

**Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee (GMDAC)
Minutes – May 24, 2018**

Holiday Inn Chicago SW – Countryside Conference Center
Countryside, IL

Members Present:

Barbara Burton, M.D., Genetics, Lurie Children’s Hospital
Timothy Geleske, M.D., Pediatrician, Private Practice
Adrienne Savant, M.D., Pediatric Pulmonologist, Lurie Children’s Hospital
Michael Msall, M.D., Developmental Pediatrician, University of Chicago
Lewis Hsu, M.D., Pediatric Hematologist, University of Illinois at Chicago
Jennifer Burton, M.S., Genetics Counselor, University of Illinois at Peoria
Rachel Katz, L.S.W., Lurie Children’s Hospital
Mary Kreiter, M.D., Pediatric Endocrinologist, Lurie Children’s Hospital
Amy Walsh, Parent of child with SCID
Cindy Mitchell, R.N., Perinatal Administrator, St. John’s Hospital
Erin Paquette, M.D., J.D., Ethicist, Lurie Children’s Hospital

Other Attendees:

George Hoganson, M.D., Geneticist, University of Illinois at Chicago
Shannon O’Brien, R.D., Metabolic Dietician, University of Illinois Medical Center
Beth Leeth, M.S., Genetic Counselor, Lurie Children’s Hospital
Tess Rhodes, R.N., Division of Specialized Care for Children
Jason Rothstein, M.P.H., Center for Jewish Genetics
Elisa Seeger, Parent of child with X-ALD
Sydney Pantaleon, Parent of child with X-ALD
Becky Kinnett and Daughter, Parent of child with X-ALD
Josh Mateffy, Parent of child with Zellweger Syndrome
Lauren Hitchens, Genetics Nurse Practitioner – Lurie Children’s Hospital
Erika Vucko, Genetics Nurse Practitioner – Lurie Children’s Hospital
Talana Hughes – Sickle Cell Disease Association of Illinois
Nancy Maruyama – SIDS of Illinois

IDPH Representatives:

Matt Charles, Chief Division of Laboratories
Nancy Giurato, Chief Newborn Screening Lab
Joel Price, Molecular Lab Supervisor (CF and SCID)
Jennifer Crew, Molecular Test Development for SCID, Liaison with Perkin Elmer
Khaja Basheerrudin, Lab Supervisor MS/MS and LSD
Claudia Nash, Newborn Screening Program Manager
Shannon Harrison RN, Newborn Screen Follow-Up Nursing Supervisor
Laura Ashbaugh RN, Newborn Screen Follow-Up Nursing Supervisor
Ginger Mullin, Newborn Hearing Screen Program Coordinator
Rebecca Barnett, Grants Coordinator

Welcome and Introductions:

Meeting was called to order at 10:36 A.M., and Claudia Nash welcomed attendees. Attendees introductions were made. Ms. Nash stated she has retired from IDPH, but she is working on contract

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through July 2018. In the absence of a committee chairperson, Claudia Nash reviewed terms of current members. Claudia stated Dr. Msall's term has expired and asked individuals whose term is expiring to continue to serve until their position is filled. Member positions currently pending approval are Dr. Julie Fleischer, Dr. George Hoganson, Shannon O'Brien, R.D., Beth Leeth, M.S., Dr. Melanie Makhija and a pediatric neurologist yet to be named.

Review of Bylaws, Committee Members, Terms and Vacancies:

The committee bylaws were distributed for review. Claudia Nash proposed overview of the following for consideration: adding additional member to reflect EHDI as part of this committee, address frequency of meetings (two meetings per year with one being by teleconference), length of membership, review of current subcommittees, and change the name of this committee to reflect addition of hearing loss. Suggestion was made to change name to "Newborn Screening." Further proposed changes or comments may be emailed to Claudia Nash by mid-June. Proposed changes will be reviewed and voted on at the next meeting.

Election of Chair:

Claudia Nash asked if any current members were interested in the committee chairperson role. Claudia provided an overview of the chair duties including agenda topics and facilitate meeting and meeting representatives. Dr. Barbara Burton and Dr. Erin Paquette expressed interest. Committee members anonymously voted and written votes were tabulated. Dr. Barbara Burton was elected as committee chair. Meeting called to order by Dr. Barbara Burton.

