INFORMATION FOR PARENTS/GUARDIANS:
My Baby Has a Positive Carnitine Uptake Defect Newborn Screening Result

What is newborn screening?
These are routine tests done soon 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 carnitine uptake defect (CUD).

What does it mean if my baby has a positive CUD newborn screening result?
This result does **not** mean that your baby has CUD. It means that more testing is needed because your baby **might** have CUD. Babies with CUD can grow and develop normally if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has CUD.

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 CUD until follow up testing has been done.

What is carnitine uptake defect (CUD)?
CUD 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 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 CUD, their body can not switch to using fat for energy. Babies can get very sick if their bodies are not able to use fat for energy when needed.

What causes CUD?
CUD happens when an enzyme called "carnitine transporter" is either missing or not working properly. This enzyme's job is to carry carnitine, a naturally occurring protein in the body, inside certain cells of the body. Once inside these cells, carnitine helps the body use fat as energy. If the carnitine can not get inside these cells, the body is unable to use fat as energy. Carnitine is also important for healthy muscles and a healthy heart.

How do I find out if my baby has CUD?
Blood and urine tests are done to find out if a baby who is screen positive actually has CUD.

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 follow up testing as soon as possible.

Why screen for CUD?
Babies who have CUD are at risk for a metabolic crisis. 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 CUD is to prevent a metabolic crisis.

How is CUD treated?
Babies with CUD are treated and monitored by a specialist called a metabolic geneticist. Lifelong dietary treatment with a carnitine supplement is required. Infants and children with CUD should not go a long time without eating. With early detection and careful treatment, children with CUD usually live healthy lives with typical growth and development.

How does a baby get CUD?

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

Where can I get more information?

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

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

NOTE TO PARENTS/GUARDIANS: This information is only applicable if your baby has had a positive newborn screening result for carnitine uptake defect (CUD). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.