NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Homocystinuria

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 homocystinuria. **This result does not mean that your baby has homocystinuria but means the chance is increased and more testing is needed.**

What is homocystinuria?

Proteins we eat are broken down by the body into different amino acids (the building blocks of protein). These amino acids are broken down further by enzymes in our cells. People with homocystinuria have trouble processing the amino acid called ***homocysteine*** and this leads to high levels of homocysteine and a related amino acid called ***methionine***. This buildup of homocysteine and methionine can cause serious health problems if the condition is not diagnosed and treated early.

The newborn screen test measures the level of methionine and homocysteine. Your baby's screen showed higher-than-expected levels of both amino acids.

What are the signs and symptoms of homocystinuria?

Babies with homocystinuria are usually healthy at birth. However, without treatment, the buildup of homocysteine can cause problems in growth, development, learning, vision, and bone health. People with homocystinuria are also at increased risk of developing blood clots.

What causes homocystinuria?

In most cases of homocystinuria, the enzyme needed to breakdown homocysteine is missing or not working properly. This enzyme is called ***cystathionine beta-synthase (CBS)***. The enzyme is made in our cells by a gene called CBS. 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 the CBS gene. A baby born with homocystinuria has received (inherited) two non-working copies of the CBS gene, one from each parent. Parents of a child with homocystinuria 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 homocystinuria. Testing may include a repeat newborn screen, another blood test to measure amino acid levels called plasma amino acids, and genetic testing.

How is homocystinuria treated?

The goal of treatment is to lower the levels of homocysteine and methionine in the body. A diet low in methionine and a special medical formula is often recommended in children with homocystinuria. Dietitians and specialists will support families as they learn about the needed dietary changes. In some cases, dietary supplementation with Vitamins B6 is highly effective.

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.

Treatment is coordinated by specialists at BC Children's Hospital

What is the outcome of treatment?

With early treatment many children have normal growth and intelligence. Early treatment also lowers the risk of the other health concerns associated with homocystinuria.

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 homocystinuria can be found at:

- **http://www.newbornscreening.info/Parents/aminoacidisorders/CBS.html**

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.