

VLCAD At a Glance

VLCAD is one of several ***Fatty Acid Oxidation Disorders (FAOD)** in which there is an inability to break down certain fats, caused by an enzyme deficiency. This results in a decreased ability to go for a long time without food or calories (fasting).

Learn more about fasting:

- During fasting, fatty acids are an important source of energy for liver and other tissues. If they are not converted to energy, it results in lethargy (lack of energy) and hypoglycemia (low blood sugar).
- Energy from fat keeps us going whenever our bodies run low of our main source of energy (glucose.) This might happen if we miss a meal or when we sleep.
- Once sugar or glucose is gone, the body tries to use fat without success. This leads to low blood sugar (known as hypoglycemia) and the buildup of harmful substances in the blood.

About one in every 30,000 babies is born with VLCAD.

Before VLCAD was included on the newborn screening test, three types of VLCAD were recognized by the severity of the condition:

- A severe early onset form that typically presented in the first months of life and included cardiac disease (cardiomyopathy) and multi-organ failure.
- An early childhood onset form with an enlarged liver that did not include an enlarged heart (cardiomyopathy), and presented with low blood sugar (hypoglycemia).

Signs of hypoglycemia:

- Weak, shaky, dizzy with clammy cold skin
- If not treated it can result in coma and possible death

- A milder, later childhood or adult onset form that affects muscles. It presents with intermittent breakdown of muscle tissue (rhabdomyolysis), muscle cramps and/or pain, and/or exercise intolerance.

Now that VLCAD can be detected through newborn screening, the determination of the type of VLCAD is determined through genetic and biochemical testing. With treatment from infancy, children with all types of VLCAD are expected to live a full life. However, they are still at risk for complications in times of illness, stress, and prolonged or extreme physical activity.

Children with VLCAD do not have any distinct physical features.

Note: There is wide variability within individuals who have this condition.

Fatty Acid Oxidation Disorders *At a Glance

We have enzymes that break down fats in our bodies. This process is known as fatty acid oxidation. When the body can't break down fatty acids to make energy, Fatty Acid Oxidation Disorders (FAOD) occur.

MCAD and VLCAD are the most common of the FAOD, but many different disorders are included in this group.

- Medium Chain Acyl-CoA Dehydrogenase (MCAD) Deficiency
<http://www.ncbi.nlm.nih.gov/books/NBK1424/>
- Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency
<http://www.ncbi.nlm.nih.gov/books/NBK6816/>
- Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency
<http://ghr.nlm.nih.gov/condition/long-chain-3-hydroxyacyl-coa-dehydrogenase-deficiency>
- Trifunctional Protein (TFP) Deficiency
<http://www.newbornscreening.info/Parents/fattyacid disorders/TFP.html>
- Carnitine Transport Defect (Primary Carnitine Deficiency)
<http://www.newbornscreening.info/Parents/fattyacid disorders/Carnitine.html>
- Carnitine-Acylcarnitine Translocase Deficiency (Translocase)
<http://ghr.nlm.nih.gov/condition/carnitine-acylcarnitine-translocase-deficiency>
- Carnitine Palmitoyl Transferase I & II (CPT I & II) Deficiency
<http://ghr.nlm.nih.gov/condition/carnitine-palmitoyltransferase-i-deficiency>
- 2,4 Dienoyl-CoA Reductase Deficiency
<http://www.fodsupport.org/list.htm>

- Short Chain Acyl-CoA Dehydrogenase (SCAD) Deficiency
<http://ghr.nlm.nih.gov/condition/short-chain-acyl-coa-dehydrogenase-deficiency>
- Short Chain L-3-Hydroxyacyl-CoA Dehydrogenase (SCHAD) Deficiency (now called 3-Hydroxy Acyl CoA Dehydrogenase Deficiency (HADH)
<http://www.fodsupport.org/list.htm>
- Electron Transfer Flavoprotein (ETF) Dehydrogenase Deficiency (GAII & MADD)
<http://www.fodsupport.org/list.htm>
- 3-Hydroxy-3 Methylglutaryl-CoA Lyase Deficiency (HMG)
<http://ghr.nlm.nih.gov/condition/3-hydroxy-3-methylglutaryl-coa-lyase-deficiency>
- Unclassified FAODs

Fatty acids are a major source of energy for the heart and muscles. Energy from fat keeps us going when our bodies run low on our main source of energy (glucose). This might happen when we miss a meal or sleep. Once sugar or glucose is gone, the body tries to use fat without success. This leads to low blood sugar (known as hypoglycemia) and the buildup of harmful substances in the blood.

