NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Maple Syrup Urine Disease (MSUD)

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 Maple Syrup Urine Disease (MSUD). **This result does not mean that your baby has MSUD but means the chance is increased and more testing is needed.**

What is MSUD?

MSUD is a rare inherited disease. It is a type of amino acid disorder. Amino acids are the building blocks of proteins. People with MSUD cannot breakdown the following amino acids: leucine, isoleucine and valine which as a group are called ***branched chain amnio acids (BCAA).*** Because of this, harmful amounts of these amnio acids and related compounds build up in the body. This buildup can cause serious health problems if the condition is not diagnosed and treated early.

The newborn screening test measures the level of these BCAA. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms of MSUD?

Babies with MSUD are usually healthy at birth. However, without treatment, harmful substances build up and put the child at risk of having a metabolic crisis. Signs of a **metabolic crisis** include lethargy (very sleepy), poor feeding with weight loss, irritability, and vomiting. If severe and left untreated, seizures and loss of consciousness can occur.

Most of the time, symptoms present in infancy but there are also milder forms that present in later childhood.

What causes MSUD?

An enzyme needed to breakdown these BCAA is not working properly in people affected with MSUD. This enzyme is called ***branched chain ketoacid dehydrogenase (BCKAD) complex***. The enzyme is made in our cells by several genes. A gene is a set of instructions on how to make a protein or enzyme. Changes in a gene, called disease-causing variants result in little or no functioning enzyme being made.

We all have two copies of each of the genes needed to make the BCKAD enzyme. A baby born with MSUD has received (inherited) two non-working copies of a specific gene, one from each parent. Parents of a child with MSUD 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 MSUD. Testing may include a repeat newborn screen, a blood test to measure plasma amnio acids, and genetic testing.

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 MSUD treated?

The goal of treatment is to prevent metabolic crises with a special low protein diet and medications prescribed by your dietitian and specialist. In addition, babies with MSUD must not go a long time without eating. So, frequent feeds are recommended. Children may need to go to the hospital when sick with a cold 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 can prevent metabolic crises and their effects. With early diagnosis and treatment, children with MSUD have improved growth and development.

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 MSUD can be found at:

- **www.newbornscreening.info/Parents/aminoaciddisorders/MSUD.html**
- **www.msud-support.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.