NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Propionic Acidemia (PA)

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 Propionic Acidemia (PA). **This result does not mean that your baby has PA but means the chance is increased and more testing is needed.**

What is PA?

PA is a rare inherited condition. Babies with PA cannot properly break down certain amino acids (the building blocks of protein) and certain types of fat. Because of this too much propionic acid and other harmful substances build up in the body. This buildup can cause serious health problems if the condition is not diagnosed and treated early.

The newborn screen measures the amount propionylcarnitine and other related substances which are increased in babies with PA. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms of PA?

Babies with PA are usually healthy at birth. But without treatment, the build up of harmful substances puts the child at risk of having an episode of metabolic crisis. Signs of a **metabolic crisis** include lethargy (very sleepy), poor feeding, floppiness, irritability, vomiting, and seizures. A metabolic crisis could result in injury to the brain, heart, and other organs. If severe and left untreated, delays in development and loss of consciousness can occur.

A metabolic crisis in babies with PA can be triggered by events such as fasting, fever, or illness. Symptoms often present in the first weeks of life.

What causes PA?

In children affected with PA, an enzyme called ***propionyl-CoA carboxylase (PCC)*** is missing or not working properly. The PCC enzyme is made by two genes called PCCA and PCCB. 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 gene. A baby born with PA has received (inherited) two non-working copies of a gene, one from each parent. Parents of a child with PA 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 PA. Testing may include a repeat newborn screen, urine organic acids and genetic testing.

How is PA treated?

The goal of treatment is to try to prevent an episode of metabolic crisis. Babies with PA must not go a long time without eating. Dietitians and specialists will support families as they learn about the required dietary considerations and measures to avoid fasting. Children may need to go to the

hospital when sick with a cold or flu if they are unable to eat and drink. In addition, a low protein diet, special formula, supplements, and medications may be recommended.

Treatment is coordinated by specialists at BC Children's Hospital.

What is the outcome of treatment?

Treatment helps to prevent life-threatening episodes and other symptoms and gives babies the best chance possible for normal 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 PA can be found at:

- **www.newbornscreening.info/Parents/organicac-iddisorders/PA.html**
- **www.oaanews.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.