My Baby Has a Positive Newborn Screening Result:
Carnitine Palmitoyl Transferase - Type 1A 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 carnitine palmitoyl transferase type 1A (CPT1A).

What does it mean if my baby has a positive newborn screening test for CPT1A?
If your baby is of Hutterite background, we know already from the newborn screen that they have CPT1A. For babies of other backgrounds, this result does not mean that your baby has CPT1A. It means that more testing is needed because your baby might have CPT1A. Babies with CPT1A can be healthier if the condition is diagnosed early, so it is important to have follow-up testing done quickly.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way.

What is CPT1A?
CPT1A 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 has used all its stores of glucose for energy and switches to using fat as an energy source. When a baby has CPT1A, 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. Babies with untreated CPT1A may damage their liver, heart and brain.

CPT-1A deficiency is found in about one in every 1200 babies who are of Hutterite background in Manitoba. CPT-1A deficiency is rare in other ethnic groups and the incidence is not well defined. About 7 out of 10 Inuit babies can have a mild variant of this condition, which requires no long term treatment and with generally an entirely normal outcome.

What causes CPT1A?
CPT1A happens when an enzyme called “carnitine palmitoyl transferase 1A” is not working properly. The job of this is to certain fats in the food we eat into energy. It also helps break down fat already stored in the body.

How do I find out if my baby actually has CPT1A?
Blood and urine tests can be done to find out if a baby who screened positive actually has CPT1A. Sometimes, other tests are needed. If your baby is of Hutterite background, no additional tests are needed.
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 if needed.

Why screen for CPT1A?
Babies with CPT1A 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 newborn screening for CPT1A is to identify an affected baby right after birth before symptoms appear and with early treatment prevent a metabolic crisis. Ultimately this will help babies and older people with CPT1A live healthier lives.

How is CPT1A treated?
Babies with CPT1A are treated and monitored by a team of specialists including a metabolic doctor, a metabolic genetic counsellor and a dietician. The treatment for CPT1A includes frequent feeding and avoiding fasting (do not go a long time without eating). A special low fat diet, cornstarch or other medications may be given for older children.

How does a baby get CPT1A?
CPT1A is an inherited (genetic) disease. A baby with CPT1A inherited two non-working copies of the CPT1A gene, one from each parent. People who have one non-working copy of the CPT1A gene are called “carriers.” Carriers of CPT1A are healthy and do not have symptoms of this condition.

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 CPT1A, 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 CPT1A. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.