# Meeting Minutes

Universal Newborn Screening Advisory Committee
(a.k.a Genetic and Metabolic Diseases Advisory Committee)
December 16, 2020
10:30 am-2:00 pm

WebEx Virtual Meeting

**Members Present:**
Aisha Ahmed, M.D., Lurie Children's Hospital
Javeed Akhter, M.D., Advocate Children's Hospital
Carrie Balian, EHDI Parent
Barbara Burton, M.D., Lurie Children's Hospital – *Committee Chair*
Glynnis Cailteux, R.N., Kankakee County Health Department
Rich Dineen, C.G.C., RUSH
Julie Fleischer, M.D., SIU School of Medicine
Lauren Hitchens, A.P.N., Lurie Children's Hospital
George Hoganson, M.D., UIC
Lewis Hsu, M.D., UIC
Ashish John, M.D., Memorial Health System
Rachel Katz, L.S.W., Lurie Children's Hospital
Janine Yasmin Khan, M.D., Lurie Children's Hospital
Mary Kreiter, M.D., Lurie Children's Hospital
Beth Leeth, M.S., Lurie Children's Hospital
Shannon O'Brien, R.D., University of Illinois Medical Center
Robin Stoner, Audiologist, RUSH

**Other Attendees:**
TaLana Hughes, M.P.H., Sickle Cell Disease Association of Illinois
Leena Mithal, M.D., Lurie Children's Hospital
Esha Phatak, UIC – Division of Specialized Care for Children
Tess Rhodes, UIC Division of Specialized Care for Children

**IDPH Representatives:**
Laura Ashbaugh, Newborn Screening Follow-up Nursing Supervisor
Rebecca Barnett, Grants Coordinator
Khaja Basheeruddin, Newborn Screening Laboratory Supervisor
Onesia Bishop, Newborn Screening Laboratory
Leslie Chapman, IDPH Laboratory
Jennifer Crew, Acting Newborn Screening Laboratory Manager
Hector Diaz, Newborn Screening Laboratory Supervisory
Joan Ehrhardt, Newborn Screening Genetic Counselor
Shannon Harrison, Newborn Screening Follow-up Nursing Supervisor
Chrissy Knepler, Newborn Screening Follow-up Nursing Supervisor
Ginger Mullin, Newborn Hearing Screen Program Coordinator
Mohammad Nasir, Newborn Screening Laboratory
Yin Peng, Newborn Screening Laboratory Scientist
Joel Price, Newborn Screening Laboratory
Pam Smith, Newborn Screening Program Manager
Charles Williams, IDPH Laboratory

**Welcome. Housekeeping, and Introductions**

Rebecca Barnett opened the meeting at 10:31. Members were welcomed, and housekeeping items covered. Pam Smith welcomed members to the first virtual UNSAC meeting. Pam stated she is retiring at the end of the year. Joan Ehrhart will be temporarily assigned to the role of Newborn Screening (NBS) Follow-up Section Manager.

Dr. Leena Mithal of Lurie Children's Hospital was introduced to the members. Dr. Mithal was invited to present to the committee later in the program on the Importance of DBS Access for the Diagnosis of Congenital Cytomegalovirus in Children with Compatible Symptoms. Joan thanked the committee members for their attendance, service and improving the IDPH NBS Program. Joan welcomed new members. Agenda items were discussed for today's meeting, and the agenda turned over to chairman, Dr. Barbara Burton.

**Approval of October 23, 2019 Meeting Minutes**

Dr. Barbara Burton welcomed members and guests, and noted that meeting minutes were sent out in advance. No corrections, additions or errors were noted. A vote was held, and the meeting minutes were approved.

**Committee Member Status**

Joan Ehrhardt stated there are no current, pending or vacant positions. One parent/community member representative from La Rabida could not be reached. IDPH may need recommendations from members for a parent nominee in the coming year.

