

Virginia Newborn Screening Advisory Committee
Thursday, November 9, 2023
10:00 AM – 2:00 PM

Location: Fairfield Library, 1401 N Laburnum Ave, Richmond, VA 23223.

Virtual Platform: Zoom Meeting Registration: https://www.zoomgov.com/webinar/register/WN_ojc6I175TQS0UgfJxuJung

*** Note: Members cannot participate remotely until after a physical quorum has adopted an electronic meeting policy ***

Draft Meeting Minutes

Committee Member Attendance	Voting Record			
	Y=Yes, N=No, A=Abstain			
Bold = Present, In Person * = Remote Participation <i>Italicized = Absent</i>	Adopt 6/22 Minutes	Adopt Bylaws	Adopt EMG	Convene Workgroup for GAMT and MPSII
Dr. Bill Wilson, UVA, Chair	Y	Y	Y	Y
Abraham Segres, VHHA	Y	Y	Y	Y
Dr. Christina Grant, CNMC	Y	Y	Y	Y
Julie Murphy, Parent*				Y
Karen Shirley, Henrico Doctors Hospital	Y	Y	Y	Y
Lisa Shaver, CHOR at VCU	Y	Y	Y	Y
Dr. Christian Chisholm, UVA, ACOG	Y	Y	Y	Y
Dr. Richard Bennett, Community Pediatrician	Y	Y	Y	Y
<i>Dr. Sylvia Lee, Community Pediatrician</i>				
Jana Monaco, NORD, Parent	Y	Y	Y	Y
Dr. Hind Al Saif, VCU	Y	Y	Y	Y
Dr. Samantha Vergano, EVMS/CHKD	Y	Y	Y	Y
<i>Dr. Brooke Vergales, Neonatologist, UVA</i>				
Kim Pekin, CPM	Y	Y	Y	Y
Eileen Coffman, Registered Dietitian	Y	Y	Y	Y
Dr. Marta Biderman Waberski, INOVA	Y	Y	Y	Y
<i>Tina Cavucci, MOD</i>				
Amy Brooks, Genetic Counselor	Y	Y	Y	Y
<i>Dr. Nayef Chahin, Virginia Chapter AAP</i>				
Dr. Alicia Prescott, DOD, Portsmouth Naval Medical Center	Y	Y	Y	Y
Rebecca White, American College of Nurse Midwives (Proxy: Regina Foran, CNM)	Y	Y	Y	Y

VDH Staff Present: Christen Crews, Mary Lowe, Jennifer Macdonald, Marcus Allen, Victoria Goyne, Clair Seckner*, Rafael Randolph
DCLS/DGS Staff Present: Emily Hopkins, Dr. Denise Toney, Leigh Emma Lion, Paul Hetterich, Chris Nixon, Richard Haughton, Aki Harada, Gretchen Cote, Keith Kellam

