



## 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 palmitoyl transferase deficiency, type 2

**Acronym:** CPT II deficiency

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

## What is CPT II deficiency?

CPT II deficiency stands for “carnitine palmitoyl transferase deficiency – type 2.” It is one type of fatty acid oxidation disorder. People with CPT II 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.

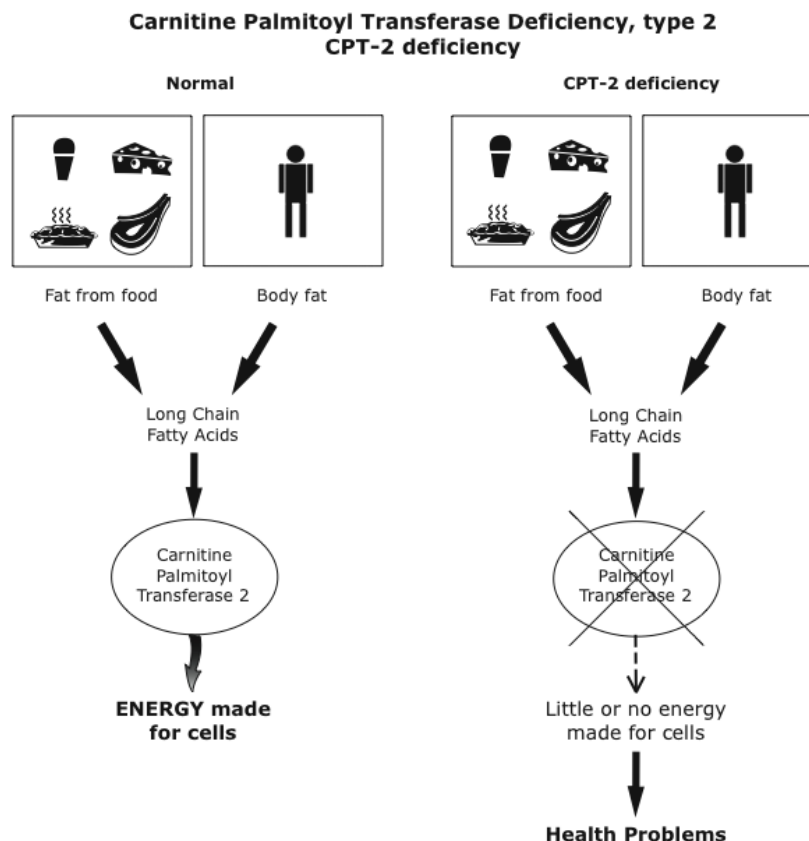
A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

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 CPT II deficiency?**

CPT II deficiency is caused by problems with an enzyme called “carnitine palmitoyl transferase 2” (CPT2). In people with CPT II deficiency, the carnitine palmitoyltransferase 2 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. 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.

When the normal 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 CPT II deficiency is not treated, what problems occur?**

A small number of babies with CPT II deficiency show symptoms shortly after birth. Others don't start showing the effects until later in infancy. Most people do not have symptoms until their teen years or early adulthood. This is called the "classic" type of CPT II deficiency.

Babies and children who show early signs of CPT II deficiency have episodes of illness called metabolic crisis. Some of the first signs 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 CPT II deficiency can develop:

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

### **CPT II deficiency in newborns**

Rarely, a baby with CPT II deficiency shows symptoms in the newborn period. Newborns with symptoms usually have many episodes of metabolic crisis. Other health effects in newborns can be:

- enlarged heart with irregular heartbeat
- enlarged liver
- muscle weakness

Some babies are also born with:

- kidney cysts
- cataracts
- defects of the brain

Without treatment, babies who have symptoms in the newborn period usually die very early in life.

### **CPT II deficiency in infants and children**

A small number of children with CPT II deficiency start showing symptoms in late infancy or early childhood. If untreated, some of the effects can be:

- repeated metabolic crises
- learning problems
- delays in walking and other motor skills
- liver problems
- muscle weakness
- enlarged heart and irregular heartbeat
- kidney problems

Without treatment, many children with CPT II deficiency die early in life.

Symptoms of metabolic crisis in babies and children with CPT II deficiency often happen after having nothing to eat for more than a few hours. Symptoms are also more likely when a baby or child with CPT II deficiency gets sick or has an infection.

Some children with CPT II deficiency have never had a metabolic crisis or other related health problems and are only found to be affected after a brother or sister has been diagnosed.

