Genetic Fact Sheets for Parents

Fatty Acid Oxidation Disorders

Screening, Technology, and Research in Genetics is a multi-state project to improve information about the financial, ethical, legal, and social issues surrounding expanded newborn screening and genetic testing – http:// www.newbornscreening.info

Disorder Name: **Short chain 3-hydroxyacyl-CoA dehydrogenase deficiency**
Acronym: **M/SCHADD**

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

What is M/SCHADD?

M/SCHADD stands for “medium/short chain 3-hydroxyacyl CoA dehydrogenase deficiency.” This disorder is also known as “3-hydroxyacyl CoA dehydrogenase deficiency.” It is one type of **fatty acid oxidation disorder**. Some people with M/SCHADD have problems using fat for energy. However, most babies with newborn screening results showing M/SCHADD never have any 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.

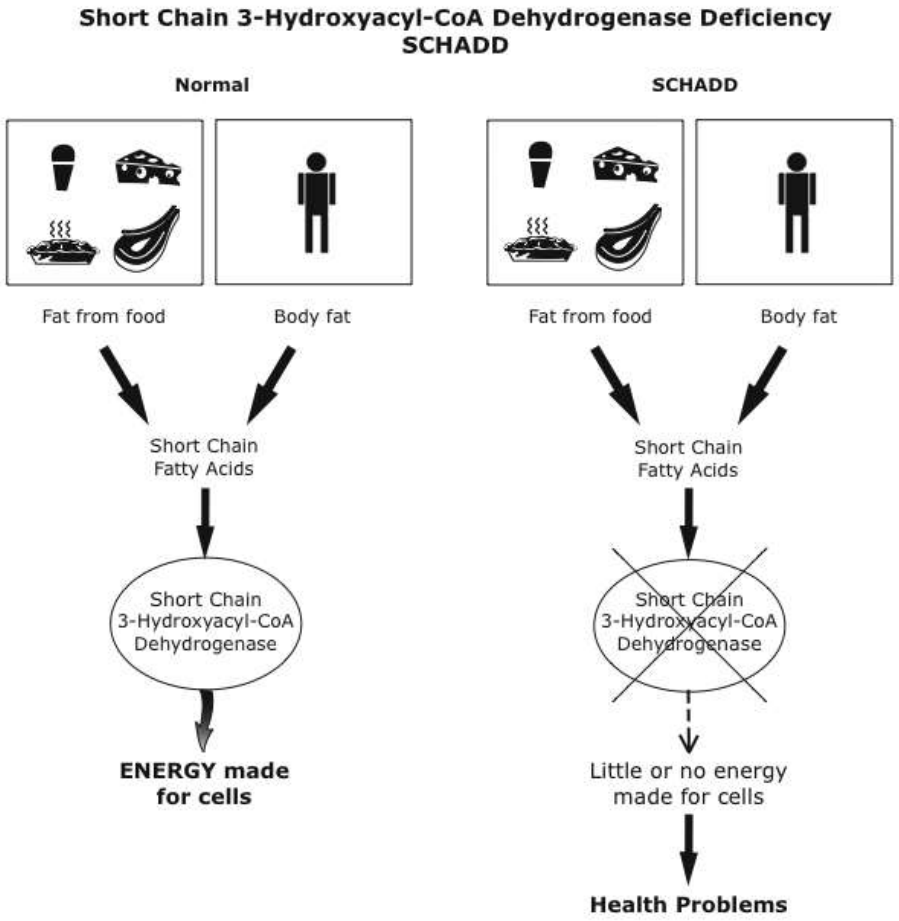
A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

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 M/SCHADD?

M/SCHADD is caused by problems with the enzyme “short chain 3-hydroxyacyl CoA dehydrogenase” (SCHAD). In people with M/SHADD, the SCHAD enzyme is either missing or not working properly. This enzyme’s job is to help change certain fats in the food we eat into energy for the body. It also helps us use 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 M/SCHADD cannot break down fat for energy. Most babies found to have M/SCHADD on newborn screening do not seem to have this problem and do not develop symptoms.

If M/SCHADD is not treated, what problems occur?

The symptoms of M/SCHADD are highly variable and not well understood. Things that cause stress, such as lack of sleep, lack of food, illness, or infection are thought to trigger episodes of illness called metabolic crises in some children with M/SCHADD but not in others.

