



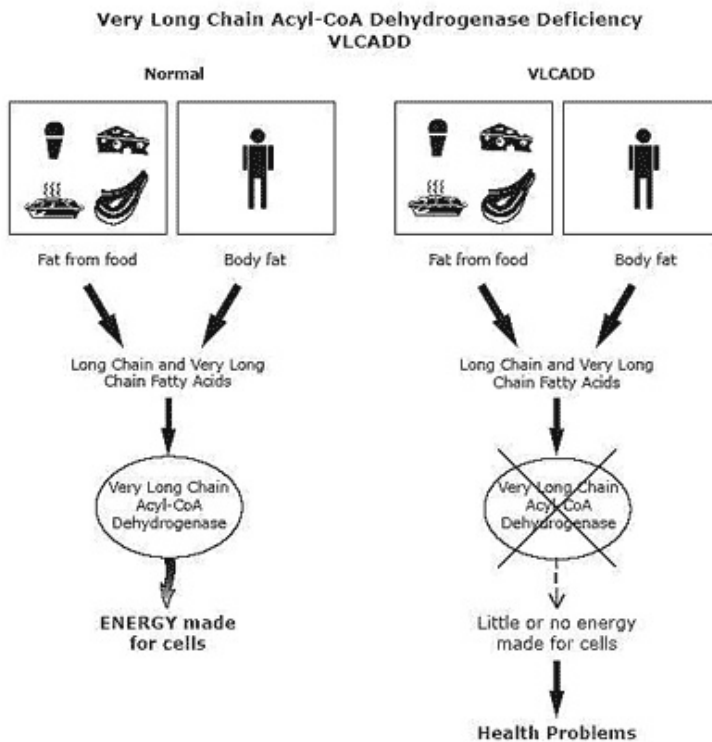
## VERY LONG CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY (VLCADD)

### A fact sheet for parents/carers

#### What is VLCADD?

VLCADD stands for “very long chain acyl-CoA dehydrogenase deficiency”. It is one type of fatty acid oxidation disorder. People with VLCADD have problems breaking down certain types of fat into energy for the body.

#### What causes VLCADD?



VLCADD occurs when an enzyme, called “very long chain acyl-CoA dehydrogenase” (VLCAD) 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.

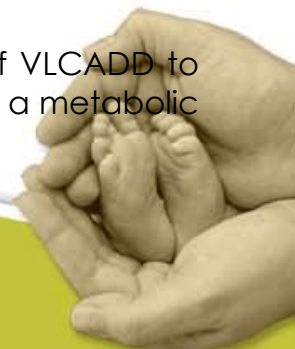
Energy from fat keeps us going whenever our bodies run low of their main source of energy, a type of sugar called glucose. Our bodies rely on fat when we don’t eat for a stretch of time – like when we miss a meal or when we sleep. When the VLCADD enzyme is missing or not working, the body cannot break down 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 VLCADD is not treated, what problems occur?

VLCADD is variable and can cause mild effects in some people and more serious health problems in others. Symptoms may start in infancy or not until adulthood. There are three forms of VLCADD: “Early”, “Childhood” and “Adult”.

It is common for babies and children with the early and childhood types of VLCADD to have episodes of illness called metabolic crises. Some of the first symptoms of a metabolic crisis are:



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- extreme sleepiness
- behavior changes
- irritable mood
- poor appetite

Some of these other symptoms may also follow:

- fever
- nausea
- diarrhea
- vomiting
- hypoglycemia

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

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

Periods of hypoglycemia can happen with or without the other symptoms. Hypoglycemia can cause a child to feel weak, shaky or dizzy with clammy, cold skin. If not treated, it can lead to coma, and possibly death.

Either hypoglycemia or a full metabolic crisis can occur:

- after going too long without food
- during illness or infection
- after heavy exercise

Symptoms of early and childhood VLCADD often happen after a period of having nothing to eat for more than a few hours. Symptoms are also more likely when a child with VLCADD gets sick or has an infection.

### **Early VLCADD**

About half of babies diagnosed with VLCADD have the "early" type. They usually start to show effects between birth and 4 months. In addition to metabolic crises, babies can also have:

- enlarged heart, irregular heartbeat and other heart problems
- enlarged liver and other liver problems
- muscle problems

If not treated, babies with early VLCADD usually die young.

### **Childhood VLCADD**

About one third of people with VLCADD have the childhood type. They usually show





symptoms in late infancy or early childhood. Episodes of hypoglycemia or full metabolic crisis happen during illness or after long periods of not eating. Other effects can include:

- enlarged liver
- other liver problems
- muscle weakness, especially after exercise

Heart problems are usually not seen in childhood VLCADD.

