SCAD: Short-Chain acyl-CoA Dehyrdogenase Deficiency

Short-chain acyl-CoA dehydrogenase deficiency, also known as SCAD deficiency, is a type of genetic condition categorized as fatty acid oxidation disorders (also known as a FAODs). 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.

The SCAD defect creates a loss of function toward the end of fat metabolism. Fortunately, due to the enzyme position in the breakdown of fat, SCAD deficiency may lead to less severe symptoms or even no symptoms (asymptomatic). Your doctor will work with you to help you better understand what SCAD means for you.

Alternative Names

- SCAD deficiency

Cause and Genetics

People usually have two copies of the ACADS gene, one inherited from each parent. SCAD deficiency occurs when there are changes in both copies of the ACADS gene (autosomal recessive inheritance). Someone who has a change in only one copy of the ACAD9 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 SCAD deficiency. Both males and females can have SCAD deficiency.

There are two common changes in the ACADS gene. These changes still alter the way the ACADS protein works, but the protein made is usually still able to do its job well enough that the person with SCAD deficiency doesn't get sick. Some rare changes in the ACADS gene cause more of an effect on protein function, but people with these changes rarely get sick from SCAD deficiency. There is one rare genetic change that can cause the biggest effect on the ACADS protein when combined with one of the common changes.

Frequency

SCAD deficiency occurs in babies around the world. Some sources say that approximately 1 in 40,000 to 1 in 100,000 babies has SCAD deficiency, while others report higher numbers.

Signs and Symptoms

Most people with SCAD deficiency do not show any signs or symptoms of the condition. Reports of people with SCAD deficiency that did have signs or symptoms included:

- Poor muscle tone (hypotonia)
- Muscle weakness/breakdown (myopathy) and storage of fat in muscles
- Failure to thrive
- A buildup of acid in the body that can cause nausea, vomiting, fast breathing and extreme tiredness (metabolic acidosis)
- A buildup of acid in the body in newborns that causes increased muscle tone (hyperreflexia)

- Coma from high levels of ammonia in the blood (hyperammonemic coma)

Unlike other FAODs, SCAD deficiency does not cause low blood sugar (hypoglycemia) or low ketones (hypoketosis).

Diagnosis

Most cases of SCAD deficiency are identified by routine newborn screening. If newborn screening is suggestive of SCAD 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 genes that cause FAODs

Many newborn screening programs do not report when a baby has high levels of butyrylcarnitine (a sign of SCAD deficiency) since most SCAD deficiency is not considered harmful (benign). High levels of butyrylcarnitine are still sometimes reported on newborn screening because this, along with high levels of ethylmalonic acid, can happen in other metabolic conditions besides SCAD deficiency. Children who have signs or symptoms of FAODs along with high levels of these metabolites may be referred for further testing to rule out other conditions.

Treatment and Management

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

Most people with SCAD deficiency do not require a specific treatment for SCAD, but if symptoms do occur a patient/caregiver should contact their doctor.

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: Short-chain acyl-CoA dehydrogenase deficiency - About the Disease
- The INFORM Network: SCAD Deficiency | Fatty Acid Oxidation Disorders Diagnosis
- Baby's First Test: Newborn screening information for short-chain acyl-CoA dehydrogenase 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.