

Universal Newborn Screening Advisory Committee

(a.k.a Genetic and Metabolic Diseases Advisory Committee)

April 11, 2025

9:00 am – 12:30 pm CST

Approved Meeting Minutes

Attendance.

	UNSAAC Member Name	Specialty	Affiliation	Attended
1	Able, Kasie	Parent & Family Advocate	North Central Region	
2	Ahmed, Aisha	Immunologist	Lurie Children's Hospital	X
3	Akhter, Javeed	Pediatric Pulmonologist & Immunologist	Advocate Christ	
4	Baker, Josh	Metabolic Geneticist (Chair)	Lurie Children's Hospital	X
5	Balian, Carrie	Parent – EHDI	Chicago area	X
6	Burton, Jen	Genetic Counselor	UICOM Peoria	X
7	Coates, Ryan	Pediatric Neurologist	UIC	
8	Fleischer, Julie	Geneticist	SIU	
9	Harris, Keondra	Caregiver	South Central Region	X
10	Horne, Christine	Advance Practice Nurse	UIC Medical Center	X
11	John, Ashish	Pediatrician	Memorial Medical Center	
12	Katz, Rachel	Social Worker	Lurie Children's Hospital	X
13	Khan, Janine	Neonatologist	Lurie Children's Hospital	X
14	Stoner, Robin	Audiologist	Rush University	X
15	Tsai, Anne	Metabolic Geneticist	UIC	
16	Whitehead, Jax	Pediatric Endocrinologist	Lurie Children's Hospital	X
	Quorum met			10 of 16
	IDPH Staff Name	Lab/NBS		Attended
1	Ashbaugh, Laura	Newborn Screening		X
2	Barnett, Rebecca	Newborn Screening		X
3	Dhiman, Vineet	Laboratory		X
4	Dougherty, Matt	Assistant General Counsel		X
5	Ehrhardt, Joan	Newborn Screening		X
6	Gilbert, Steve	Laboratory		X
7	Harrison, Shannon	Newborn Screening		X
8	Jordan, Kate	Early Hearing Detection and Intervention		X
9	Kappell, Tate	Laboratory		X
10	Knepler, Chrissy	Newborn Screening		X
11	Komis, Warren	Laboratory		X
12	Mullin, Ginger	Early Hearing Detection and Intervention		X
13	Nasir, Mohammad	Laboratory		X
14	Smith, Matt	Chief Policy Officer		X
15	Stalets, Melissa	Office of Health Promotion		X
	Ex Officio Members	Agency		Attended
1	Hughes, TaLana	Sickle Cell Disease Association of Illinois (SCDAI)		X
2	Pollock, Maria	Illinois Rare Disease Commission (IRDC)		X
3	Zielinski, Ebonie	Division of Specialized Care for Children (DSCC)		X

I. Welcome and housekeeping.

9:07 a.m. Joan Ehrhardt welcomed members and provided housekeeping reminders. Voting was postponed until the Committee reached quorum.

II. Approve April 2024 meeting minutes.

The Committee met quorum to vote on minutes. Christine Horne motioned to approve the April 2024 meeting minutes. Jen Burton seconded. No one opposed. Meeting minutes were approved at 9:57 a.m.

III. Committee member status.

Joan stated the committee has two member terms that will expire this year, Rachel Katz in June, and Josh Baker in December. Arlene Carlos' application is pending approval. Beatriz Menendez resigned. Aisha Ahmed, Robin Stoner, and Javeed Akter terms have expired. Joan encouraged these members to reach out if they would like to renew their terms.

IV. IDPH reports.

A. Newborn Screening Laboratory. Tate Kappell, the Newborn Screening Section Chief presented laboratory updates to the Committee.

Staffing. The laboratory is hiring 18 Clinical Laboratory Technician/Trainee positions. The CDC Foundation contract is ending this June, so the lab will lose CDC staff. Tate is hopeful to train new employees prior to CDC staff leaving. MSMS (metabolic screening/NeoBase 2) is most affected by staffing shortages.

