NORTH CAROLINA NEWBORN SCREENING PROGRAM

Guanidinoacetate Methyltransferase (GAMT) Deficiency Fact Sheet for Parents

All babies born in North Carolina are screened at birth to look for certain health problems that can be treated if caught early. The newborn screening result showed that your baby might have guanidinoacetate methyltransferase (GAMT) deficiency. Your baby will be referred to a specialist for more testing to know for sure.

There are usually no signs of GAMT deficiency at birth.

What happens next?

Babies with GAMT deficiency are healthier if treatment begins early, so it is important to have follow-up testing done quickly to find out whether your baby has this condition. Diagnostic tests include testing for guanidinoacetate (GUAC), an amino acid, and low creatine in the blood after a positive newborn screening test. Genetic testing can also be helpful.

What is GAMT deficiency?

GAMT deficiency is a rare genetic condition where the body does not produce enough of the enzyme GAMT. GAMT is needed to make creatine. Creatine provides energy to all cells in the body and is needed for muscle and brain development. Without treatment, GAMT deficiency can damage the brain and muscles. Early diagnosis and treatment can help children with GAMT deficiency live healthy lives.

What are the symptoms of GAMT deficiency?

Symptoms, or signs, of GAMT deficiency may appear anytime from 3 months to 3 years of age. They may include:

- Delayed sitting or walking
- Delayed speech
- Muscle weakness
- Uncontrolled movements
- Seizures or epilepsy

How is GAMT deficiency treated?

GAMT deficiency is a treatable condition. Early diagnosis and consistent treatment have been shown to prevent development of intellectual disability and other symptoms of GAMT deficiency. Oral supplements of creatine and ornithine and a protein restricted diet are typically prescribed. Children with GAMT deficiency should see their regular doctor, a doctor who specializes in GAMT deficiency, and a dietitian.

Where do I go for more information?

Use your phone's camera to scan the QR codes below.

Association for Creatine Deficiencies

Newborn Screening Information Center

Baby's First Test

UNC Pediatric Genetics and Metabolism

NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

State of North Carolina Department of Health and Human Services Division of Public Health
www.ncdhhs.gov
https://slph.dph.ncdhhs.gov/newborn/default.asp
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