**IDPH Reports**

**Newborn Screening Laboratory**

- **Covid19 response** – Jennifer Crew stated she is the acting NBS chief for the lab and supervisor of the diagnostic virology lab. IDPH was the first lab in the nation to begin screening for COVID 19. Lab was able to hire and reassign molecular staff to run the virology lab to test for COVID 19. They also felt direct effects of COVID 19. Several staff had COVID or were exposed to COVID, so they were quarantined at home. Several staff members had to alter their schedules due to childcare needs. Dr. Yin Peng was detained in China, being quarantined, which hindered MSMS cutoff changes and other tasks for several months. Many indirect effects were felt, since staff was dispersed in the lab due to social distancing. Conference rooms were used to report COVID 19 results and data entry. PE LIMS staff was working from home, so response times were increased. SMA newborn screening implementation was pushed back from April to June due to all these changes. There have been delays in hiring new staff due to HR and CMS working from home and increased timing of response and duties.
- **Less than 24-hour sample collection** – Jennifer Crew stated that in August IDPH changed the way the NBS lab reports out unsatisfactory NP specimens in the NICU protocol. NICU admissions should have NBS collected upon admission and repeat at 48-72 hours of age. These early collections lead to false positive results due to factors such as timing, stress, transfusions, special feedings, and antibiotics. These factors affect results for several disorders. Therefore, IDPH no longer report results on specimens that are not valid at <24 hours of age. Eight percent of specimens are NP unsatisfactory specimens received in the lab. Interpretation is reported out as unsatisfactory. Valid results are reported out as usual. Shannon Harrison will share the memo with committee members.
- **ALD screening** – Khaja Basharuddin provided an update on NBS for X-linked adrenoleukodystrophy (ALD). Screening for ALD began on June 18, 2019 in Illinois. IDPH lab measures C26:LPC from dried blood spots using HPLC and MSMS. Laboratory procedure was explained. So far, 238,000+ specimens have been screened from the start date until December 10, 2020. Results for one-year June 2019-June 2020 were shared in handouts. Of 166,507 screens, this includes ten (10) cases with clinical follow-up information: 4 males with X-ALD (categorized as presumptive and monitored clinically per recommendations), 5 female carriers of ALD, 1 was diagnosed with peroxisomal disorder (Zellweger spectrum). Reference the ALD Data Summary. To date, IDPH estimates 1 in 33,000 babies has ALD.
- **SMA screening** – Vineet Dhiman provided background on spinal muscular atrophy (SMA). Illinois began screening for SMA on June 29, 2020. Vineet discussed the Illinois screening platform and data. IDPH uses a PCR probe that is multiplexed into the SCID screening assay. It tests for presence or absence of the SMN1 gene. If not detected, we report the result out as positive. The assay does not distinguish or identify carriers. Validation included 10,000 specimens to test our assay and laboratory information management system (LIMS). Projected start date was April 2020, but this was delayed due to COVID. Data: 8 positive specimens; 4 had SMA 2 or 3; 4 were identified as carriers. All of our diagnosed cases had a value of 0-no PCR signal. All 4 carriers had a weak signal on PCR or a result of Cq ≥ 35, possibly due to a mutation in the primer exon 7 junction. On validation, the Lab confirmed some carriers per the CDC. We received 2 of these as control materials. We need more data points to verify the mutation location. SMN2 screening data will potentially help specialists provide better clinical care (e.g., facilitate insurance approvals). IDPH has approval to purchase instruments in hopes to add SMN2 screening within the next year.

  A severe combined immune deficiency (SCID) screening reporting change went into effect with SMA screening. We changed from TREC copies to Cq value for SCID screening. Most states around the nation use the Cq value, and it is the gold standard. Also changed from 250 TREC copies to Cq equivalent value of 200 TREC copies (Cq ≥ 35). It reduced our false positive results for SCID.

