

Meeting Minutes

Universal Newborn Screening Advisory Committee (a.k.a Genetic and Metabolic Diseases Advisory Committee)

April 3, 2024

9:30 am – 11:30 am CST Hybrid (Virtual by Webex and In Person at OSF in Peoria)

Attendance

Member Name	Specialty	Affiliation	In Person	Virtual
Able, Kasie	Parent & Family Advocate	North Central Region		-
Ahmed, Aisha	Immunologist	Lurie Children's Hospital		-
Akhter, Javeed	Pediatric Pulmonologist & Immunologist	Advocate Christ		X
Balian, Carrie	Parent – EHDI	Chicago area		X
Baker, Josh	Geneticist (Chair)	Lurie Children's Hospital		X
Harris, Keondra	Caregiver	South Central Region		X
Horne, Christine	Advance Practice Nurse	UIC Medical Center		X
Hsu, Lewis (pending)	Pediatric Hematologist	UIC		X
Jackson, Heather (pending)	Local Public Health Nurse	Fayette County Health Department		-
John, Ashish	Pediatrician	Memorial Medical Ctr.		-
Khan, Janine	Neonatologist	Lurie Children's Hospital		X
Katz, Rachel	Social Worker	Lurie Children's Hospital		X
Menendez, Beatriz	Genetic Counselor	UIC		X
Stoner, Robin	Audiologist	Rush University		X
Arduini, Katie (pending)	Registered Dietitian	Lurie Children's Hospital		P
Burton, Jen (pending)	Genetic Counselor	UICOM Peoria	P	
Coates, Ryan (pending)	Pediatric Neurologist			-
Tsai, Anne (pending)	Metabolic Geneticist	UIC		P
Fleischer, Julie (pending)	Geneticist	Pending		-
Whitehead, Jax (pending)	Pediatric Endocrinologist	Pending		P
Ex Officio Members				
Alexander Smith	Ex Officio Member	IDPH, Perinatal Advisory Council		
Ebonie Zielinski	Ex Officio Member	UIC-Division of Specialized Care for Children	X	
TaLana Hughes,	Ex Officio Member	Sickle Cell Disease Association of Illinois		X
Sarah Jurgensmeyer	Ex Officio Member	Illinois Society of Genetic Professionals	NA	-

Guests and Members of the Public

Nickki Gulden, Nurse UIC, Shelby Miller, Dietitian UIC

IDPH Staff

In person: Rebecca Barnett, IDPH, Joan Ehrhardt, IDPH NBS Follow Up Program, Chrissy Knepler, IDPH NBS Follow Up Program, Shannon Harrison, IDPH NBS Follow Up Program, Laura Ashbaugh, IDPH NBS Follow Up Program
Virtual: Margo Asbury, IDPH NBS Lab, Kasey Burger IDPH NBS Follow Up Program, Tate Kappell, IDPH NBS Lab, Ginger Mullin IDPH EHDI Program, Christine Gilley, IDPH NBS Follow Up Program, Khaja Basheeruddin, IDPH NBS Lab, Mohammad S Nasir, IDPH NBS Lab, Joel Price, IDPH NBS Lab, George Dizikes, IDPH Lab, Vineet Dhiman, IDPH NBS Lab, Kathleen (Kate) Jordan, IDPH EHDI Program, Josh Geltz IDPH Lab (at 10:21)

Welcome

The meeting was called to order at 9:30 am with welcome and housekeeping reminders. There was quorum.

Approve Minutes (December Meeting)

December 2023 meeting minutes approval: Christine moved to approve, with no changes; seconded by Carrie. Minutes were approved with none abstaining and none opposed. (9:35)

Committee Member Status

Joan provided an update. Several applications are pending. If all are approved, the Committee will have all 20 positions filled. Two additional current member terms will expire in February 2024.

