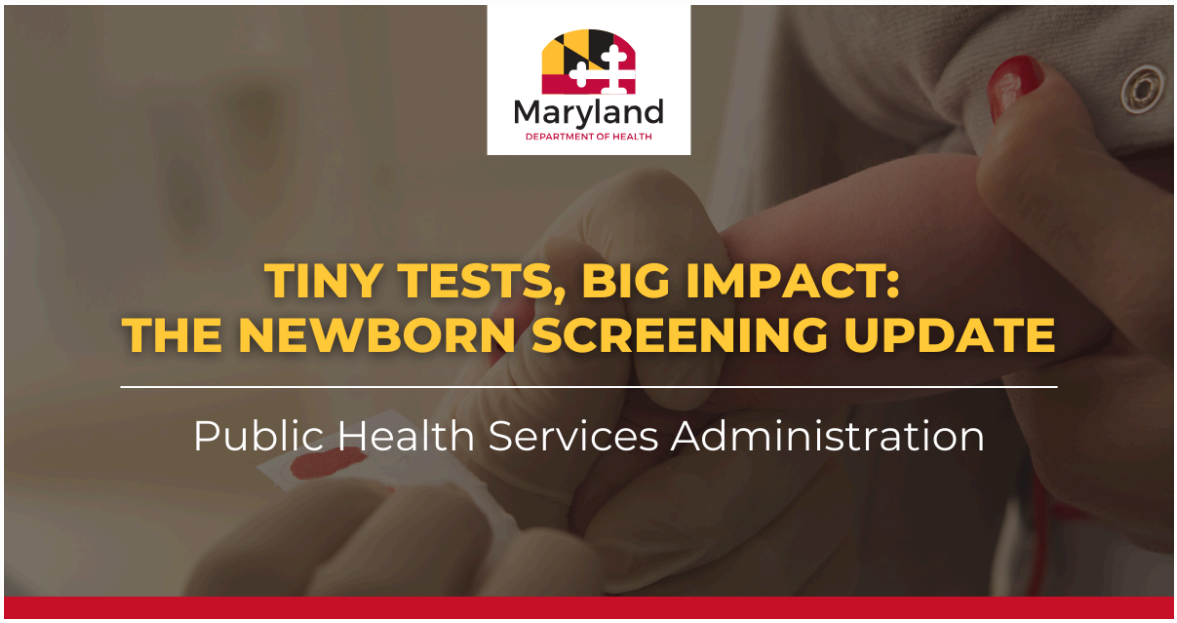
February 2026



🩸New Sickle Cell Trait Resource for Providers

We are pleased to share a valuable new resource to help you counsel families about Sickle Cell Trait (SCT).

Quick Identification

Remember that Sickle Cell Trait is reported on the Newborn Screening Report as FAS under hemoglobin.

Helpful Handout Available

Pediatric Hematologist Corinna Schultz, M.D., from Delaware, has generously provided us with a practical handout designed to aid communication with parents.

This resource covers two key areas:

- "Things to know about sickle cell trait"

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 patients and families.

Provider Contact Information
Please complete this form to provide the

- "Talking points for parents of infants with sickle cell trait"

Find the complete handout **here**.

💡 Action Item Reminder

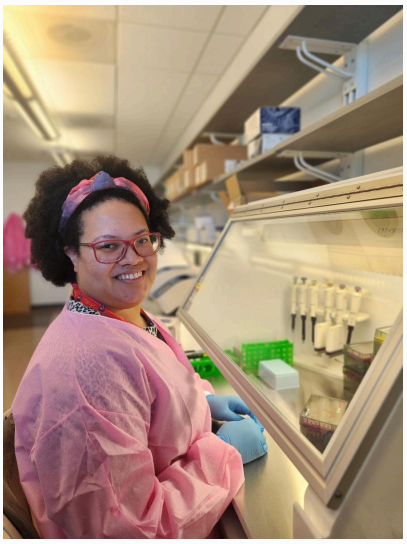
Please ensure documentation of Sickle Cell Trait in the health record—it should be noted in both the History and the Problem List.

direct phone number for a clinical staff member at your facility/practice. This will prevent delays in notifying providers of critical results.

Employee Corner: Sharing Our Why

Meet Chantel Wilson

Public Health Laboratory Scientist II



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

A: Two and a half years.

Q: What are your principal duties at the MDH laboratory?

A: My principal duties in the lab include accessioning and punching of newborn dried blood spot (DBS) specimens, performing DNA extraction, and using real-time PCR (qPCR) to simultaneously screen newborns for Spinal Muscular Atrophy (SMA) and Severe Combined Immunodeficiency (SCID). We also combine PCR with flow-cell fluorometry in the second-tier CFDNA test to screen newborns for 64 CFTR gene variants that cause Cystic Fibrosis. For those tests, I'm also responsible for analyzing and importing test results into STARLIMS, reagent preparation, equipment maintenance, inventory, and other quality assurance tasks. More recently, I've also been responsible for punching DBS specimens and sending those punches to the Wisconsin Newborn Screening Lab for the third-tier CFTR test. This test uses next-generation sequencing to screen for an expanded panel of CFTR variants that cause Cystic Fibrosis.

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

A: I've always wanted a career where I could use my scientific knowledge to directly
impact my local community. It's been rewarding for me to see that the work I do can, for instance, help a newborn almost immediately see a pediatric neurologist at Johns Hopkins and get started with a life-changing treatment for SMA. To me, NBS means doing all I can for the next generation.

Q: Was there anything that surprised you about NBS here in Maryland?