### **CPT II deficiency in adults**

Most people with CPT II deficiency have “classic” CPT II deficiency, or the “muscle” type.

Symptoms of classic CPT II deficiency usually start between 15 and 30 years of age. Periods of muscle weakness are common. Breakdown of muscle fibers can happen. Symptoms usually happen during heavy exercise or after going without food for a long period of time. Signs of muscle breakdown include:

- muscle aches
- muscle weakness
- cramps
- reddish-brown color to the urine

Adults who are not treated can develop kidney failure.

Classic CPT II deficiency does not cause metabolic crises or heart problems.

## What is the treatment for CPT II 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 CPT II deficiency.

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

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

Infants and young children with CPT II 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 CPT II 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 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 an experienced dietician.

Children with CPT II deficiency cannot use certain building blocks of fat called "long chain fatty acids." Your dietician can help create a food plan low in these fats. Much of the rest of the fat in the diet will likely be in the form of medium chain fatty acids.

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 CPT II 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-Carninite**

Some children and adults 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 any supplements without checking with your doctor.

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

Always call your health care provider right away when your child has any of the following:

- poor appetite
- low energy or excessive sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- persistent muscle pain, weakness, or reddish-brown color to the urine

Babies and children with CPT II deficiency need to eat extra starchy food and drink more fluids during any illness - even if they may not feel hungry – or they could have a metabolic crisis. Children who are sick often don't want to eat. If they won't or can't eat, they 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.

#### **6. Avoid long periods of exercise and avoid getting cold**

Long periods of strenuous exercise can trigger symptoms in both children and adults. So can cold weather. Muscle effects can include:

- muscle aches
- cramps
- weakness
- reddish-brown urine

If muscle symptoms occur, prompt treatment is needed to prevent kidney damage. Children or adults with muscle symptoms should:

- drink fluids right away
- eat something starchy or sugary
- get to a hospital for treatment

To help prevent muscle symptoms:

- avoid prolonged or heavy exercise

- keep the body warm
- eat starchy or sugary foods before and during periods of moderate exercise

## **What happens when CPT II deficiency is treated?**

Treatment can help prevent or control symptoms in some children with CPT II deficiency. Children who are treated early may be able to live healthy lives with typical growth and development. Some children do continue to have episodes of metabolic crisis and other health problems despite treatment.

When treated, adults with the classic form of CPT II deficiency are expected to live healthy lives.

## **What causes the CPT2 enzyme to be absent or not working correctly?**

Genes tell the body to make enzymes. The CPT2 gene instructs the body to make the carnitine palmitoyltransferase 2 enzyme. Everyone has two copies of the CPT2 gene. People with CPT II deficiency have changes, also called variants in both copies of their CPT2 genes that cause them to not work correctly. Because of the variants in the CPT2 genes, the CPT2 enzyme either does not work properly or is not made at all.

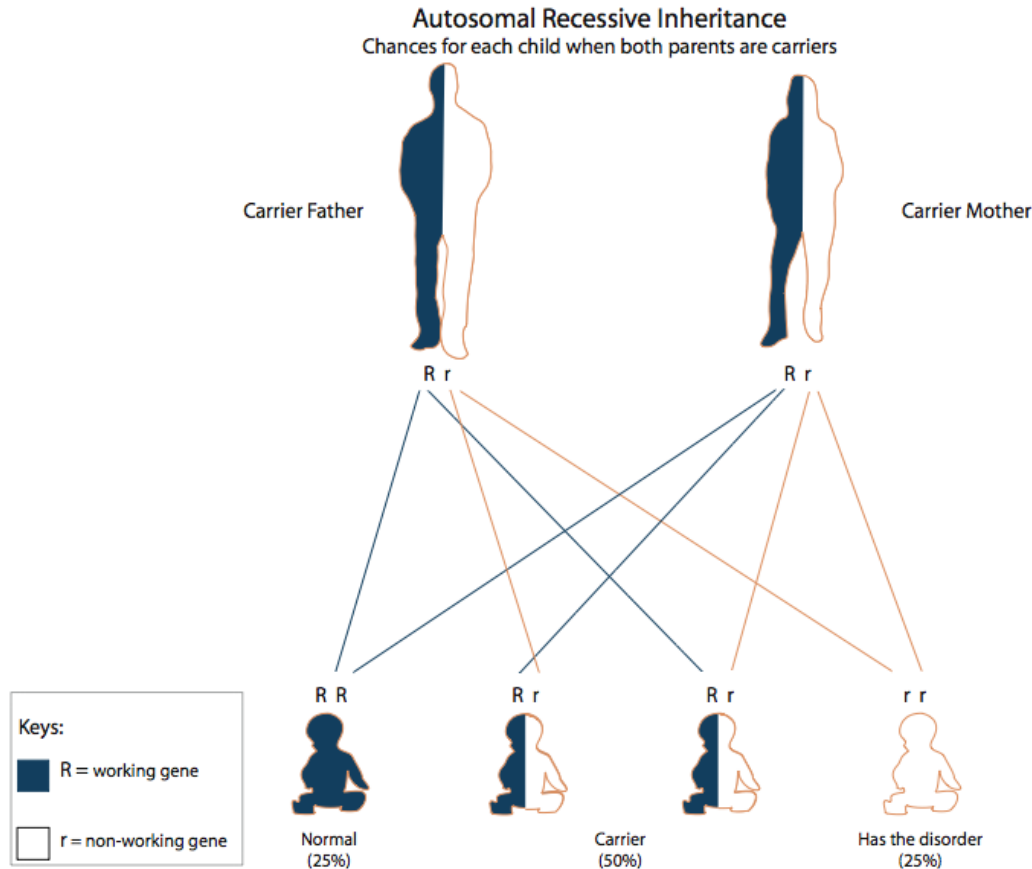
## **How is CPT II deficiency inherited?**