Some children with VLCADD have never had symptoms and are only found to be affected after a brother or sister has been diagnosed.

### Adult VLCADD

About one fifth of people with VLCADD have the adult type. They usually show symptoms starting in the teen years or in adulthood. Periods of muscle weakness are common. Breakdown of muscle fibers can occur. This usually happens during heavy exercise or after going without food for a long period of time.

Signs of muscle breakdown are:

- muscle aches
- weakness
- cramps
- reddish-brown color to the urine.

Adults with muscle symptoms who do not get treatment can develop kidney failure. People with the adult form of VLCADD usually do not have heart problems, hypoglycemia or metabolic crises.

### What is the treatment for VLCADD?

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 VLCADD. 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 VLCADD:

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

Infants and young children with VLCADD 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 VLCADD 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 diet is recommended. 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, etc.) and protein (lean meat and low-fat dairy foods). Any diet changes should be made under the guidance of an experienced dietician.

People with VLCADD 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.

## 3. MCT oil and L-carnitine

Medium Chain Triglyceride oil (MCT oil) is often used as part of the food plan for people with VLCADD. 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.

Do not use any medication or supplement 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, weakness, or reddish-brown color to the urine



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Children with VLCADD 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 or a metabolic crisis. When they become sick, children with VLCADD often 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.

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

Long periods of exercise can also trigger symptoms. Problems occurring during or after exercise can include:

- muscle aches
- weakness
- cramps
- reddish-brown color to the urine.

If muscle symptoms happen, prompt treatment is needed to prevent kidney damage. Children and 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 VLCADD is treated?**

With prompt and careful treatment, people with the childhood and adult forms of VLCADD can often live healthy lives with typical growth and development. Before diagnosis through newborn screening was available, the early form of VLCADD was fatal. Now, with immediate and ongoing treatment, many infants with VLCADD are surviving.

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

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

**How is VLCADD inherited?**

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



# NEWBORN SCREENING

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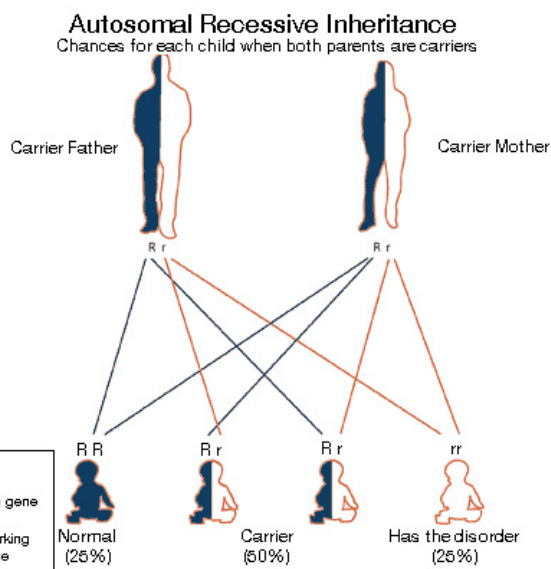
Everyone has a pair of genes that make the VLCAD enzyme. In children with VLCADD, neither of these genes works correctly. These children inherit one non-working gene for the condition from each parent.

Parents of children with VLCADD are rarely affected with the disorder. Instead, each parent has a single non-working gene for VLCADD. They are called carriers. Carriers do not have VLCADD 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 VLCADD. 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 VLCADD. Genetic counselors can answer your questions about how VLCADD is inherited, options 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?



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

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

## Having VLCADD

The brothers and sisters of a baby with VLCADD have a chance of being affected, even if they haven't had symptoms. Finding out whether other children in the family have VLCADD is important because early treatment may prevent serious health

problems. Talk to your doctor or genetic counselor about testing your other children for VLCADD.

## VLCADD Carriers

Brothers and sisters who do not have VLCADD 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 VLCADD 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 VLCADD.





Some states do not provide newborn screening for VLCADD. However, expanded newborn screening through private labs is available for babies born in states that do not screen for this condition. 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 VLCADD in a newborn baby. In this case, special diagnostic testing should be done in addition to newborn screening. During pregnancy, women carrying fetuses with VLCADD may be at increased risk to develop serious medical problems. A small number of women are known to 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 VLCADD 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 of a child with VLCADD can have special testing to check for this disorder. Ask your metabolic doctor whether your other children should be tested for VLCADD.

### **How many people have VLCADD?**

VLCADD is thought to be a rare disorder. The actual incidence is unknown.

### **Does VLCADD happen more frequently in a certain ethnic group?**

No, VLCADD does not happen more often in any specific race, ethnic group, geographical area or country.

### **Where can I find more information?**

Fatty Oxidation Disorders (FOD) Family Support Group: <http://www.fodsupport.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>

