

May 2025



## **TINY TESTS, BIG IMPACT: THE NEWBORN SCREENING UPDATE**

Public Health Services Administration

### **Celebrating our Dedicated Lab Professionals**

We want to give a heartfelt shout-out to our incredible Newborn Screening (NBS) laboratory team!

From April 20th to 26th, 2025, we proudly celebrated National Laboratory Professionals Week, recognizing the vital contributions of our dedicated staff.

The week kicked off with an insightful lecture by Julie Cohen MSc from the Kennedy Krieger Institute. Her presentation on Adrenoleukodystrophy shed light on the profound impact this disorder has on infants and their families, underscoring the critical importance of the work done in our lab.

The week's celebrations culminated in a lab-wide staff appreciation lunch, a wonderful opportunity to acknowledge the hard work, expertise, and commitment of each member of our team.

Thank you to all our laboratory professionals for your unwavering dedication to the health of newborns in Maryland! Your commitment to accuracy and

#### **ICYMI- In Case You Missed It**

##### **Provider Request Form**

Click [here](#) to update specimen information, submit a newborn screening refusal form, order lab slips, or request sickle cell trait results.

##### **Webinar**

Click [here](#) to view recordings of the recent NBS webinars.

##### **Educational Materials**

Click [here](#) to order Newborn Screening educational materials for your

efficiency makes a real difference in the lives of countless families.



patients and families.

### Provider Contact Information

Please complete [this form](#) to provide the direct phone number for a clinical staff member at your facility/practice. This will prevent delays in notifying providers of critical results.

## Timely Newborn Diagnosis Depends on You: Tips for Accurate Lab Slip Documentation

We want to highlight a critical issue impacting newborn screening. Missing or inaccurate information on lab slips cause significant delays, impacting timely diagnosis and follow-up care. Please ensure that all of the fields are fully and accurately completed prior to submission to the Maryland State lab.

### Recommendations to Improve Accuracy:

- **Trained Staff Only**: Please ensure only trained personnel are responsible for completing the newborn screening process.
- **No Parent Completion**: To maintain accuracy, parents should not complete the lab slips.
- **Minimize Distractions**: Completing slips in a quiet, focused environment can help reduce errors.
- **Utilize Resources**: Keep templates or guides readily available at collection stations for quick reference.
- **Implement Verification Steps**: Let's double-check information at multiple points:
  - Before collecting the specimen.
  - Immediately after collection.
  - Before the sample is shipped to the lab.
- **Pediatrician Information**: Please confirm the lab slip includes the correct name and contact information for the baby's pediatrician who will provide care after discharge.

Your attention to these details is vital in ensuring timely care for our newest arrivals.

Thank you for your commitment to the health and well-being of newborns in Maryland.

## Coming Soon: Infantile Krabbe Leukodystrophy

Get ready! Maryland will soon be expanding its newborn screening panel to include Infantile Krabbe Leukodystrophy. Here's what providers need to know about this important addition:

Incidence: Approximately 1 in 100,000 births in the U.S.

Prevalence: Fewer than 500 diagnosed cases in the U.S.

Signs and Symptoms: Irritability, feeding difficulties, developmental delay, muscle stiffness, seizures, loss of motor skills

Cause: Krabbe disease is caused by changes in the *GALC* gene, which lead to a deficiency of the enzyme galactocerebrosidase. This enzyme is essential for protecting the myelin sheath that insulates nerve cells. Without it, myelin breaks down, causing severe and progressive damage to the brain and nervous system.

Treatment: There is no cure, but early treatment may improve outcomes. Options include stem cell (bone marrow) transplantation in presymptomatic infants. Supportive care may also include physical therapy, medications to manage seizures and spasticity, and nutritional support.

## Employee Corner: Sharing Our Why

### Meet Arie Willoughby-Spriggs

Q: What is your official title?

A: Public Health Scientist Level III

Q: What are your principal duties at MDH?

