NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Galactosemia (GALT)

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 has a positive newborn screen result for galactosemia. This result does not mean that your baby has galactosemia but means the chance is increased and more testing is needed.

What is Galactosemia?

Galactose is a type of sugar found in breast milk, most non-soy baby formulas and other foods. A small amount is also made naturally in the body. Babies with galactosemia cannot breakdown galactose because the enzyme needed to do this is missing or not working properly. As a result, galactose builds up in the body and can cause serious health problems if the baby is not diagnosed and treated early. The missing enzyme is called ***GALT*** (galactose-1-phosphate uridyl transferase). Some babies have a milder form of the condition in which there is some GALT activity.

The newborn screen test measures the level of GALT enzyme activity and amount of total galactose in your baby's bloodspot card. Your baby's screen showed a **low enzyme activity level**.

What are the early signs and symptoms of Galactosemia?

Babies with galactosemia are normal at birth, but serious health issues may develop without treatment. Early signs are hypoglycemia (low blood sugar), feeding problems, poor weight gain, liver injury, sleepiness, and infection.

What causes the disease?

The GALT enzyme is made by a gene called the GALT gene. A gene is a set of instructions (like a recipe) on how to make an enzyme. Changes in the GALT gene, called disease causing variants result in little or no functioning enzyme being made.

We all have two copies of the GALT gene. A baby born with galactosemia has received (inherited) two non-working copies of the GALT gene, one from each parent. Parents of a child with galactosemia are usually carriers. A carrier is a person with one working and one non-working copy of the 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 galactosemia. Testing may include a repeat newborn screen, blood test to assess your baby's liver function, and genetic testing of the GALT gene.

How is Galactosemia treated?

A galactose-restricted diet is effective in preventing many of the complications of galactosemia, including liver problems. Treatment is coordinated by specialists at BC Children's Hospital.

What is the outcome of treatment?

Early diagnosis and treatment ensure the best outcome for your baby by reducing the chance of life-threatening symptoms. Children with galactosemia continue to be followed by a metabolic specialist for treatment and monitoring.

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.

What is its incidence?

Galactosemia is estimated to affect about 1 in every 40,000 babies born.

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 galactosemia can be found at:

- **https://galactosemia.org/**
- **https://www.galactosemia.com/**

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.