Virginia Newborn Screening Advisory Committee
Thursday, December 7, 2017
10:00 a.m. – 3:00 PM

The Division of Consolidated Laboratory Services (DCLS)
600 North 5th St. Room T21/T23
Richmond, VA 23219
Conference call-in phone number: 1-866-842-5779 Code: 804-648-4480

AGENDA

Members (check = present):

- n Dr. Bill Wilson, UVA, Chair
- n Abraham Segres, VHHA (proxy = Dr. Bill Wilson)
- n Sarah Viall, CNMC
- n Julie Murphy, Parent
- ¤ Karen Shirley, HCA-Va, Chippenham Hospital
- n Lisa Shaver, Children's Hospital of Richmond at VCU
- n Amber Price, American College of Nurse Midwives
- n Rachel Gannaway, Genetic Counselor, VCU
- n Dr. Christian Chisholm, UVA, ACOG
- ¤ AAP, TBD
- ¤ Pediatrician, TBD
- n Christy Keppel, MOD
- n Jana Monaco, NORD, Parent
- n Dr. Hind Al Saif, VCU
- n Dr. Samantha Vergano, EVMS/CHKD (proxy = Eileen Coffman)
- n Dr. Brooke Vergales, Neonatologist, UVA
- n Kim Pekin, CPM
- n Barb Goodin, Dietician, UVA
- ¤ DoD, TBD
- ¤ INOVA, TBD

VDH & DCLS Staff

- n Willie Andrews
- n Jennifer Macdonald

Interested Parties: Hannah Gosey, Kim Outten, Cassidy Gosey, Krystal Hayes, Hayley Hayes, Rebecca Horsting (Patient Worthy), Terri Klein (MPS Society), Michael Schleter, Virginia Pallante (VCU), Pranoot Tanpaiboon (CNMC), Bryan Carlton, Jen Hutcheson, Christina Owens, Laura Duncan (VCU), Kelly Jones, Katie Crosby (CNMC), John Gibson (Biogen), Chris Nixon (DCLS), Richard Haughton (DCLS), Rhonda West (DCLS), Dena Potter (DGS), Denise Toney (DCLS), Kim Turner (DCLS) Ciera Plum (VDH), Katharine Duni (VDH), Jennifer Brickey (VDH), Shamaree Cromartie (VDH), Marcus Allen (VDH), Daphne Miller (VDH), Lillie Chandler (VDH), Christen Crews (VDH)

10:00 – 10:20	Welcome: Dr. Bill Wilson, Chair A. Welcome to DCLS: Dr. Denise Toney a. Dr. Denise Toney, lab director of DCLS, welcomed participants to meeting acknowledging full room and contribution to saving babies. Committee is particularly important to all citizens to provide opportunity to have voice heard as we move forward with making decisions that affect the Commonwealth. b. Safety procedures and facilities were explained in the event needed during this meeting. B. Roll Call a. Dr. Wilson welcomed all attendees to the meeting and began roll call. Time for public comment will occur later in the meeting. b. Voting members introduced themselves c. Eileen Coffman will act as Dr. Samantha Vergano' s proxy for

	voting matters. d. Dr. Bill Wilson will act as Abraham Segres proxy for voting matters. C. Introductions of Members and Interested Parties a. Interested parties introduced themselves. D. Review of Agenda E. Approval of June 29, 2017 Meeting Minutes a. Meeting minutes were approved. F. Travel Reimbursement (members only)
10:20 – 10:35	Public Comment • Jennifer Hutchinson, Brian, Carlton (son)- spoke in support of testing for MPS-1 at birth, shared Carlton's diagnostic journey – Carlton was normal at birth and he was a healthy growing child. The first indication was delayed walking at 19 months. After this time period, by age 3 he was treated for multiple issues including respiratory, umbilical hernia, joint mobility. He began OT and PT for hypotonia that proceeded through elementary school and was referred to cardiologist (heart murmur). At age 6, another specialist addressed respiratory issues. He was seen by multiple neurologists and diagnosed with cornea dystrophy. He did well with school, continued PT and OT for limited range of therapy. By age 8, Carlton's second grade teacher asked if there is anything she should know because he couldn't sit cross legged when requested. Asperger's was questioned, but ruled out. After 2 more years of continued aches/ailments, physical therapist requested X ray of shoulders. Radiologist report came back with findings noticeably seen in LSDs. They were referred to geneticist at UVA, and diagnosed with MPS-1 Hurler-Scheie. Carlton began to receive enzyme therapy and he will continue to life. After beginning treatment, and immediate improvement was noticed but other issues cannot be reversed. He has had multiple surgeries including on his spine. If treatment had started earlier, he would have had better outcome and shorter diagnostic odyssey. • Crystal Hayes, daughter Hayley, spoke in support of adding Pompe to Virginia's panel. Crystal is a Virginia registered nurse and works at Henrico Doctors Hospital. Haley sent to MCV at 6 months due to failure to gain weight, she had low muscle tone but was meeting milestones. It took approximately 2 weeks for diagnosis of Pompe Infantile. In 2006, treatment for Pompe had just been approved 2 months prior, and she was advised life expectancy was 1 year. They opted to start treatment immediately and included medications every 3-4 hours, breathing treatments, NG tube, IV infusion every 2 weeks. Hayley is now being seen at Duke University as they are the leaders in Pompe. Crystal works as a nurse in the newborn nursery and understands the importance of newborn screening and if her daughter had been diagnosed at birth, then she could be walking (confined to wheel chair). Crystal is administrator of large Pompe group on Facebook including representation of families from different states and seeking support. In reference to the adult form, she has seen adults be misdiagnosed for 10-20 years before finally diagnosed in 30s/40s. Hayley is now in the 6th grade and

