

Minutes

Universal Newborn Screening Advisory Committee
(a.k.a Genetic and Metabolic Diseases Advisory Committee)
December 1, 2021 VIRTUAL

Attendance

Member Name	Specialty	Affiliation	Present
Ahmed, Aisha	Immunologist	Lurie	-
Akhter, Javeed	Pediatric Pulmonologist & Immunologist	Advocate Christ	X
Balian, Carrie	Parent – EHDl	Chicago area	X
Burton, Barbara	Geneticist (Chair)	Lurie	X
Cailteux, Glynis	LHD Representative	Kankakee Health Dept.	X
Dineen, Rich	Genetic Counselor	Rush	X
Fleischer, Julie	Geneticist	SIU	-
Hitchins, Lauren	Advance Practice Nurse	Lurie	X
Hoganson, George	Geneticist	UIC	-
Hsu, Lewis	Pediatric Hematologist	UIC	X (partial)
John, Ashish	Pediatrician	Memorial Medical Ctr.	X
Khan, Janine	Neonatologist	Lurie	-
Katz, Rachel	Social Worker	Lurie	X
Kreiter, Mary	Pediatric Endocrinologist	Lurie	X
Leeth, Beth	Genetic Counselor	NMH	X
O'Brien, Shannon	Nutritionist	UIC	-
Paquette, Erin	Hospital Ethicist	Lurie	X
Stoner, Robin	Audiologist	Rush	X
Yoshii, Akira	Pediatric Neurologist	UIC	X (partial)
VACANT	Parent		-

UNSAC applicants, ex officio members, and partners: Kasie Able (Parent), Joshua Baker (Geneticist), Keondra Harris (Local Public Health), Beatriz Menendez (Genetic Counselor); TaLana Hughes (Sickle Cell Disease Association of Illinois), Tess Rhodes (UIC Division of Specialized Care for Children), Jason Rothstein (Norton And Elaine Sarnoff Center for Jewish Genetics)

IDPH Staff: Laura Ashbaugh, Rebecca Barnett, Khaja Basheeruddin, Leslie Chapman, Hector E. Diaz, Vineet Dhiman, Joan Ehrhardt, Pearlie Gardley, Shannon Harrison, Crystal Jordan, Chrissy Knepler, Karen Mancera-Cuevas (at 10:41), Ginger Mullin, Mohammad Nasir, Yin Peng, Joel Price, Charles Williams, Ashley Mochizuki (Genetic Counseling Student Intern)

Public: TB [presumed Twila Brase] of [Citizen's Council for Health Freedom](#) (MN)

Welcome: UNSAC attendees were welcomed and the meeting began at 9:06. Meeting housekeeping items were reviewed.

Approval of May 2021 Meeting Minutes: No corrections, comments, or objections were voiced. Minutes were approved.

UNSAC Member Status: The status of members with terms ending in 2021 was reviewed, and members were acknowledged for their service. Dr. Burton confirmed that she will be stepping down after this meeting and not serving another term on the committee. She will continue as interim Chair until the spring 2022 meeting, when a new Chair will be elected. Dr. Burton shared her thoughts as outgoing Chair of the UNSAC. A packet with newborn screening (NBS) data used to be handed out at in-person meetings, and she would like this to be provided to UNSAC members as a routine. Another item to be addressed has to do with clinical follow-up data that is provided to the NBS follow-up program related to NBS clinical diagnosis and long term follow-up data. No output is seen from the long term follow-up data by IDPH NBS Program. Dr. Burton described the general duties of the Chair, in cooperation with IDPH NBS Program. This includes, for example, occasional requests from the NBS Program to address questions and concerns of practitioners about newborn screening.

Joan thanked all members whose terms are expiring early in 2022 and noted prospective members in attendance who have applications pending the IDPH Director's signature. Carrie Balian indicated that she would like to serve another term.

IDPH Reports

Newborn Screening Laboratory

NBS Diagnosed Conditions (Data) = Joan discussed provisional data listed for Follow-Up Program. This was displayed visually during the webinar, and not distributed as a handout. Dr Burton requested a copy of data. Joan stated that NBS Program needs to compare the data system (laboratory information management system (LIMS)) output to clinical follow-up summaries to verify diagnostic status and case numbers. Ginger Mullin discussed the drop in birth rate, which has dropped to 130,000 births/year. The dropping birth rate would be expected to account for some drop in case numbers. Dr. Burton requested that data be shared with the committee members "when cleaned up."

Krabbe Algorithm – Joan stated that the updates to the Krabbe newborn screening algorithm were presented at an earlier meeting. There were no questions

Newborn Screening Follow-Up Program

Staffing –Crystal shared that the NBS Follow-up Program currently has 2 vacancies for blood spot: one Office Specialist (OS) and one Nurse (RN1) position; there are 23 staff currently. At this time Program is preparing to fill both positions, as well as hiring an NBS Data Coordinating. Awaiting HR approval.

Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) - Joan gave an overview of the federal committee, which is administered by Health Resources and Services Administration (HRSA) and makes recommendations for all state NBS programs. Illinois screens for all conditions on the recommended uniform screening panel (RUSP), as well as a few others not on the RUSP. The ACHDNC recently discussed challenges the committee is currently facing, as well as those of the newborn screening community. The advances in technology are increasing both potential NBS methodologies and novel clinical treatments. This is resulting in a "bottleneck" of conditions to be considered for RUSP inclusion at the federal level. Plus, increasing pressure on state NBS systems to expand, with variable resources at the state level.

FY22 Grant Programs - Rebecca provided an overview of the grant programs' history, challenges, and revisions to the current FY22-FY24 cycle. We had a competitive applications year for FY22. There are 15 awards to support genetic counseling clinic services (1.7 million award pool), 9 Sickle Cell Disease (SCD) Follow-up awards totaling \$500,00, and 6 awards to local public health entities for Genetic Education and Follow-up (1.05 million awarded of 2.794 million pool). Rebecca discussed some of the challenges which had been encountered by grantees, including changes to the genetics program staffing and administration. Some of the health departments did not have patients referred for genetics consult. This year our program developed a regional collaborative group (Illinois Regional Genetic Services Workgroup (IRGSW)) and allocated funding to regional leads to participate in updating the program. IDPH did not get a huge response from local public health entities. Prospective genetic counseling and sickle cell centers could apply for different grant components. Grants started July 1, 2021.

In addition to the NBS-funded grant programs, there are 2 programs which are supported by general revenue funds: *Preventing Sleep-Related Infant Deaths* awarded to SIDS of IL, and Comprehensive Sickle Cell Clinical Care Program at UIC. Questions were asked and answered regarding the grants programs.

Administrative Rules – Joan shared that the draft revised NBS Administrative Rules were reviewed and approved by the State Board of Health without revision or comment. They are currently pending review by Joint Committee on Administrative Rules. Dr. Burton asked if the requested language providing for release of dried blood spot NBS sample to providers is included. Joan confirmed that it is.

Early Hearing Detection and Intervention Program

Ginger Mullin provided programmatic updates and review of EHDI screening and referral data. The EHDI program has identified covid-related gaps and delays in screening, referral, and evaluation. The program is working to address these gaps. Carrie Balian provided an update on family support activities. The committee thanked Ginger, Carrie and the rest of the EHDI program and partners for all their hard work.

Other Reports

Newborn Screening and Laboratory Subcommittee – George Hoganson, subcommittee chair, was not present today. In his absence, Chrissy gave a brief overview of the subcommittee's last meeting's topics:

- Krabbe NBS algorithm changes
- Vendor issues with PerkinElmer LIMS changes
- 30-day unsatisfactory specimen (UNSAT)/corrections protocol changes
- Cystic fibrosis (CF) molecular screening automatic if meconium ileus marked on card
- Staffing changes
- Spinal muscular atrophy (SMA) and adrenoleukodystrophy (ALD) updates

Newborn Screening Hemoglobinopathy Collaborative - Shannon shared that this group continues to be active, typically meeting quarterly and promoting networking and collaboration among clinical specialists, support groups, and IDPH NBS.

Reports from Partners

Norton & Elaine Sarnoff Center for Jewish Genetics – Jason – (brief) discussed their organization's education and subsidized screening ~300 per year, about 1/3 = carrier of gene variant for disorder on the panel; offered decoding genetic testing webinars – live w/genetic counselor (JUF.org)

Sickle Cell Disease Association of Illinois – Talana (brief) described the legislative activities: sitting, meeting, educating re: appropriations, needs for assistance, 504 and IEP connections, comprehensive in-out of care. Due to covid concerns, SCDAI held curbside celebration this year. SCDAI provides navigation for adults, among their other services.

Illinois Society of Genetic Professionals - none

Sudden Infant Death Services of Illinois, Inc. - none

Division of Specialized Care for Children – Tess (RN) discussed her experience of 26yr+ at DSCC and DSCC services; answering requests for in-home support, telehealth and tele-therapy; assistance with transportation problems. DSCC – MCO cooperation. If a need is not being met by MCO, family may approach DSCC to enroll for coverage of needed service. Rich/Tess discussed suggestions regarding links on home page to regional offices. Child/family connections. Tess briefly provided an update on the medical food coverage program. Getting MCO connections established has been a challenge. LilsDietary – more grocery than DME, for example.

Other New Business – No new business was introduced.

Next Meeting Date

Date TBA – VIRTUAL Spring 2022 meeting. UNSAC will plan for a virtual spring meeting due to the current Covid19 surges and projected increased risk in transmission and rise in cases due to the recently recognized omicron variant. IDPH will continue to monitor the situation, in hopes that the Fall meeting may be in person. Joan will send out a poll early in the new year with the goal of scheduling both meetings for 2022.

Emergency contact information - Rebecca stated the emergency forms must be completed for all in-person meetings due to travel. There is less of a need with virtual meetings.

Public Comments – Dr. Burton asked about the status of GAMT legislation previously introduced in Illinois. Joan stated it looks like it died in committee; she has not seen any new initiative for it. Dr. Akhter stated he requested severe combined immune deficiency SCID NBS data from IDPH since the cutoff was changed in June 2020. Shannon Harrison provided him with this SCID data regarding the cutoff change and screening change that occurred when the SCID assay was multiplexed with the SMA assay.

Meeting Adjourned at 11:26 A.M.