Approval of Minutes- July 21, 2016 Meeting:

Talana Hughes from SCDAI stated that camp name was incorrect. Name should be changed to Camp North Star Reach. Meeting minutes approved.

Overview of IDPH Programs- Power Point Presentation:

Power Point presentation by IDPH representatives on Newborn Screening in Illinois. Presentation provided overview of NBS Laboratory and Follow-up Team, Early Hearing Detection and Intervention Act, Birth Rate/Volume of specimens, Perinatal Network, Statewide Grants/Medical Specialists, Quality Improvement Collaborative, Newborn Screening Process, State and National Collaboration Partners, NewSTEPS APHL site visit and future challenges implementing new disorders. Discussion by group regarding the reporting newborn screen results for specimen of baby < 24 hours of age. It was suggested that the program only report results that are valid prior to 24 hours of age such as hemoglobin, galactosemia and biotinidase.

IDPH Reports:

Newborn Screening Follow-Up Program

Claudia Nash reported that two additional nurses were hired for the follow-up section, and one will work on Saturdays. The follow-up team is experiencing frequent staff turnover as staff seek promotions and wage increases. They currently have 5 staff vacancies. The follow-up team is continuing to work on electronically linking the newborn screening data system with the Illinois Vital Records System which is where hospitals enter the birth certificate data. They are also working with UPS to set up Saturday delivery of NBS cards to the IDPH laboratory.

Newborn Screening Laboratory

Matt Charles reported the IDPH Chicago laboratory suffered a flood in January 2018 due to frozen pipes causing \$2 million damage and requiring major remodeling and updating of the facility. The incident occurred on a Sunday, but no instruments or timelines of testing were compromised during this incident.

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The laboratory is working to update their LIMS system and are awaiting FDA approval of Perkin Elmer NeoBase 2 kit which includes screening for adrenoleukodystrophy (ALD). Jennifer Crew reported she has received two whole genome instruments and is working on SCID NextGen sequencing. Jennifer reported this is part of a multi-state collaborative headed by New York state. The laboratory stated they are also working with Perkin Elmer on a continuity of operations plan in case of an actual building destruction or a natural disaster.

Early Hearing Detection and Intervention Program (EHDI)

Ginger Mullin reported on the progress of the EHDI program. The EHDI program continues to work toward screening no later than 1 month of age, diagnosis no later than 3 months of age and intervention no later than 6 months of age. Illinois birth rate is decreasing while the number of infants diagnosed with a congenital hearing loss increase to a rate of 2.0/1000. A focus of the program is to address timeliness of services and reporting. The program has reached the lowest loss to follow-up (LTF) rate since the beginning of the program. The current LTF for birth cohort 2016 is 35% utilizing the Centers for Disease Control and Prevention metrics. A primary goal for the program over the next year is to integrate with the Illinois Vital Records data system.

Other Reports:

Newborn Screening and Laboratory Subcommittee

There has been no recent meetings or activity. Dr. George Hoganson expressed interest in reviewing the APHL site visit report and suggested the group meet after to discuss the report. Laboratory staff will notify subcommittee members to schedule a meeting.

Lysosomal Storage Diseases Subcommittee

Claudia Nash reported the subcommittee has been meeting regularly by teleconference. Krabbe and MPSII screening has been successfully implemented and follow up is going smoothly.

Newborn Screening Cystic Fibrosis Collaborative

Claudia Nash reported CF collaborative started in 2008, and Dr. Savant is now leading the collaborative. This group has not met for several months as no crucial issues have arose. The CF listserv remains open for members to exchange information and ask questions. The collaborative can schedule a meeting if there are issues to discuss.

Newborn Screening Hemoglobinopathy Collaborative

Shannon Harrison reported the Hemoglobinopathy Collaborative continues to meet every other month by teleconference. There are currently 11 hematology centers as well as 2 centers in St Louis on the calls along with SCDAI and STORM. The collaborative discusses QI projects, webinars, events, and issues with referrals, medication coverage, etc. A pharmacist with the state of Illinois has also attended calls to discuss prescriptions for Medicaid and managed care patients.