10:00-10:10	<p>Welcome: Dr. Bill Wilson, Chair</p> <ul style="list-style-type: none"> A. Call Meeting to Order B. Roll Call of Board Members C. Review of Agenda
10:10-10:20	<p>Public Comment</p> <p>Kelly Danoy shared that in 2015 and 2020 the committee made decision not to include Krabbe Disease to the screening panel. It impacted her family as her daughter had a delayed diagnosis of Krabbe Disease (early infantile onset). A timely diagnosis would have improved supportive care for her daughter, and the NBS AC has abilities to implement and help children born with this disease.</p>
10:20-10:30	<p>Administrative Updates, Christen Crews</p> <ul style="list-style-type: none"> A. Review and Vote on Proposed Revisions to Bylaws (EMG) (adopted) B. Review and Vote on Electronic Meeting Guidelines (adopted) <ul style="list-style-type: none"> • Member Julie Murphy joined meeting remotely at this time C. Approval of Meeting Minutes 6/30/2022 (approved)
10:40-11:00	<p>Legislative Updates, Christen Crews</p> <ul style="list-style-type: none"> A. NBS Program Funding Model Workgroup Update – Workgroup convened as mandated by the 2023 General Assembly (GA) session in HB2224 introduced by Delegate Kathleen Murphy. The workgroup had 3 meetings, and the priority of workgroup was to address barriers of cost and equitability for NBS. Key stakeholders were screened on newborn reimbursement and barriers to screening. The findings of the workgroup were to not recommend one model, but to consider all models. The report has been submitted for review and, once published to the General Assembly, VDH staff will share with the workgroup and Committee members. B. 12VAC5-71 Periodic Regulation Review Update – inconsistencies with recommendations, priorities, and providers. Promulgate CCHD regulations to new chapter will be presented to Board of Health in April or June 2024, will propose adding a new section to NBS regulations supporting electronic messaging with HL7.
11:00-11:30	<p>Program Business, Mary Lowe and Emily Hopkins</p> <ul style="list-style-type: none"> A. ACHDNC Meeting Updates – Mary Lowe shared that the RUSP is a list of disorders which currently has 37 disorders and VA screens for 35. The ACHDNC adopted an expedited review process for disorders previously reviewed and not included. The expedited review process allows for faster renomination and review if there exists a minimum of one material change or change in scope of screening; if voted for expedited review, the vote for inclusion to the RUSP must be held within 9 months. B. New Disorder Workgroups <p>The NBS AC voted to convene a Workgroup to review considering adding 2 new disorders on the RUSP: MPSII and GAMT. The process of how a new disorder is added to Virginia’s NBS panel was reviewed by Christen Crews: When a new disorder is added to the RUSP, workgroup reviews the disorder to research, educate, and provide recommendations to the larger NBS AC. Subject Matter Experts (SMEs) are invited to present on the new disorders. The workgroup will provide the Board of Health with recommendations of the workgroup and NBS AC. The recommendations will be shared with the Governor’s Office for review.</p> C. Specialist Roundtable Meetings – Improve communication and care coordination for infants identified through NBS by convening annual specialty specific meetings with representation from each regional consultant group.

	<p>These meetings will be collaborative with representation from specialty consultants, VDH NBS Follow-Up, and DCLS with the goal is to identifying pain points and solutions for care coordination and follow-up.</p> <p>D. Electronic Messaging Updates: Emily Hopkins, DCLS, provided updates on electronic messaging, currently 22 hospitals engaged in some form of electronic messaging activity; DCLS is currently receiving 53% of sample orders electronically. DCLS allows for flexibility while holding to a standardized process; hospitals may build their own or elect to purchase an interface for sample order submission and result capture. DCLS is in the final year of a HRSA-funded grant project to enable partner with a software vendor: iConnect Consulting. Seeking to expand available solutions; efforts continue to onboard with hospitals. There is not a defined figure of cost to hospital to implement system, as it depends on technical team and how they can incorporate and prioritize what the hospital is able to do. The risk outweighs additional cost for timeliness in reporting NBS results. The courier process for bringing NBS results to hospital was explained and the NBS Connect electronic portal (MD, NP, PA, Midwives). It was suggested to consider bringing DMAS into the discussion. DOD doctors are not licensed in state where practicing due to frequently moving, and DCLS is working on expanding access to more providers.</p>
11:45-12:30	<p>Working Lunch, Staff</p> <p>A. GALT Discussion, Leigh Emma Lion – Reviewed the AC meeting in December 2021, discussion around GALT was held in which the notes indicate that consultants requested a review of GALT data and consideration of modifying the current critical algorithm from 3x ABN GALT. Discussions occurred around historical data without critical TGAL on first screen and a “classic galactosemia” diagnosis. Sometimes infants are switched to soy without accurate documentation on the DBS collection card. The third card is not useful for diagnosis to the genetic consultants, and they suggested improving communication/education to pediatricians to not stopping breastfeeding and switching to soy due to an abnormal GALT without talking to Genetics. It was suggested to do a similar referral review to Genetics as the LSD 1st tier for multiple screens for opt-out of sequencing with 2X ABN GALT. The NBS nurses are available 7 days a week if providers have questions, and can connect with Genetics if needed. It was suggested to highlight contact information for Follow-Up on the reports for providers and potentially update formatting on report. This will be discussed further for improvements at the Genetics roundtable specialist meeting.</p> <p>B. LSD Variant Re-classification Review, Gretchen Cote – DCLS – discussed 2nd tier LSD sequencing platform NBSVI in May 2022 initiated update to NBSVI and began issuing amended reports for reclassified variants. 176 reviewed, 42 variants reclassified. 622 amended reports issued to submitter and care provider on record. Critical rates since reclassifications: Pompe – 39.53% down from 56.16% in 2021; MPSI 34.91% down from 49.32% in 2021. One additional variant has been reclassified through routine checks for new information, resulting in 2 amended reports. Preliminary on why program does not classify a variant as a “pseudodeficiency” and can be discussed further in specialist roundtable meeting.</p> <p>C. Data Review, Emily Hopkins – 11/1/22 thru 10/31/23 data</p> <ul style="list-style-type: none"> • 87,037 infants screened, 101,758 samples received, increase to unsatisfactory samples at 2.74% (up 37% since 2022)

