Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee (GMDAC)
Minutes July 21, 2016
University of Illinois at Springfield, Springfield, IL

Members Present:
Celia Anguiano, Parent
Jennifer Burton, M.S., University of Illinois at Peoria
Ramsay Fuleihan, M.D., Lurie Children's Hospital
Janine Khan, M.D., Lurie Children's Hospital
Cindy Mitchell, R.N., St. John's Hospital
Lainie Friedman Ross, M.D., PhD., University of Chicago
Adrienne Savant, M.D., Lurie Children's Hospital
Cathy Wicklund, M.S. Northwestern University

Members Not Present:
Barbara Burton, M.D., Lurie Children's Hospital
Glynis Caliteux, R.N., Kankakee County Health Department
Timothy Geleske, M.D., Private Practice
George Hoganson, M.D., University of Illinois at Chicago
Rachel Katz, L.S.W., Lurie Children's Hospital
Mary Kreiter, M.D., Lurie Children's Hospital
Michael Msall, M.D., University of Chicago
Alexis Thompson, M.D., Lurie Children's Hospital
Amy Walsh, Parent
Steven White, Office of the Cook County Medical Examiner

Other Attendees:
Deb Boylan, R.N., Cardinal Glennon Children's Hospital
Sibyl Cox, R.D., SIU School of Medicine
Brook Croke, M.S., Genetic Counselor
Julie Fleischer, M.D., SIU School of Medicine
Christie Hoell, M.S., Genetic Task Force of Illinois
Lewis Hsu, M.D., University of Illinois at Chicago
Tess Rhodes, R.N., Division of Specialized Care for Children
Jason Rothstein, M.P.H., Center for Jewish Genetics
Brad Tinkle, M.D., Advocate Medical Group

IDPH Representatives:
Matt Charles, Chief Division of Laboratories
David Culp, Ph.D., Deputy Director
Conny Moody, Deputy Director
Hector Diaz, Newborn Screening Laboratory Staff
Genetics/Newborn Screening Program
Jean Becker, Nurse Consultant
Shannon Harrison, Nurse Consultant
Ginger Mullin, Newborn Hearing Screening Program Coordinator
Claudia Nash, Program Manager

IDPH Representatives (continued):

Bianca Sanchez, Graduate Student Intern
Heather Shryock, Data Manager
Nikki Woolverton, Grants Manager

Introductions and Announcement of New Members

In the absence of a committee chairperson, Dr. Lainie Friedman Ross graciously agreed to chair this meeting. The meeting was called to order at 10:40 a.m. with introductions of three new committee members in attendance, five new members not in attendance and two nominees in attendance. Due to delays in new appointments, the committee has not met in 21 months. Since a quorum was not present at this meeting, no action could be taken to nominate or elect a new chairperson, or approve minutes from the previous meeting. The committee bylaws were distributed for review. Claudia Nash asked the members to consider the existing standing committees and whether additional committees should be added. Once a chair is elected, subcommittee chairs will need to be appointed and subcommittee members reinstated or selected.

Subcommittee Reports

Newborn Screening and Laboratory Subcommittee- no recent activity

Lysosomal Storage Diseases Subcommittee

Jean Becker provided a handout from the most recent teleconference meeting held May 20, 2016. A subsequent face-to-face meeting was also held at Lurie Children's Hospital June 2, 2016. Data from November 2014-May 2016 were reviewed, which indicate nearly 190,000 specimens have been screened for five LSDs (Fabry, Gaucher, Niemann-Pick, MPS I and Pompe), 589 had abnormal findings and 32 cases were diagnosed. Regarding lysosomal storage disease screening , the IDPH laboratory will implement a new assay for IDUA using lactone as an inhibitor of beta-glucuronidase which has iduronidase-like activity, which should reduce the false positive rate for MPS1. Lab staff are reviewing data on GLA levels in low birth weight and term infants as a function of chronologic age, and have seen that low birth weight infants have higher GLA levels than term infants although these gradually return to levels comparable to term infants by about one month of age. Screening for Krabbe has not yet begun due to delays in securing a contract with a vendor to perform molecular testing. Preliminary testing for MPS II has been conducted at the IDPH laboratory, and the next step will be to complete a full scale validation study, prior to statewide implementation of screening. Staff from Cardinal Glennon Children's Hospital in St. Louis reported that Missouri has been screening for Krabbe, Fabry, Gaucher and MPS II and have also received some referrals from Illinois.

