Delayed Implementation of Newborn Screening for Guanidinoacetate Methyltransferase (GAMT) Deficiency

GS 130A-125(b)

Report to the

Joint Legislative Oversight Committee on Health and Human Services

by

North Carolina Department of Health and Human Services

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Background

The Recommended Uniform Screening Panel (RUSP) is a list of biochemical and genetic disorders promulgated by the Secretary of the United States Department of Health and Human Services. The RUSP serves as a guide for state newborn screening (NBS) programs when adding conditions to their mandated screening panels. General Statute 130A-125(b) directs the North Carolina Department of Health and Human Services (DHHS) to include screening for conditions added to the RUSP within three (3) years of the addition. Furthermore, when a delay adding a RUSP-identified condition to the Newborn Screening Program exceeds three years, DHHS must submit a report to the Joint Legislative Oversight Committee on Health and Human Services every six months following the three-year delay.

Guanidinoacetate Methyltransferase (GAMT) deficiency was added to the RUSP on January 4, 2023. As of July 4, 2026, the NC NBS program has not initiated screening for GAMT deficiency. The purpose of this report is to explain the delay and provide a timeline for implementation.

The process to onboard screening for additional disorders is complex as newborn screening is more than a simple test. The public health laboratory testing is a critical, core component, but it is just one piece of a broader public health system working for families. Every state newborn screening program has six essential parts: screening, follow-up, diagnosis, management, evaluation, and education, and DHHS must ensure that the full system is in place before screening can begin.

When GAMT deficiency was added to the RUSP, the NC NBS Program was on the precipice of launching screening for both Mucopolysaccharidosis Type I and Pompe Disease, which began on February 13, 2023. In addition, the Program was in the early stages of a plan to implement screening for Mucopolysaccharidosis Type II (MPS-II), which had been added to the RUSP on August 2, 2022. An assessment performed by the NC NBS Program revealed that implementation of screening for MPS-II and GAMT deficiency would require additional laboratory staffing, development of new educational materials, updates to the Program's information technology system, new laboratory equipment, expanded testing supplies, and a major renovation to the laboratory facility. An increase in the NBS fee was not needed to support the addition of the conditions.

From the second half of 2023 through early 2025, the NBS Program started several hiring and procurement actions to achieve implementation of MPS-II and GAMT deficiency. In addition, the Program began revising educational materials, updating the information system, and working with DHHS on laboratory renovation plans. The new laboratory positions were created and filled. New educational materials have been developed and are ready for use. The information system has been reconfigured, and the laboratory renovations are completed.

The NBS Program successfully implemented screening for MPS-II in August of 2025; however, there have been delays in newborn screening for GAMT deficiency. As described in the annual report to the joint legislative oversight committee in March 2026, several procurement actions that began in 2024 were not completed until 2026, and some requests remain in progress. These purchase requests largely include new laboratory equipment and testing supplies. The most important contract delay, which was due to delays within the state procurement process, involved

the purchase of certain equipment required to implement screening for GAMT deficiency. The procurement process for this equipment began in August 2024, the vendor contract was not completed until February 2026, and the first equipment was delivered in March 2026. After installation was complete, the NBS Program immediately began the regulatory-required test validation. Historically, these test validations can take six (6) to eight (8) months to complete. While the Program is working to expedite the GAMT deficiency validation, the process is ongoing and anticipated to be complete with the initiation of screening by October 2026.