INFORMATION FOR PARENTS/GUARDIANS:

My Baby Has a Positive Guanidinoacetate Methyltransferase Deficiency Newborn Screening Result

What is newborn screening?

These are routine tests done shortly after birth on every baby born in Ontario. A small sample of blood is taken from the heel of your baby and is tested for rare, treatable diseases, including guanidinoacetate methyltransferase (GAMT) deficiency.

What does it mean if my baby has a positive GAMT deficiency newborn screening result?

This result does **not** mean that your baby has GAMT deficiency. It means that more testing is needed because your baby **might** have GAMT deficiency. Babies with GAMT deficiency are healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out if your baby has this condition.

You may feel worried about your baby's screen positive result. Many parents in this situation feel this way. Remember, we do not know for sure that your baby has GAMT deficiency until follow up testing has been done.

What is GAMT deficiency?

GAMT deficiency is a very rare inherited (genetic) disease that affects how the body makes creatine. Creatine is a source of energy for our cells, in particular the brain and muscles. Creatine is important for normal growth and development.

If the body does not have enough creatine health problems like seizures can develop along with low muscle tone, muscle weakness, movement disorders, and intellectual disabilities.

GAMT deficiency is very rare. It is thought to affect between 1 in every 550,000 and 1 in every 2,500,000 babies born in Ontario. There are fewer than 150 people diagnosed with GAMT deficiency in the whole world.

What causes GAMT deficiency?

GAMT deficiency happens when the enzyme guanidinoacetate methyltransferase is either missing or not working properly. The job of this enzyme is to create creatine from guanidinoacetate. When the guanidinoacetate methyltransferase enzyme is not working properly, creatine levels are low and guanidinoacetate (GAA) levels are high.

How do I find out if my baby has GAMT deficiency?

Blood and urine tests are done to find out if a baby who screened positive actually has GAMT deficiency.

When can my baby have these tests?

Your baby's doctor or a health care professional at a Newborn Screening Treatment Centre will call you to talk about the results of your baby's positive newborn screen and arrange more testing as soon as possible.

Why screen for GAMT deficiency?

Babies who have GAMT deficiency look normal when they are born but will develop serious health problems and intellectual disability if they are not treated. The goal of newborn screening for GAMT deficiency is to identify and treat babies with GAMT deficiency early to prevent health and development problems.

How is GAMT deficiency treated?

Babies with GAMT deficiency are treated and monitored by a team of health care specialists including a metabolic doctor and a dietician. The treatment is lifelong and can include certain supplements, medication and a special diet. Babies with GAMT deficiency are checked often for their creatine and GAA levels, development and other health issues associated with GAMT deficiency.

How does a baby get GAMT deficiency?

GAMT deficiency is an inherited (genetic) disease. A baby with GAMT deficiency inherited two non-working copies of the *GAMT* gene, one copy from each parent. People who have one non-working copy of the *GAMT* gene are called "carriers." Carriers of GAMT deficiency are healthy, do not have, and will never develop, symptoms of GAMT deficiency.

Where can I get more information?

For more information on newborn screening, please talk to your local health care provider or visit the Parents section of our website at www.newbornscreening.on.ca.

For more information on GAMT deficiency, please visit the Association for Creatine Disorders website at https://creatineinfo.org/.

NOTE TO PARENTS/GUARDIANS: This information is only for parents whose baby has had a positive newborn screening result for GAMT deficiency. Please remember that this fact sheet was written for information purposes only. The fact sheet should not replace professional medical advice, diagnosis or treatment.

415 Smyth Road
Ottawa Ontario K1H 8M8
Toll-free: 1877-NBS-8330 / Local: (613)738-3222
www.newbornscreening.on.ca @NBS_Ontario