NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Phenylketonuria (PKU) – Amino Acid Disorder

Your baby had a newborn screen. This test was done on a blood sample collected by heel prick shortly after birth. This screen tests for a number of rare conditions. 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 PKU. This result does not mean that your baby has PKU but means the chance is increased and more testing is needed.

What is PKU?

PKU is an amino acid disorder. Amino acids are the building blocks of proteins. People with PKU cannot breakdown a particular amino acid called ***phenylalanine***. Because of this, phenylalanine builds up in their body.

What are the signs and symptoms of PKU?

In the first few weeks of life, babies with PKU usually show no signs of disease. However, without treatment, phenylalanine builds up in the body and causes irreversible harm to the baby's brain resulting in intellectual disability.

What causes the disease?

The enzyme needed to breakdown phenylalanine does not work properly in people affected with PKU. This enzyme is called ***phenylalanine hydroxylase*** (PAH). It is made in our cells by a gene called the PAH gene.

A gene is a set of instructions (like a recipe) on how to make a protein or enzyme. Some changes in the PAH gene, called disease causing variants result in the enzyme not working at all or working at a very low level.

We all have two copies of the PAH gene. A person with one working and one non-working PAH gene is called a carrier. People usually don't know they are a carrier because carriers are unaffected. A baby born with PKU has received a non-working PAH gene from both parents.

What happens next?

A specialist 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 specialist will examine your baby and discuss with you further testing options to confirm if your baby has PKU. Testing may include measuring the levels of phenylalanine and tyrosine in a blood sample and genetic testing of the PAH gene. Other tests may also be considered.

What is the treatment of the disease?

Treatment involves a special diet that is low in protein (low in phenylalanine) and sometimes medication.

If a diagnosis of PKU is confirmed, then a dietitian from the BGCS team will help support you as you learn about the special PKU diet. Babies with PKU have their health and development checked regularly with follow up tests used to monitor and adjust treatments.

What is the outcome of treatment?

Babies who are diagnosed and treated early with PKU typically have normal growth and development.

What is its incidence?

PKU affects about 1 out of every 12,000 babies born in BC.

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 family doctor. 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 PKU can be found at:

- **http://www.newbornscreening.info/Parents/aminoaciddisorders/PKU.html**
- **http://www.nspku.org**
- **http://www.geneclinics.org/**
- **http://www.marchofdimes.com/professionals/14332_1219.asp**

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.