Fatty Acid Oxidation Disorders

What are fatty acid oxidation disorders?
Fatty acid oxidation disorders are inherited conditions that affect the way a person’s body breaks down certain fats (fatty acids). A person with a fatty acid oxidation disorder cannot breakdown their stored fat for energy. Consequently, the body begins to fail once the food the person has eaten runs out. In addition, fatty acids build up in the blood. In the case of fatty acid oxidation disorders, the inability to break down fats for energy and the build up of fatty acids can causes serious health problems.

How do fatty acid oxidation disorders affect a child?
The symptoms of these conditions vary and depend on the type of fatty acid oxidation disorder. Without treatment, children with an fatty acid oxidation disorder may experience periods of poor feeding, lack of energy, difficulty breathing, low blood glucose (sugar) and vomiting. These episodes can become serious enough to lead to developmental delay, seizures, coma and even sudden death.

What causes fatty acid oxidation disorders?
Fatty acid oxidation disorders are genetic conditions caused by changes in certain genes. These genes are responsible for making enzymes. These enzymes are responsible for breaking down fatty acids. When there is an alteration in these genes, enzyme levels go down and fatty acids build up in the blood.

Fatty acid oxidation disorders are inherited in an autosomal recessive pattern, which means two copies of a gene must be changed for a person to be affected with a fatty acid oxidation disorder. Most often, the parents of a child with an autosomal recessive condition are not affected because they are “carriers”, with one copy of the changed gene and one copy of the normal gene.

When both parents are carriers, there is a one-in-four (or 25%) chance that both will pass a changed gene on to a child, causing the child to be born with the condition. There also is a one-in-four (or 25%) chance that they will each pass on a normal gene, and the child will be free of the condition. There is a two-in-four (or 50%) chance that a child will inherit a changed gene from one parent and a normal gene from the other, making it a carrier like its parents. These chances are the same in each pregnancy for the same parents.

Is there a test for fatty acid oxidation disorders?
Yes. Babies can be tested (newborn screening) for fatty acid oxidation disorders before they leave the hospital. The baby’s heel is pricked and a few drops of blood are taken. The blood is sent to the state laboratory to find out if it has more than a normal amount of fatty acids.

There are various types of fatty acid oxidation disorders. The following is a list of fatty acid oxidation disorders that can be screened for:
- 2, 4 Dienoyl CoA Reductase deficiency
- Carnitine/Acylcarnitine Translocase deficiency (CAT)
- Carnitine Palmitoyl Tranferase deficiency Type I (CPT-I)
- Carnitine Palmitoyl Tranferase deficiency Type II (CPT-2)
- Carnitine Uptake Defect (CUD)
- Long/Very Long Chain Acyl CoA Dehydrogenase deficiency (LCAD/VLACD)
- Long Chain Hydroxy Acyl CoA Dehydrogenase deficiency (LCHAD)
Medium Chain Acyl CoA Dehydrogenase deficiency (MCAD)
Multiple Acyl CoA Dehydrogenase deficiency (GA-II)
Short Chain Acyl CoA Dehydrogenase deficiency (SCAD)
Trifunctional Protein deficiency (TFP)

Can fatty acid oxidation symptoms be prevented?
In many cases, therapy with a special diet and/or prescription medication is used. Children and adults with a fatty acid oxidation disorder require follow-up care at a medical center or clinic that specializes in this condition.

DISCLAIMER: The information contained on this page is not intended to replace the advice of a genetic metabolic medical professional.

Resources:
FOD (Fatty Oxidation Disorder) Family Support Group
805 Montrose Drive
Greensboro, NC 27410
Phone: 336-547-8682
Email: deb@fodsupport.org
www.fodsupport.org

United Mitochondrial Disease Foundation
8085 Saltsburg Road, Suite 201
Pittsburgh, PA 15239
Phone: 412-793-8077
Fax: 412-793-6477
Email: info@umdf.org
www.umdf.org

References: