

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://ghr.nlm.nih.gov/ghr/page/Home>

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/diseasedescriptions.php>

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 email: info@umdf.org

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

STAR-G Hawaii Department of Health

<http://www.newbornscreening.info/Parents/fattyaciddisorders/CPT1.html>