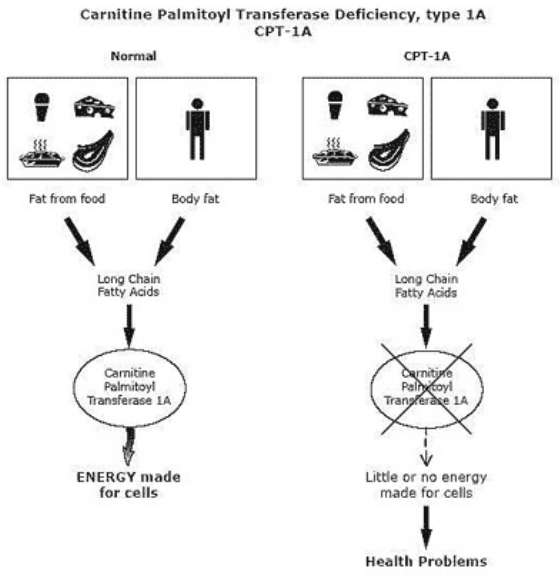
CARNITINE PALMITOYL TRANSFERASE (CPT 1)
A fact sheet for parents/carer

What is CPT-1A deficiency?

CPT-1A deficiency stands for "carnitine palmitoyl transferase - type 1A deficiency". It is one type of fatty acid oxidation disorder. People with CPT-1A deficiency have problems breaking down fat into energy for the body.

What causes CPT-1A?



CPT-1A deficiency occurs when an enzyme, called "carnitine palmitoyl transferase 1A" (CPT-1A) 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 CPT-1A 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-1A deficiency is not treated, what problems occur?

CPT-1A deficiency can cause episodes of illness caused metabolic crises. Children with CPT-1A 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

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- high levels of ammonia in the blood

If a metabolic crisis is not treated, a child with CPT-1A deficiency can develop:

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

Between episodes of metabolic crisis, people with CPT-1A deficiency are usually healthy. However, repeated episodes may cause brain damage that can result in learning problems or mental retardation.

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 CPT-1A 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 CPT-1A deficiency have never had symptoms and are only found to be affected after a brother or sister has been diagnosed.

What is the treatment for CPT-1A deficiency?

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 CPT-1A 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 CPT-1A deficiency:

1. Avoid going a long time without food

Infants and young children with CPT-1A 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 CPT-1A deficiency can

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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 CPT-1A deficiency.

People with CPT-1A 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 Triglyceride oil (MCT oil) is often used as part of the food plan for people with CPT-1A deficiency. This special oil has medium chain fatty acids that can be used for energy. A metabolic doctor or dietician can guide you in how to use this supplement. You will need to get a prescription from your doctor to get MCT oil.

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 CPT-1A 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 CPT-1A deficiency is treated?

With prompt and careful treatment, children with CPT-1A 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.

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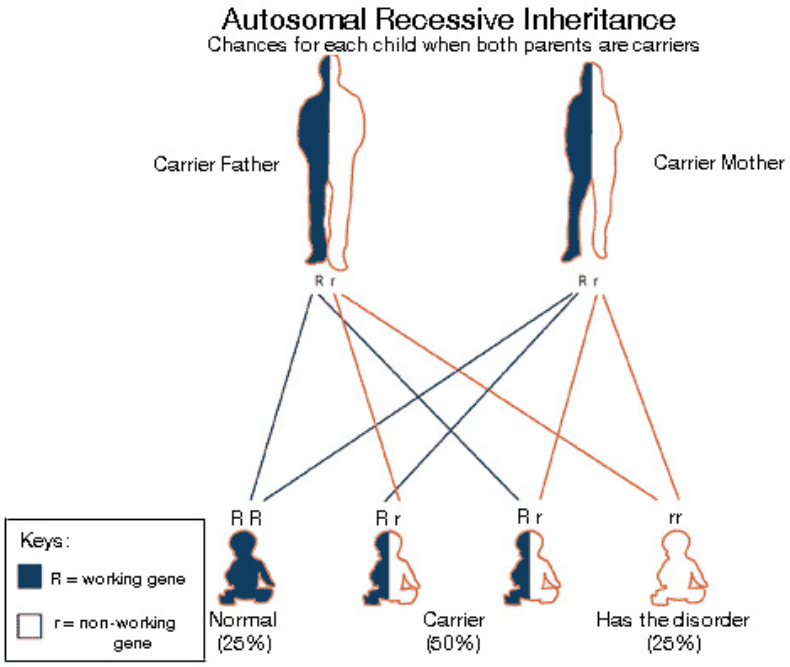
If repeated episodes of metabolic crisis occur, there is a chance for permanent learning disabilities or mental retardation.

What causes the CPT-1A enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. People with SCHADD have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the SCHAD enzyme either does not work properly or is not made at all.

How is CPT-1A deficiency inherited?



CPT-1A deficiency is inherited in an autosomal recessive manner. It affects both boys and girls equally. Everyone has a pair of genes that make the CPT-1A enzyme. In children with CPT-1A deficiency, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent. Parents of children with CPT-1A deficiency are rarely affected with the disorder. Instead, each parent has a single non-working gene for CPT-1A deficiency. They are called carriers. Carriers do not have the condition because the other gene of this pair is working correctly. When both parents are carriers for CPT-1A deficiency, there is a 25% chance in each pregnancy for the child to have CPT-1A 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 CPT-1A deficiency. Genetic counselors can answer your questions about how CPT-1A deficiency is inherited, choices during future pregnancies, and how to test other family members. Ask your doctor about a referral to a genetic counselor.

What other testing is available?

CPT-1A 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 CPT-1A deficiency.

Can other members of the family have CPT-1A or be carriers?

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Having CPT-1A deficiency

The brothers and sisters of an affected baby have a chance of having CPT-1A deficiency, even if they haven't had symptoms. Finding out whether other children in the family have CPT-1A deficiency is important because early treatment may prevent serious health problems. Talk to your doctor or genetic counselor about testing your other children for CPT-1A deficiency.

CPT-1A deficiency carriers

Brothers and sisters who are not affected with CPT-1A 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. Each of the parents' brothers and sisters has a 50% chance to be a CPT-1A deficiency 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 CPT-1A deficiency. Some states do not offer newborn screening for CPT-1A deficiency. However, expanded newborn screening through private laboratories is available for babies born in states that do not screen for this condition.

When both parents are carriers, newborn screening results are not adequate to rule out CPT-1A 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-1A 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-1A 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 CPT-1A deficiency

To make sure they do not have the condition, brothers and sisters of a child with CPT-1a deficiency can have special tests done on a skin sample.

How many people have CPT-1A deficiency?

CPT-1A deficiency is found in about one in every 1200 babies in the North American Hutterite community. It is also more common among the Native American Inuit people of Canada and Alaska. CPT-1A deficiency is rare in other ethnic groups around the world. The actual

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incidence is unknown.

Does CPT-1A deficiency happen more frequently in a certain ethnic group?

CPT-1A 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.

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

Children Living with Inherited Metabolic Diseases (CLIMB)

http://www.climb.org.uk

Genetic Alliance

http://www.geneticalliance.org

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