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: Carnitine acylcarnitine translocase deficiency
## Acronym: CAT



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

## What is CAT deficiency?

CAT deficiency stands for “carnitine acylcarnitine translocase deficiency.” It is one type of **fatty acid oxidation disorder**. People with CAT deficiency have problems breaking down fat into energy for the body.

**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.

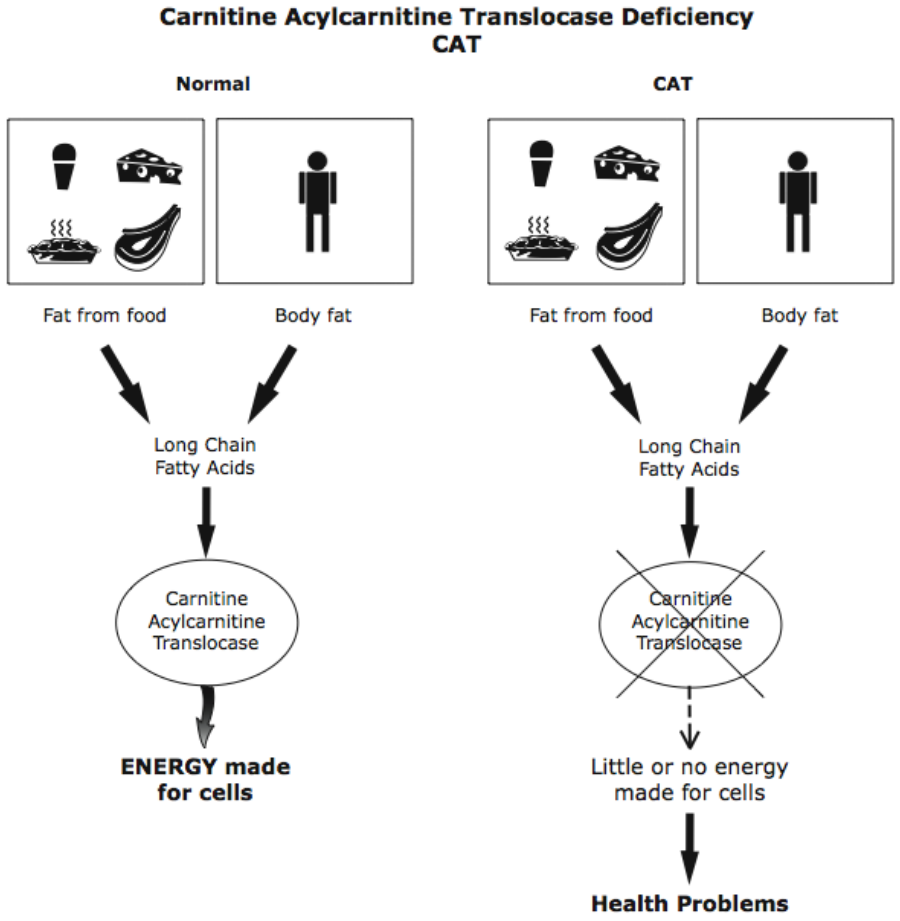
A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

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 CAT deficiency?

CAT deficiency occurs when an enzyme, called “carnitine acylcarnitine translocase” (CAT), is either missing or not working properly. This enzyme’s job is to help change certain fats in the food we eat into energy. It also helps to break 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 for energy when we don't eat for a stretch of time – like when we miss a meal or when we sleep.

When the CAT enzyme is missing or not working, the body cannot use fat for energy, and must rely solely on glucose. Although glucose is a good source of energy, there is a limited amount available. Once the glucose has been used up, the body tries to use fat without success. This leads to low blood sugar, called hypoglycemia, and to the build up of harmful substances in the blood.

## If CAT deficiency is not treated, what problems occur?

CAT deficiency can cause episodes of illness called metabolic crises.
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
- hypoglycemia

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

- muscle weakness
- seizures
- breathing problems
- coma, sometimes leading to death

There are two types of CAT deficiency. The most common type happens in newborns. A milder, less common type happens in older infants and children.

### CAT deficiency in newborns

Newborns with CAT deficiency often show symptoms within the first week of life. Episodes of metabolic crisis are common. High levels of ammonia in the blood can occur. This can cause serious brain damage.

Newborns with CAT may also have:

- low muscle tone (floppy muscles and joints) and muscle weakness
- enlarged liver
- heart problems and enlarged heart
- breathing problems

Babies who are not treated usually die of heart problems, breathing problems, liver failure, or high levels of ammonia in the blood.

