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

What is newborn screening?
These are routine tests done shortly after birth on every baby born in Manitoba. 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 (VLCAD) deficiency.

What does it mean if my baby has a positive newborn screening test for VLCAD deficiency?
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 deficiency?
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 deficiency, his/her body cannot switch to using fat for energy. Babies can get very sick if their bodies are not able to use to use fat for energy when needed. Some babies with VLCAD who have problems with their heart, liver and muscles require special treatments.

Some people with VLCAD deficiency 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 deficiency and who will not.

What causes VLCAD deficiency?
VLCAD deficiency 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 actually has VLCAD deficiency?
Blood and urine tests are done to find out if a baby who screened positive actually has VLCAD deficiency. Sometimes, other tests are done.
When can my baby have these tests?
Your baby’s doctor or a health care professional at the Winnipeg Children’s Hospital 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. Babies with VLCAD are especially prone to heart problems including rhythm disturbances and heart failure. The goal of screening for VLCAD deficiency is to identify an affected baby before any symptoms appear and to begin treatment promptly that can prevent a metabolic crisis. This will ultimately help all affected babies and other affected people with all types of VLCAD live healthier lives.

How is VLCAD deficiency treated?
Babies with VLCAD deficiency are treated and monitored by a team of metabolic specialists including a metabolic doctor, metabolic dietician and a dietician. Pediatric cardiologists are also essential in their care. The treatment for VLCAD deficiency includes frequent feeding and avoiding fasting (do not go a long time without eating (generally 6-8 hours in infancy) as well as special heart medications if necessary. Sometimes, a special low fat diet is given. Some babies are given medication, carnitine or cornstarch.

How does a baby get VLCAD deficiency?
VLCAD deficiency is an inherited (genetic) disease. A baby with VLCAD deficiency 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 deficiency are healthy and do not have symptoms of VLCAD.

Where can I get more information?
For more information on newborn screening, please talk to your local health care provider or visit the Winnipeg Metabolic Program website www.wrha.mb.ca/prog/genetics or the newborn screening section of the Cadham Provincial Laboratory website www.gov.mb.ca/health/publichealth/cpl/baby.html.

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.