IDPH Reports

Early Hearing Detection and Intervention (EHDI) Program: Ginger Mullin provided programmatic updates. The current funding structure, being a blend of different federal grant funding programs (Centers for Disease Control and Prevention (CDC), Health Resources and Services Administration (HRSA)) was discussed, and a forecast of future grant opportunities and funder priorities. The national technical assistance center (grant funded) is moving to Gallaudet University after many years with current center. This is anticipated to change EHDI program focus nationally. An increased focus by HRSA on longer term outcomes, like literacy, for children identified with hearing impairments through universal newborn hearing screening puts pressure on EHDI Programs in Illinois and other states to expand beyond their traditional mission and established activities and partners. At IDPH, program staffing is a challenge, with long timelines to fill vacancies. Ginger provided program data on outcomes. Annual data for 2022 has been shared with CDC, as a grant deliverable. The Illinois birth rate continues to decline substantially, being about 125,000. The refer rate is depressed since COVID. Issues related to equipment, training and oversight were identified. The Illinois lost to follow-up rate is estimated at 17%. While this needs to be improved, is better than the national average of about 20%.

Carrie Ballian provided an update on **Illinois Hands & Voices** activities to support families. Carrie also shared national updates on work toward adding congenital cytomegalovirus infection (cCMV) to newborn screening panels. Minnesota has implemented cCMV NBS statewide. Illinois EHDI Program is collaborating with the Adverse Pregnancy Outcomes Reporting System on activities to improve cCMV identification and tracking. Committee members asked questions and discussed various strategies for publicizing EHDI needs and increasing knowledge and awareness about EHDI aims and improving outcomes. Dr. Baker suggested a publication of Illinois' experience and data. Ginger states we don't have anyone on staff with that expertise at this time. Dr. Hsu asked if the program could partner with community health workers. Ginger asked that the Committee consider what action might be taken to provide additional stable funding to strengthen the EHDI program's capacity and sustainability. She also discussed various strategies and approaches EHDI Program has taken to improve system outcome measures.

Newborn Screening Laboratory: Tate Kappell provided updates from the IDPH Newborn Screening Laboratory. The lab currently has vacancies for 19 technicians which they want to post soon. They have posted a microbiology and whole genome sequencing supervisor to help with expansion of sequencing. Another position to help with Tate's position in direction and leadership of lab is to be posted. **Neobase2 (NB2)** kit for amino acid, organic acid and fatty acid oxidation disorders is estimated go live of June or July 2024 depending on the vendor Revvity. **GAMT deficiency** is now included on the Recommended Uniform Screening Panel (RUSP) for all states. IDPH will look at adding GAMT deficiency to the Illinois newborn screening panel once NB2 is live. The NBS Lab plan is to multiplex it to NB2 working with our vendor. Metachromatic leukodystrophy (MLD) screening requirements are being investigated. IDPH NBS Lab is talking with a couple of other states about 1st and 2nd tier tests. They are hoping to get something in the works by end of year. Duchenne muscular dystrophy (DMD) newborn screening potential is also under discussion now. IDPH NBS Lab is waiting to see an FDA approved kit if DMD were to be added federally to the RUSP. Tate added that the biggest hurdles for adding new tests are staffing and funding.

Cystic Fibrosis (CF) molecular panel update was provided by Vineet Dhiman. The old recommendations were for NBS systems to use the 23-mutation panel. New recommendations by ACMG in April 2023 are to screen for 100 *CFTR* mutations. This gives a better representation of diverse populations. Current Illinois CF mutation panel includes 74 mutations with 46 of the 100 ACMG recommended mutations. NBS Lab is exploring an upgraded CF panel to include all 100 recommended mutations. But it would not include all 74 on the current panel. Vineet analyzed retrospective data of all positive CF cases from March 2018-December 2023 and presented it to this group. He has been talking with our vendor to see if we can include a couple of additional

mutations that are seen more often in Illinois. NBS Lab has plans to move forward to this new enhanced panel. Illinois is to beta-test the new panel and assess any changes in workflow in the next year or two. Jennifer asked if discussion has been had to get away from variant panel screening and moving to whole gene sequencing in Illinois. Vineet stated they have explored but will not be able to in the near future. However, these guidelines state still to do a screening panel before gene sequencing. The NBS Lab wants to meet ACMG recommendations. It was pointed out that this enhancement will triple the cost of the CF screen. Dr. Akhter asked about Wisconsin newborn screening program which uses the Illumina panel but has a much smaller birth rate than Illinois. Wisconsin also has an established infrastructure for molecular analysis already set up where Illinois does not at this time. Dr. Tsai also stated how WI and IL have very different ethnicities. Jen Burton stated she had three cases in which a baby had one mutation identified but had CF clinically, so it is a rare occurrence. Vineet stated there is no panel on the market that captures all disease-causing mutations for CF. More than 400 have been described as disease-causing in the gene variant database CFTR2.