- **Krabbe algorithm revisions** – Khaja Basharuddin discussed proposed changes to the Krabbe screening protocol. If we have low GALC activity, we will refer to specialists to help diagnose Krabbe disease. Currently, our specimen is sent to PE 2nd tier testing for psychosine levels, 30 KB deletion and gene sequencing. Current protocol and reporting were explained to the Committee. Three new proposed changes were discussed. Examples of abnormal reports were viewed and discussed.
- **Proposed cutoff changes** – Dr. Yin Peng discussed proposed MSMS cutoff changes, found in the handout "Phase 1 Report for Neobase cutoff Revision". These changes will decrease our false positive rate of NBS results. Compared to the CLIR database, new cutoffs did not identify all MSUD cases; missed 4 cases of 168 in the data. Dr. Onesia Bishop mentioned the study being Phase 1. IDPH will transition to Phase 2 once we change to Neobase 2 next year.
- **Mailer Revision** – Jennifer Crew stated IDPH is currently working on updating the IDPH Newborn Screening Mailer report that is sent to hospitals and physicians. We are reviewing other state reports to decide on what would work best for Illinois. Templates were shown. PerkinElmer developed a comments section that is currently being used on our reports to provide more clarification about the results. Examples of other state reports were shown. Projected date of new mailer is 2021. Members also suggested adding QR codes for physicians to be able to use to identify comments.

**Newborn Screening Follow-Up Program**

- **Covid19 impact (COOP and Clinical Follow-up)** – Chrissy Knepler stated how COVID has affected the blood spot program. Many hospitals and contact personnel changed to working remotely, so IDPH had to change its usual protocols, (e.g. emailing instead of faxing reports). Many hospital contacts changed or staff was furloughed. Therefore, it was more time consuming for IDPH staff to locate contacts and get follow-up communication. Many local health department nurses replaced home visits with phone calls, which increased our lost to follow-up rates. Families and parents are afraid to take their newborns to clinics and hospitals for repeat specimens, referrals, etc. The follow-up program also began working remotely. Immediate follow-up is still ongoing, but some of our follow-up activities have been delayed. EHDI has also had issues with delays in getting follow-up testing and referrals due to parents not wanting to bring their newborns into hospitals.
- **Staffing updates** – Pam Smith described staffing issues due to COVID and due to changes in our staffing model. The EHDI program created a new position to assist with grants management. The blood spot program was approved by the Director's Office to change from a clerical model to a registered nurse model for follow-up of abnormal newborn screening results. In past 45 days, 4 new nurses have been hired, and we are currently interviewing for 5 other nursing positions. Hiring has been delayed due to COVID, and resources were stretched with nurses accepting emergency positions. With the new registered nurse model, our nursing supervisors will be able to dedicate more time towards education and outreach, as opposed to the extensive oversight from the clerical model. Office specialists (entry level) have continued to promote out of the program, which is counterproductive. We also lost our NBS data manager during this time, as well as the entire program having to transition to working remotely. We are currently working at <50% of our standard staffing. With nine new hires expected, we have to focus on training them. Then, we will hire a new data manager. We are committed to providing the data needed by our specialists and workgroups.
- **Recommended CCHD NBS algorithm** – Joan Ehrhardt reviewed current and recommended revisions of the national NBS CCHD screening algorithm (included in handouts). Illinois adopted the nationally recommended screening algorithm in 2011, but Illinois is not currently collecting any screening data to gauge quality measures. When comparing both screening algorithms, published research concluded the false positive rate did not significantly increase. It simplifies the screening process, and the new algorithm is recommended. In Illinois, we currently have new rules in final review with our legal team that will require facilities to report their screening results and follow-up with the IDPH Newborn Screening Follow-up Program. It is expected that the AAP will endorse this new screening algorithm in 2021. Illinois would like to be proactive and institute this expected change at the same time that we begin collecting screening results from facilities. Dr. Khan at NMH speculated that the majority of CCHD is diagnosed prenatally. However, they also discover other, non-critical heart disease. Dr. Khan will reach out to Joan regarding sharing of screening data at NMH.
- **Grant funding** – Rebecca Barnett reviewed the genetics grants reports (included in the packet). This includes the genetics education services, genetic counseling, sickle cell grant-funded activities. The grant cycle runs from July 1-June 30 each year. She and Joan met with the grantees regarding moving to competitive grant implementation. There has been a decline in services due to COVID, but IDPH allowed alternative services (e.g. telehealth, online/web-based education, etc.) to meet program requirements. FY2020 grant program data was reviewed with the Committee (reference materials included).
- **Other Activities:** Joan Ehrhardt provided a few additional updates. State of Illinois legal staff has also been impacted, and the NBS Administrative Rules draft is currently with IDPH legal review before it can move on to JCAR. The Illinois Society for Genetic Professionals (ISGP) asked IDPH, NBS for its genetics plan released in 2007. They want to review it, update the plan to align with their strategic plan. Per feedback, the 2007 plan is pretty solid, but ISGP would like to see some updates. We are currently working with the GC program at NW to establish a genetic counseling rotation.

