Guanidinoacetate Methyltransferase (GAMT) Deficiency

What is newborn screening?

Newborn screening (NBS) is done soon after birth to check for health conditions that can be hidden at birth. To do the screening, a nurse takes a few drops of blood from your baby's heel. This blood sample is done on all newborns.

Please remember that just because the newborn screen flags for a disorder, it does not mean that your child has a diagnosis of that disorder. It means that your child should have further testing.

KEY POINTS:

- **You have just heard that your baby might have Guanidinoacetate Methyltransferase (GAMT) Deficiency. Please understand that the newborn screening is just that: a screening test. Further testing is needed before a diagnosis can be made.**
- **When GAMT is detected early and treatment is started, many babies with the condition are able to live longer lives with improved health and development.**

What does my baby's newborn screen show?

Your baby's newborn screen flagged for a condition called Guanidinoacetate Methyltransferase deficiency. It is commonly called GAMT deficiency, and sometimes, just "GAMT". Not all babies with an out-of-range screening result will go on to get a diagnosis of GAMT deficiency. However, since some babies will be diagnosed with GAMT, it is important to have all follow-up tests done as soon as possible, so that treatment can be started, if needed. Detecting the condition early and beginning treatment might help prevent or delay symptoms associated with GAMT deficiency.

What is GAMT deficiency?

GAMT deficiency is a rare inherited condition. Less than 150 people with GAMT deficiency have been identified. GAMT deficiency affects the body's ability to make creatine. Creatine is important for muscles and for the brain. Without a good supply of creatine, the brain and muscles have a hard time storing energy and a hard time using energy when needed. This can cause neurological problems such as intellectual disability, limited speech development, seizures, behavioral problems, and involuntary muscle movements. However, early diagnosis and treatment is thought to improve the health and development in children affected by GAMT deficiency.

What early signs are seen with GAMT deficiency?

People with untreated GAMT deficiency can develop symptoms anytime between age 3 months to 3 years. Early signs of GAMT deficiency may include:

- Being late to sit on own
- Being late to start walking
- Late to start talking
- Poor muscle tone (hypotonia)
- Involuntary movements (tremors or tics)
- Epilepsy/seizures

Without treatment, people with GAMT deficiency can go on to have intellectual disability, behavior disorders (such as hyperactivity and autism), and might only say a few words.

What are the next steps?

Your baby's doctor will contact you to arrange for your baby to have additional testing. It is very important that you bring your baby for the needed testing as soon as possible.

Follow-up testing will involve a blood draw and collecting a urine sample to check your baby's blood and urine for signs of GAMT deficiency. Genetic testing may also be recommended. A specialized MRI type test, called magnetic resonance (MR) spectroscopy, may also be used to see if creatine levels are reduced in the brain.

Children with GAMT will need to be followed by a team of healthcare workers. They should see their regular doctor (pediatrician) and healthcare providers who specialize in GAMT disease. Your baby's doctor will help arrange for a clinic visit with specialists familiar with GAMT. The healthcare team will discuss symptoms and how to monitor and treat your child in more detail.

What if I still have questions?

SCAN HERE for Information about *NEWBORN SCREENING* including TIPS for preparing for blood draws and collecting urine samples on newborns;, and more

We understand that this can be an overwhelming and emotional process. Many families have questions and concerns. The Connecticut Newborn Screening Network (the Network) is available to put you in touch with the best resource. To reach the Network, you can call 860-837-7870, Monday-Friday, 8:30am-4:30pm. We also recommend the website www.babysfirsttest.org and the Association for Creatine Deficiencies https://creatineinfo.org as resources.

This fact sheet contains general information and is for information purposes only. Every child is different and some of this information may not apply to your child specifically. This sheet does not replace medical advice, diagnosis, or treatment from your child's healthcare provider.

Connecticut Department of Public Health
Connecticut Newborn Screening Program • 860.920.6628
Connecticut Newborn Screening Network • 860.837.7870