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

Everyone has two copies of the CPT2 gene that make the CPT2 enzyme. In children with CPT II deficiency, neither of these genes works correctly. These children inherit one non-working CPT2 gene for the condition from each parent.

Parents of children with CPT II deficiency are rarely affected with the disorder. Instead, each parent has a single non-working CPT2 gene. They are called carriers for CPT II deficiency. Carriers do not have CPT II deficiency because their other gene is working correctly. In most cases, carriers of CPT II deficiency do not have any symptoms of the disease. However, in a small number of cases, carriers of CPT II deficiency have been reported with symptoms of the disorder.

When both parents are carriers, there is a 25% chance in each pregnancy for the child to have CPT II 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 inherit two working genes.



Genetic counseling is available to families who have children with CPT II deficiency. Genetic counselors can answer your questions about how CPT II deficiency 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 CPT II deficiency can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes, also called variants, in the CPT II genes. In some 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 is not necessary to diagnose your child. It can be helpful for carrier testing or prenatal diagnosis, discussed below.



## What other testing is available?

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

## Can you test during a future pregnancy?

If both gene changes have been found in your child with CPT II 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, an enzyme test can be done on cells from the fetus. Again, the sample needed for this test is obtained by either CVS or amniocentesis.

Parents may 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 CPT II deficiency. A genetic counselor can talk with you about your choices and answer questions about prenatal testing or testing your baby after birth.

## Can other members of the family have CPT II deficiency or be carriers?

### Having CPT II deficiency

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

### CPT II deficiency carriers

Brothers and sisters who do not have CPT II 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 CPT II deficiency, your brothers and sisters have a 50% chance to be a CPT II deficiency carrier. It is important for other family members to be told that they could be carriers. There is a chance they are also at risk to have children with CPT II deficiency. Most commonly, carriers of CPT II deficiency do not have any symptoms of the disease. However, a small number of carriers of CPT II deficiency have been reported to have symptoms of this disorder.

Some states do not offer newborn screening for CPT II deficiency. However, expanded newborn screening through private labs is available 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 CPT II 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 CPT II 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 CPT II 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**

Brothers and sisters of an affected child can be tested for CPT II deficiency using either a DNA test or a special enzyme test.

### **Carrier Testing**

If both gene changes in the CPT2 genes have been found in the child with CPT II 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 CPT II deficiency?**

CPT II deficiency is rare. The actual incidence is unknown.

## **Does CPT II deficiency happen more often in a certain ethnic group?**

CPT II deficiency does not happen more often in any specific race, ethnic group, geographical area, or country.

## **Does CPT II deficiency go by any other names?**

CPT II deficiency is also called:

- CPT-2 deficiency
- Carnitine palmytoyltransferase 2 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>

Baby's First Test

<http://www.babysfirsttest.org>

MedlinePlus

<https://medlineplus.gov/genetics/condition/carnitine-palmitoyltransferase-ii-deficiency/>

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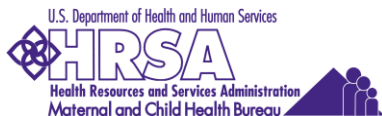
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