Alberta Health Services
Alberta Newborn Screening

Talking with Parents > When talking to a parent whose baby has received an abnormal screen result, let them know that an abnormal screen result doesn't mean that the infant has the condition. It means that they have a higher chance of having the condition, and they need more tests to find out for sure. A printable sheet for parents with more condition specific information can be used to support your conversation, visit ahs.ca/nms and under Quick Reference click on *What conditions are screened for?*

Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency

(metabolic condition: fatty acid oxidation disorder) *Information for Health Professionals*

Also known as:

- acyl-CoA dehydrogenase very long chain deficiency
- very long-chain acyl coenzyme A dehydrogenase deficiency

What are fatty acid oxidation disorders?

Fatty acid oxidation disorders (FAOD) are a group of inherited conditions in which fatty acids cannot be broken down to provide the body with energy. Fatty acids are a key source of energy during fasting or stress, such as during an illness. An inability to metabolize fatty acids leads to an accumulation of toxic metabolites in the body along with cellular energy deficiency which in combination can cause serious health problems.

What is VLCAD deficiency?

Very long chain acyl-CoA dehydrogenase (VLCAD) is one of the mitochondrial enzymes required in the breakdown of fatty acids to produce energy. VLCAD breaks down fatty acids longer than 16 carbon atoms in length. People with VLCAD deficiency are unable to break down these long chain fatty acids to produce energy.

What causes VLCAD deficiency?

VLCAD deficiency is caused by pathogenic variants in the ACADVL gene resulting in absent or decreased enzyme activity.

How common is VLCAD deficiency?

The incidence of VLCAD deficiency is about 1 in every 40,000 to 100,000 infants born worldwide.

What are the clinical features of VLCAD deficiency?

Although infants with VLCAD deficiency may appear normal at birth, during a period of fasting (such as during a common illness), an infant may present with lethargy, hypoglycemia, hypotonia, vomiting, diarrhea and seizures. This can progress quickly to coma and death if treatment is not started quickly. Some children may present later with muscle weakness and fatigue and can develop significant heart problems. The first episode usually occurs within the first few months of life. However, milder forms of VLCAD deficiency may present later in childhood or adulthood.

What is the screening test for VLCAD deficiency?

A specific pattern of metabolites is detected on the newborn blood spot screen. Newborn blood spot screening will not detect all infants with VLCAD. Infants with clinical symptoms need timely assessment and diagnostic testing even if their screen result is normal.

How is the diagnosis confirmed?

The diagnosis of VLCAD deficiency is confirmed by detecting specific metabolites in urine or on blood acylcarnitine analysis. Further testing may include enzyme analysis and/or molecular genetic analysis of the ACADVL gene. The Clinical and Metabolic Genetics Program will arrange diagnostic testing.

How is VLCAD deficiency treated?

VLCAD deficiency is treated by avoidance of fasting and a diet which is low in long chain fatty acids. Prompt treatment of a metabolic crisis with intravenous fluids and glucose is necessary. Effective treatment can prevent metabolic crisis and improve the outcome of this disorder. The treatment is lifelong.

Is VLCAD deficiency inherited?

VLCAD deficiency is inherited as an autosomal recessive disorder. Parents of a child with VLCAD deficiency are carriers of the condition and have a 1 in 4 chance of having another affected child in each subsequent pregnancy. VLCAD deficiency carriers are healthy. Genetic counselling is available to families with VLCAD deficiency.

Additional resources are available through:

Clinical & Metabolic Genetics Program (Edmonton)

8-53 Medical Sciences Building
8440 – 112 St. NW
Edmonton, AB T6G 2H7
Phone: 780-407-7333
Fax: 780-407-6845

Emergency consultations:
Phone 780-407-8822 and ask for the specialist on call for metabolic diseases.

Clinical & Metabolic Genetics Program (Calgary)

Alberta Children's Hospital
28 Oki Drive NW
Calgary, AB T3B 6A8
Phone: 403-955-7587
Fax: 403-955-3091

Emergency consultations:
Phone 403-955-7211 and ask for the specialist on call for metabolic diseases.

Early screening and follow-up care – every baby, every time

For more information about the Alberta Newborn Screening Program, visit www.ahs.ca/newbornscreening