxidation disorders (LC-FAODs) are a group of inherited, genetic disorders. LC-FAODS occur when there is a defect in one or more enzymes that help break down fat for energy. LC-FAODs prevent the body from using fats normally.



Fat from the diet and fat released from body stores are a main source of energy for cells. In the blood, fat is mostly present as fatty acids, which are building blocks of fat. Fatty acids are chains of carbon atoms that come in different lengths. Long-chain fatty acids (LCFAs) are typically 16 or 18 carbons long. To produce energy from fatty acids, the body must break them down by a chain of reactions called the "fatty acid oxidation cycle."

The first step in breaking down LCFAs involves an enzyme called VLCAD. The next steps are completed by a group of three enzymes called the trifunctional protein (TFP). One of the enzymes in the TFP is called LCHAD. The end product of the fatty acid oxidation cycle is a two-carbon fatty acid called acetyl-CoA, which cells use for energy.

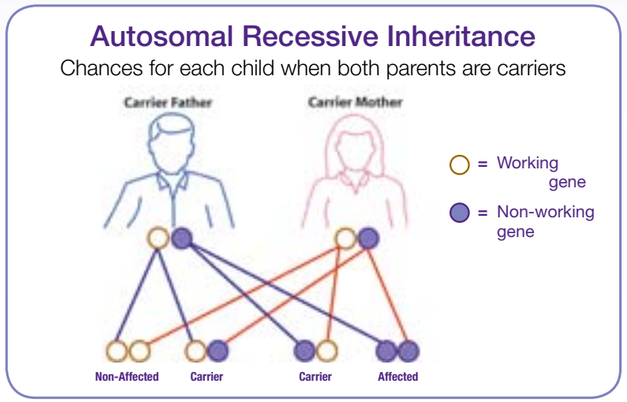
Medical problems associated with LC-FAODs can range from mild to life-threatening. Severe LC-FAODs include TFP deficiency, LCHAD deficiency, and one form of VLCAD. Problems begin very early in infancy and start with poor feeding, vomiting and weakness (called lethargy) and can quickly lead to seizures, coma and/or heart problems. On the other hand, people with the mildest form of VLCAD deficiency are often well until they are teens or adults, when severe muscle weakness and pain develop during exercise.

Finding out that your child has a LC-FAOD can be overwhelming. Fortunately, nutrition management is available to help prevent many of these problems. Take comfort in knowing that your metabolic healthcare team can provide you with the best advice to help manage the disorder.

Incidence, Genetics and Newborn Screening

Of the LC-FAODs, VLCAD deficiency is the most common, and occurs in about 1 in every 30,000 births. LCHAD deficiency and TFP deficiency are much rarer.

LC-FAODs are inherited as “recessive disorders.” In recessive disorders, both the mother and father are “carriers,” and they do not typically have any symptoms of the disorder (see diagram). With each pregnancy there is a 1 in 4 (25%) chance that the infant will be affected. A genetic counselor can help explain the inheritance of LC-FAODs and the risks to future infants.



While carriers typically do not have any symptoms, female carriers of LCHAD deficiency and TFP deficiency can develop “HELLP syndrome” during pregnancy if their growing infant is affected with one of these disorders. In HELLP syndrome, a mother can develop high blood pressure, stomach pain and bleeding as the amount of fat increases in her liver. Your metabolic healthcare team can provide guidance if you are at risk for HELLP syndrome.

LC-FAODs can be screened for at birth through a simple blood test. For more details on newborn screening (NBS) in the US, visit http://genes-r-us.uthscsa.edu; for information on NBS in Canada, see http://www.cadth.ca.

Management of Long-Chain Fatty Acid Oxidation Disorders

There is no cure for LC-FAODs, but they can be managed with a modified diet and special medical formulas specifically designed for persons with LC-FAODs. The modified diet for LC-FAODs means avoiding long “fasts” and eating a diet that is very low in fat, but supplemented with medium chain triglycerides (MCTs).

A fast is the time between meals when we are not eating. During a fast, the body first uses glucose (sugar) stores for energy. However, glucose stores are small and eventually the body switches to using fat stores as the main energy source. This releases LCFAs, which cause problems in people with LC-FAODs. Limiting the fasting time reduces the amount of LCFAs released. In early infancy, fasting is often limited to less than 4 hours. As the infant grows older, fasting times gradually increase. Sometimes, raw cornstarch is eaten before bedtime. Cornstarch breaks down slowly in the stomach to provide a slow and steady supply of glucose in the blood. This may help increase the allowed time for fasting overnight. The metabolic healthcare team can help you set safe fasting times.

A medical formula is a very important part of the diet for LC-FAODs. The fat in LC-FAOD formulas is very low in LCFAs, but includes MCTs. MCTs contain fatty acids that are medium-chain, or less than 12 carbons long. The body can use MCTs without the enzymes needed to break down LCFAs. The MCTs in the medical formula allows people with LC-FAODs to safely get energy from fat. Medical formulas may also provide protein, carbohydrates, vitamins and minerals the body needs for normal growth.

People with LC-FAODs eat a diet that is very low in fat because LCFAs are the main type of fat in foods, breast milk, and most formulas. A limited amount of fat is allowed in the diet. For infants, breast milk or regular infant formula may be given in precise amounts along with the LC-FAOD medical formula. As the baby grows and can eat solid foods, the breast milk or regular infant formula will be removed from the diet and replaced with foods.

Since all foods with fat contain LCFAs, people with LC-FAODs must limit their intake of foods high in fat. Typically the amount of fat in the diet is counted. MCT oil may be added to the medical formula or to foods to safely provide additional energy. Also, small amounts of certain oils may also be added to the diet to prevent deficiency of essential fatty acids (EFA), which are long-chain fats that people must get from the diet.

Nutricia North America provides a range of medical formulas. Please contact us for more information. Your dietitian will help you decide which products are best.

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If you or your child is sick

During an illness or after an injury, the body releases extra glucose and breaks down fat stores to provide the extra energy needed to recover. This can raise the level of LCFAs in the blood and lead to serious medical problems. Early signs that require immediate medical attention include vomiting, lethargy, muscle pain and/or changes in mental status. During any illness, it is very important to notify your metabolic clinic immediately. The fasting time may be shortened and the diet may be adjusted to increase carbohydrate and decrease fat. This can help slow down the breakdown of fat stores. Your clinic will give you an emergency letter – if you notice symptoms of high LCFAs in the blood, take this letter with you to the emergency room. During serious illness, a hospital stay may be necessary.

Resources

- Star-G website: http://www.newbornscreening.info
- Fatty Acid Oxidation Disorders Family Support Group: http://www.fodsupport.org

Nutricia North America would like to thank Sandy van Calcar, PhD, RD, University of Wisconsin-Madison for her consultation.

For more information about Nutricia products and great recipes, visit **www.medicalfood.com**

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