A: To process, prepare, assess, and analyze data from dried blood spots. The spots are collected from the babies within the first 24 hours to 7 days of life. We try to comply with the 72-hour turnaround time that is guaranteed to our clients. I do my part by making sure all steps are done as fast as we can meet them. I also have other duties that aid in the processes, including tracking and ordering inventory so that we always have what is needed to complete testing. Lastly, I also assist in validations, calibrations, etc. to aid in new testing in the lab.

Q: How long have you been with the NBS program?

A: 4 years

Q: What attracted you to NBS and what does NBS mean to you?



A: I was attracted to NBS after receiving my master's degree in public health. I felt that it would be a great place to be in the public health sector. I wanted to see if I could do more for the community coming from the clinical healthcare system. I love babies. Being here is a full circle moment for me. I have worked as a CNA on a mother/ baby unit, a medical technologist testing mom's and babies' blood, and now as a PHLSIII testing for newborn diseases.

Q: What is your prediction on the future of NBS?

A: I see a more up to date, automated work force. In order to expand to new assays, we need the latest testing methodologies, and I can see us moving into that direction. With new leadership, and having our team get accustomed to new ways of work completion, we are moving in the right direction. With new analyzers, new techniques, and workflows, we will be able to provide more quality care, in a faster and timely manner. This will increase the survival of a baby's life.

## Newborn Screening Collection Tip of the Month

- What is "unsatisfactory 2" on the newborn screening report?
  - UNS 2, or unsatisfactory 2 means the specimen appears scratched or abraded.

Common Causes of UNS2	Tips on How to Prevent UNS2
Touching/rubbing the heel on the filter paper	Apply the blood drop to the filter paper, ensuring the heel and skin do not touch the card.
Dragging the capillary tube across the collection site	Do not use capillary tubes for collection.
Improper handling or positioning of the infant's foot	Ensure proper collection site warming to promote free-flowing blood.  Use a light touch and avoid scraping or dragging motions.
Improper handling of specimen after collection	Allow the specimen to dry completely before folding the protective flap.  Handle specimen cards with care to prevent bending or folding.

**Proper Specimen**

**Improper Specimen**



## Lab transitions instrumentation to simplify workflow and improve specificity for Lysosomal Disorders

Currently, our laboratory utilizes Digital Microfluidics (DMF) technology to screen for Pompe, MPS I, and Fabry disease, while MPS II screening is performed using Fluorometry. With the anticipated addition of Krabbe disease to the panel on July 1, 2025, we're streamlining our processes by consolidating the testing for these lysosomal disorders onto a single, advanced platform: Liquid Chromatography Tandem Mass Spectrometry (LC-MS/MS).

What are the benefits of this change?

The LC-MS/MS method employs a sophisticated column to separate the products of enzymatic reactions before detection. This separation offers significant advantages, including:

- **Improved Specificity:** By effectively removing interfering substances, the column ensures clearer and more accurate identification of each disorder's specific markers.
- **Enhanced Reporting:** This increased specificity leads to more reliable and precise reporting of screening results.
- **Increased Laboratory Efficiency:** Transitioning to LC-MS/MS will enable valuable staff cross-training across different areas of the laboratory, ultimately boosting overall efficiency and flexibility.

This move to LC-MS/MS represents a significant step forward in our commitment to providing the most accurate and efficient newborn screening possible for the families of Maryland. We are excited about the

improvements this technology will bring to the detection of these important disorders.



## Heart Corner (CCHD)

Just a friendly reminder! In our last newsletter, we shared a significant update from the American Academy of Pediatrics (AAP) concerning critical congenital heart disease (CCHD) screening in newborns.

The AAP has recently updated its clinical practice guidelines, strongly emphasizing the crucial role of pulse oximetry and early intervention as the primary way to screen and identify newborns who may be at risk for CCHD.

We encourage you to take a moment to review these important revised guidelines, which you can find [here](#).

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