Disorder name: Short chain acyl-CoA dehydrogenase deficiency
Acronym: SCADD

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This fact sheet contains general information on SCADD. Every child with SCADD is different and some of this information may not apply to your child specifically. Not all is known about SCADD and, at present, there is no standard treatment plan. Certain treatments may be recommended for some children but not others. Children with SCADD should be followed by a metabolic doctor in addition to their primary doctor.

What is SCADD?

SCADD stands for “short chain acyl-CoA dehydrogenase deficiency.” It is one type of fatty acid oxidation disorder. Some people with SCADD cannot break down fat into energy for the body. However, most babies with newborn screening results showing SCADD never have symptoms.
**Fatty Acid Oxidation Disorders:**

Fatty acid oxidation disorders (FAODs) are a group of rare inherited conditions. They are caused by enzymes that do not work properly.

A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

The symptoms and treatment vary between different FAODs. They can also vary from person to person with the same FAOD. See the fact sheets for each specific FAOD.

FAODs are inherited in an **autosomal recessive** manner and affect both males and females.

**What causes SCADD?**

SCADD is caused by problems with an enzyme called “short chain acyl-CoA dehydrogenase” (SCAD). In people with SCADD, this enzyme is either missing or not working properly. This enzyme’s job is to break down certain fats from the food we eat into energy. It also breaks down fat already stored in the body.
Energy from fat keeps us going whenever our bodies run low of their main source of energy, a type of sugar called glucose. Our bodies rely on fat when we don’t eat for a stretch of time – like when we miss a meal or when we sleep.

Some people with SCADD cannot properly break down fat for energy. However, most people with SCADD do not seem to have this problem and do not ever develop symptoms.

If SCADD is not treated, what problems occur?

SCADD is highly variable and not well understood. Most babies found to have SCADD through newborn screening never have symptoms. In fact, so far, there have been only about 20 people with SCADD reported to have health effects. Things that cause stress, such as lack of sleep, going without food for too long, illness, or infection are thought to trigger episodes of illness called metabolic crisis in some children but not others.

For the small number of people with SCADD who show effects, the condition occurs in two different types: one found in infants, the other found in adults.

SCADD in infants
This type of SCADD is found in newborns and infants. Symptoms, when they happen, often start between the first week and 3 months of life.

Some of the first symptoms of a metabolic crisis are:
- extreme sleepiness
- behavior changes
- irritable mood
- poor appetite

Other symptoms then follow:
- fever
- diarrhea
- vomiting
- increased levels of acidic substances in the blood, called metabolic acidosis

If a metabolic crisis is not treated, a child with SCADD can develop:
- breathing problems
- seizures
- coma, sometimes leading to death

Other effects of SCADD seen in some infants and children are:
- poor weight gain
- delays in learning
- delays in walking and other motor skills
- hyperactivity
- decreased or increased muscle tone
- muscle weakness
- enlarged liver
- enlarged spleen

Symptoms of a metabolic crisis often happen after having nothing to eat for more than a few hours. Symptoms are also more likely when a child with SCADD gets sick or has an infection.

Many children with this condition have never had any effects and may only be found to have SCADD after a brother or sister has been diagnosed. Most children diagnosed through newborn screening never develop any symptoms related to SCADD.

**SCADD in adults**
The second type of SCADD is found in adults. The adult type of SCADD affects just the muscles. It can cause ongoing muscle problems, pain, and weakness. Adults with SCADD can also have episodes of nausea, vomiting, and shortness of breath. The muscle problems often get worse after heavy exercise or exertion.

**What is the treatment for SCADD?**

Your baby’s primary doctor may work with a metabolic doctor to care for your child. Your doctor may also suggest that you meet with a dietician familiar with SCADD.

Certain treatments may be advised for some children but not others. Babies found to have SCADD on newborn screening, but who have not shown any effects, may not need treatment. When necessary, treatment is usually needed throughout life. The following are treatments recommended for some, but not all, children with SCADD:

1. **Avoid going a long time without food**
   Some babies and young children with SCADD may need to eat often to avoid a metabolic crisis. These children should not go without food for more than four to six hours. In fact, some babies may need to eat even more often than this. They may also need to be fed during the night. Your metabolic doctor will tell you whether your child needs to be fed more often than normal. If so, you will receive an eating plan tailored to your child’s specific needs.

   Your metabolic doctor will continue to advise you on how often your child should eat as he or she gets older. When they are well, most teens and adults with
SCADD can go without food for up to 12 hours. People who have had symptoms may need to continue the other treatments throughout life.

2. **Diet**
A low fat, high carbohydrate food plan may be advised for some children with SCADD. Carbohydrates give the body many types of sugar that can be used as energy. In fact, for children needing this treatment, most food in the diet should be carbohydrates (bread, pasta, fruit, vegetables, etc.) and protein (lean meat and low-fat dairy foods). Any diet changes should be made under the guidance of a dietician familiar with SCADD.

Ask your doctor whether or not your child needs to have any changes in his or her diet.

3. **L-Carnitine and Riboflavin**
Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps the body create energy. It also helps the body get rid of harmful wastes. Your doctor will decide whether or not your child needs L-carnitine. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor.

A few children with SCADD have been helped by riboflavin (vitamin B2) supplements. Ask your metabolic doctor whether your child should take riboflavin.

Do not use any medications or supplements without checking with your doctor.