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
- low blood sugar, called hypoglycemia

If a metabolic crisis is not treated, a child with M/SCHADD can develop:
- breathing problems
- seizures
- swelling of the brain
- coma, sometimes leading to death

Other effects of M/SCHADD can include:
- irregular heart beat and other heart problems
- enlarged heart
- liver problems
- muscle problems
- high levels of insulin in the blood in some babies

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 person with M/SCHADD gets sick or has an infection.

What is the treatment for M/SCHADD?

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 M/SCHADD.

Certain treatments may be advised for some children but not others. When necessary, treatment is usually needed throughout life. The following are treatments that may be recommended for some children with M/SCHADD:

1. Avoid going a long time without food

Some infants and young children with M/SCHADD may need to eat frequently to prevent a metabolic crisis. Your metabolic doctor will tell you how often your child needs to be fed. In general, it is often suggested that infants that need to avoid fasting be fed every four to six hours. Some babies may need to eat even more frequently than this. It is important that these infants be fed during the night. If needed, your metabolic doctor and dietician will give you an appropriate feeding plan for your infant. Your doctor may also give you a 'sick day' plan tailored to your child's needs for you to follow during illnesses or other times when your child will not eat.

2. Diet
Sometimes a low fat, high carbohydrate food plan is recommended. 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 M/SCHADD.

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

3. L-carnitine supplements and other medications
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 your child needs L-carnitine. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor.

Babies with high insulin levels may need medication. Your doctor will let you know if your child needs to take medication for this.

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

4. Call your doctor at the start of any illness
If your child has shown previous symptoms of M/SCHADD, always call your health care provider when he or she has any of the following:

- poor appetite
- low energy or excessive sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- persistent muscle pain or weakness

Children who have had symptoms of M/SCHADD need to eat extra starchy food and drink more fluids during any illness - even if they may not feel hungry - or they could develop a metabolic crisis. Children who are sick often don't want to eat. If they won't or can't eat, children with symptoms of M/SCHADD 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 M/SCHADD is treated?

It is not known how effective treatment is in preventing health problems. It is hoped that with prompt and careful treatment, children with M/SCHADD will be able to live healthy lives with typical growth and development.

The goal of treatment is to prevent death and serious health problems. However, children who have repeated episodes of hypoglycemia or metabolic crisis may develop brain damage. This can result in learning problems, intellectual disabilities, or other lifelong effects.

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

Genes tell the body how to make enzymes. The HADH gene instructs the body to make the SCHAD enzyme. Everyone has two copies of the HADH gene. People with M/SCHADD have changes, also called variants, in both copies of their HADH genes that cause them to not work correctly. Because of the variants in the HADH genes, the SCHAD enzyme either does not work properly or is not made at all.

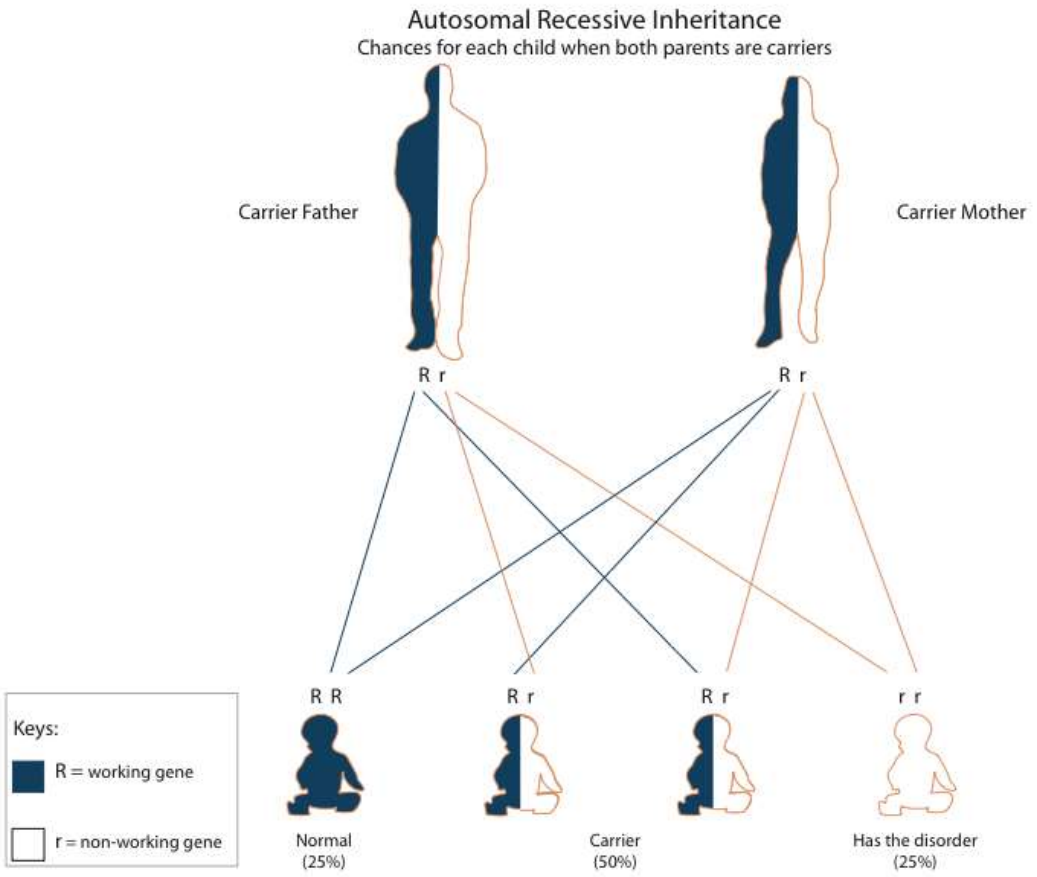
How is M/SCHADD inherited?

