

2022- Public Health System Impact Assessment for Guanidinoacetate methyltransferase deficiency- NBS

Initial Survey of the Advisory Committee on Heritable Disorders in Newborns and Children's Public Health System Assessment

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The purpose of this survey is to inform the Advisory Committee on Heritable Disorders in Newborns and Children (Committee) about states' ability to add newborn screening (NBS) for Guanidinoacetate methyltransferase (GAMT) deficiency using information gathered from most of the state and territorial NBS programs in the U.S. Your input will provide valuable information and aid the deliberations of the Committee. Please refer to the (GAMT) deficiency screening factsheet to help you answer the following questions about the ability of your state or territory to add screening for GAMT deficiency to your NBS program. Please consult with others, as needed, including laboratory and follow-up staff, medical professionals and specialists, to complete the survey. When unsure about a response, please provide your best estimate. If you were to answer every question, we estimate it will take an average of 10 hours to complete this form.

1. Within the last three years, your state has: (check all that apply)

- ☐ Included GAMT deficiency as part of the routine NBS panel.
- ☐ Planned, implemented, or completed any type of pilot study or pilot evaluation for GAMT deficiency.
- ☐ Issued a mandate or state-level decision to start screening for GAMT deficiency.
- ☒ None of the above

2. Which of the following entities provide NBS laboratory services for your state's NBS program?

- ☐ Your own state's public health or NBS laboratory
 - ☐ A state university laboratory for which there is an intra-state agency agreement
 - ☐ A contracted regional NBS laboratory
 - ☐ A contracted commercial laboratory
 - ☐ Other – please specify: _____
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NBS programs consider many factors when deciding to add a condition to their NBS panel. The following question asks you to consider, in general, how much the following factors would be an issue when considering adding GAMT deficiency to your NBS panel.

3. Please indicate if the following implementation factors for GAMT deficiency would present a major challenge, a minor challenge, or would not be a challenge, given the current status of the NBS Program in your state.

	Major Challenge	Minor Challenge	Not a Challenge
Availability of a validated screening test in your state	☐	☐	☐
Ability to conduct short-term follow-up for out-of-range screening results, including tracking and follow-up testing	☐	☐	☐
Identifying specialists in your state (or region) who can treat newborns and children with GAMT deficiency	☐	☐	☐
Availability of treatment for in your state GAMT deficiency	☐	☐	☐
Increasing your NBS fee	☐	☐	☐
Addressing administrative challenges (please specify in comments section)	☐	☐	☐

Please share additional comments below.

4. Please describe any additional overarching challenges.

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For questions 5-7 please assume that GAMT deficiency has been authorized for addition to your state's panel and funds for laboratory testing and follow-up have been made available.

5. The following question considers the various resources needed (e.g. human resources, facilities, etc.) by your NBS program in order to implement screening for GAMT deficiency.

5a.

	Have already	Do not have but can get within 1 year	Cannot get within 1 year
Screening method for GAMT deficiency: MS/MS using a derivatized or non-derivatized assay	☐	☐	☐
Quantity and type of laboratory equipment needed to screen for GAMT deficiency	☐	☐	☐
Laboratory technical expertise to screen for GAMT deficiency	☐	☐	☐
Sufficient number of technical staff to screen for GAMT deficiency	☐	☐	☐
LIMS capacity and instrumentation interface	☐	☐	☐
Sufficient number of NBS staff to notify and track NBS results	☐	☐	☐
Access to appropriate diagnostic services after an abnormal or out of range screening result is reported (e.g., diagnostic testing, clinical evaluations)	☐	☐	☐
Genetic counselors, or other staff with the necessary expertise, to cover the expected caseload	☐	☐	☐
Specialists to cover expected GAMT deficiency caseload	☐	☐	☐
Treatment centers for expected GAMT deficiency caseload	☐	☐	☐
Follow-up protocols for GAMT deficiency cases	☐	☐	☐

Please share additional comments below.

5b.

	Have already	Do not have but can get within 1 year	Cannot get within 1 year
Availability of the screening test in the state university laboratory for which there is an intra-state agency agreement, or contracted regional laboratory, or commercial laboratory	☐	☐	☐
LIMS capacity and instrumentation interface	☐	☐	☐
Sufficient number of NBS staff to notify and track NBS results	☐	☐	☐
Access to appropriate diagnostic services after an abnormal or out of range screening result is reported (e.g., diagnostic testing, clinical evaluations)	☐	☐	☐
Genetic counselors, or other staff with the necessary expertise, to cover the expected caseload	☐	☐	☐
Specialists to cover expected GAMT deficiency caseload	☐	☐	☐
Treatment centers for expected GAMT deficiency caseload	☐	☐	☐
Follow-up protocols for GAMT deficiency cases	☐	☐	☐

Please share additional comments below.

5c. Would you conduct a second-tier test for GAMT deficiency?

- ☐ Yes, we could be ready in the next year
 - ☐ Most likely, but we would not be ready in the next year
 - ☐ Yes, but we would have to contract the second-tier test
 - ☐ No we do not think it is necessary to conduct second-tier testing
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6. Please indicate the degree* to which these factors impede or facilitate your ability to adopt screening for GAMT deficiency in your state.

