NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Glutaric Aciduria Type 1 (GA1)

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 a condition called Glutaric aciduria type 1 (GA1). **This result does not mean that your baby has GA1 but means the chance is increased and more testing is needed.**

What is GA1?

GA1 is a rare inherited condition that causes a problem in protein metabolism (breakdown). Proteins are made up of building blocks called amnio acids. Babies with GA1 cannot properly breakdown, the following amnio acids: lysine, hydroxylysine, and tryptophan. This is because the enzyme needed to do this is missing or not working properly. This enzyme is called ***glutaryl-CoA dehydrogenase.*** As a result, an acid called glutaric 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 of glutarylcarnitine which is increased in babies with GA1. Your baby's screen showed a higher-than-expected level.

What are the signs and symptoms of GA1?

Babies with GA1 are usually healthy in the newborn period. 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 and unresponsive), poor feeding, floppiness, irritability, jitteriness, and vomiting. If left untreated, seizures and loss of consciousness can occur.

The risk of a metabolic crisis is highest during infancy and can be triggered by events such as fasting, fever, or illness. The risk of having a metabolic crisis decreases as the child grows older.

What causes GA1 deficiency?

The glutaryl-CoA dehydrogenase enzyme is made by a gene called GCDH gene. A gene is a set of instructions on how to make a protein or enzyme. Changes in the GCDH gene, called disease-causing variants result in little or no functioning enzyme being made.

We all have two copies of the GCDH gene. A baby born with GA1 has received (inherited) two non-working GCDH genes, one from each parent. Parents of a child with GA1 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 GA1. Testing may include a repeat newborn screen, urine organic acids and genetic testing of the GCDH gene.

How is GA1 treated?

The goal of treatment is to try to prevent a metabolic crisis. Babies with GA1 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 is recommended.

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

What is the outcome of treatment?

With early diagnosis and treatment, most individuals with GA1 remain healthy.

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 GA1 can be found at:

- **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.