



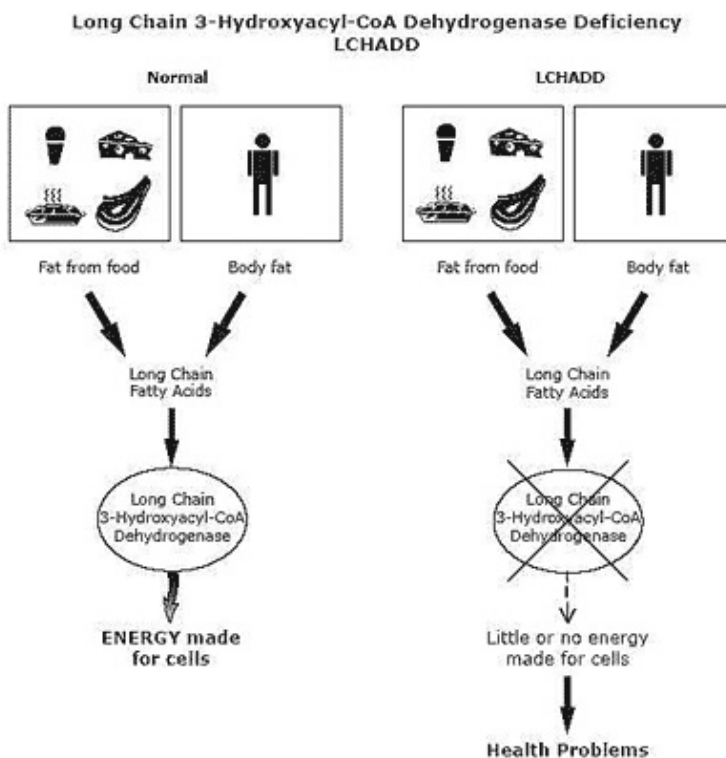
## LONG CHAIN 3-HYDROXYACYL-CoA DEHYDROGENASE DEFICIENCY (LCHADD)

### A fact sheet for parents/carers

#### What is LCHADD?

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 LCHADD?



LCHADD occurs when an enzyme, called “long chain 3-hydroxyacyl-CoA dehydrogenase” (LCHAD), 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 well, 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 two. LCHADD causes episodes of hypoglycemia. The first symptoms of hypoglycemia are:

- extreme sleepiness or fatigue
- weakness





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- nausea
- vomiting
- irritability or jitteriness
- behavior changes

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

- breathing problems
- swelling of the brain
- seizures
- 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:

- poor weight gain
- delays in learning
- delays in walking and other motor skills
- enlarged liver and other liver problems
- enlarged heart and other heart problems
- vision loss due to build-up of pigment in the retina
- anemia
- nerve problems
- bouts of muscle weakness and pain, especially after heavy exercise or illness

Some children with LCHADD have never had symptoms and are only found to be affected 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 is usually needed throughout life. The following are treatments often recommended for children with LCHADD:

#### 1. Avoid going a long time without food

Infants and young children with LCHADD 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. It is important that infants be fed during the night. They may need to be woken up to eat if they do not wake up on their own. 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 LCHADD 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 familiar with LCHADD. 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 the 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. MCT oil, L-carnitine and other supplements**

Medium Chain Triglyceride oil (MCT oil) is often used as part of the food plan for people with LCHADD. This special oil has medium chain fatty acids 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 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 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 or medications without checking with your doctor.

## **4. Call your doctor at the start of any illness**

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

- poor appetite
- low energy or excessive sleepiness
- vomiting
- diarrhea
- an infection
- a fever
- persistent muscle pain or weakness, or reddish-brown color to the urine





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Children with LCHADD need to eat extra starchy food and drink more fluids during any illness - even if they may not feel hungry – or they could develop hypoglycemia. 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.

**5. Avoid prolonged exercise or exertion.**

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

- muscle aches
- cramps
- weakness
- reddish-brown color to the urine (caused by breakdown of muscle fibers)

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

- drink fluids right away
- eat something starchy or sugary
- get to a hospital for treatment

**To help prevent muscle symptoms:**

- avoid prolonged or heavy exercise
- keep the body warm
- 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.

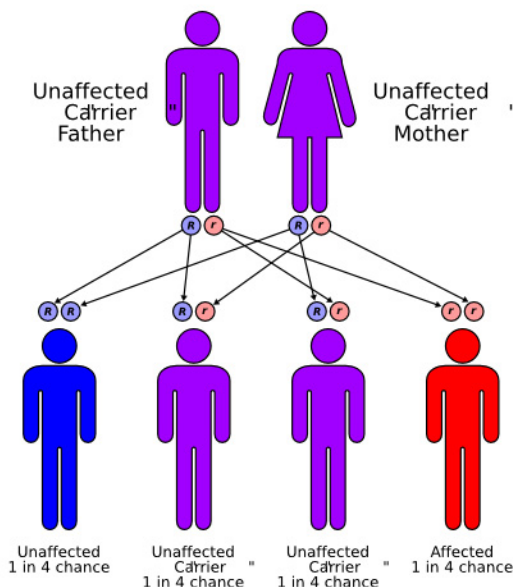
**What causes the LCHAD enzyme to be absent or not working properly?**

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.

**How is LCHADD inherited?**

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





Everyone has a pair of genes that make the LCHAD enzyme. In children with LCHADD, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent. Parents of children with LCHADD are rarely affected with the disorder. Instead, each parent has a single non-working 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% chance in each pregnancy for the child to have LCHADD. 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 LCHADD. Genetic counselors can answer your questions about how LCHADD 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?

LCHADD can also be confirmed by a special test called a "fatty acid oxidation probe" using a skin sample. Talk to your doctor or your genetic counselor if you have questions about testing for LCHADD.

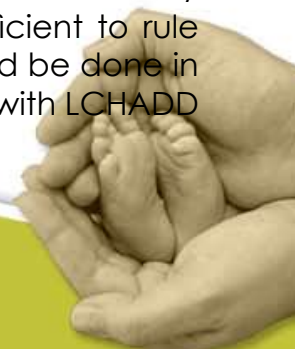
Can other members of the family have LCHADD or be carriers?

### 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 only be done in people over 18 years of age. Each of the parents' brothers and sisters has a 50% chance to be an LCHADD carrier. It is important for other family members to be told that they could be carriers. Expanded newborn screening through private lab is available for babies born that do not screen for LCHADD. To learn more about expanded newborn screening, see How to obtain MS/MS. 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





are at risk to develop serious medical problems. Some of these women develop:

- excessive vomiting
- abdominal pain
- high blood pressure
- jaundice
- abnormal fat storage in the liver
- 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.

### **How many people have LCHADD?**

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

### **Does LCHADD happen more often in a certain ethnic group?**

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

### **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>

