



LCHADD

(Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency)

What is it?

LCHADD stands for long chain 3-hydroxyacyl-CoA dehydrogenase deficiency. It is one type of fatty acid oxidation disorder. People with LCHADD have problems breaking down fat into energy for the body.

What causes it?

LCHADD occurs when an enzyme called long chain 3-hydroxyacyl-CoA dehydrogenase (LCHADD) is either missing or not working properly. This enzyme's job is to break down certain fats from the food we eat into energy. It also breaks 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 normal LCHAD enzyme is missing or not working, the body cannot break down 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 build up of harmful substances in the blood.

If LCHADD is not treated, what problems occur?

LCHADD can cause mild effects in some people and more serious health problems in others. Babies and children with LCHADD

usually begin to show symptoms sometime from birth through age 2. LCHADD causes episodes of hypoglycemia (low blood sugar). The first symptoms of a hypoglycemia are:

- 1) Extreme sleepiness.
- 2) Weakness.
- 3) Nausea.
- 4) Vomiting.
- 5) Irritability or jitteriness.
- 6) Behavior changes.

If hypoglycemia is not treated, a child with LCHADD can develop:

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

Symptoms often happen after having nothing to eat for more than a few hours. Symptoms are also more likely to occur when a person with LCHADD gets sick or has an infection.

Between episodes of hypoglycemia, people with LCHADD are usually healthy. However, repeated episodes can cause brain damage. This can result in learning disabilities or mental retardation.

Babies and children who are not treated may have:

- 1) Poor weight gain.
- 2) Delays in learning.
- 3) Delays in walking and other motor skills.
- 4) Enlarged liver and other liver problems.
- 5) Enlarged heart and other heart problems.
- 6) Vision loss due to build-up of pigment in the retina.

- 7) Anemia.
- 8) Nerve problems.
- 9) Bouts of muscle weakness and pain, especially after heavy exercise or illness.

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

What is the treatment for LCHADD?

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 LCHADD.

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 LCHADD:

Avoid Going a Long Time Without Food

Babies and young children with LCHADD need to eat often to avoid hypoglycemia. 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.

Young children with LCHADD 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 LCHADD can go without food for up to 12 hours without problems. They do need to continue 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.

People with LCHADD 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 may 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, L-Carnitine and other Supplements

Medium chain triglyceride oil (MCT oil) often is used as part of the food plan for people with LCHADD. 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. You will need to get a prescription from your doctor to get MCT oil.

Some children may be helped by taking L-carnitine. This is a safe and natural substance that helps the body make energy. It also helps the body get rid of harmful wastes. Your doctor will decide whether your child needs L-carnitine. Unless you are advised otherwise, use only L-carnitine prescribed by your doctor.

In addition to the above supplements, some doctors suggest taking DHA (docosahexanoic acid), which may help prevent loss of eyesight. Ask your doctor whether your child should use this supplement.

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) Infection
- 6) Fever
- 7) Persistent muscle pain, weakness, or reddish-brown color to urine

Children with LCHADD need to have extra starchy food and drink more fluids during an illness – even if they may not feel hungry – or they could develop hypoglycemia (low blood sugar). Children who are sick often don't want to eat. If they won't or can't eat, children with LCHADD may need to be treated in the hospital to prevent problems.

Ask your metabolic doctor if you should carry a special travel letter with medical instructions for your child's care.

Avoid Prolonged Exercise or Exertion

Long periods of exercise also can trigger symptoms. Effects of exercise may include:

- 1) Muscle aches.
- 2) Weakness.
- 3) Cramps.
- 4) Reddish-brown color to urine (caused by breakdown of muscle fibers).

If muscle symptoms happen, 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 carbohydrates before and during periods of moderate exercise.

What happens when LCHADD is treated?

With prompt and careful treatment, children with LCHADD can often live healthy lives with typical growth and development.

Even with treatment, some people with LCHADD continue to have episodes of hypoglycemia. This can lead to learning problems or mental retardation. And, even with treatment, some people still develop vision, muscle, liver or heart problems.

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

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

Is LCHADD inherited?

LCHADD is inherited and affects both boys and girls equally.

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

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

Having LCHADD

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

LCHADD Carriers

Brothers and sisters who do not have LCHADD 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 LCHADD 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 LCHADD.

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

During pregnancy, women carrying fetuses with LCHADD may be at increased risk to develop serious medical problems. Some of these women develop:

- 1) Excessive vomiting.
- 2) Abdominal pain.
- 3) High blood pressure.
- 4) Jaundice.
- 5) Abdominal fat storage in the liver.
- 6) Severe bleeding.

All women with a family history of LCHADD should share this information with their obstetricians and other health-care providers before and during any future pregnancies.

Knowing about these risks allows early treatment.

Can other family members be tested?

Diagnostic Testing

Brothers and sisters can be tested for LCHADD by DNA testing or other special tests.

Carrier Testing

If both gene changes have been found in the child with LCHADD, other family members can have DNA testing to see if they are carriers.

How may people have LCHADD?

LCHADD is a rare disorder. The actual incidence is unknown.

Does LCHADD happen more frequently in a certain ethnic group?

Although LCHADD happens in every ethnic group, it happens more often in people who have ancestors from Finland.

Does LCHADD go by any other names?

LCHADD is also called LCHAD deficiency

A variant of LCHADD is called trifunctional protein deficiency (TFP). Please see TFP fact sheet for information on this condition.

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.**

Acknowledgement

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Disclaimer

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