NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Isovaleric Acidemia (IVA)

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

What is IVA?

IVA is a rare inherited organic acid disorder. People with IVA are unable to breakdown an amino acid called ***leucine***. Amino acids are the building blocks of proteins. Because of this inability to breakdown leucine, harmful substances (such as organic acids) build up in the body. This buildup can cause serious health problems if the condition is not diagnosed and treated early.

The newborn screen test measures the level of isovalerylcarnitine which is increased in babies with IVA. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms of IVA?

Babies with IVA are usually healthy at birth. However, without treatment, harmful substances build up and put the child at risk of having a **metabolic crisis**. Signs of a metabolic crisis include lethargy (very sleepy), poor feeding with weight loss, irritability, difficulty staying warm, vomiting and the odour of sweaty feet. If severe and left untreated, seizures and loss of consciousness can occur.

A metabolic crisis can be triggered by events such as fever or illness and the first episode most often happens in infancy. The risk of having a metabolic crisis decreases as the child grows older. Some forms of IVA do not present until childhood.

What causes IVA?

The enzyme needed to breakdown leucine is missing or not working properly in people affected with IVA. This enzyme is called ***isovaleryl-CoA dehydrogenase (IVD)***. The enzyme is made in our cells by a gene called IVD. 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 IVD gene. A baby born with IVA has received (inherited) two non-working copies of the IVD gene, one from each parent. Parents of a child with IVA 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 IVA. Testing may include a repeat newborn screen, a urine test to measure organic acids, and genetic testing.

How is IVA treated?

The goal of treatment is to try to prevent a metabolic crisis. Babies with IVA must not go long periods of 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 special low protein diet and supplements are often recommended.

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?

Early treatment often allows babies with IVA to lead healthy lives with normal growth and development. A few children may develop some symptoms despite treatment. As children get older, the risk of metabolic crises decreases.

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 IVA can be found at:

- **www.newbornscreening.info/Parents/organicaciddisorders/IVA.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.