NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Very Long Chain Acyl-CoA dehydrogenase deficiency (VLCAD)

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This is done to identify a number of rare but treatable conditions which usually do not have any symptoms early on. Knowing if your child has one of these conditions early in life is important, because early diagnosis allows for early treatment and better health outcomes.

Your baby had a positive newborn screen result for VLCAD deficiency. **This result does not mean that your baby has VLCAD deficiency but means the chance is increased and more testing is needed to confirm**.

What is VLCAD deficiency?

VLCAD deficiency is a type of fatty acid oxidation disorder. Our bodies normally breakdown the sugars, fats, and proteins we eat to make energy. Our cells need this energy to work properly. The main energy source is usually sugar (carbohydrates). But during long periods of time without food (fasting), our body relies more on stored fat for energy. People who have VLCAD deficiency cannot breakdown certain fats called ***long chain fatty acids*** to make energy. When fasting (such as during a cold or flu) babies with VLCAD deficiency do not make enough energy for proper cell function and can become very ill.

The newborn screen measures the amount of long chain fatty acids which are increased in babies with VLCAD deficiency. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms?

Babies with VLCAD deficiency are usually healthy at birth and remain healthy if they do not fast. During prolonged fasting episodes, a child may develop low blood sugar levels which could lead to a metabolic crisis. A ***metabolic crisis*** can present as lethargy (very sleepy and unresponsive), floppiness, and irritability. If severe and left untreated, this can progress quickly to seizures and loss of consciousness. Some babies may develop heart, liver, and muscles problems.

In most cases, symptoms present in the first few months of life but there are also milder forms that do not present until later childhood or even in adulthood.

What causes VLCAD deficiency?

The enzyme needed to breakdown long chain fatty acids is not working properly in people affected with VLCAD deficiency. The VLCAD enzyme is made in our cells by a gene called the ACADVL gene.

A gene is a set of instructions on how to make a protein or enzyme. Changes in the ACADVL gene, called disease-causing variants result in little or no functioning enzyme being made.

We all have two copies of the ACADVL gene. A baby born with VLCAD deficiency has received (inherited) two non-working copies of the ACADVL gene, one from each parent. Parents of a child with VLCAD deficiency are usually carriers. A carrier is a person with one working and one non-working copy of a gene. People usually don't know they are a carrier because carriers are unaffected.

What happens next?

Your primary care provider will review more information with you. If needed a specialist doctor in the Biochemical Genetics Clinical Service (BGCS) will contact you to answer questions and organize a visit for your family at BC Children's Hospital in Vancouver.

The doctor will meet your baby and discuss with you further testing options to confirm if your baby has VLCAD deficiency. Testing may include a repeat newborn screen, additional blood and urine screens and genetic testing of the ACADVL gene.

How is VLCAD deficiency treated?

The goal of treatment is to try to prevent an episode of metabolic crisis. Babies with VLCAD must not go long periods of time without eating. Dietitians and specialists will support

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.

families as they learn about the required dietary considerations and measures to avoid fasting. Children with VLCAD may need to go to the hospital when sick with a cold or flu if they are unable to eat and drink.

Treatment is coordinated by specialists at BC Children's Hospital.

What is the outcome of treatment?

Treatment is very effective at preventing metabolic crises. With early treatment and careful monitoring most babies with VLCAD will lead healthy lives with normal growth and development. As children get older, the risk of metabolic crisis decreases.

How do you feel?

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. If you are feeling overwhelmed or have questions, you can talk with your health care provider to discuss supports available.

Where can I get more information?

Talk to your primary care provider. You may also call the BGCS Newborn Screening Team at 604-875-2623.

Information about the BC Newborn Screening Program can be found at:
www.newbornscreeningbc.ca

Information about VLCAD deficiency can be found at:

- **www.newbornscreening.info/Parents/fattyaciddisorders/VLCADD.html**
- **www.fodsupport.org/**

This fact sheet provides basic information only. It does not take the place of medical advice, diagnosis, or treatment. Always talk to your health care provider about specific health concerns.