



SCADD (Short Chain Acyl-CoA Dehydrogenase Deficiency)

What is it?

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.

What causes it?

SCADD occurs when an enzyme called short chain acyl-CoA dehydrogenase (SCAD) is either missing or not working properly. This enzyme's job is to break down certain fats in 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 symptoms. 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 crises in some children but not others.

For the small number of people with SCADD who show effects, the condition occurs in two different forms: one found in infants and 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 third month of life.

Some of the first symptoms of a metabolic crisis are:

- 1) Extreme sleepiness.
- 2) Behavior changes.
- 3) Irritable mood.
- 4) Poor appetite.

Other symptoms then follow:

- 1) Fever
- 2) Diarrhea
- 3) Vomiting
- 4) Increased levels of acidic substances in the blood, called metabolic acidosis

If a metabolic crisis is not treated, a child with SCADD can develop:

- 1) Breathing problems.
- 2) Seizures.
- 3) Coma, sometimes leading to death.

Other effects of SCADD seen in some infants and children include:

- 1) Poor weight gain.
- 2) Delays in learning.
- 3) Delays in walking and other motor skills.
- 4) Hyperactivity.
- 5) Low muscle tone.
- 6) Muscle weakness or muscle tightness.
- 7) Enlarged liver.
- 8) Enlarged spleen.

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

Many children with this condition never had any effects and may be found to have SCADD only after a brother or sister has been diagnosed.

SCADD in Adults

The second type of SCADD is found in adults. The adult form of SCADD affects just the muscles. It can cause ongoing muscle problems, pain and weakness. Adults with SCADD also can 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 will work with a metabolic doctor to care for your child. Your doctor also may 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 usually is needed throughout life. The following are treatments often recommended for children with SCADD:

Avoid Going a Long Time Without Food

Some babies and young children with SCADD 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 also may need to be fed during the night. They need to be awakened to eat if they do not wake up on their own.

When they are well, most teens and adults with SCADD can go without food for up to 10 to 12 hours. People who have had symptoms do need to continue other treatments throughout life.

Diet

A low-fat, high-carbohydrate diet 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.

Ask your doctor whether your child needs to have any changes in his or her diet.

L-carnitine and Riboflavin

Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps the body make energy. It also helps the body get rid of harmful wastes. Your doctor will decide whether 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 medication without checking with your doctor.

Call Your Doctor at the Start of Illness

Call your health-care provider when your child has any of the following:

- 1) Poor appetite
- 2) Low energy or excessive sleepiness
- 3) Vomiting
- 4) Diarrhea
- 5) Infection
- 6) Fever
- 7) 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.

Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

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 to make various enzymes. People with SCADD have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the SCAD enzyme may not work properly or may not be made at all. Only a small number of babies found to have SCADD through newborn screening will actually have symptoms.

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 these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with SCADD rarely have the disorder. Instead, each parent has a single nonworking gene for SCADD. They are called carriers. Carriers do not have SCADD because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have SCADD. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

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 be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent 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.

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.

Can other family members be tested?

Diagnostic Testing

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

Carrier Testing

Carrier testing for SCADD may be available to other family members. Your metabolic doctor or genetic counselor can advise you about carrier testing for SCADD.

How may people have SCADD?

About one in 40,000 to one in 100,000 people have SCADD.

Does SCADD happen more frequently 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:

- 1) SCAD deficiency.
- 2) ACADS deficiency.
- 3) SCADH deficiency.

Where can I find more information?

Fatty Oxidation Disorders (FOD) Family Support Group

www.fodsupport.org

Organic Acidemia Association

www.oaanews.org

United Mitochondrial Disease Foundation

www.umdf.org

Children Living with Inherited Metabolic Diseases (CLIMB)

www.climb.org.uk

Genetic Alliance

www.geneticalliance.org

Children's Special Health Services (CSHS)

State Capitol Judicial Wing

600 E. Boulevard Ave., Department 301

Bismarck, ND 58505-0269

Toll Free: 800.755.2714

701.328.2436

Relay TDD: 701.328.3975

CSHS website: www.ndhealth.gov/CSHS

North Dakota Department of Health website:

www.ndhealth.gov

Family support resources available from CSHS:

- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

Family Resources

Family to Family Network

Center for Rural Health

University of North Dakota

School of Medicine and Health Sciences

P.O. Box 9037

Grand Forks, ND 58202-9037

Toll Free: 888.434.7436

701.777.2359

Fax: 701.777.2353

E-mail: NDF2F@medicine.nodak.edu

www.medicine.nodak.edu/crh

Pathfinder Services of ND

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1600 2nd Ave. SW, Ste. 19

Minot, ND 58701

Toll Free: 800.245.5840

701.837.7500

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Edgeley, ND 58433
Toll Free: 888.522.9654
701.493.2634
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www.geocities.com/ndfv

**This fact sheet has general information.
Every child is different and some of these facts
may not apply to your child specifically.
Certain treatments may be recommended for
some children but not others. All children
should be followed by a metabolic doctor in
addition to their primary-care provider.

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