Carnitine Palmitoyl Transferase Deficiency (CPT-II)
A fatty acid oxidation disorder

What is it?
Carnitine Palmitoyl Transferase Deficiency (also known as CPT-II) is an inherited fatty acid oxidation disorder. People with CPT-II cannot properly break down 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 CPT-II must eat on a very regular basis and should not go long without food.

What are the symptoms?
People with CPT-II can appear normal at birth. The majority of people with CPT-II present with hypoglycemia, high ammonia levels, fatigue, vomiting, and seizures after an episode of poor feeding, infection, or diarrhea. These symptoms can progress very quickly to coma, cardiac arrest, brain damage, or even death if not treated quickly. Many symptoms of CPT-II can be prevented by immediate treatment and lifelong management. People with CPT-II typically receive follow-up care by a team of professionals that is experienced in treating people with metabolic disorders.

Inheritance and frequency
CPT-II is inherited in an autosomal recessive manner. This means that for a person to be affected with CPT-II, he or she must have inherited two non-working copies of the gene responsible for causing CPT-II. 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 CPT-II. Typically, there is no family history of CPT-II in an affected person. CPT-II is a rare fatty acid oxidation disorder; the total number of people affected with CPT-II is not known.

How is it detected?
CPT-II 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?
CPT-II is treated by eating frequently and avoiding fasting, and sometimes a special 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
e-mail:  info@umdf.org
website:  http://www.umdf.org

American College of Medical Genetics
Newborn Screening ACT Sheets and Confirmatory Algorithms
http://www.aacmg.net/resources/policies/ACT/condition-analyte-links.htm

Cardinal Glennon Children’s Hospital
St. Louis, Missouri 314-577-5639
Website:  http://pediatrics.slu.edu/index.phtml?page=geneticsdiv

Children’s Hospital at University Hospital and Clinics
Columbia, Missouri 573-882-6991
Website:  http://www.genetics.missouri.edu/

Children’s Mercy Hospital
Kansas City, Missouri 816-234-3290
Website:  http://www.childrens-mercy.org/content/view.aspx?id=155

St. Louis Children’s Hospital
St. Louis, Missouri 314-454-6093
Website:  http://www.peds.wustl.edu/genetics/