New disorders update (GAMT/MLD/ALD triplex). In July 2024, the lab transitioned to a new assay, NeoBase2, and noticed an increase in the number of abnormal callouts. The NBS lab anticipated needing additional data to further refine cutoffs after going live with the NB2 assay; however, several thousand more samples are needed to better refine cutoffs. Current cutoffs are available on the IDPH website for reference. To better refine cutoffs and reduce false positives, prompt feedback from specialists on case diagnoses are needed. The Lab Subcommittee is also needed to routinely discuss matters such as these to provide both NBS program and clinical perspectives.

Neobase 2 (NBS2) cutoff updates. Two, new disorders were added to the panel, GAMT and MLD. DMD will likely be the next disorder added by the NBS program by using an FDA-cleared kit (already out). GAMT deficiency will no longer be added to Neobase2. No state is screening for MLD, however, we are exchanging information with New York to work on the assay. The lab would like to develop a 2-tiered screening (inhouse) and 3rd-tiered screening (send out). The lab has developed a multiplexed assay to screen for MLD, GAMT, and X-ALD in one test. Currently, the lab is running a study to determine feasibility on a large sample scale. Initiating validation will depend on when the lab can acquire new instrumentation required for the test.

Hemoglobinopathies update. The lab will transition to a newer HPLC platform for hemoglobinopathy screening. Providers should not notice any significant changes in hemoglobin results.

B. Newborn Screening Follow-Up Program.

Staffing. The Health Assessment and Screening section has been slightly restructured to include several programs: NBS Follow Up, Early Hearing Detection and Intervention (EHDI), NBS and genetic-related grants, and the School Age Vision and Hearing programs. Over the last two (2) years, we have been working to fill several positions. Recently, we filled: four (4) clerical titles and two (2) RN-1 positions. We will be reposting the data coordinator position and hope to post the NBS Genetic Counselor and Program Planner II (EHDI) positions soon.

Administrative Rules. The NBS blood spot sample retention code is at the final step with the Joint Committee on Administrative Rules (JCAR). For GAMT deficiency rulemaking, we are preparing for State Board of Health (SBOH)

review; MLD is also preparing for SBOH review. IDPH NBS is hoping to increase the screening fee to cover these costs. Joan briefly reviewed all new disorders that will be implemented.

Hemoglobinopathy Collaborative. Shannon reported the HGB Collaborative continues to meet bi-monthly to discuss education, research, and grant funding efforts.

Board Member Question: Javeed Akhter asked whether IDPH plans to change the current CF panel with the new CF Foundation guidelines. Vineet Dhiman stated the lab is researching the possibility of sequencing an increased CF gene panel with current staffing and funding shortages.

Grant Updates. Rebecca Barnett provided updates for each of the newborn screening grant programs, including Genetic Counseling, Sickle Cell Follow Up, and Genetics Education and Follow Up. These three programs received a Grants Accountability and Transparency Act (GATA) exemption to offer a continuous, three-year grant cycle that started July 1, 2024, and will end June 30, 2027. Having a continuous grant cycle will substantially reduce grant processing for both IDPH and grantees. Rebecca shared program data and accomplishments over the last grant cycle (FY22-FY24). We now have two (2) grant programs receiving general revenue funding, including the Comprehensive Sickle Cell Clinical Care grant and the new, Sickle Cell Prevention, Care, and Treatment grant program. Both programs were funded starting FY25 and are expected to renew for FY26. The new, Sickle Cell Prevention, Care, and Treatment program will provide, develop, and expand sickle cell services, targeting adolescents and adults.

