Virginia Newborn Screening Advisory Committee
Thursday, November 14, 2019
10:00 a.m. – 12:00 PM

The Division of Consolidated Laboratory Services (DCLS)
600 North 5th St. Room T21/T23
Richmond, VA 23219

AGENDA

Members (check = present):

- ☒ Dr. Bill Wilson, UVA, Chair
- ☐ Abraham Segres, VHHA
- ☒ Sarah Viall, CNMC
- ☐ Julie Murphy, Parent
- ☒ Karen Shirley, HCA-Va, Chippenham Hospital
- ☐ Lisa Shaver, Children's Hospital of Richmond at VCU
- ☐ Amber Price, American College of Nurse Midwives
- ☒ Rachel Gannaway, Genetic Counselor, VCU
- ☐ Dr. Christian Chisholm, UVA, ACOG
- ☐ Dr. Michael Martin, Virginia Chapter AAP
- ☐ Dr. Sylvia Lee, Community Pediatrician
- ☒ Marie Pokraka, MOD
- ☒ Jana Monaco, NORD, Parent
- ☒ Dr. Hind Al Saif, VCU
- ☒ Dr. Samantha Vergano, EVMS/CHKD
- ☒ Dr. Brooke Vergales, Neonatologist, UVA
- ☒ Kim Pekin, CPM
- ☒ Barb Goodin, Dietician, UVA
- ☒ Dr. Stephanie Smith, DOD, Naval Medical Center Portsmouth
- ☐ INOVA, TBD

VDH & DCLS Staff

- ☒ Willie Andrews
- ☒ Jennifer Macdonald

Interested Parties: Tammy McKinley (CPM), Marta Biderman-Waberski, Christie Burnette, Erica Price, Eileen Coffman (CHKD), Jennifer Dent (VCU), Chris Nixon (DCLS), Richard Haughton (DCLS), Gretchen Cote (DCLS), Shayna Barnes (DCLS), Marilyn Bibbs-Freeman (DCLS), Paul Hetterich (DCLS), Leigh Emma Lion (DCLS), Jennifer Brickey (VDH), Shamaree Cromartie (VDH), Marcus Allen (VDH), Jennie Dinh (VDH), Lillie Chandler (VDH), Vickie Woon (VDH), Nicole Scarborough (VDH), Christen Crews (VDH)

10:00 – 10:20	Welcome: Dr. Bill Wilson, Chair A. Welcome to DCLS: Dr. Marilyn Bibbs-Freeman a. Dr. Marilyn Bibbs-Freeman, new deputy director at DCLS, welcomed participants to meeting and contribution to saving babies. Safety procedures and facilities were explained in the event needed during this meeting. B. Role Call a. Dr. Wilson welcomed all attendees to the meeting and began roll call. Time for public comment will occur later in the meeting. C. Introductions of Members and Interested Parties a. Introductions were made by all parties present in person and on the phone D. Review of Agenda E. Approval of June 29, 2017 Meeting Minutes a. Approved F. Travel Reimbursement (members only)

10:20 – 10:25	Public Comment - No public comment
10:25-11:00	Virginia NBS program updates: Willie Andrews, Jen Macdonald A. National front: ACHDNC meeting updates a. The Secretary Advisory Committee for newborn screening has been disbanded due to the "Newborn Screening Saves Lives" Act not yet reauthorized. The program is optimistic that the committee will resume when the bill passes. B. Virginia Programmatic Updates 1. Sickle Cell Screening/Program Update a. Sharmaree Cromatrie shared receiving applying and successfully being award 2 grants focusing on the sickle cell population. The first grant, in partnership with the CDC, establishes infrastructure in Virginia for Sickle Cell Disease surveillance. Surveillance will be achieved by linking with the DCLS newborn screening laboratory data, vital records, and emergency room data. The second grant, in partnership with CNMC, will randomly select and interview 30 families diagnosed with a sickle cell trait. 2. CYSHCN Updates a. Marcus Allen- manages the Children Youth and Special Health care needs program, funded by the Title V grant. Approximately 50% of funding goes to children with special needs. The program includes 4 programs- Care Connection for Children (CCC) and Sickle Cell Program relate directly to this meeting, CCC provide case management for children with various conditions on panel. The program recently launched medical home and transition modules after 2 years of hard work. There are tracks for providers and parents/youth in the community. Marcus Allen will provide link to modules to be shared with the committee. 3. EHDI a. Jennie Dinh, Newborn Screening Program Administrator, provided updates on EHDI activities. EHDI has been finalizing cCMV protocols to be initiated by September 2020. They will be working with infectious disease providers in the area to find case studies. The program also recently submitted a HRSA grant for programmatic funding and finished a Shared Plan of Care document resource for providers. 4. DBS a. Christen Crews shared successful pilot and implementation of a REDCap database with the Genetic referral centers for all metabolic and lysosomal storage disorder results. This resource provides a searchable database for consultants, automatic email notification of a new critical, ability to view open case load, diagnose infants/close cases electronically, and view diagnosis reports. It also provides more timely follow-up and saves on

