NORTH DAKOTA
DEPARTMENT of HEALTH

## NEWBORN SCREENING FACT SHEET

# CPT-2 Deficiency
## (Carnitine Palmitoyl Transferase Deficiency, CACT, Type 2)

### What is it?
CPT-2 deficiency stands for carnitine palmitoyl transferase deficiency – type 2. It is one type of fatty acid oxidation disorder. People with CPT-2 deficiency have problems using fat as energy for the body.

### What causes it?
CPT-2 deficiency occurs when an enzyme, called carnitine palmitoyl transferase – type 2 (CPT-2) 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 or 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 buildup of harmful substances in the blood.

### If CPT-2 deficiency is not treated, what problems occur?
A small number of babies with CPT-2 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 classic form of CPT-2 deficiency.

Babies and children who show early signs of CPT-2 deficiency have episodes of illness called metabolic crises. Some of the first signs of a metabolic crisis are:
1) Extreme sleepiness.
2) Behavior changes.
3) Irritable mood.
4) Poor appetite.

Other symptoms then follow:
1) Fever
2) Diarrhea
3) Vomiting
4) Hypoglycemia

If a metabolic crisis is not treated, a child with CPT-2 deficiency can develop:
1) Breathing problems.
2) Seizures.
3) Coma, sometimes leading to death.

#### *CPT-2 Deficiency in Newborns*
Rarely, a baby with CPT-deficiency shows symptoms in the newborn period. Newborns with symptoms usually have many episodes of metabolic crisis. Other health effects in newborns can be:
1) Enlarged heart with irregular heartbeat.
2) Enlarged liver.
3) Muscle weakness.

Some babies are also born with:
1) Kidney cysts.
2) Cataracts.
3) Defects of the brain.

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

### *CPT-2 Deficiency in Infants and Children*

A small number of children with CPT-2 deficiency start showing symptoms in late infancy or early childhood. If untreated, some of the effects can be:
1) Repeated metabolic crises.
2) Learning problems.
3) Delays in walking and other motor skills.
4) Liver problems.
5) Muscle weakness.
6) Enlarged heart and irregular heartbeat.
7) Kidney problems.

**Without treatment many of these children die early in life.**

Symptoms of metabolic crisis in babies and children with CPT-2 deficiency often happen after having nothing to eat for more than a few hours. Symptoms also are more likely when a baby or child with CPT-2 deficiency gets sick or has an infection.

Some children with CPT-2 deficiency have never had a metabolic crisis or other related health problems and are found to be affected only after a brother or sister has been diagnosed.

### *CPT-2 Deficiency in Adults*

Most people with CPT-2 deficiency have the adult form. This is also called classic CPT-2 deficiency or the muscle form.

Symptoms of classic CPT-2 deficiency usually start between ages 15 and 30. 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:
1) Muscle aches.
2) Muscle weakness.
3) Cramps.
4) Reddish-brown color to urine.

Adults who are not treated can develop kidney failure.

Classic CPT-2 deficiency does not cause metabolic crises or heart problems.

## What is the treatment for CPT-2 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-2 deficiency.

Certain treatments may be advised for some children but not others. When necessary, treatment usually is needed throughout life. The following are treatments often recommended for children with CPT-2 deficiency.

### *Avoid Going a Long Time Without Food*

Babies and young children with CPT-2 deficiency need to eat often to avoid having a metabolic crisis. They should not go without food for more than four to six hours. Some babies may need to eat even more often than this. It is important that babies be fed during the night. They need to be awakened to eat if they do not wake up on their own.

Children with CPT-2 deficiency should have a starchy snack before bed and another during the night. They need another snack first thing in the morning. Raw cornstarch mixed with water, milk or other drink is a good source of long-lasting energy. This is sometimes suggested for children older than age 1. Your dietician can give you ideas for good nighttime snacks.

When they are well, most teens and adults with CPT-2 deficiency can go without food for up to 12 hours. They do need to continue the other treatments throughout life.

### *Diet*

Sometimes a low-fat, high carbohydrate diet 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.

Children with CPT-2 deficiency cannot use certain building blocks of fat called long chain fatty acids. A 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.

### *MCT Oil and L-carnitine*

Medium chain triglyceride oil (MCT oil) often is used as part of the food plan for people with CPT-2 deficiency. This special oil has medium chain fatty acids that can be used in small amounts for energy. Your metabolic doctor or dietician can guide you in how to use this supplement.

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.

### *Call Your Doctor at the Start of Any Illness*

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

1) Poor appetite
2) Low energy or excessive sleepiness
3) Vomiting
4) Diarrhea
5) An infection
6) A fever
7) Persistent muscle pain or weakness

Babies and children with CPT-2 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 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.