	has many years ahead of her. We should be able to help as many kids as possible. • Kim Outten spoke in support of adding MPS-1 to Virginia's panel. Her daughter, Hannah, was born 21 years ago and when she was 6 weeks old, mom noticed something wrong and had concerns. The next two months, were filled with doctor appointments and she was diagnosed at 2 months with first ear infection (no exposure to daycare). Hannah had a bump on spine appeared not at birth. She had respiratory issues, and they were scared to lay her down. Family history of brother who passed away from CCHD at 18 days old. Hannah had feeding issues and diagnosed with failure to thrive. Hannah started having severe reflux episodes – choking, vomiting, coughing and then turned blue. Hannah had an Upper GI completed and then was sent to neurology. Two weeks later, they met with neurologist and was diagnosed with MPS-1. Neurologist told them to take her home and love her, make her comfortable for as long as she was here. Hannah's pediatrician said would not give up on Hannah and referred her to Duke for full evaluation. They found a redundant palate that was causing breathing and feeding problems, she had surgery and a BMT transplant. Her younger daughter, Cassidy, was tested in utero and began treatment and had her transplant shortly after birth.
10:35- 11:35	Virginia NBS program updates: Willie Andrews, Jen Macdonald A. November 2017 SACHDNC meeting updates a. Review programmatic updates – shared November meeting highlights b. NewSTEPS did a presentation on working toward NBS timeliness goals, Virginia data was highlighted showcasing improvements in Virginia's timeliness through deploying site visits and other outreaches. The number of specimens collected at less than 48 hours was increased and we also decreased the transit time from submitter to laboratory. c. Implications of carrier testing through NBS- discussion whether or not to report out carriers, Virginia does report out carriers or receive carrier diagnosis when receiving diagnostic results back from abnormal or critical results. d. SMA Evidence Review: Phase 2 report- Public health system impact assessment completed by the Association of Public Health Laboratories (APHL), guest speaker at lunch will discuss SMA (spinal muscular atrophy). A workgroup is reviewing the science and treatment of SMA, it is thought that it will be reviewed for voting to add to the RUSP in February 2018. e. Clinical and Public Health Impact of SCID screening- 94% of all infants being screened in the US, focus shifting to QA/QI, education, LT follow-up, transfusions, and insurance coverage. B. 2017 APHL Newborn Screening and Genetic Testing Symposium overview a. New Orleans meeting in September 2017, DCLS/VDH represented with 7 posters and oral presentation on DCLS Capstone Project (partnership with UVA grad students to improve process with report card generation and data analysis).

