NORTH CAROLINA NEWBORN SCREENING PROGRAM

Guanidinoacetate Methyltransferase (GAMT) Deficiency Fact Sheet for Providers

What is GAMT deficiency?

Guanidinoacetate methyltransferase (GAMT) deficiency is an autosomal recessive condition caused by deficient GAMT activity, impairing the ability to synthesize creatine by methylation of guanidinoacetate (GUAC). Without creatine, the body cannot use and store energy. This inability to use and store energy primarily affects brain and muscle function. Children who receive early and ongoing treatment for GAMT deficiency can live healthy lives.

What are the symptoms of GAMT deficiency?

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

- Developmental delay
- Cognitive impairment
- Seizures
- Delayed speech
- Behavioral problems
- Muscle weakness and hypotonia
- Involuntary movements

How is GAMT deficiency diagnosed?

The diagnosis of GAMT deficiency is based on increased levels of GUAC and low levels of creatine in the plasma and urine resulting in an elevated GUAC-to-creatine ratio. Genetic testing for variants in the GAMT gene may be required to confirm the diagnosis.

How is GAMT deficiency treated?

Treatment aims to reduce GUAC production and supply the creatine that is not produced by the body. Patients are typically prescribed oral supplements of creatine monohydrate and L-ornithine. Depending on the patient, a provider may also recommend diet restrictions and sodium benzoate supplementation to further minimize GUAC accumulation. Treatments may include the following:

- Creatine monohydrate and L-ornithine supplements
- Medications to treat seizures
- Speech, occupational, and behavior therapy
- Sodium benzoate
- Special diet low in protein (specifically arginine)

NORTH CAROLINA NEWBORN SCREENING PROGRAM

Mucopolysaccharidosis I Fact Sheet for Providers

How do I handle an abnormal screening for GAMT deficiency?

- Inform the family of the newborn screening result and ascertain clinical status.
- Provide the family with the GAMT deficiency parent handout developed by the North Carolina Newborn Screening Program.
- Evaluate the newborn (newborns are expected to be asymptomatic).
- Follow the instructions faxed by the North Carolina Newborn Screening Coordinator regarding follow-up testing, referrals, and treatment. Reach out to the Newborn Screening Coordinator if you have additional questions.
- Report final diagnostic outcome to the North Carolina Newborn Screening Program.

Where do I go for more information?

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

ACT Sheet

Gene Reviews

National Organization for Rare Disorders

Medline Plus

Where do I send parents for 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

NCDHHS is an equal opportunity employer and provider.

This fact sheet was supported by Health Resources and Services Administration (HRSA), Department of Health and Human Services (HHS) as part of a financial assistance award totaling $344,348 with 100 percent funded by HRSA/HHS. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA/HHS or the U.S. Government.