Provider action sheet for guanidinoacetate methyltransferase deficiency (GAMT)

If the California Newborn Screening (NBS) Program has identified an infant in your care who has a screen-positive result for guanidinoacetate methyltransferase deficiency (GAMT).

What steps do I need to take?

1. **Consult** with a California Children's Services Metabolic Special Care Center (SCC) specialist. They will ask for your assessment of the infant's current health status and if the infant has any signs of the disorder. Further evaluation and confirmatory tests will be arranged by the SCC.
2. **Contact** the family to explain the positive newborn screening test result. For information on how best to communicate, consult the **Health Resources & Services Administration Newborn Communication Guide** (https://bit.ly/HRSAGuide) or search online for "HRSA" "heritable" "communication").
3. **Review** the accompanying, "Family action sheet for guanidinoacetate methyltransferase deficiency (GAMT)" with the family and ensure they understand.
4. **Inform** the family that this result requires more testing and evaluation. Emphasize that you, with NBS Program Area Service Center (ASC) staff and the SCC specialist, will guide them through the next steps of confirmatory testing and follow-up services.
5. **Advise** parents to (1) follow the plan for confirmatory testing right away, (2) keep their infant's appointments with the SCC specialist, and (3) start treatment if indicated. The family should receive services from a team of specialists, including genetic counseling.

Clinical information

GAMT is an autosomal recessive disorder caused by pathogenic variants in the *GAMT* gene which is responsible for making GAMT enzyme. This enzyme helps convert guanidinoacetate (GUAC) into creatine which is needed for the body to store and use energy. Deficiency in the GAMT enzyme results in creatine deficiency and in the accumulation of GUAC in the cells. This leads to severe organ and tissue damage, particularly of the brain and muscles.

The clinical presentations of GAMT are non-specific and can appear anytime from 3 months to 3 years of age. Symptoms may include developmental delay, cognitive impairment, seizures, delayed speech, muscle weakness, and involuntary movements.

Early and lifelong treatments include creatine monohydrate and ornithine supplements, sodium benzoate, and arginine-restricted diet to increase cerebral creatine levels and decrease GUAC levels.

Please visit these sites. Search for the site name and "GAMT".

- **HRSA** (https://newbornscreening.hrsa.gov/conditions/guanidinoacetate-methyltransferase-deficiency)
- **GeneReviews** (https://www.ncbi.nlm.nih.gov/books/NBK3794)

Questions?

For follow-up questions, please call your NBS Program Area Service Center.

For program questions, please email NBS Program staff at NBS@cdph.ca.gov or visit the NBS Program website (www.cdph.ca.gov/NBS).

California Department of Public Health
Newborn Screening Program (www.cdph.ca.gov/NBS)