	<ul style="list-style-type: none"> ○ Most common rejection reasons are oversaturated, improperly collected and old samples. ● Worked with Follow-Up team to update Rejection education sheet, will incorporate QR code, increasing education and outreach efforts. ● Avg time to collection to time of receipt has slightly decreased over time to 1.63 days.
12:30-1:50	<p>Programmatic Updates, Staff</p> <p>A. Follow-Up, Mary Lowe, Review of updates to Follow-Up staff including 2 senior nurses -Victoria Goynes (Short Term Follow-Up Senior/Nurse Educator and Clair Seckner (Long Term Follow-Up Senior Nurse), 3 short term follow-up nurses, program support tech, and part-time critical follow-up nurse. The new critical follow-up nurse role will provide more timely follow-up on reported criticals (nurse currently in orientation). Provided updates on expanded EMR access, education and Outreach – education and specimen collection training, birth hospitals and outpatient settings. Virtual education sessions available now, contact email: newborn_screening @vdh.virginia.gov. Recent acquisition of training infant feet for hands-on collection practice will be used on on-site training of nurses, phlebotomists, etc.</p> <p>B. Treatment Sample Updates, Paul Hetterich – Upcoming improvements to treatment sample process (PKU, MSUD, HCU) Received an Association of Public Health Laboratories (APHL) Continuous Quality Improvement Award for this project. Created new collection card for treatment samples with fewer fields to reduce manual entry errors. Sample of new card was shared with committee members.</p> <ul style="list-style-type: none"> ● Treatment sample reports can be viewed in the Newborn Screening Portal. Work is in progress to allow additional provider types, such as dietitians, access to the portal. Once this capability is rolled out in 2024, DCLS plans to discontinue faxing of treatment sample reports to providers (reports will still be mailed to providers and will be viewable in the portal). Members expressed concerns regarding not knowing when new results are available, suggested an email alert to the provider (system is not currently able to do this function, DCLS will explore). It was shared that the clients who use the treatment sample program do not always make the provider aware when submitting a card to DCLS. ● Correction factor of 1.3 for elevated phenylalanine results, used to approximate plasma phenylalanine values, will be discontinued. ● It was questioned how many blood spots are needed for testing. Only 1 is needed for treatment samples; however, it is the same filter paper so there will be 5 circles still on the card. <p>C. CFF Grant and NexGen CFTR Sequencing, Gretchen Cote – reviewed the expanded 2nd tier for Cystic Fibrosis (CF) with Next Generation Sequencing (NGS) to go live early 2024. The program received a 2-year Screening improvement Program Award from the CF Foundation to assist in implementation of the new testing methodology. The custom NGS test will allow for future expansion and multiplexing additional gene targets. The sequencing targets 700+ variants classified as CF – causing or varying clinical consequence in CFTR2 database. Expected average turnaround time is 7 to 10 days from sample received. Only significant CF variants will be included, not all, as if screened for open variant it would expand what is reporting and affect the turnaround time. This will replace existing assay with the NGS panel, still be</p>

	<p>referring 2 variants as screen critical; 1 variant abnormal; 0 variant still abnormal.</p> <p>D. Open Discussion</p> <ul style="list-style-type: none"> • Christen Crews reviewed appointment terms of the Committee members and read section in bylaws, the staff will communicate membership terms more clearly in the future.
1:50-2:00	<p>Next Meeting Date and Adjournment; Dr. Bill Wilson, Chair</p> <p>A. Next meeting date: TBD, Spring 2024, poll for dates.</p> <p>B. Dr. Wilson adjourned the meeting at 1:17pm.</p>

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