NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Tyrosinemia Type 1 (TYR1)

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 Tyrosinemia Type 1 (TYR1). **This result does not mean that your baby has TYR1 but means the chance is increased and more testing is needed.**

What is TYR1?

TYR1 is a rare inherited disorder. It is a type of amino acid disorder. Amino acids are the building blocks of proteins. People with TYR1 cannot properly breakdown the amino acid ***tyrosine***. Because of this, harmful amounts of tyrosine and its by-products, such as ***succinylacetone*** build up in the body.

The newborn screen test measures the level of succinylacetone. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms of TYR1?

Babies with TYR1 are usually healthy at birth. However, without treatment, harmful substances build up and cause liver and kidney injury. Early symptoms can include poor weight gain, lethargy (very sleepy and unresponsive), vomiting, diarrhea, and irritability. Without early diagnosis and treatment, this condition could progress to liver failure and loss of life.

What causes TYR1?

An enzyme needed to breakdown tyrosine is not working properly in people affected with TYR1. This enzyme is called ***fumarylacetoacetase (FAH)***. The enzyme is made in our cells by a gene called FAH. 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 FAH gene. A baby born with TYR1 has received (inherited) two non-working copies of the FAH gene, one from each parent. Parents of a child with TYR1 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 TYR1. Testing may include a repeat newborn screen, a urine test to measure organic acids, and genetic testing.

How is TYR1 treated?

Children with TYR1 are treated with a medication called Nitisinone. This medication helps to prevent the build up of harmful substances like succinylacetone. A special low protein diet is also recommended. Dietitians and specialists will support families as they learn about the special diet.

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

What is the outcome of treatment?

Early treatment helps to prevent serious health problems such as liver and kidney disease and neurological problems.

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.

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 TYR1 can be found at:

- **www.newbornscreening.info/Parents/aminoaciddisorders/Tyrosinemia.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.