### *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:

1) Muscle aches.
2) Cramps.
3) Weakness.
4) Reddish-brown urine.

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

1) Drink fluids right away.
2) Eat something starchy or sugary.
3) Get to a hospital for treatment.

To help prevent muscle symptoms:

1) Avoid prolonged or heavy exercise.
2) Keep the body warm.
3) Eat starchy or sugary food before and during periods of moderate exercise.

## What happens when CPT-2 deficiency is treated?

Treatment can help prevent or control symptoms in some children with CPT-2 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-2 deficiency are expected to live healthy lives.

## What causes the CPT-2 enzyme to be absent or not working correctly?

Genes tell the body to make various enzymes. People with CPT-2 deficiency have a pair of genes that do not work correctly. Because of the changes in this pair of genes, the CPT-2 enzyme does not work properly or is not made at all.

## Is CPT-2 deficiency inherited?

CPT-2 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-2 enzyme. In children with CPT-2 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-2 deficiency are rarely affected with the disorder. Instead, each parent has a single nonworking for CPT-2 deficiency. They are called carriers for CPT-2 deficiency. Carriers do not have CPT-2 deficiency because the other gene of this pair is working correctly.

When both parents are carriers, there is a 25 percent chance in each pregnancy for the child to have CPT-2 deficiency. There is a 50 percent chance for the child to be a carrier, just like the parents. And, there is a 25 percent chance for the child to have two working genes.

## Can other members of the family have CPT-2 deficiency or be carriers?

### *Having CPT-2 Deficiency*

The brothers and sisters of a baby with CPT-2 deficiency have a chance of being affected, even if they haven't had symptoms. Finding out whether any other children in the family have CPT-2 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-2 deficiency.

### *CPT-2 Deficiency Carriers*

Brothers and sisters who do not have CPT-2 deficiency still have a chance to be carriers like their parents. Except in special cases, carrier testing should be done only in people older than 18.

Each of the parents' brothers and sisters has a 50 percent chance to be a CPT-2 deficiency carrier. It is important for other family members to be told that they could be carriers. There is a small chance they are also at risk to have children with CPT-2 deficiency.

When both parents are carriers, newborn screening results are not sufficient to rule out CPT-2 deficiency in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening.

## Can other family members be tested?

### *Diagnostic Testing*

Brothers and sisters of an affected child can be tested for CPT-2 deficiency using either a DNA test or a special enzyme test.

### *Carrier Testing*

If both gene changes have been found in the child with CPT-2 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 may people have CPT-2 Deficiency?

CPT-2 deficiency is rare. The actual incidence is unknown.

**Does CPT-2 deficiency happen more frequently in a certain ethnic group?**
CPT-2 deficiency does not happen in any specific race, ethnic group, geographical area or country.

**Does CPT-2 deficiency go by any other names?**
CPT-2 deficiency is sometimes also called CPT-II 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

Children Living with Inherited Metabolic Diseases (CLIMB)
www.climb.org.uk

Genetic Alliance
www.geneticalliance.org

Children's Special Health Services (CSHS)
State Capitol Judicial Wing
600 E. Boulevard Ave., Department 301
Bismarck, ND 58505-0269
Toll Free: 800.755.2714
701.328.2436
Relay TDD: 701.328.3975
CSHS website: www.ndhealth.gov/CSHS
North Dakota Department of Health website:
www.ndhealth.gov

Family support resources available from CSHS:
- Guidelines of Care Info
- Family Support Packet
- Financial Help Packet
- Insurance Fact Sheet

***Family Resources***

Family to Family Network
Center for Rural Health
University of North Dakota
School of Medicine and Health Sciences
P.O. Box 9037
Grand Forks, ND 58202-9037
Toll Free: 888.434.7436
701.777.2359
Fax: 701.777.2353
E-mail: NDF2F@medicine.nodak.edu
www.medicine.nodak.edu/crh

Pathfinder Services of ND
Pathfinder Family Center
1600 2nd Ave. SW, Ste. 19
Minot, ND 58701
Toll Free: 800.245.5840
701.837.7500
Relay TDD: 701.837.7501
E-mail: ndpath01@ndak.net
www.pathfinder.minot.com

Family Voices of North Dakota, Inc.
P.O. Box 163
Edgeley, ND 58433
Toll Free: 888.522.9654
701.493.2634
Fax: 701.493.2635
www.geocities.com/ndfv

**This fact sheet has general information. Every child is different and some of these facts may not apply to your child specifically. Certain treatments may be recommended for some children but not others. All children should be followed by a metabolic doctor in addition to their primary-care provider.

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## Disclaimer

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