When a FAOD is diagnosed and treated at birth, the prognosis for most FAOD is excellent. Symptoms and treatments can vary between different FAOD and even can vary within the same FAOD. If undiagnosed and/or untreated, disorders can lead to serious complications.

Complications include problems with the liver, heart, eyes, general muscle development, and possible death.

Symptoms vary with FAOD from complex to no symptoms:

- Chronic bouts of low blood sugar (hypoglycemia) leading to many hospitalizations within the first years of life
- Lack of energy (lethargy)
- Muscle weakness
- Liver abnormalities
- Life threatening heart problems

Treatment consists of dietary interventions specific to the particular FAOD.

Children with FAOD do not have any distinct physical features.

VLCAD - Things to Think About

1. Medical / Dietary Needs

What you need to know

It is important for an individual with VLCAD or other FAOD to eat regularly and **not** go without food or calories for more than 8-12 hours. This is especially important during times of high metabolic stress, such as illness. This means frequent feeds as infants and may involve a cornstarch supplement at bedtime when they are older if they have the severe form of VLCAD.

A low fat (<30% of total energy from fat) diet is recommended and in the more severe form this may mean an almost non-fat diet. Children with the more severe forms may take supplements of medium-chain triglycerides (MCT) oil and walnut or similar oils in order to get their essential fats without taking in very long chain fats. Carnitine supplements may benefit some children. It is important to be sensitive to cultural differences in diet.

Excess food intake is stored in the body as very long chains fats, so controlling the overall calories is also important to avoid obesity.

Be aware, or ask a parent, if the child has a medical alert bracelet.

What you can do

During the school day, a student may require low fat/high carbohydrate foods throughout the day. Parents may leave cereal or other food with the teacher or nurse to use as needed. Multiple snacks during the day may be necessary.

Ensure good hydration and carbohydrate intake during periods of exercise. Low blood sugar (hypoglycemia) must be avoided.

Ensure sick day plans are in place for illness or other times when a child cannot eat.

Notify a parent if you notice changes. Documenting is helpful.

NOTE: A child may appear hydrated and still be heading to a crisis. They still require calories to prevent or help them through the metabolic crisis/stress.

2. Education Supports

What you need to know

It is important to have HIGH EXPECTATIONS for learning for children who have VLCAD.

Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

A 504 plan for diet and/or gym may be required. If an individual with VLCAD or any other FAOD has an acute metabolic event that leads to a brain injury, they may have secondary academic consequences.

Secondary academic consequences due to brain injury may include:

- Loss of developmental milestones
- Acquiring aphasia (an impairment in language ability)
- Attention deficit disorder

What you can do

An IEP may be necessary if the child has had a brain injury. They may need a 504 plan for diet and gym.

3. Behavior & Sensory Support

No specific behavior issues

4. Physical Activity, Trips, Events

What you need to know

- It is very important that individuals with VLCAD have access to fluids and snacks. They need to bring snacks or drinks on field trips that last more than a few hours.
- Because children, especially older children, are at risk for muscle breakdown (rhabdomyolysis) during periods of strenuous exercise, care must be taken during gym class to provide adequate fluids and even supplemental carbohydrates before any strenuous exercise.
 - Some children with FAOD's may not be able to participate in a typical gym class.
- If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming (www.nepassage.org).

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What you can do

- If the episodes of rhabdomyolysis (muscle cramps and fatigue after long periods of exercise) become frequent or severe, a modified physical education plan may become necessary.

5. School Absences & Fatigue

What you need to know

Individuals with VLCAD should not have increased absences except in the severe form in which they may need to be hospitalized for emergency treatment after an illness.

In addition, older children and adults with VLCAD may have muscle cramps and fatigue after long periods of exercise related to muscle breakdown (rhabdomyolysis).

What you can do

For the reason above, it is even more important to make sure that they have sufficient carbohydrate intake before strenuous exercise.

