NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Long Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD) and Trifunctional Protein Deficiency (TFP)

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This is done to identify a number of rare but treatable conditions which usually do not have any symptoms early on. Knowing if your child has one of these conditions early in life is important, because early diagnosis allows for early treatment and better health outcomes.

Your baby had a positive newborn screen result for Long Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency or Trifunctional Protein (TFP) Deficiency. **This result does not mean that your baby has LCHAD or TFP deficiency but means the chance is increased and more testing is needed to confirm.**

What are LCHAD and TFP deficiency?

They are a type of fatty acid oxidation disorder. Our bodies normally breakdown the sugars, fats, and proteins we eat to make energy. Our cells need this energy to work properly. The main energy source is usually sugar (carbohydrates). But during long periods of time without food (fasting), our body relies more on stored fat for energy. People who have LCHAD or TFP deficiency cannot breakdown certain fats called ***long chain fatty acids*** to make energy. When fasting (such as during a cold or flu) babies with LCHAD or TFP deficiency do not make enough energy for proper cell function and can become very ill.

The newborn screen measures the amount of long chain fatty acids which are increased in babies with LCHAD and TFP deficiency. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms?

With regular feeding, babies with LCHAD or TFP deficiency usually appear healthy in the newborn period. However, during prolonged fasting episodes, a child may develop low blood sugar levels which could lead to a ***metabolic crisis***. A metabolic crisis can present as lethargy (very sleepy and unresponsive), floppiness, and irritability. If severe and left untreated, this can progress quickly to seizures and loss of consciousness. Some children may have additional health problems due to LCHAD or TFP deficiency which are monitored and managed if they arise.

In most cases, symptoms present in infancy or early childhood but there are also milder forms that do not present until later in life.

What causes LCHAD and TFP deficiency?

One of the enzymes needed to breakdown long chain fatty acids is not working properly in people affected with the condition. The enzymes needed are made in our cells by two genes called HADHA and HADHB. A gene is a set of instructions on how to make a protein or enzyme. Changes in the gene, called disease-causing variants result in the enzyme not working properly or not being made at all.

We all have two copies of the HADHA and HADHB genes. A baby born with LCHAD or TFP deficiency has received (inherited) two non-working copies of a gene, one from each parent (i.e., the baby has two non-working copies of the HADHA gene or of the HADHB gene). Parents of a child with LCHAD or TFP are usually carriers. A carrier is a person with one working and one non-working copy of a gene. People usually don't know they are a carrier because carriers are unaffected.

What happens next?

Your primary care provider will review more information with you. If needed a specialist doctor in the Biochemical Genetics Clinical Service (BGCS) will contact you to answer questions and organize a visit for your family at BC Children's Hospital in Vancouver.

The doctor will meet your baby and discuss with you further testing options to confirm if your baby has LCHAD or TFP deficiency. Testing may include a repeat newborn screen, additional blood and urine screens, and genetic testing of the HADHA and HADHB genes.

Provincial Health Services Authority

Newborn Screening BC is a collaboration of Provincial Lab Medicine Services, BC Children's Hospital and BC Women's Hospital and Health Centre, and Perinatal Services BC, all part of the Provincial Health Services Authority.

How is LCHAD or TFP deficiency treated?

The goal of treatment is to try to prevent an episode of metabolic crisis. Babies with LCHAD or TFP must not go long periods of time without eating. Dietitians and specialists will support families as they learn about the required dietary considerations and measures to avoid fasting. Children may need to go to the hospital when sick with colds or flu if they are unable to eat and drink.

Treatment is coordinated by specialists at BC Children's Hospital.

What is the outcome of treatment?

Treatment aims to prevent metabolic crises and other symptoms and help children with LCHAD and TFP to lead the healthiest lives possible.

How do you feel?

For most parents, this is an unexpected result. You may feel scared and upset which are normal feelings. If you are feeling overwhelmed or have questions, you can talk with your health care provider to discuss supports available.

Where can I get more information?

Talk to your primary care provider. You may also call the BGCS Newborn Screening Team at 604-875-2623.

Information about the BC Newborn Screening Program can be found at
www.newbornscreeningbc.ca

Information about LCHAD and TFP deficiency can be found at:

- **www.fodsupport.org/**

This fact sheet provides basic information only. It does not take the place of medical advice, diagnosis, or treatment. Always talk to your health care provider about specific health concerns.