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 1A
Acronym: CPT1A

This fact sheet contains general information about CPT1A deficiency. Every child is different and some of this information may not apply to your child specifically. Not all is known about CPT1A deficiency and, at present, there is no standard treatment plan. Certain treatments may be recommended for some children but not others. Children with CPT1A deficiency should be followed by a metabolic doctor in addition to their primary doctor.

What is CPT1A deficiency?

CPT1A deficiency stands for “carnitine palmitoyl transferase - type 1A deficiency.” It is one type of fatty acid oxidation disorder. People with CPT1A 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.

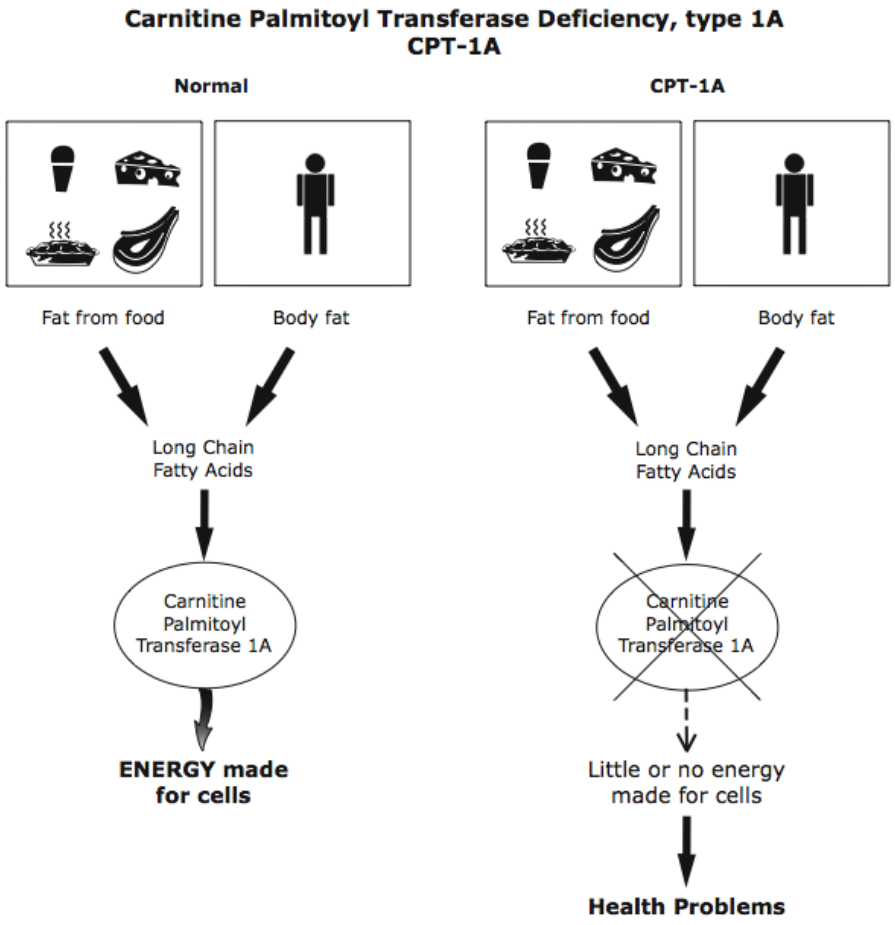
A number of enzymes are needed to break down fats in the body (a process called fatty acid oxidation). Problems with any of these enzymes can cause a fatty acid oxidation disorder. People with FAODs cannot properly break down fat from either the food they eat or from fat stored in their bodies.

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 CPT1A deficiency?

CPT1A deficiency is caused by problems with the enzyme, “carnitine palmitoyl transferase 1A” (CPT1A). In people with CPT1A deficiency, the CPT1A 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 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 when we don't eat for a stretch of time – like when we miss a meal or when we sleep.

When the CPT1A 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 CPT1A deficiency is not treated, what problems occur?

CPT1A deficiency can cause episodes of illness caused metabolic crises. Children with CPT1A deficiency usually start showing symptoms between the ages of 8 and 18 months, although effects can occur earlier. 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
- high levels of ammonia in the blood

If a metabolic crisis is not treated, a child with CPT1A deficiency can develop:
- breathing problems
- seizures
- coma, sometimes leading to death

Between episodes of metabolic crisis, people with CPT1A deficiency are usually healthy. However, repeated episodes may cause brain damage that can result in learning problems or intellectual disabilities.