### CAT deficiency in children

Children with the mild type of CAT deficiency usually start having symptoms before age three. They are at risk to have episodes of metabolic crisis, but usually do not have heart problems.

In both types of CAT deficiency, symptoms often happen after having nothing to eat for more than a few hours. Symptoms are also more likely when a person with CAT deficiency gets sick or has an infection.

## What is the treatment for CAT deficiency?

Your baby's primary doctor will work with a metabolic doctor to care for your child. Your doctor may also suggest that you meet with a dietician familiar with CAT deficiency.

Certain treatments may be helpful for some children but not others. When necessary, treatment is usually needed throughout life. The following are treatments sometimes recommended for children with CAT deficiency:

**1. Avoid going a long time without food**

Infants and young children with CAT deficiency 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 be fed every four to six hours. Some babies need to eat even more frequently than this. It is important that infants be fed during the night. They may need to be woken up to eat if they do not wake up on their own. Your metabolic doctor and dietician will give you an appropriate feeding plan for your infant. Your doctor will 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.

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, many teens and adults with CAT deficiency can go without food for up to 12 hours without problems. The other treatments usually need to be continued throughout life.

**2. Diet**

Sometimes a low-fat, high carbohydrate diet is advised. In fact, for children needing this treatment, most foods in the diet should be carbohydrates (bread, pasta, fruit, vegetables, etc.) and protein (lean meat and low-fat dairy food).

People with CAT deficiency cannot use particular building blocks of fat called "long chain fatty acids." A dietician can help you create a food plan low in these fats. Any diet changes should be made under the guidance of a dietician experienced with CAT deficiency.

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

**3. Medium Chain Triglycerides**

Medium Chain Triglycerides (MCT) are often used as part of the food plan for people with CAT deficiency. MCTs are available in MCT oil and in triheptanoin (DOJOLVI ®) - a medication made of MCTs. These supplements have medium chain fatty acids that can be used in small amounts for energy. Your metabolic doctor or dietician can tell you how to use these supplements. You will need to get a prescription from your doctor to get these MCTs supplements.

**4. L-Carnitine**

Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps body cells make 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. Do not use L-carnitine without checking with your doctor.

**5. Call your doctor at the start of any illness**

Always 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

Children with CAT deficiency 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 CAT deficiency may need to be treated in the hospital to prevent a metabolic crisis. Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

## What happens when CAT deficiency is treated?

Prompt and careful treatment may help prevent or control symptoms in children with CAT deficiency. However, some children continue to have metabolic crises and other health problems despite treatment. Even with treatment, there is a risk of death, especially in newborns with symptoms.

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

Genes tell the body to make various enzymes. The SLC25A20 gene instructs the body to make the enzyme carnitine-acylcarnitine translocase (CAT enzyme). Everyone has two copies of the SLC25A20 gene. People with CAT deficiency have changes, also called variants in both copies of their SLC25A20 genes that cause them to not work correctly. Because of the variants in the SLC25A20 genes, the CAT enzyme either does not work properly or is not made at all.