C. Virginia Programmatic Updates
   1. EHDI
      a. Daphne Miller- Early Hearing Detection Intervention Coordinator, focused in last year on updating protocols to provide to PCPs, hospitals, and audiologists. Started a learning community in NOVA includes parents, early intervention- pilot, meeting quarterly but moving to monthly, will be shared across state in the next year. Goal is to create material to give to parents on what to do for the first 100 days- what you should do (ophthalmologist, genetic counseling, early intervention, etc.). Working on implementing tele audiology (HRSA grant)
   2. CCHD
      a. Christen Crews shared information regarding the new CCHD follow-up and referral process that will start January 2018. Jen Macdonald shared about partnering with midwives for CCHD screening and donation of pulse oximeters.
   3. Sickle Cell Screening/Program Update
      a. Shamaree Cromartie shared updating the parent brochure and carrier trait letter. Shamaree shared conversations with parents and confusions between trait and disease.
   4. 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. 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. Dr. Wilson travels to one of centers to provide services (rural area). In process of adding medical home and transition modules to UVA website, April 30, 2018 will be first statewide CYSHCN statewide meeting.
   5. DBS
      a. Christen Crews shared new initiative with rebranding site visits to Newborn Screening Education updates. A hospital with significant quality visit received 2 education sessions and improved significantly. More education visits will be scheduled with application for continuing nursing education (CNE) credits.
   6. NewSTEPS 360 grant updates
      a. Jen Macdonald: NewSTEPS- technical assistance newborn screening collaboration program, funded by HRSA, provides data services, technical assistance and QA/QI. De-identified case data is added monthly into national data repository.
      b. Willie Andrews: Electronic messaging- VA was awarded grant in 2016- using Health Information Technology to improve timeliness through electronic demographic and order submission and result reporting. Willie discussed normal process with manual

data entry and pilot project in place with 13 hospitals across the state. As a part of the grant, introduced to Oz systems that is used as a conduit with hospital EMR to communicate with state laboratory. 4 hospitals are actively submitting demographics electronically, next phase will include receiving results electronically .

c. Jen Macdonald: NYMAC Collaboration- New York Mid-Atlantic Consortium for Genetic and Newborn Screening Services- education project focused on timeliness and best methods to reach providers. Shared past projects, current projects looking at hospital unsatisfactory specimens and transit time data for hospitals participating in education.

7. Periodic CF review
    a. Richard Haughton, Principal Molecular Scientist, reviewed Periodic Cystic Review in July 2017. Historically, Virginia was batching mutation analysis and resulting in turn-around time of 2 weeks. Virginia implemented workflow changes and have results in a week or less. Richard explained CF testing in Virginia, IRT/DNA using 96% percentile floating IRT cut-off or >170ng/ml. 2nd tier testing looks for 39 of most common mutations found in CF. If 1 mutation or 2 mutation, require sweat chloride test. Data and case study resulting in delayed diagnosis was shared.
8. Periodic SCID review
    a. Periodic meeting occurred in November 2017, discussed national SCID meeting in August 2017. Review of SCID methodology, screening algorithm, and follow-up reporting. Referral to immunology is by Initial Critical or Critical by consecutive ABN/UNSAT (Currently 2 x UNSAT, will move to x3 in 2018). Repeat requested if ABN, UNSAT, < 24 hours, transfused. Data was reviewed including 2 SCID babies identified. Richard discussed case studies that were shared at the meeting with the SCID babies being diagnosed within 7 and 8 days of life. The impact of heparin on inhibiting PCR reaction was shared and it was related to high number of UNSATs by specific submitters that initiated QA/QI.
9. NBS Data Review
    a. Jen Macdonald shared preliminary data for 2017 (Q1-Q3)
    b. NBS Timeliness Initiatives
        i. Willie Andrews shared the Secretary's Advisory Committee on Heritable Disorders recommendations for newborn screening
        ii. Transit Time:
            1. Sample to be collected between 24-48 hours, and to be received by laboratory by 24 hours after collection. Lab provides courier service to

	hospitals 6 days a week (Sunday-Friday). **2.** QA/QI improvements began in 2013- added courier to all hospitals, added Sunday (wasn't available at the time), monitor and visit top offenders, distribute quarterly report cards (monthly in 2018), UPS courier services for midwives and birth centers (2018) **3.** Shared new/redesigned report card- reviews different parameters: samples sent, transit time, rank to other hospitals unsatisfactory samples, providing averages, added new metric (samples over 4 days in transit), ultimate goal is to have all samples received within 1 day, set transitional goal of 2 days for Virginia. Limitation to identify midwives associated with birth centers, will work with Kim Pekin to create midwife. New diagnosed cases report accompanies each report card to show impact of newborn screen collection. **4.** Reviewed additional data parameters for time critical results and QA/QI measures for improving workflow and decreasing time to report time critical results and all results reported at less than 7 days of life
11:35 - 12:30	Old Business A. CAH screening: 2nd tier screening update a. Chris Nixon, Tandem Mass Spectrometry Principal Scientist, reviewed main goal of 2nd tier CAH implementation is to reduce CAH false positives. Single enzyme extract of 17-OHP is inexpensive but has high false positive rate. Developing an optimized DBS extraction procedure using mass spectrometry and chromatography to separate and extract. Space has been acquired, proposed implementation Q1 2018. Currently working on validation processes and IT build-out for daily utilization and optimizing process workflow B. Addition of Pompe Disease to Virginia's NBS Panel, vote on recommendation a. Jen Macdonald reviewed background of LSD workgroup convened on 2/24/2017, NBSAC reviewed on 6/29/2017. Process includes ACHDNC disorder review, current scientific literature, review of other state screening data, ID infrastructure needs, economic evaluation, ID benefits and Risks, summary of Findings. If committee does vote to add disorders to panel, next step is commissioner report, moves to board of health, regulatory process, planning and implementation. b. Christen Crews reviewed overview of workgroup sessions with recommendation to add Pompe to Virginia's panel with 2nd tier reflex