paper without needed faxes to communicate. Screenshots demonstrating REDcap were shared with the committee.

5. Electronic messaging
    a. Willie Andrews provided update on the electronic messaging project in Virginia. The goals are to improve timeliness through electronic demographic and order submission and result reporting. Willie discussed normal process with manual data entry and reviewed the pilot project in place. 10 hospitals are actively submitting demographics electronically, and one hospital (Sentara Norfolk General) is the first hospital to receive electronic results back.
    b. Data analysis of project has shown that electronic transmittal of demographics has decreased the percent of samples received with missing or erroneous information from 4.6% to 0.4% of participating hospitals. Additionally, Sentara Norfolk General provided data that prior to receiving results electronically, it took approximately 10.8 days before the results would appear in an infant's EMR (lab receive sample, test sample, send to hospital via courier, scanned into chart). It has been reduced to 3-6 days depending on test results. One week reduction = life saving.
    c. New opportunity for NBS E-messaging in Virginia through APHL grant. DCLS is partnering with iConnect consulting to develop e-messaging tools to create more options for sending demographics/receiving results. The grant is a 4 year project and DCLS is seeking hospital partners.
    d. Another project is to add order entry capability for smaller groups (pediatricians, midwives, treatment samples) to the web portal.
6. NBS Data Review
    a. Willie Andrews shared preliminary data for 2019 (through 10/31/2019
        i. Of note, 2,733 infants have been diagnosed and 1.86% of total samples received were noted to be "unsatisfactory" and unable to be tested.
        ii. Diagnosed cases by disorder/trait were provided to committee attendees- including new data on Lysosomal Storage Disorders- the first two babies with MPS-1 were identified in September 2019.
        iii. The transit time (time of collection to time of receipt by the lab) has decreased from 2.94 days in 2013 to 1.56 days in 2019 Q3. The significant reduction- almost 50%- is attributed to expansion to 7 days a week, quality improvement projects with hospitals, and the laboratory working all holidays. The transit times for each hospital across the state were shared.