## How is CAT deficiency inherited?

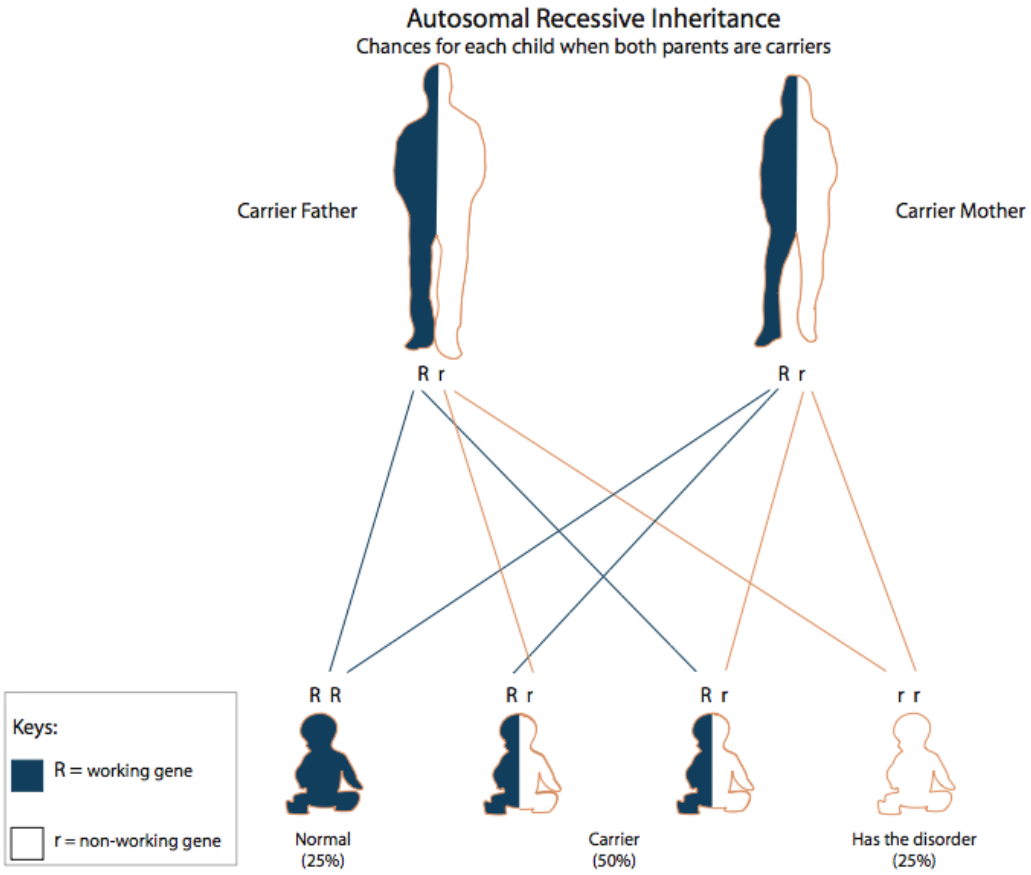
CAT deficiency is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has two copies of the SLC25A20 gene that makes the CAT enzyme. In children with CAT deficiency, neither of these genes works correctly. These

children inherit one non-working SLC25A20 gene for the condition from each parent.

Parents of children with CAT deficiency rarely have the disorder. Instead, each parent has a single non-working SLC25A20 gene for CAT deficiency. They are called carriers. Carriers do not have the condition because their other SLC25A20 gene is working correctly.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CAT deficiency. 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 CAT deficiency. Genetic counselors can answer your questions about how CAT deficiency is inherited, options 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 CAT deficiency can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes (variants) in the SLC25A20 genes that cause CAT deficiency.

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?

CAT deficiency can also be confirmed by a special enzyme test on a skin sample. Talk to your doctor or genetic counselor if you have questions about testing for CAT deficiency.

## Can you test during a future pregnancy?

If both gene changes (variants) have been found in the child with CAT deficiency, 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 special tests 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 CAT deficiency. 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 CAT deficiency or be carriers?

### CAT deficiency

The brothers and sisters of an affected baby have a chance of having CAT deficiency, even if they haven't had symptoms. Finding out whether other children in the family have CAT deficiency is important because early treatment may prevent serious health problems. Talk to your doctor or genetic counselor about testing your other children.

### CAT deficiency carriers

Brothers and sisters who do not have CAT deficiency 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 CAT deficiency, your brothers and sisters have a 50% chance to be a 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 CAT deficiency.

Some states do not offer newborn screening for CAT deficiency. 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 CAT deficiency in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

During pregnancy, women carrying fetuses with CAT deficiency 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 CAT deficiency 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 for CAT deficiency

Brothers and sisters of an affected child can be tested for CAT deficiency using either DNA testing or a special enzyme test to determine if they also have this disorder.

### Carrier testing

If both gene changes in the SLC25A20 genes have been found in the child with CAT deficiency, 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 CAT deficiency?

CAT deficiency is very rare. The incidence in the United States in thought to be between 1 in 750,000 to 1 in 2,000,000. The incidence in Asian is thought to be between 1 in 60,000 and 1 in 400,000.

## Does CAT deficiency happen more often in a certain ethnic group?

No, CAT deficiency does not happen more often in any specific race, ethnic group, geographical area, or country.

## Does CAT deficiency go by any other names?

CAT deficiency is also known as:

- carnitine acylcarnitine carrier (CAC)
- CACT 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

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

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

MedlinePlus
https://medlineplus.gov/genetics/condition/carnitine-acylcarnitine-translocase-deficiency/

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| **Document Info** | **Created by:** | www.newbornscreening.info |
| | **Reviewed by:** | HI, CA, OR, and WA metabolic specialists |
| | **Review date:** | September 31, 2007<br>June 30, 2013<br>April 24,2020<br>March 1, 2023 |
| | **Updated on:** | March 1, 2023 |



U.S. Department of Health and Human Services HRSA Health Resources and Services Administration Maternal and Child Health Bureau

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