**Early Hearing Detection and Intervention (EHDI) Program**

Ginger Mullin provided EHDI Program updates to committee members. The EHDI Program is a collaborative effort between IDPH, HHS and DSCC. A program overview and review of funding was given.

- **HRSA grant with UIC-DSCC** – Esha Phatak from UIC DSCC provided the background of the EHDI grant, funded through March 2024. The goal is to establish a workgroup (under this committee) to improve the number of children receiving diagnostic testing by 3 months of age and follow-up services by 6 months of age. DSCC will work with DHS and IDPH to achieve these goals by 2024. The workgroup membership will have representation from DSCC, Early Intervention, IDPH, Health Start, Guide By Your Side, Chicago Hearing Society, IL Society of the Deaf, Families for Hands and Voices, Maternal, Infant and Early Childhood Home Visiting Program. Please contact Esha if you are interested in being a part of the work group. The committee members unanimously voted to create this working group.
- **Dr. Leena Mithal, presentation** (after genetic professional discussion). Dr. Mithal summarized key points of congenital CMV infection, which can cause hearing loss and neurological disability. The critical time to make a definitive diagnosis from saliva/urine is within the first 3 weeks of life to tell if it is congenitally infection. Antiviral meds are available but have side effects if not acquired congenitally. A newborn screening sample, residual dried blood spot (DBS), is very useful in identifying cases in children >3 weeks of age. Data from DBS is not very useful for universal screening, but it is very useful in identifying cases of children >3 weeks of age with compatible symptoms. The issue is getting access to DBS cards and extend the timeframe that these DBS can be kept. Currently, IDPH only keeps normal cards for 4 months of age. Some states are keeping cards for many months to years now for this area and for research or to make a retrospective diagnosis. Congenital CMV was reviewed with the group. The Committee is sympathetic to the Cause in releasing cards/residual sample. Dr. Bishop reiterated the need for rule changes to release the cards. There is also a logistical issue of storage capacity with IDPH. Dr Burton and Dr Khan would like IDPH to look into the issue of storage capacity and retention and present at the next meeting.

**Other Reports**

- **Newborn Screening and Laboratory Subcommittee** – Dr. Hoganson stated the Subcommittee met last week. All areas of discussion were covered in that meeting.
- **Newborn Screening Hemoglobinopathy Collaborative** – Shannon Harrison stated 11 Illinois and two St. Louis centers participate, as well as representatives from Sickle Cell Disease Association of Illinois (SCDAI), and Sickle Treatment and Outcomes Research in the Midwest (STORM) in bimonthly meetings. The Collaborative has met a little less due to the pandemic.

**Reports from Partners**

- **Norton & Elaine Sarnoff Center for Jewish Genetics** – no report
- **Sickle Cell Disease Association of Illinois** – Talana Hughes stated they try to work with hematology clinics and stay connected with families. SCDAI is seeking legislative support to get funding for SCD to increase patients' access to services.
- **Illinois Society of Genetic Professionals** – no report
- **Sudden Infant Death Services of Illinois, Inc**. – no report
- **Division of Specialized Care for Children** – Tess Rhodes displayed their website on how they can help families. DSCC will follow-up with the families requesting services. Tess reviewed 3 programs at DSCC and available services and resources listed on their website. There is also a referral form for genetics, metabolic and hearing available on the website.

**Other New Business**
No new business

**Next Meeting Date**
Next meeting date will be determined. Historically, committee meetings are held twice a year (once in the fall, once in the spring). IDPH stated the goal is to meet twice a year. IDPH will schedule a virtual meeting in April or May.

**Emergency contact information -** Rebecca Barnett requested new members complete the emergency contact information sheet and for returning members to update their emergency contact information if needed.

**Public Comments**
None

**Meeting adjourned at 2:14 p.m**.