11:00 – 11:45	Old Business A. CAH screening: 2nd tier screening update a. Chris Nixon, Tandem Mass Spectrometry Principal Scientist, updated that 2nd tier CAH was implemented in October 2019. He reviewed main goal of 2nd tier CAH implementation is to reduce CAH false positives. The daily workflow includes eliminating confirmation in duplicate and reflexing initial abnormals to 2nd tier to keep turn around time to 2 days. The program will continue to monitor data; however, the total number of out of range results reported (Critical + Abnormal) decreased from 150 in July 2019 to 34 in October 2019. B. X-ALD and SMA Implementation Updates a. Jen Macdonald shared that X-ALD and SMA have entered the 2nd stage of the regulatory process. The opportunity for public comment will be available in the next couple of months. b. Internal efforts include weekly meetings and the laboratory is preparing for validation. Changes are being developed in the laboratory's information management system (LIMS) and the goal is to implement screening for the disorders statewide in June/July 2020. SMA workgroup this afternoon and X-ALD workgroup to be scheduled soon with genetics (TBD: January 2020). c. Dr. Wilson questioned if 2nd Tier testing will be used for SMA- advised only 1st tier (newborn screening lab will not provide SMN2 copies). Richard Haughton, Principal Molecular Scientist, explained that SMA screening will be multiplexed with SCID. A probe will be used to detect homozygous exon 7 deletion- as recommended by the ACHDNC when submitted to be added to the RUSP. This is expected to identify 95% of SMA cases, it is estimated that 5% will not be caught through newborn screening. The testing will not detect SMA carriers. There is an estimated incidence of 1 in 10,000 births. C. cCMV screening implementation a. EHDI and DCLS have collaborated and developed regulations for cCMV with an effective date of September 2020. b. Staff are writing protocols for providers c. Start up and lab costs included in October 1, 2019 fee increase d. Protocol: If infant fails hearing screen in hospital, then test for cCMV through saliva collection i. Meridian is only FDA approved testing with PCR- dried urine on filter paper is not yet FDA approved. e. Plan is to transport with DBS samples with cooling (provides 7 day window for testing, only 48 hours if at room temperature) D. Holiday/Weekend Reporting a. Infants diagnosed as a result of expansion of work days were shared – 25 time sensitive disorders in 2018 and 34 so far in 2019. E. DGS NBS Web Portal

	a. Willie Andrews discussed the NBS web portal that was implemented on May 1, 2019. Currently, MD, Np, and midwives with Virginia licenses can request access. As of 11/4/2019, 9,241 NBS reports have been viewed by providers across the Commonwealth. b. Opportunities to expand include cCMV results and manual order entry for NBS samples
11:45 – 12:00p	New Business A. Recent Fee Increase a. Virginia Newborn Screening Program is dependent on fee for service for all operations. The fee recently increased to $138 per collection kit on October 1, 2019. No new fee increase is currently being planned b. Kim Pekin questioned Medicaid increasing reimbursement rate for newborn screening- it is currently approximately $40 per infant. Jen Macdonald will discuss with other Maternal Child Health efforts currently in place. c. Stakeholders can advocate for the program to receive funding (i.e. general funds to supplement to decrease need for fee increases). The 2020 General Assembly session is long (60 days)- if the program receives a bill, may reach out to program's stakeholders to comment. B. Other Funding Opportunities a. DCLS/VDH is always seeking additional funding opportunities to assist with startup and QA/QI initiatives. Recent funding through awarded grants were reviewed. C. Other Discussion a. Barb Goodin (UVA) shared that she is working with Laura Duncan (VCU) and Eileen Coffman (CHKD) on a bill for mandated insurance coverage for metabolic formula. It may be limited to disorders on the NBS panel (amino acids/PKU). b. MPS-1: A question was raised regarding the enzyme assay for MPS-1 and the large number of initial positives i. Chris Nixon explained that when the enzyme assay was validated, the lab brought in an approved FDA assay and external dried blood spot samples from other states (Virginia does not do pilot testing). Over 2,000 dried blood spot extracts were used for analysis on initial assay. ii. Data has shown that the GAA enzyme (Pompe) decreases as the infant ages- as a result, the lab re-evaluated cut-offs and established age based cut-offs once acquired enough samples from the population iii. Data from IDUA do not show the same age based trend. The algorithm includes reflexing to 2nd tier gene sequencing if the MPS-1 enzyme is critically low, multiple 2 X Abnormal results or 2 X unsatisfactory results.

	iv. Unsatisfactory results of the enzyme can be caused by layering, sample quality (heat/humidity, transit, drying process, etc). v. Follow-up is reaching out to consultants if an infant has a normal enzyme with other samples that would cause the infant to be sequenced. This has cut the number of MPS-1 samples to be sequenced in half. The program will re-evaluate the current algorithm after analyzing data at a year. vi. Missouri has shared that they have implemented seasonal cut-offs to address enzyme degradation with heat/humidity. vii. Sarah Viall at CNMC shared that Maryland does not do 2nd Tier testing for MPS-1 at this time which results in a much higher number of referrals. D. 2020 meeting dates a. TBD: June 11 / June 18 and November 12
12:00p	Adjournment