4. **Call your doctor at the start of any illness**
Call your health care provider when your child has any of the following:
- poor appetite
- low energy or excessive sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- persistent muscle pain or weakness

Some children with SCADD may need to eat extra starchy food and drink more fluids during an illness – even if they may not feel hungry – to prevent a metabolic crisis. Children who are sick often don’t want to eat. If they won’t or can’t eat, some children with SCADD may need to be treated in the hospital to prevent problems. If needed, your doctor will give you a ‘sick day’ plan for you to follow during illness or other times when your child will not eat.
What happens when SCADD is treated?

It is not known how effective treatment is in preventing problems. Treatment may help prevent or control symptoms in some children. Children who need treatment and are treated early may be able to live healthy lives with typical growth and development. Some children, though, may continue to have learning delays, muscle weakness, and other health problems despite treatment.

What causes the SCAD enzyme to be absent or not working correctly?

Genes tell the body how to make enzymes. The ACADS gene instructs the body to make the SCAD enzyme. Everyone has two copies of the ACADS gene. People with SCADD have changes, also called variants, in both copies of their ACADS genes that cause them to not work correctly.

How is SCADD inherited?

SCADD is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has a pair of genes that make the SCAD enzyme. In children with SCADD, neither of their ACADS genes works correctly. These children inherit one non-working ACADS gene from each parent.

Parents of children with SCADD rarely have the disorder. Instead, each parent has a single non-working ACADS gene. They are called carriers. Carriers do not have SCADD because their other ACADS gene is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have SCADD. There is a 50% chance for the child to be a carrier, just like the parents. And, there is a 25% chance for the child to have two working genes.
**Genetic counseling** is available to families who have children with SCADD. Genetic counselors can answer your questions about how SCADD is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

**Is genetic testing available?**

Genetic testing for SCADD can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes (variants) in the pair of genes that cause SCADD. DNA testing is often used to diagnose SCADD in children. In some affected children, both gene changes can be found. However, in other children, neither or only one of the two gene changes can be found, even though we know they are present.

DNA testing results will not help establish if your child will develop symptoms from SCADD. However, it can be helpful for carrier testing or prenatal diagnosis, discussed below. Talk with your metabolic doctor or genetic counselor if you have questions about DNA testing for SCADD.
What other testing is available?

SCADD can be confirmed by an enzyme test using a blood or skin sample. Talk to your doctor or your genetic counselor if you have questions about testing for SCADD.

Can you test during a future pregnancy?

If both gene changes have been found in your child with SCADD, DNA testing can be done during future pregnancies. The sample needed for this test is obtained by either CVS or amniocentesis.

If DNA testing would not be helpful, testing during pregnancy can be attempted by performing an enzyme test on fetal cells. Again, the sample needed for these tests is obtained by either CVS or amniocentesis.

Parents may either choose to have testing during pregnancy or wait until birth. Parents may also choose to use assisted reproductive techniques to decrease the chance that their future children would have SCADD. A genetic counselor can talk to you about your choices and answer other questions about prenatal testing or testing your baby after birth.

Can other members of the family have SCADD or be carriers?

**Having SCADD**
The brothers and sisters of an affected baby have a chance of having SCADD, even if they haven’t had symptoms. Talk with your doctor or genetic counselor about testing your other children for SCADD.

**SCADD Carriers**
Brothers and sisters who do not have SCADD still have a chance to be carriers like their parents. Except in special cases, carrier testing should only be done in people over 18 years of age.

If you are a parent of a child with SCADD, your brothers and sisters have a 50% chance to be a SCADD carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with SCADD.

Some states do not offer newborn screening for SCADD. However, expanded newborn screening is available through private labs for babies born in states that do not screen for this condition. Your healthcare provider or genetic counselor can help you obtain expanded newborn screening.
When both parents are carriers, newborn screening results are not sufficient to rule out SCADD in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

During pregnancy, women carrying fetuses with SCADD may be at increased risk to develop serious medical problems. Some women carrying fetuses with Fatty Acid Oxidation Disorders have developed:

- excessive vomiting
- abdominal pain
- high blood pressure
- jaundice
- abnormal fat storage in the liver
- severe bleeding

All women with a family history of SCADD should share this information with their obstetricians and other health care providers before and during any future pregnancies. Knowing about these risks allows better medical care and early treatment if needed.

Can other family members be tested?

**Diagnostic testing**
Brothers and sisters can be tested for SCADD using a blood or skin sample.

**Carrier testing**
If both gene changes have been found in your child with SCADD, other family members can have DNA testing to see if they are carriers.

If DNA testing would not be helpful, other methods of carrier testing may be available. Your metabolic doctor or genetic counselor can answer your questions about carrier testing.

How many people have SCADD?

SCADD was originally thought to be very rare. However, newborn screening for this disorder revealed that SCADD is more common than previously believed and is thought to affect between 1 in 35,000 to 50,000 newborns.

Does SCADD happen more often in a certain ethnic group?

SCADD does not happen more often in any specific race, ethnic group, geographical area, or country.
Does SCADD go by any other names?

SCADD is also sometimes called:
- SCAD deficiency
- ACADS deficiency
- SCADH deficiency

Where can I find more information?

Fatty Oxidation Disorders (FOD) Family Support Group
http://www.fodsupport.org

Organic Acidemia Association
http://www.oaanews.org

United Mitochondrial Disease Foundation
http://www.umdf.org

Metabolic Support UK
https://www.metabolicsupportuk.org

Genetic Home Reference

Baby’s First Test
http://www.babysfirsttest.org
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This project is supported by a grant from the Maternal and Child Health Bureau, Health Resources and Service Administration, Genetic Services Branch, MCH Project #: UH7MC30774-01-00 http://mchb.hrsa.gov

Short chain acyl-CoA dehydrogenase deficiency
Created by www.newbornscreening.info  11  Review Date: 5/14/2020