Symptoms often happen after having nothing to eat for more than a few hours. During long periods without eating, the glucose in the body is used up. This causes hypoglycemia. The body then tries to use fat for energy, leading to the build up of harmful substances in the blood. Symptoms are also more likely when a person with CPT1A deficiency gets sick or has an infection. Prompt emergency treatment of infants and children with CPT1A can help prevent metabolic crises or lessen their severity.

Babies and children who are not treated can have:
- learning problems
- delays in walking and other motor skills
- liver, heart, or kidney problems

Some children with CPT1A deficiency have never had symptoms and are only found to be affected after a brother or sister has been diagnosed. CPT1A deficiency in people of Inuit (Native American Alaskan) or First Nation (Canada) ethnicity very rarely causes symptoms.

What is the treatment for CPT1A 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 CPT1A 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 children with CPT1A deficiency:

1. Avoid going a long time without food
Infants and young children with CPT1A 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. 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 CPT1A 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 a dietician experienced with CPT1A deficiency.

People with CPT1A 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 fat in the diet will likely be in the form of medium chain fatty acids.

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

3. Medium Chain Triglyceride oil (MCT)
Medium Chain Triglycerides (MCT) are often used as part of the food plan for people with CPT1A 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. 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

Children with CPT1A deficiency need to eat extra starchy food and drink more fluids than usual when they are sick – 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 serious health problems. Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

What happens when CPT1A deficiency is treated?

With prompt and careful treatment, children with CPT1A deficiency often live healthy lives with typical growth and development. After 5 years of age, metabolic crises tend to happen less often and are not as severe.

If repeated episodes of metabolic crisis occur, there is a chance for permanent learning disabilities or intellectual disabilities.

What causes the CPT1A enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. The CPT1A gene instructs the body to make the CPT1A enzyme. Everyone has two copies of the CPT1A gene. People with CPT1A deficiency have changes, also called variants in both copies of their CPT1A genes that cause them to not work correctly. Because of the variants in the CPT1A genes, the CPT1A enzyme either does not work properly or is not made at all.

How is CPT1A deficiency inherited?

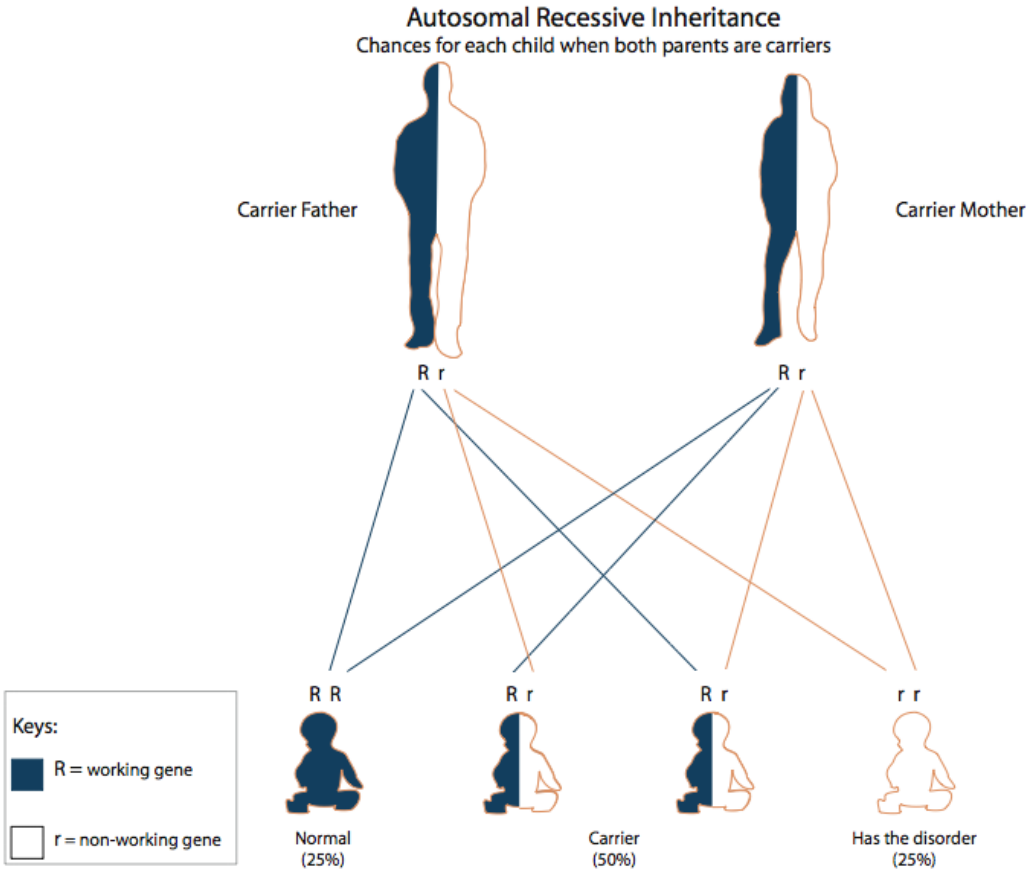
CPT1A deficiency is inherited in an autosomal recessive manner. It affects both boys and girls equally.