A: I was surprised to see the way the NBS laboratory uses automation to process a large amount of specimens in a relatively short amount of time and with high precision and accuracy. Many laboratories use multiple personnel and take at least 4 hours for tasks like DNA extraction, mastermix preparation, manual pipetting, and qPCR. However, with the use of a liquid handling robot, a quick and simple heat-based extraction, optimized reagents, and a qPCR machine that can process 384 wells at once, the SCID-SMA test can be performed on 360 patient specimens at once and the results will be ready in 2 hours.

Q: What is your prediction for the future of NBS in Maryland?

A: I think in the future we'll see the use of next-generation sequencing for most of the tests on the newborn screening panel in Maryland.

Coming Soon!

Maryland is expanding its newborn screening program! Following the addition of MLD and DMD to the federal Recommended Uniform Screening Panel on December 16, 2025, the state is moving to begin routine testing for these conditions. Per Maryland law, these screenings will be integrated into the standard panel by June 16, 2027. Stay tuned for future updates regarding follow-up procedures and reporting guidelines.

🩸 Specimen Collection Tip of the Quarter

Category	Description / Cause / Appearance / Prevention
Unsatisfactory #5: Serum Rings, Dilution, Discoloration, or Contamination	**Ensuring high-quality newborn screening specimens is essential for accurate results.**

What Does This Mean?	Occurs when a blood spot shows visible serum rings, appears watery or unusually colored, or is contaminated by substances like alcohol, formula, lotion, or water.
Why It Matters	Interferes with lab testing, leading to: Inaccurate or invalid results; Specimen rejection; Delays in diagnosis and treatment; Repeat collections for the baby.
Common Causes	Alcohol not fully dried before puncture; Excessive squeezing or “milking” of the heel; Blood mixing with formula, water, or lotion; Touching the card with contaminated gloves; Applying blood to a wet or dirty card.
What It Looks Like	Clear or pale rings around the spot; Watery or uneven appearance; Very light-colored spots; Yellow or brown discoloration; Greasy or smeared look.
How to Prevent It	Let alcohol dry completely; Use free-flowing drops of blood; Avoid squeezing the heel; Keep the card clean and dry; Do not touch the collection circles; Air-dry cards flat and completely.

Newborn Screening Update: TGAL Testing

Due to intermittent supply chain constraints and reagent backorders, **Total Galactose (TGAL) testing is temporarily unavailable** through the Maryland Department of Health (MDH).

The Newborn Screening Program **continues to monitor for Classic Galactosemia** through **GALT enzyme activity testing**, which remains fully operational and uninterrupted. As a result, this temporary disruption **is not expected to delay the identification or referral of infants at risk for classical galactosemia**, a time-critical condition.

During this period, the laboratory is unable to screen for **Galactokinase Deficiency (GALK)** or **Epimerase Deficiency (GALE)**. Both conditions are classified in regulation as **non–time-critical screened conditions**.

Please monitor your babies for the following and refer to a metabolic specialist if necessary.

- Classical Galactosemia: diarrhea, vomiting, gaining weight slowly (failure to thrive), poor feeding, or excessive sleepiness (lethargy),
- GALE: yellow skin or eyes (jaundice), floppy arms and legs (hypotonia), poor feeding or sucking, vomiting, weight loss.
- GALK: delayed growth, cloudy eyes (cataracts) or difficulty feeding.

We will share updates as soon as TGAL testing resumes.

Condition	GALT Enzyme Activity	TGAL (Total Galactose)	Clinical Onset	Clinical Manifestations
GALT (Classic Galactosemia)	Very Reduced or Absent	Very Elevated	Day 3–7	Liver failure, sepsis (E. coli), jaundice, feeding refusal, and long-term cognitive/ speech delays.
GALK (Galactokinase Deficiency)	Normal	Very Elevated	Day 14–28+	Rapid-onset cataracts, permanent vision loss if left untreated.
GALE (Epimerase Deficiency)	Normal	Elevated	Day 3–10	Range of Severity: Symptoms can mirror GALT (liver failure, cataracts, growth failure). Peripheral forms may be asymptomatic.

Newborn Screening Office Hours: Bi-Monthly Virtual Sessions

What are Office Hours?
A time to talk directly with the Newborn Screening (NBS) team, ask questions, and share ideas.

Why join?
- Ask questions about data, reports, or processes
- Get clear answers about policies and procedures
- Share feedback to help improve the program
- Learn about new NBS updates and initiatives

Who you'll meet:
EHDI, CCHD, and Bloodspot program staff

When:
- Every 2nd and 4th Wednesday of the month from 12:00–1:00 PM
- Next meeting is February 11th, 2026

Video call link: https://meet.google.com/fgi-emtv-hvn
Or dial: (US) +1 619-832-0534 PIN: 514 674 158#

What to expect:
- Open discussion
- Informal and supportive setting
- Time for quick questions or bigger conversations

❤️ Heart Corner: Spotlight on Critical Congenital Heart Disease (CCHD)

Children's National and The American Academy of Pediatrics (AAP) have created free educational resources for providers and families on the importance of CCHD screening for newborns.

🎥 Featured Resources: CCHD Family and Provider Materials

- Critical Congenital Heart Disease Screening: Provider Education
- Your Newborn Baby's Heart Screening

Please feel free to share the parent education flyer. For more information on CCHD screening, please visit this webpage.

Want something included in the next quarterly newsletter? Email mdh.cyshcn@maryland.gov

health.maryland.gov/phpa/cyshcn/Pages/home.aspx