	testing. c. Vote: "Virginia shall initiate regulatory action to implement newborn screening for Pompe Disease. Screening methodology shall include: 1st tier screening in conjunction with 2nd tier reflex testing." 15 Y 0 N. C. Addition of MPS-1 to Virginia's NBS Panel, vote on recommendation a. Christen Crews reviewed overview of workgroup sessions with recommendation to add MPS-1 to Virginia's panel with 2nd tier reflex testing. Kim Pekin questioned concerns/impact of added cost to newborn screening fee, Dr. Wilson agreed would be beneficial if additional funding came from general funds and support from state legislature as it is mandated. Vote: All 14 voting members were in favor of adopting recommendation. b. Vote: "Virginia shall initiate regulatory action to implement newborn screening for MPS-1. Screening methodology shall include: 1st tier screening in conjunction with 2nd tier reflex testing." 15 Y 0 N.
12:30	Break and Lunch Set-up
12:45 – 1:30	Working Lunch - Guest Speaker Dr. Crystal Proud, Pediatric Neurologist, CHKD will be speaking on Spinal Muscular Atrophy (SMA) • Reviewed several cases of patients in clinic and challenges with SMA, presentations of low tone, proximal weakness, extremely expressive facial movements, no neuromuscular response on reflex check, etc. • Several different phenotypes- o Zero- noted at birth, presumably symptomatic in utero, may not survive neonatal period o 1- never able to sit o 2- able to sit o 3- able to walk o 4- mid to late adult onset, milder disease • Has 26 patients in her clinic ranging from a few weeks old to 78 years • Presentation/Diagnostic Considerations- 5q SMA o Weakness- neurogenic vs myopathic vs dystrophic o Pattern of weakness- proximal vs distal o Labs – CPK o EMG/NCS o Reassuring neuroimaging (confirm no structural reason for their weakness) o 1 in 6-11,000 births o Carrier: 1 in 38 – 1 in 70 • Interventions include addressing mobility, joint contractures (PT/OT), respiratory insufficiency, nutrition, scoliosis, osteoporosis. Requires multidisciplinary care • Antisense oligonucleotide technology allows SMN2 gene to create more functional protein (Spinraza), approximately $125,000 a dose, benefit of efficacy of drug outweighs burden of cost

	• Proposing to add it to the newborn screening panel in Virginia
1:30 -3:00	New Business A. Expansion to 6 days/week a. Willie Andrews: In Q1 2018, expand current schedule Monday-Friday (Saturday half day) to Monday-Saturday. Lab is working with HR to expand staff to dedicated Saturday workers. Expansion to 7 days including Sunday coverage will occur in near future. b. Dr. Wilson requesting back line phone number for when reporting critical newborn screening results to consultants. B. Periodic Regulation Review, formation of subcommittee or actual start of review a. Jen Macdonald: Regulation review still option for this term, closed for public comment but open for review. VDH Director of Regulatory and Legislative Affairs has recommended to do both regulation review and addition of new disorders at the same time. i. Requesting forming of work group of committee to do review of regulation, Jen Macdonald will email voting members to solicit stakeholders C. Addition of X-ALD to Virginia's Panel, formation of subcommittee a. X-ALD has been approved and added to RUSP, discussed doing reviews of X-ALD and SMA together. b. Jen Macdonald will email voting members for to solicit stakeholders and conduct reviews D. 2018 meeting dates and potential agenda items a. June 7 b. November 15
	Meeting adjourned at 2:00 PM.