M/SCHADD is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has two copies of the HADH gene that make the SCHAD enzyme. In children with M/SCHADD, neither of these HADH genes works correctly. These children inherit one non-working HADH gene from each parent.

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

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have M/SCHADD. 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 M/SCHADD. Genetic counselors can answer your questions about how M/SCHADD 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 M/SCHADD may be available. Genetic testing, also called DNA testing, looks for changes (variants) in the HADH genes that cause M/SCHADD. Ask your metabolic doctor or genetic counselor about genetic testing for M/SCHADD.

DNA testing is not necessary to diagnose your child. It can be helpful for carrier testing or prenatal diagnosis, discussed below.

What other testing is available?

M/SCHADD can be confirmed by a special test called a "fatty acid oxidation probe" using a skin sample. Talk to your doctor or your genetic counselor if you have questions about testing for M/SCHADD.

Can you test during a future pregnancy?

It may be possible to test for M/SCHADD during pregnancy either by DNA testing or through a special test called a "fatty acid oxidation probe" using cells from the fetus. The sample needed for these tests is obtained by either CVS or amniocentesis.

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

Can other members of the family have M/SCHADD or be carriers?

Having M/SCHADD

The brothers and sisters of a baby with M/SCHADD have a small chance of having this condition, even if they haven't had symptoms. Finding out whether other children in the family have M/SCHADD may be important because early treatment may prevent serious health problems. Talk to your doctor or genetic counselor about testing your other children for M/SCHADD.

M/SCHADD Carriers

Brothers and sisters who do not have M/SCHADD 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 M/SCHADD, your brothers and sisters have a 50% chance to be a M/SCHADD carrier. It is important for other family members to be told that they could be carriers. There is a very small chance they are also at risk to have children with M/SCHADD.

Some states do not offer newborn screening for M/SCHADD. However, expanded newborn screening through private labs is available for babies born in states that do not screen for M/SCHADD. Your healthcare provider or genetic counselor can help you obtain expanded newborn screening.

When both parents are M/SCHADD carriers, newborn screening results are not sufficient to rule out M/SCHADD in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

During pregnancy, women carrying fetuses with M/SCHADD 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 M/SCHADD 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 M/SCHADD by DNA testing using a blood sample.

Carrier Testing

Carrier testing for M/SCHADD may be available. Ask your metabolic doctor or genetic counseling whether carrier testing is possible.

How many people have M/SCHADD?

M/SCHADD is very rare. The actual incidence is unknown.

Does M/SCHADD happen more often in a certain ethnic group?

M/SCHADD does not happen more often in any specific race, ethnic group, geographical area, or country.

Does M/SCHADD go by any other names?

M/SCHADD is sometimes also called:

- SCHAD deficiency
- SCHADD
- HADHSC deficiency
- HADH deficiency
- HAD deficiency
- L-3-alpha-hydroxyacyl-CoA dehydrogenase, short chain, 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
https://ghr.nlm.nih.gov/condition/3-hydroxyacyl-coa-dehydrogenase-deficiency

Baby's First Test
http://www.babysfirsttest.org

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