MCKAT: Medium Chain 3-ketoacyl-CoA Thiolase Deficiency Deficiency

Medium chain 3-ketoacyl-CoA thiolase deficiency, also known as MCKAT 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. This prevents the body from creating needed energy during times of stress, illness, fasting, and exercise, which can lead to medical symptoms.

Alternative Names

- MCKAT Deficiency

Frequency

MCKAT deficiency is very rare, and the exact frequency is unknown. MCKAT deficiency is the rarest FAOD. The gene that causes MCKAT deficiency and the way it is inherited is not currently known.

Signs and Symptoms

There is limited information about the signs and symptoms of MCKAT deficiency since so few cases have been reported. In the first reported case, a 2-day-old baby showed signs of:

- Vomiting
- Dehydration
- Acidic blood (metabolic acidosis)
- Liver disease
- Severe muscle breakdown (rhabdomyolysis)
- Reddish-brown urine (myoglobinuria)

Older patients have shown other signs and symptoms, including:

- Low blood sugar (hypoglycemia)
- Vomiting
- Floppiness/poor muscle tone (hypotonia)
- Coma if the time between feedings is too long (fasting intolerance)
- Enlarged heart (cardiomyopathy)

In one case, the first sign of MCKAT deficiency was sudden death.

Diagnosis

There is very limited information on how babies with MCKAT were diagnosed. The only extensive reports were taken from the first case of MCKAT which occurred in a 2-day old baby. Testing showed high levels of lactic acids, ketones and dicarboxylic acids. Testing on skin cells showed that certain fats made little energy. Testing also showed low MCKAT activity level and reduced MCKAT protein.

Testing a baby for MCKAT before birth is not currently possible, as the gene that causes MCKAT is unknown.

Treatment and Management

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

There are no established treatments for MCKAT deficiency. Individuals with symptoms like dehydration, low blood sugar, and heart malfunctions should be treated immediately as they could have one of the many other fatty oxidation disorders. It is important to stay in close contact with your child's doctor as new treatments may develop over time.

Clinical Trials

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

Resources

- The INFORM Network: MCKAT Deficiency | Fatty Acid Oxidation Disorders Diagnosis
- Baby's First Test: Newborn screening information for medium-chain ketoacyl-CoA thiolase deficiency | Baby's First Test

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.