Everyone has two copies of the CPT1A gene that make the CPT1A enzyme. In children with CPT1A deficiency, neither of their CPT1A genes works correctly. These children inherit one non-working CPT1A gene from each parent.

Parents of children with CPT1A deficiency are rarely affected with the disorder. Instead, each parent has a single non-working CPT1A gene for CPT1A deficiency. They are called carriers. Carriers do not have the condition because their other CPT1A gene is working correctly.

When both parents are carriers for CPT1A deficiency, there is a 25% chance in each pregnancy for the child to have CPT1A 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 CPT1A deficiency. Genetic counselors can answer your questions about how CPT1A 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 CPT1A deficiency can be done on a blood sample. Genetic testing, also called DNA testing, looks for changes (variants) in the CPT1A genes that cause the condition. In most affected children, both gene changes can be found. However, in some 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. Talk with your metabolic doctor or genetic counselor if you have questions about DNA testing for CPT1A deficiency.

What other testing is available?

CPT1A deficiency can be confirmed by a special enzyme test on a skin sample. Your doctor or genetic counselor can answer your questions about testing for CPT1A deficiency.

Can you test during a future pregnancy?

If both gene changes (variants) have been found in your child with CPT1A 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 done by an enzyme test on cells from the fetus. 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 CPT1A deficiency. A genetic counselor can talk to you about your choices and answer questions about prenatal testing or testing your baby after birth.

Can other members of the family have CPT1A deficiency or be carriers?

Having CPT1A deficiency

The brothers and sisters of an affected baby have a chance of having CPT1A deficiency, even if they haven't had symptoms. Finding out whether other children in the family have CPT1A deficiency is important because early treatment may prevent serious health problems. Talk to your doctor or genetic counselor about testing your other children for CPT1A deficiency.

CPT1A deficiency carriers

Brothers and sisters who are not affected with CPT1A 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 CPT1A 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 CPT1A deficiency.

Some states do not offer newborn screening for CPT1A 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 adequate to rule out CPT1A 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 CPT1A 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 CPT1A 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 CPT1A deficiency

To make sure they do not have the condition, brothers and sisters of a child with CPT1A deficiency can have special tests done on a skin sample. DNA testing may also be done if both variants have been identified in the affected sibling.

Carrier testing for CPT1A deficiency

Carrier testing may be available to other family members. Ask your metabolic doctor or genetic counselor about carrier testing for family members.

How many people have CPT1A deficiency?

CPT1A deficiency is found in about one in every 1,200 babies in the North American Hutterite community. It is also more common among the Native American Inuit people of Canada and Alaska. CPT1A deficiency is rare in other ethnic groups around the world. The actual incidence is unknown.

Does CPT1A deficiency happen more often in a certain ethnic group?

CPT1A deficiency can be seen in every ethnic group and geographical area. However, it is more common in the North American Hutterite community and also among the Native American Inuit people of Canada and Alaska.

Does CPT1A deficiency go by any other names?

CPT1A deficiency is sometimes also called:

- CPT1 deficiency
- CPT1 deficiency (liver)

There is another type of CPT deficiency called CPT1B, or “muscle type.” CPT1B is not discussed in this fact sheet.

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-i-deficiency/

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