

INFORMATION FOR PARENTS/GUARDIANS:

My Baby Has a Positive Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency/Trifunctional Protein 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 two diseases called long chain acyl-Co-A dehydrogenase deficiency (LCHAD) and trifunctional protein deficiency (TFP).

What does it mean if my baby has a positive LCHAD/TFP newborn screening result?

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

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 LCHAD or TFP until more testing has been done.

What are LCHAD and TFP?

LCHAD and TFP are rare inherited (genetic) diseases that cause 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 LCHAD or TFP, 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 LCHAD and TFP also have problems with their development, heart, eyes, nerves and liver.

What causes LCHAD and TFP?

LCHAD happens when an enzyme called long chain 3-hydroxyacyl-co-A dehydrogenase is either missing or not working properly. TFP happens when a group of enzymes called trifunctional protein (which includes LCHAD) is either missing or not working properly. The job of these enzymes is to break down the "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 LCHAD or TFP?

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

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 LCHAD and TFP?

Babies with LCHAD and TFP 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 LCHAD and TFP is to prevent a metabolic crisis and to help people with LCHAD and TFP live healthier lives.

How are LCHAD and TFP treated?

Babies with LCHAD and TFP are treated and monitored by a team of specialists including a metabolic doctor and a dietician. The treatment for LCHAD and TFP 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 LCHAD or TFP?

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

Where can I get more information?

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

For more information on LCHAD and TFP, 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 long chain 3-hydroxyacyl-co-A dehydrogenase deficiency (LCHAD)/trifunctional protein deficiency (TFP). Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