C. IDPH Newborn Screening Budget Picture.

Matt Smith is the Chief Policy Officer at IDPH who provided the federal landscape updates impacting Illinois. Robert F Kennedy Jr assured senators he would not significantly change the vaccination programs at the CDC. In the meantime, the current administration is trying to reshape the federal government causing several lawsuits. The President has issued several executive orders (currently 111) within the first 100 days in office. The Department has been trying to adjust to the many changes, such as withdrawing from the World Health Organization (WHO). Matt focused updates relevant to current changes (within the last two weeks) and updates on the congressional budget process. IDPH has been issued termination notices for 14 COVID grant funding streams, part of a larger, \$1.06 billion package that started earlier. The Attorney General filed suits, along with 23 other states to request a temporary restraining order (TRO) against the US Department of Health and Human Services (HHS)'s termination of over \$11 billion in total grants. TRO granted until mid-April when further hearings will occur to determine whether to do a preliminary injunction. On March 27, 2025, HHS Secretary, Robert F. Kennedy, Jr. announced the Department would be restructuring agencies and reducing staff and number of divisions, making room for the new "Administration for a Healthy America". In mid-March we came close to a federal budget shutdown, so Congress passed a Continuing Resolution extending Federal appropriation levels through September 30, 2025. Congress is working to pass a new budget through the reconciliation process, which allows the Senate to pass bills without having a filibuster-proof majority. Matt went over the chambers' reconciliation instructions for committees to make cuts. Over the next ten years, the House and Senate need to cut HHS funds as instructed. From HHS's overall, annual budget of \$1.7 trillion, the House needs to cut \$880 billion, and the Senate needs to cut \$1 billion over a ten-year period. Most of HHS's funding includes Medicaid and Medicare spending. Since the budget resolutions have just passed, committees will meet to recommend cuts. Then, they will consider the overall bill on the floor. Once bills start to be considered, we'll have a better sense of spending changes.

D. Early Hearing Detection and Intervention Program.

Ginger Mullin presented updates for the EHDI program. EHDI is currently 2/3 staffed, having two vacant positions since November 2023. Ginger anticipates filling one of those positions in the next two months and is hopeful the other position will be posted soon.

EHDI applied for the competitive, five-year CDC grant, due March 6. The EHDI program has been under the CDC and the National Center on Birth Defects and Developmental Disabilities section that was eliminated. Also, all

CDC-EHDI staff are gone except one commissioned officer, who we anticipate being re-assigned. The EHDI grant is funded through federal partners, including the CDC (over 25 years) and Health Resource Service Administration (HRSA). HRSA released the notice of award for year two (2) of their five (5)-year grant.

Kate Jordan shared the data snapshot indicating the Illinois birth rate has been declining; however, the incidence of hearing loss has not been declining. The EHDI team works hard to meet or exceed our national benchmarks for screening no later than one (1) month of age, diagnosing no later than three (3) months of age, and enrolling into early intervention no later than six (6) months of age. The NIH reported the most common sensory defect is hearing loss (1.7 of 1000 babies in 2022).

Carrie Balian has a son diagnosed through the Early Hearing Detection Intervention (EHDI), Newborn Screening program. In addition to an UNSAC committee member, she is the coordinator for the Guide By Your Side program through Hands and Voices of Illinois. This program provides parent to parent support and resources for families and continues to support and collaborate with EHDI. Carrie provided updates on new guidance from the Family Centered Early Intervention (FCEI). The FCEI provides support for deaf and hard of hearing services. Their concept and principles reflect a consensus of an international multi-disciplinary co-production team. Carrie provided the FCEI categories by outlining support and structure concepts to improve early infant service outcomes. The EHDI program has translated CMV resources into ten (10) additional languages for non-English speaking families. Carrie reviewed several grant objectives to expand services to ensure early diagnosis and intervention – in addition to screening. Funding uncertainty is concerning, since this program is currently managed by a small team. Each team member contributes a very important role in meeting the 1-3-6 national benchmarks to ensure positive outcomes. The program continues to explore alternative funding sources; however, this work and what's requested through these grants is not sustainable. Carrie advocated for support to develop a plan to protect the future of the EHDI program. Newborn hearing screening is part of the Illinois RUSP and confirms about 300 babies with hearing loss each year, while other disorders like MLD only confirms about 3 cases per year. Currently, 10 states designate a portion of their newborn screening fees to support their EHDI programs.

The EHDI program has a three-part ask for the Committee to consider. EHDI is requesting the following services to be included in the Newborn Screening fee increase.

