NC DEPARTMENT OF HEALTH AND HUMAN SERVICES

JOSH STEIN • Governor
DEVDUTTA SANGVAI • Secretary
KELLY KIMPLE • Director, Public Health
YVONNE COPELAND • Director, Division of Child and Family Well-Being

Memo

To: North Carolina Newborn Screening Partners

From: Scott M. Shone, PhD, HCLD(ABB), Public Health Laboratory Director
Anne O. Odusanya, DrPH, MPH, Assistant Director, Division of Child and Family Well-Being

Date: September 18, 2026

Re: Implementation of Newborn Screening for New Conditions and Reporting Updates

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Beginning September 19, 2026, the North Carolina Newborn Screening Program will screen all dried blood spot specimens submitted to the North Carolina State Laboratory of Public Health (NCSLPH) for Guanidinoacetate methyltransferase (GAMT) deficiency and Infantile Krabbe Disease.

GAMT deficiency is a metabolic disorder where the body cannot make creatine resulting in a toxic buildup of guanidinoacetate. Symptoms and onset of GAMT deficiency vary and affect the muscles, brain, and nervous system. Screening results that suggest elevated risk for GAMT deficiency will be communicated to the newborn's health care provider by the UNC Division of Genetics and Metabolism. The UNC Program will provide consultation services and recommendations for follow-up testing.

Infantile Krabbe Disease is caused by deficiency of the enzyme galactocerebrosidase and primarily affects the nervous system. Screening results that suggest elevated risk for Krabbe disease will be communicated to the newborn's health care provider by the Division of Child and Family Well-Being (DCFW) Follow-up Program. The DCFW Follow-up Program will provide consultation services and recommendations for follow-up testing.

Newborn screening for GAMT deficiency and Infantile Krabbe Disease will be integrated into the standard newborn screening panel, and results will be included on all NCSLPH Newborn screening reports. Reports are provided in hard copy form and are also available via the NCSLPH Clinical and Environmental Lab Results (CELR) online portal.

In addition to the new disorders, the North Carolina Newborn Screening Program updated results reporting for amino acid disorders to account for Total Parenteral Nutrition (TPN) interference. Attached to this memo is an additional page outlining these reporting updates. Please review these details if you are involved in the care of infants receiving TPN.

For questions about the newborn screening laboratory testing, please contact the NCSLPH Newborn Screening Laboratory at 919-733-3937.

Newborn Screening Reporting Updates for TPN Interference

North Carolina Newborn Screening (NBS) results reporting has been updated to account for Total Parenteral Nutrition (TPN) interference for the following Amino Acid conditions:

- Phenylketonuria (PKU)
- Homocystinuria/Hypermethioninemia (HCY/MET)
- Maple Syrup Urine Disease (MSUD)
- Argininemia (ARG)

How This Changes Newborn Screening Results

If a specimen has elevated N-Acetyl-Tyrosine (NAT) – a component of TPN – the NBS Final Report will show **"TPN"** instead of an interpretation for the Amino Acid conditions listed above. The report will also include this comment:

AA/AC – Elevated N-acetyl Tyrosine (NAT) suggests interference from total parenteral nutrition (TPN). Amino Acid concentrations may be falsely elevated in babies receiving TPN. Please collect a new filter paper specimen on DHHS Newborn Screening Filter Paper Form and Resubmit 48–72 hours after TPN is discontinued. Presence of elevated NAT does not rule out a metabolic disorder. Perform diagnostic testing as clinically indicated.

What You Need to Do

1. Collect a new filter paper specimen (DHHS NBS Filter Paper Form) **48–72 hours after TPN is discontinued** and submit to the North Carolina State Laboratory of Public Health for testing.
2. Order **diagnostic testing and contact the UNC Division of Genetics & Metabolism if there are concerns for an inborn error of metabolism** – elevated NAT does not rule out a metabolic disorder.

How This Changes Newborn Screening Follow-up

Specimens with TPN interference **will not be reported** to the Newborn Screening Follow-up Program.

Questions or Concerns

Contact UNC Division of Genetics & Metabolism:

- Weekdays 9AM-5PM: 919-962-4995 or 919-445-0845
- After Hours (URGENT ONLY): Call the UNC Hospitals Paging Operator at 984-974-1000 – *Request to speak with the Pediatric Geneticist on Call*