



CTD/CUD (Carnitine Transporter Deficiency) (Carnitine Uptake Deficiency)

What is it?

CTD stands for carnitine transporter deficiency. It is one type of fatty acid oxidation disorder. People with CTD have problems using fat as energy for the body.

What causes it?

CTD occurs when an enzyme called carnitine transporter (CT) is either missing or not working properly. This enzyme's job is to carry a substance called carnitine into our cells. Carnitine helps the body make energy from the fat in food. 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 CT enzyme is missing or not working well, the body cannot use fat for energy. Instead, it 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 CTD is not treated, what problems occur?

There are two main forms of CTD: one begins in infancy, the other in childhood.

CTD in Infants

Babies with CTD first show symptoms between birth and age 3. CTD can cause bouts of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are:

- 1) Extreme sleepiness.
- 2) Behavior changes.
- 3) Irritable mood.
- 4) Poor appetite.

Other symptoms then follow:

- 1) Fever
- 2) Nausea
- 3) Diarrhea
- 4) Vomiting
- 5) Hypoglycemia

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

- 1) Breathing problems.
- 2) Swelling of the brain.
- 3) Seizures.
- 4) Coma, sometimes leading to death.

Babies who are not treated may have other effects:

- 1) Enlarged heart
- 2) Enlarged liver
- 3) Muscle weakness
- 4) Anemia

Repeated episodes of metabolic crisis can cause brain damage. This can result in learning problems or mental retardation.

Symptoms of a metabolic crisis often happen after having nothing to eat for more than a few hours. Symptoms are also more likely when a child with CTD gets sick or has an infection.

CTD in Children

Children with CTD appear perfectly normal until symptoms begin, usually between the ages of 1 and 7. Some of the effects of childhood CTD are:

- 1) Enlarged heart.
- 2) Muscle weakness.
- 3) If left untreated, risk of heart failure and death.

Children with this type of CTD do not have episodes of hypoglycemia or metabolic crises. Their intelligence is not affected.

Some children with CTD deficiency never have symptoms and are found to be affected only after a brother or sister is diagnosed.

What is the treatment for CTD?

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 CTD.

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 CTD:

L-Carnitine

The main treatment for CTD is lifelong use of L-carnitine. This is a safe and natural substance that helps the body cells make energy. It also helps the body get rid of harmful wastes. L-carnitine can reverse the heart problems and muscle weakness that happen in children with CTD.

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.

Avoid Going a Long Time Without Food

Babies and young children with CTD need to eat often to avoid problems. 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 CTD should have a starchy snack before bed and another during the night. They may 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, if needed.

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

Diet

Sometimes, in addition to L-carnitine treatment, a low-fat, high-carbohydrate diet is recommended. Any diet changes should be made under the guidance of a dietician. Ask your doctor whether your child needs to have any changes in his or her diet.

If Your Baby Has CTD, Call Your Doctor at the Start of Any Illness

Always call your health-care provider when your baby 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 with CTD 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 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 CTD is treated?

With prompt and careful treatment, children with CTD usually live healthy lives with typical growth and development. Treatment with L-carnitine can often reverse heart enlargement and muscle weakness.

Babies with CTD who have repeated episodes of metabolic crisis may have permanent brain damage. This can cause learning disabilities or mental retardation.

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

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

How is CTD inherited?

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

Everyone has a pair of genes that make the CT enzyme. In children with CTD, neither of these genes works correctly. These children inherit one nonworking gene for the condition from each parent.

Parents of children with CTD rarely are affected with the disorder. Instead, each parent has a single nonworking gene for CTD. They are called carriers. Carriers do not have CTD 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 CTD. 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 CTD or be carriers?

Having CTD

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

CTD Carriers

Brothers and sisters who do not have CTD 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 CTD 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 CTD.

When both parents are carriers, newborn screening results are not sufficient to rule out the condition 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 for CTD

Brothers and sisters of a baby with CTD can be tested using a special test done on a skin sample.

CTD Carrier Testing

Carrier testing may be available to other family members. Ask your metabolic doctor or genetic counseling if you have questions about carrier testing.

How may people have CTD?

About one in every 40,000 babies in the United States is born with CTD.

Does CTD happen more frequently in a certain ethnic group?

CTD has not been found to occur more often in any particular race, ethnic group, geographical area or country.

Does CTD go by any other names?

CTD is sometimes also called:

- 1) System carnitine deficiency (SCD).
- 2) Primary carnitine deficiency.
- 3) Carnitine uptake defect (CUD).

Where can I find more information?

Fatty Oxidation Disorders (FOD) Family Support Group
www.fodsupport.org

Organic Acidemia Association
www.oaanswers.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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