1. Increase \$25 - to sustain the EHDI program.
2. Increase \$7 - to add parent-to-parent support to keep the language and literacy on track of the deaf and hard of hearing children.
3. Increase \$5 - to support the Cytomegalovirus (CMV) infrastructure to meet education and documentation requirements. The work relative to CMV has been unfunded. The statute is insufficient in documenting children with CMV, and the program is not allowed to provide any follow up.

Action Items: the EHDI program will draft a letter (for UNSAC review) to the IDPH Director (cc the Rare Disease Commission) requesting to be added to the NBS fee structure. Joan Ehrhardt will connect with Maria Pollock to discuss next steps in bringing this request to legislators for consideration.

V. Old Business

A. Re-establish NBS Lab Subcommittee and identify Chair.

Jen Burton nominated Dr. Anne Tsai for the NBS Lab Subcommittee Chair. Christine Horne seconded nomination. No one opposed nomination. The Committee passed nomination of Dr. Anne Tsai as NBS Lab Subcommittee Chair.

B. Re-establish NBS Expansion Subcommittee and identify Chair.

Josh Baker would like to table the re-establishment of the NBS Expansion Subcommittee given the national aspect.

C. Other Old Business.

Jen Burton requested newborn screening data reports to be provided again to committee members. These reports included data, such as, number of callouts for each disorder. Joan Ehrhardt stated the Newborn Screening program lost data system knowledge from staff turnover and longstanding vacancies. The data is not easily accessible in the newborn screening system, and we do not have a current data coordinator to generate these reports.

Jen Burton would like to revisit discussions on restructuring abnormal callouts to initially include consultants or specialty centers. There continues to be many delays in referrals and misinformation from providers to families. One consideration would be to create a workgroup consisting of several consultants from different specialties to discuss best approaches in updating the callout structure. Jen agreed a possible short-term solution could be implementing an MOU with hospitals to expedite consultant referrals. Joan would like to discuss creating a workgroup with leadership. The portal is still on the list for development. Laura Ashbaugh suggested scheduling an informal, small group discussion with UIC and Lurie's on current processes for each disorder.

VI. New Business. No new business.

VII. Reports from Partners

A. Illinois Society of Genetic Professionals. No report.

B. Division of Specialized Care for Children (DSCC).

Ebonie Zielinski reported that DSCC received 648 genetic and 567 hearing referrals in FY24. The next grant cycle will focus on updating DSCC's resources, supports, and tip sheets to make the process easier. They continue to have education series about maximizing health insurance benefits. The next training topic (on May 13 at 6 p.m.) will be about available division of developmental disabilities waivers. Reference the DSCC website for more information. The Family Advisory Committee has started a transportation subcommittee to investigate transportation issues experienced by families. Based on these findings, the subcommittee makes recommendations to HFS to improve these services. The Youth Advisory Council is coming together to develop educational social media messaging targeting the youth perspective in transitioning to adult health care services.

C. Perinatal Advisory Committee. No report.

D. Sickle Cell Disease Association of Illinois (SCDAI).

TaLana Hughes is the executive director for SCDAI and has a 22-year-old daughter born with sickle cell disease. SCDAI was awarded funding to participate in the new, Sickle Cell Prevention, Care, and Treatment program to increase access and awareness for individuals and their families affected by sickle cell disease in Illinois. SCDAI hosted an annual, Sickle Cell Advocacy Day (in Springfield) and plan to continue this event every November. TaLana is very encouraged about the new gene therapies and curative options for individuals with sickle cell disease. This year, the national organization, Sickle Cell Disease Association of America (SCDAA) is hosting their annual, sickle cell convention in Rosemont, IL.

E. Norton & Elaine Sarnoff Center for Jewish Genetics. No report.

F. Illinois Rare Disease Commission.

Maria Pollock is chair of the Illinois Rare Disease Commission over the last two years. Maria expressed the challenges in accessing services for rare disorders thinks our committees can work together to make a difference. Maria invited everyone to the next RDC monthly meeting on Wednesday.

VIII. Next Meeting Date. TBD (Virtual, In-person, or Hybrid).

IX. Public Comments. No public comments.

X. Adjourn. Joan Ehrhardt closed meeting at 12:44 p.m.