GUANIDINOACETATE METHYLTRANSFERASE DEFICIENCY

NEWBORN SCREENING (NBS) IN FLORIDA

What is GAMT deficiency?

Guanidinoacetate methyltransferase (GAMT) deficiency is an inherited condition which affects the body's ability to produce creatine. With GAMT deficiency, the body does not have enough of the enzyme GAMT to break down guanidinoacetate (GUAC) into creatine, which leads to a deficiency of the substance. Without a sufficient amount of creatine, the brain and muscles of the body will have difficulty properly using and storing energy.

Incidence

GAMT deficiency affects 1 in 250,000 to 1 in 550,000 individuals.

Signs and Symptoms of GAMT

The signs and symptoms of GAMT deficiency can vary among those diagnosed with the disease.

Symptoms can include:

- Intellectual disability
- Seizures or epilepsy
- Speech delays
- Involuntary movements
- Developmental delays
- Behavioral abnormalities
- Hyperactivity

The onset of symptoms is usually between three months and two years of age. Early identification and treatment can lead to better health outcomes.

FLORIDA'S METHOD OF SCREENING

First Tier Screening

Screening for GAMT deficiency is completed by an analysis of amino acids using tandem mass spectrometry testing.

Second Tier Screening

Samples with out-of-range first tier results will undergo additional testing of GUAC and creatine.

What to expect if the NBS has out-of-range results for GAMT?

Each out-of-range result is verified by nursing staff with the NBS Follow-up Program. The following actions are needed, depending on the result:

- **Borderline Results:** A repeat NBS will be requested for infants with an initial out-of-range screening result. Once received, the results of the repeat specimen would be evaluated for additional steps.
- **Presumptive Positive Results:** If the repeat specimen remains out-of-range results for GAMT, the infant will be referred to one of three contracted genetics referral centers for diagnostic evaluation.

It is important to remember that out-of-range results do NOT mean an infant has GAMT deficiency. Additional testing is needed to confirm or rule out a diagnosis.

GAMT INFORMATION AND RESOURCES

Gene Reviews
NCBI.NLM.NIH.gov/books/NBK3794

Genetic and Rare Diseases Information Center
RareDiseases.Info.NIH.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency

Genetics Home Reference
MedlinePlus.gov/genetics/condition/guanidinoacetate-methyltransferase-deficiency

Baby's First Test
BabysFirstTest.org/newborn-screening/conditions/guanidinoacetate-methyltransferase-deficiency

Florida Department of Health
Division of Children's Medical Services
Florida Newborn Screening Program
850-245-4201 | CMS.NBS@FLHealth.gov

For additional information, visit
FloridaNewbornScreening.com