Newborn Screening Follow-Up Program: Joan Ehrhardt discussed the program make up of the Health Assessment and Screening Section and current vacancies which are seen in all program areas. Joan discussed the NBS Follow Up Program records archive project in preparation for program offices to be moved. Rebecca Barnett provided an update on the NBS and Genetics Grants Programs for Genetic Centers, Sickle Cell Centers, and Local Health Departments.

Other Reports

There was no activity in the NBS Lab Subcommittee. Shannon reported for the NBS Hemoglobinopathy Collaborative which continues to meet bi-monthly via Webex. This group is open to all hematology centers in Illinois and St. Louis, Missouri. This group discusses sickle cell and other hemoglobinopathy related educational efforts, out-reach, grant funding, research projects, state screening and follow-up issues.

Old Business

UNSAC Recommendation to add GAMT to IL NBS Panel was made at the last meeting. Dr. Baker will write letter to IDPH Director on behalf of UNSAC in support of screening.

New Business

Jen Burton asked IDPH if any movement has been had regarding restructuring the callout model for abnormal screens due to a couple of delays. Joan stated that there had been internal discussion regarding referrals, but nothing had been decided. Dr. Hsu stated as we are having more children immigrating to IL, we are getting more children who are diagnosed with disease. Dr. Baker said that there is national recommendation that all undocumented refugees should have a screen. Dr. Tsai asked if we have calculated costs of screen for each individual, genome sequencing, and referrals such as calling police. Joan stated that the lab had to leave the webinar earlier in the meeting. They have been working on cost estimates. Joan also recognized costs for EHD, CCHD and follow-up. She stated that the NBS Program would like to ask for a fee increase for this and to support an epidemiologist. Referral has also experience headaches for DHS no longer being able to go out to homes and do home visits or even phone calls on patients. Chrissy explained DCFS guidelines with the Committee, other alternatives used by the NBS Program, as well as education that is needed. Jen Burton also brought up the issue with NICUs functioning with metabolic coverage. Chrissy Knepler stated that IDPH can only make recommendations but cannot dictate to NICUs on how to practice. She suggested members representing the entire state be recruited for the lab subcommittee to address concerns or possibly set up separate work groups to focus on these issues. Dr. Tsai discussed talking with perinatal/OBGYN regarding testing and sequencing DNA. TaLana Hughes stated they have success in finding parents as a community-based organization with a Community Health Worker (CHW).

Reports from Partners

Illinois Society of Genetic Professionals – Joan Ehrhardt shared that their strategic plan update meeting was held on April 2, Annual Symposium will be May 20 at Prentice Women's Hospital. ISGP is partnering with Northwestern in an information workshop to be hosted in June 2024.

Division of Specialized Care for Children – Ebonie Zalinski shared information on NBS and hearing referrals. DSCC went through a site visit for accreditation to provide services and care coordination and to expand financial assistance. Many families need help with travel, transportation, equipment, etc. so if DSCC gets this funding, that will be used to help with this need.

Sickle Cell Disease Association of Illinois – TaLana Hughes focused on keeping patients and families updated specifically on new treatments, working with CHW to identify patients not linked to care, transportation, insurance, partnering with IDPH, connecting with hematologists. She shared that the Governor signed an Executive Order on March 18 to finance equitable access to SCD treatments (and other high-cost drugs), leverage national and state providers, and establish an advisory committee to accomplish this. She also mentioned being excited for grant funding that SCDAI could apply for, being IDPH funding for new SCD grants. Region 4/Midwest Genetics Network funding has ended. They were able to pull together a train the trainer guide for parents to navigate care and advocate for their children.

Next Meeting Date

TBA (Virtual option is preferred)

Public Comments *None*

Motion to adjourn at 11:36 and meeting adjourned.