	Major Barrier	Minor Barrier	Minor Facilitator	Major Facilitator	Not Applicable
Predicted run time to screen for GAMT deficiency as it relates to other workload	☐	☐	☐	☐	☐
Extent to which the screening test for GAMT deficiency can be multiplexed with screening for other conditions	☐	☐	☐	☐	☐
Other ongoing NBS program activities (e.g., addition of other conditions, other quality improvements)	☐	☐	☐	☐	☐
Estimated cost per specimen to conduct screening (personnel, equipment, reagents)	☐	☐	☐	☐	☐
Estimated cost of treatment for newborns diagnosed with GAMT deficiency	☐	☐	☐	☐	☐
Expected clinical outcomes of newborns identified by screening	☐	☐	☐	☐	☐
Expected cost-benefit of screening in your state	☐	☐	☐	☐	☐
Advocacy for screening for this GAMT deficiency	☐	☐	☐	☐	☐
Other non-NBS public health priorities within your state	☐	☐	☐	☐	☐

Please share additional comments below.

Major barrier- Will prevent testing from being implemented effectively and/or timely.
Minor barrier- May compromise testing so it is not performed effectively and/or timely.
Minor facilitator- May allow testing to be done effectively and/or timely.
Major facilitator- Will allow testing to be done effectively and/or timely.

7. Please describe any additional factors that impede or facilitate adoption of screening for GAMT deficiency in your state.

8a. What are the most significant barrier(s) to screening for GAMT deficiency in your state?

8b. What would most facilitate screening for GAMT deficiency in your state?

9. Please estimate the time it would take your NBS program to initiate screening for GAMT deficiency in your state (i.e. get authority and funds to screen for GAMT deficiency , go through administrative processes, meet with your state NBS committees and complete all activities needed to implement and commence screening for all newborns in your state)?

- ☐ 12 months or less
 - ☐ 13 to 24 months
 - ☐ 25 to 36 months
 - ☐ 37 to 48 months
 - ☐ More than 48 months
-

10. The question above related to the overall timeline. We recognize some of the activities happen in tandem and some cannot begin until a previous activity has been completed. Please estimate the total time needed, in general, for each individual activity listed below within your NBS program. If needed, please consult with laboratory and follow-up staff, medical professionals and specialists, prior to completing the survey.

10a.

	12 months or less	13 – 24 months	25 – 36 months	37 to 48 months	> 48 months	Not Applicable
Obtain authorization to screen for GAMT deficiency	☐	☐	☐	☐	☐	☐
Availability of funds to implement screening for GAMT deficiency	☐	☐	☐	☐	☐	☐
Meet with Advisory committees and other stakeholders	☐	☐	☐	☐	☐	☐
Obtain and procure equipment for screening for GAMT deficiency	☐	☐	☐	☐	☐	☐
Hire necessary laboratory and follow-up staff	☐	☐	☐	☐	☐	☐
Select, develop, and validate the screening test within your laboratory	☐	☐	☐	☐	☐	☐
Develop a screening algorithm, follow-up protocols, and train follow up staff	☐	☐	☐	☐	☐	☐
Set up reporting and results systems for added condition (e.g., LIMS)	☐	☐	☐	☐	☐	☐
Collaborate with specialists and clinicians in the community to determine which diagnostic tests will be recommended upon identification of an out of range NBS result	☐	☐	☐	☐	☐	☐
Conduct an internal validation study for GAMT deficiency	☐	☐	☐	☐	☐	☐
Pilot test the screening process within your state, after validation has taken place	☐	☐	☐	☐	☐	☐
Implement statewide screening for all newborns, including full reporting and follow-up of abnormal screens after validation and pilot testing	☐	☐	☐	☐	☐	☐

Please share additional comments below.

10b.

	12 months or less	13 – 24 months	25 – 36 months	37 to 48 months	> 48 months	Not Applicable
Obtain authorization to screen for GAMT deficiency	☐	☐	☐	☐	☐	☐
Availability of funds to implement screening for GAMT deficiency	☐	☐	☐	☐	☐	☐
Meet with Advisory committees and other stakeholders	☐	☐	☐	☐	☐	☐
Develop follow-up protocols, and train follow up staff	☐	☐	☐	☐	☐	☐
Set up reporting and results systems for added condition (e.g., LIMS)	☐	☐	☐	☐	☐	☐
Collaborate with specialists and clinicians in the community to determine which diagnostic tests will be recommended upon identification of an out of range NBS result	☐	☐	☐	☐	☐	☐
Add the screening test to the existing outside laboratory contract	☐	☐	☐	☐	☐	☐
Implement statewide screening for all newborns, including full reporting and follow-up of abnormal screens after validation and pilot testing	☐	☐	☐	☐	☐	☐

Please share additional comments below.

11. Are there any special considerations regarding GAMT deficiency that need to be taken into account when assessing the impact on the public health system? (e.g. variants of unknown significance, pseudodeficiencies, age of onset, access to specialists, access to treatment, cost of treatment, etc.). Please describe:

12. Please share any additional information regarding implementation of NBS for GAMT deficiency.

13. Please provide information about the respondent:

- ☐ Name: _____
 - ☐ Phone number: _____
 - ☐ Email address: _____
 - ☐ Job title: _____
-

14. Who did you consult with to answer these questions? Please check all that apply.

- ☐ State NBS laboratory experts
- ☐ Other NBS program staff
- ☐ State NBS advisory board
- ☐ State Title V Director
- ☐ GMAT deficiency Specialists
- ☐ Primary care providers
- ☐ Advocates within your state for GMAT deficiency screening
- ☐ Others- please specify: _____
- ☐ ☒ None of the above

End of Survey

Thank you for completing the survey!
To have your responses recorded please click on Submit button below.