NEWBORN SCREENING BC

Blood spot card screening | Parent information sheet

Guanidinoacetate Methyltransferase Deficiency (GAMT)

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 Guanidinoacetate Methyltransferase (GAMT) deficiency. This result does not mean that your baby has GAMT deficiency but means the chance is increased and more testing is needed.

What is GAMT deficiency?

GAMT deficiency is a rare inherited disorder. People with GAMT deficiency do not make enough creatine. In our cells ***creatine*** plays an important role in energy supply and storage. Without enough creatine, cells in our body, particularly brain and muscle cells, do not get enough energy to function properly. The process to make creatine in people with GAMT deficiency is not working and this results not only in low creatine levels but also in a buildup of a harmful substance called ***guanidinoacetate (GAA)***. The low creatine and high GAA levels can cause serious health problems if the condition is not diagnosed and treated early.

The newborn screen test measures the level of GAA and looks for genetic changes in the GAMT gene if the GAA level is increased. Your baby's screen showed a higher-than-expected GAA level and changes in the GAMT gene.

What are the signs and symptoms of GAMT deficiency?

Babies with GAMT deficiency are usually healthy at birth because their mothers provided them with creatine during the pregnancy. But without treatment, symptoms develop in early childhood. This condition mainly affects the brain and muscles. Symptoms include intellectual disability, seizures, speech problems, and abnormal movements. The symptoms and their severity can vary between children with GAMT deficiency.

What causes GAMT deficiency?

One of the enzymes needed to make creatine is missing or not working properly in people affected with GAMT deficiency. This enzyme is called ***guanidinoacetate methyltransferase***. The enzyme is made in our cells by a gene called GAMT. 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 GAMT gene. A baby born with GAMT deficiency has received (inherited) two non-working copies of the GAMT gene, one from each parent. Parents of a child with GAMT deficiency 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 GAMT deficiency. Testing may include a repeat newborn screen, retesting GAA and creatine levels in blood and urine samples, and possibly additional genetic testing.

How is GAMT treated?

The mainstay of treatment is an oral creatine supplement along with dietary changes to limit the production of GAA. This may include a natural protein restricted diet and supplementation with a special medical formula.

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?

When treatment starts early, there is a much better chance for normal growth, development, and intelligence.

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 GAMT can be found at:

- **https://rarediseases.info.nih.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency**
- **https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency**

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.