TEXAS Health and Human Services

Newborn Screening ACT Sheet

Guanidinoacetate Methyltransferase (GAMT) Deficiency

Condition Description: Guanidinoacetate methyltransferase (GAMT) deficiency is an autosomal recessive condition caused by deficient GAMT enzymatic activity, impairing the ability to synthesize creatine by methylation of guanidinoacetate. Without creatine, the body is unable to use and to store energy which affects brain and muscle function, causing seizures, developmental delay, impaired speech development, behavioral changes, hypotonia, and movement disorders.

Please Take the Following Immediate Actions:

- ☐ **Immediately consult with pediatric metabolic specialist (See attached list).**
- ☐ **Contact family.** Inform them of the newborn screening result. Newborns are expected to be asymptomatic.
  There is a resource for this conversation on page 6: **Next steps after positive Newborn Screening for GAMT deficiency**
- ☐ **Obtain initial labs:** plasma guanidinoacetate (GUAC) levels , plasma creatine, and plasma creatinine.
- ☐ **Collect repeat screen (between 7-14 days of life)** if the second screen has not been done.
- ☐ **FAX lab results to {SGUserFax}**

Confirmation of diagnosis: Guanidinoacetate levels in urine and plasma levels are characteristically increased. Creatine and creatinine levels in urine and plasma are (relatively) low. Molecular genetic testing may be required to confirm the diagnosis.

Clinical expectations: GAMT deficiency typically presents between 3 months to 3 years of age with developmental delay, hypotonia, seizures, and behavioral disorders such as autism or self-injury. About 30% of patients have a movement disorder such as ataxia or have other involuntary movements. Treatment is directed at promoting normal growth and development by the restoration of creatine levels and the reduction of guanidinoacetate. This is accomplished by creatine supplementation and reduction of guanidinoacetate concentrations using protein restriction, ornithine, and benzoate supplementation. Dietary therapy should be administered under the guidance of a metabolic specialist.

Additional information:
How to Communicate Newborn Screening Results https://www.hrsa.gov/sites/default/files/hrsa/advisory-committees/heritable-disorders/resources/achdnc-communication-guide-newborn.pdf
MedlinePlus https://medlineplus.gov/genetics/condition/guanidinoacetate-methyltransferase-deficiency/
Creatine Deficiency Disorders Gene Reviews https://www.ncbi.nlm.nih.gov/books/NBK3794/
NIH https://rarediseases.info.nih.gov/diseases/2578/guanidinoacetate-methyltransferase-deficiency
HRSA https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency