

INFORMATION FOR PARENTS/GUARDIANS:

My Baby Has a Positive Very Long Chain Acyl-Co-A Dehydrogenase Deficiency Newborn Screening Result

What is newborn screening?

These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from your baby and is tested for rare, treatable diseases, including very long chain acyl-Co-A dehydrogenase deficiency (VLCAD).

What does it mean if my baby has a positive VLCAD newborn screening result?

This result does **not** mean that your baby has VLCAD. It means that more testing is needed because your baby **might** have VLCAD. Babies with VLCAD can be healthier if VLCAD is diagnosed early, so it is important to have follow-up testing done quickly to find out if your baby has VLCAD.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has VLCAD until more testing has been done.

What is VLCAD?

VLCAD is a rare inherited (genetic) disease that causes a baby to have problems using fat as an energy source. The human body needs energy to perform all of its daily activities. The body's main source of energy is a type of sugar called glucose. If a baby has not eaten for a while – such as when they are sick or if they miss a meal – the body runs out of glucose and switches to using fat as an energy source.

When a baby has VLCAD, their body cannot switch to using fat for energy. Babies can get very sick if their bodies are not able to use fat for energy when needed. Some babies with VLCAD also have problems with their heart, liver and muscles.

Some people with VLCAD never develop any health problems associated with VLCAD and some do not develop problems until childhood or adulthood. However, there is no way to know who will develop problems caused by VLCAD and who will not.

What causes VLCAD?

VLCAD happens when an enzyme called very long chain acyl-co-A dehydrogenase is either missing or not working properly. This enzyme's job is to break down the "very long" sized fats that we eat, or that are stored in the body, into smaller sized fats and eventually into energy.

How do I find out if my baby has VLCAD?

Blood and urine tests are done to find out if a baby who screened positive actually has VLCAD. Sometimes, other tests are done.

When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange more testing as soon as possible.

Why screen for VLCAD?

Babies with VLCAD can appear perfectly healthy at birth. However, they are at risk for a metabolic crisis during periods of not eating (for example, illness). A metabolic crisis is a serious health condition caused by low blood sugar and/or



the build-up of harmful substances in the blood. Symptoms of a metabolic crisis are poor feeding, vomiting, lethargy, excessive sleepiness and irritability. If a metabolic crisis is not treated, breathing problems, seizures, coma, and sometimes death can occur. The goal of screening for VLCAD is to prevent a metabolic crisis and help people with all types of VLCAD live healthier lives.

How is VLCAD treated?

Babies with VLCAD are treated and monitored by a team of specialists including a metabolic doctor and a dietician. The treatment for VLCAD includes frequent feeding and avoiding fasting (do not go a long time without eating). Sometimes, a special low fat diet is given. Some babies are given medication, carnitine or cornstarch.

How does a baby get VLCAD?

VLCAD is an inherited (genetic) disease. A baby with VLCAD inherited two non-working copies of the VLCAD gene, one from each parent. People who have one non-working copy of the VLCAD gene are called "carriers." Carriers of VLCAD are healthy and do not have symptoms of VLCAD.

Where can I get more information?

For more information on newborn screening, please visit the Parent section of our website at www.newbornscreening.on.ca or talk to your local health care provider.

For more information on VLCAD, please visit the Fatty Acid Oxidation Family Support Group website at <http://www.fodsupport.org>

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for very long chain acyl-co-A dehydrogenase deficiency (VLCAD). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

