NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Biotinidase Deficiency (BIOT)

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

What is Biotinidase deficiency?

Biotinidase deficiency is an inherited condition caused by an inability to use and recycle a vitamin called ***biotin***. People with this condition do not make enough ***biotinidase enzyme*** which is responsible for recycling biotin into a useable form called "free biotin." The free biotin is needed to help metabolize some fats, proteins, and carbohydrates properly.

The newborn screen test measures the level of biotinidase enzyme activity in your baby's bloodspot card. Your baby's screen showed a **low enzyme activity level**.

What are the signs and symptoms of Biotinidase deficiency?

Babies with biotinidase deficiency are usually healthy at birth but may develop symptoms a few weeks or months after birth if not diagnosed and treated early. Symptoms include seizures, weak muscle tone (floppiness), developmental delays, vision and hearing loss, and changes to skin and hair.

What causes the disease?

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

We all have two copies of the BTN gene. A baby born with biotinidase deficiency has received (inherited) two non-working copies of the BTN gene, one from each parent.

Parents of a child with biotinidase deficiency 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 arrange a repeat newborn screen card as soon as possible. The Newborn Screening laboratory will repeat the biotinidase test on the new sample to confirm the initial result. A repeat screen is needed because there are a few sample quality issues that could cause an initial result to show a falsely low enzyme activity level. In many cases the repeat test result will be normal.

If the repeat newborn screen result is normal, then your baby is not at increased risk of biotinidase deficiency. This is a reassuring result, and no further testing is needed.

If the repeat newborn screen result is still abnormal then, a specialist doctor in the Biochemical Genetics Clinical Service (BGCS) will contact you to provide more information, 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 biotinidase deficiency. Testing may include measuring biotinidase activity level in a blood sample and genetic testing of the BTN gene.

How is Biotinidase deficiency treated?

Babies with biotinidase deficiency are treated with biotin supplementation. The treatment is life long and is very effective. If treatment is started before symptoms appear then babies with biotinidase deficiency have normal growth and development. Treatment is coordinated by specialists at BC Children's Hospital.

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?

Biotinidase deficiency is estimated to affect about 1 in every 60,000 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 biotinidase deficiency can be found at:

- **https://medlineplus.gov/genetics/condition/biotinidase-deficiency/**

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.