6. Emergency Planning

What you need to know

All individuals who have VLCAD need an emergency letter to be given to health care providers in the event of a crisis. The letter should address the following:

- When a child with VLCAD has a viral illness or otherwise has decreased intake they may become lethargic or hypoglycemic.
- Glucose through an IV is essential to prevent hypoglycemia and other complications.
- The heart muscle can be affected and cause abnormal heart rhythms that may need to be treated.
- Watch for red or rust colored urine or muscle cramps that may be a sign of muscle breakdown (rhabdomyolysis).

- Symptoms that a child with VLCAD may be in trouble may include:
 - Lack of energy
 - Lethargy
 - Muscle weakness
 - Muscle cramps
 - Any vomiting or diarrhea

What you can do

Obtain a copy of the emergency letter so treatment can begin as soon as possible.

7. Resources

New England Consortium of Metabolic Programs

<http://newenglandconsortium.org/>

The New England Consortium of Metabolic Programs brings together healthcare professionals at all levels involved in identifying and treating individuals with metabolic disorders.

The goals of the Consortium are to disseminate information, collaborate in the development of social support programs and educational materials, support organizations for parents and adults with metabolic disorders, provide training for students in medicine and related fields, jointly develop and conduct research projects, and establish uniform treatment protocols for individuals with metabolic disorders.

FOD Family Support Group

<http://www.fodsupport.org/>

"All in this together"

Genetic Fact Sheets for Parents

<http://www.newbornscreening.info/Parents/fattyacid disorders/VLCADD.html>

*Fatty Acid Oxidation Disorders - VLCAD
by Screening, Technology & Research in Genetics/Expanded Newborn Screening*

Genetics Home Reference

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<https://ghr.nlm.nih.gov/condition/very-long-chain-acyl-coa-dehydrogenase-deficiency>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

GeneReviews

<http://www.ncbi.nlm.nih.gov/books/NBK6816/>

Learn more about the genetics of VLCAD.

Note: *This printable version does not include the information found under the green button marked "Transitions" on the website. Those general pages may be printed separately.*

8. Meet a Child with VCLAD

Walking with Landon!

GEMSS would like to thank Landon and his mother for their generosity in sharing this story with us. You have made the site come to life with the addition of your thoughts and feelings. Thank you so much!



Regan describes her son Landon as a very energetic, smart, and stubborn red-head! He is 'all-boy,' she says. Landon is currently in a day-care/preschool in his southwestern town in the USA.

Landon's Newborn Screening revealed an abnormality but the follow-up test was normal. For 9 days, the diagnosis was undetermined and a blood draw on the ninth day was abnormal. At 3 weeks, he saw a geneticist and a biopsy of his skin was done. Further testing then showed that he did have the diagnosis of VLCAD, and it was the non-cardiac version. However, he is the only person known to have this exact mutation. His sister and his mother are both carriers of the mutation.

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Landon's prognosis is good and he has had four big scares when his blood sugar dropped rapidly and he was hospitalized. His family always carries glucose and they have a protocol that they follow. If his blood sugar gets below 70, they call the doctor and if it is below 65, they get to a hospital. "He has a very particular whine when his blood sugar is dropping" so they are able to tell when he is dropping.

In school, Landon is able to eat the healthy snacks they serve as they are low-fat. He knows what he can and can't eat and will let people know what is okay for him to eat. He only has 23 grams of fat every day and the school staff has been very helpful in making sure he eats and drinks according to the plan. He needs to drink Gatorade on the playground and the staff has him drink every 20 minutes or so when he is active. The school also has a letter from the geneticist outlining when to call and when to seek medical attention. Landon himself can tell when his sugar is low and will often say "I need some OJ or Gatorade – my legs are starting to hurt." The school team has listened, asked lots of questions, and worked well with the family to make sure Landon's school experience is successful.

Landon is doing well developmentally and is "always ahead of the curve," according to his mother. He was home for 3 ½ years as they were advised to keep him away from germs. He did not have a lot of interaction with other children until he got to preschool so he is developing more social skills in school.

Regan advises teachers to listen to parents. "Ask them to come in and talk," she says. She has heard some other parents say that they 'don't feel heard' and teachers sometimes 'treat them like they do not know anything.' That has not been true in her experience! Landon is even doing some teaching in his class. For example, the school was doing a unit on hospitals and the teachers were very impressed that Landon was able to tell the other children what it is like in the hospital, how they check for blood sugar, how it hurts for a minute, "but that's okay because then they give you a toy!"