Severe Combined Immune Deficiency (SCID) Newborn Screening Collaborative

Shannon Harrison reported the SCID collaborative consists of IDPH laboratory, IDPH follow-up staff and the five immunology centers in Chicago and St Louis. In the past, the NBS Program has reviewed cut off values, screening results, and diagnoses. The collaborative has not met in a year due to chairperson being on sabbatical and no issues with SCID screening or follow-up.

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Newborn Screening Expansion Subcommittee – There has been no recent activity.

Reports from Partners:

Center for Jewish Genetics

Jason Rothstein reported the Center for Jewish Genetics, now known as Norton & Elaine Sarnoff Center for Jewish Genetics, is an educational organization to educate Jewish population regarding hereditary and genetic disorders. The center facilitates referrals for 300 patients each year. Their screening program is a comprehensive screening panel of 200 disorders. They have hired a full-time community health educator who is planning to do a community needs assessment in the next two years. The center is hosting a presentation “What’s Jewish about BRCA?” on June 21, 2018 in Skokie.

Sickle Cell Disease Association of Illinois (SCDAI)

Talana Hughes reported SCDAI receiving HRSA funding for sickle cell disease education, support and follow-up for disease and trait. She stated SCDAI is assisting patients with “Linked In” at hematology centers to improve access to care, support services, assistance with medical providers, and transition to adult care.

Genetics Task Force of Illinois

No official report was given, but Beth Leeth stated the organization is changing their name to Illinois Society of Genetic Professionals.

Sudden Infant Death Services of Illinois, Inc.

Nancy Maruyama reported IDPH provides grant funding to this organization for safe sleep education and bereavement services for deaths on children from birth to age one. She stated cause of death is now referred to as “death by unsafe sleep situations” not SIDS. As part of NICHQ, SIDS conducted a survey on Safe Sleep practices from birth to age one year. The survey was given to birthing hospitals with survey questions such as “What are your sleep policies?” and “Do you have a sleep policy?” They are working with UIC epidemiology to interpret data. The Illinois SIDS rate 6 per 1000 with 50% of deaths in Cook County. SIDS of Illinois formed in 1968 and is celebrating 50 years.

Division of Specialized Care for Children

Tess Rhodes from the Division of Specialized Care for Children (DSCC) reported that DSCC provides care for children age’s birth to 21 years old and covers diagnostic testing for many disorders included in the newborn screening panel not covered by insurance. The rules have recently been changed and more medical categories are now being covered. The blood disorder category will now cover sickle cell disease services. The pulmonary disease category now includes cystic fibrosis but does exclude asthma. A gastrointestinal disease category has been added. Unless specifically listed as an exclusion, most conditions should be covered. Also, the federal poverty level was increased from \$285 to \$325. The cap will probably decrease as most have primary insurance and no deductible or co-pay. The new rules are effective on July 1, 2018. They will go back 90 days to cover lab work in preparation for appointment and will extend diagnoses up to 15 months. A diagnostic form with the new changes should be available in July.

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Public Comments:

There were four parents in attendance at the meeting. Elise Seeger from the Ayden Seeger Foundation is a mother of son who had a late diagnosis of X-ALD. The foundation started to focus on universal Newborn Screening for X-ALD. Ms. Seeger stated Illinois ALD law passed in 2015 and requested an update on the status of ALD screening implementation. Synde Pantaleon, mother of 2 ½ year old diagnosed with ALD, expressed concerns over lack of knowledge among pediatricians. Becky Kinnett son's Blake was diagnosed with ALD at age 15 and expressed frustration on why Illinois has not implemented screening yet. Josh Mateffy stated his daughter was diagnosed at 3 months of age with Zellweger's syndrome. If Illinois tested for ALD, he believes her disorder would have been found on newborn screening.

Other New Business:

No other new business discussed. Dr. Jen Burton inquired about progress of the eReports portal allowing physicians to access newborn screen report electronically. The IDPH legal team stated that new rules need to be in place for physicians to access NBS electronically.

Next Meeting Date:

The meeting was adjourned at 2:00 PM. The date for the next meeting was not established, but a late fall 2018 meeting via videoconference was proposed.