CPT1a: Carnitine Palmitoyltransferase 1 Deficiency

Carnitine palmitoyltransferase 1 deficiency, also known as CPT1a deficiency, is a type of genetic condition categorized as a fatty acid oxidation disorder (also known as a FAOD). The body usually gets its energy by breaking down, or burning, fats and sugars. People with FAODs cannot properly break down certain types of fats. For those with CPT1a deficiency, an enzyme needed to move long-chain fats into the mitochondria to be broken down for energy does not function properly. This prevents the body from creating needed energy during times of stress, illness, fasting, and exercise, which can lead to medical symptoms.

Genetics

People usually have two copies of the CPT1A gene, one inherited from each parent. CPT1a deficiency occurs when there are changes in both copies of the CPT1A gene (autosomal recessive inheritance). Someone who has a change in only one copy of the CPT1A gene is called a carrier, and they usually do not have any medical symptoms. If both parents are carriers, there is a 1 in 4 chance with each pregnancy that their child will have CPT1a deficiency. Both males and females can have CPT1a deficiency.

Frequency

The severe form of CPT1a deficiency is very rare with less than 60 cases identified worldwide. There is also a mild form of CPT1a deficiency frequently found in the Inupiaq, Yu'pik, and the Inuit populations in Alaska and Canada, as well as in Hutterite populations.

Signs and Symptoms

CPT1a deficiency can cause a range of signs and symptoms. Whether your child was diagnosed via newborn screening may also affect when, if, and how these symptoms present. Always check with your doctor if your child is doing something out of "their" normal, as every child may present with symptoms a little differently. Signs and symptoms may include:

- Life threatening low blood sugar with lack of ketones (hypoketotic hypoglycemia)
- Coma and seizures from hypoglycemia and hypoketosis
- Poor liver function
- Enlarged liver (hepatomegaly)
- Liver failure
- Organ failure

Diagnosis

Most cases of CPT1a deficiency are identified by routine newborn screening. If newborn screening is suggestive of CPT1a deficiency, additional tests may be performed including:

- Measuring biochemical markers in the blood or urine
- Performing a genetic test to look for changes in the CPT1A gene

In some cases a diagnosis of CPT1a deficiency can be made before birth. If there is a family history of CPT1a deficiency, or if parents are known carriers, testing can be performed on amniotic fluid (the fluid surrounding a baby) or chorionic villi (a specific part of the placenta).

Some pregnant mothers will experience a life threatening syndrome called HELLP syndrome during pregnancy. This may include high blood pressure, abnormal liver functions, and decreased blood clotting.

Treatment and Management

Before beginning any treatment or therapy, please consult with your physician.

Treatment and management of CPT1a deficiency may include:

- Regular eating schedules to prevent low blood sugar (hypoglycemia)
- Continuous feeding directly into the stomach in extreme cases
- Possible nutritional supplements like medium-chain triglycerides (e.g., MCT oil)
- Intravenous (IV) sugar-containing fluids called D10 to treat metabolic crisis

Parents should call their health care provider immediately if babies show symptoms like excessive sleepiness, vomiting, diarrhea, a fever, poor appetite, or an infection. Medical treatment should be sought immediately if there is loss of consciousness or severe confusion (decompensation), as these are signs of dangerously low blood sugar.

Patients should discuss the appropriate preparation for anesthesia with their metabolic team.

All patients should have an emergency protocol letter, written and signed by their doctor, that details their prescribed treatment during crisis and in emergency room settings, to manage severe episodes.

Clinical Trials

For specific details on other clinical trials, visit the MitoAction Clinical Trials page or www.clinicaltrials.gov.

Resources

- The National Institutes of Health: Carnitine-acylcarnitine translocase deficiency - About the Disease
- The INFORM Network: CPT1a Deficiency | Fatty Acid Oxidation Disorders Diagnosis
- National Organization for Rare Disorders (NORD) Carnitine Palmitoyltransferase 1A Deficiency - NORD

Connecting with others who are impacted by a rare disease allows for important information to be shared about day-to day life, prevents isolation, and gives hope. Please contact MitoAction for peer support opportunities at 888-MITO-411 or email mito411@mitoaction.org. Other resources we recommend are:

- New Patient Kit for FAODs
- Planning and Preparation
- Monthly Expert Series
- Energy in Action Podcast

MitoAction does not provide medical advice, diagnosis, treatment, or legal advice. It is essential that all those living with or caring for someone with a Mitochondrial or FAOD disease have an emergency protocol letter. These letters, which are written and signed by a doctor, share details about prescribed treatment during crises and in emergency room settings. Always check with your doctor if you or your child has concerns as everyone may present with symptoms differently. Before beginning any treatment or therapy, please consult with your physician.