Newborn Screening Cystic Fibrosis Quality Improvement Collaborative

Dr. Adrienne Savant reported that the last teleconference meeting of this committee was held May 9, 2016. Overall, the two quality improvement measures in Illinois have exceeded national standards with the sweat test quantity not sufficient rate being 3.3% (standard <10%) and percentage of families provided genetic counseling at 99% (standard>90%). The CF molecular test kit (Hologic) currently in use at the IDPH newborn screening laboratory has been recalled and the lab will begin using a kit with less mutations (23 instead of 47) due to delays in procurement of an expanded panel. Use of the ACMG 23 reduced mutation panel will likely result in a failure to identify cases more common in the most vulnerable minority populations, (mutations more common in Hispanic infants) in Illinois.

Newborn Screening Hemoglobinopathy Collaborative
Dr. Lewis Hsu reported on the most recent teleconference meeting of this collaborative, held May 25, 2016, and meeting minutes were distributed to the group. IDPH staff and collaborative members have worked with a pharmacist from the Illinois Department of Healthcare and Family Services regarding prescription drug coverage for hydroxyurea and to require pre-authorizations be completed only on an annual basis for chronic diseases.

Severe Combined Immune Deficiency Collaborative
Dr. Ramsay Fuleihan reported the severe combined immune deficiency (SCID) collaborative, which is comprised of 5 referral centers, has been meeting regularly every three months, and most recently met April 25, 2016. Dr. Fuleihan reported that 11 cases of SCID have been detected, 20 DiGeorge, 4 with trisomy 21 and 11 newborns with idiopathic T-call lymphopenia, which usually resolves. He also reported that the TREC cutoff has been lowered to 250 TREC copies/ul, which has been helpful in reducing the number of false positive screening reports.

Newborn Screening Expansion Subcommittee-no recent activity

Newborn Metabolic Screening and Treatment Code Changes
Deputy Director Conny Moody stated that legislation in 2015 resulted in an increase in the newborn screening fee to $118, and the addition of screening for X-ALD (adrenoleukodystrophy) to the Illinois panel, once the necessary prerequisites are in place. Revisions to the Newborn Metabolic Screening and Treatment Administrative Code are being prepared that will address the addition of critical congenital heart disease to the screening panel, and matters related to access to test results, sharing and access of residual samples and specimen storage and retention and changes regarding specimen collection for newborns in the NICU.

IDPH Report
Newborn Screening Laboratory
The issue of timeliness of testing was reviewed and Matt Charles indicated that the average testing turnaround time is seven days in the lab. The long range plan includes trying to expand working hours to six days on the weekend, to achieve the national timeliness goals. Matt stated the lab has implemented GSP (Genetic Screening Processor by Perkin Elmer), a new technology for endocrine, galactosemia and biotinidase testing, which has decreased testing turn-around time and is more automated than previous technology. He also stated that implementation of a method using non-derivatized sample preparation for the mass spectrometers, Neobase, will be implemented in the spring of 2017 which will be more efficient. Perkin Elmer plans to include testing for X-ALD in subsequent versions of this assay.

Newborn Screening Follow-Up Program
Claudia Nash reviewed the hospital submission data included in the packet, which indicate over 98% of specimens are received by the IDPH lab within three days of collection, since IDPH provides overnight UPS delivery. IDPH does monitor timeliness data on a quarterly basis according to the criteria defined by the Association of Public Health Laboratories NewSTEPs program. These milestones could be improved if the NBS lab were operational Saturday. Plans are proceeding to implement eReports, a system for electronic access to newborn screening results for physicians within

the next calendar year. IDPH is also finalizing development of a module that would import newborn/maternal demographics from the birth record into the newborn screening data system. This

would eliminate the need for hand entry of data on the blood collection cards by hospital staff and data entry by IDPH staff, and would result is more complete and accurate demographic data. Claudia also reported that the IDPH staff coordinate four "collaboratives" of medical subspecialists that meet regularly by teleconference to improve services to newborns and families: Hematology, Immunology (SCID), Lysosomal Storage Disorders and Cystic Fibrosis. Staff from the IDPH Follow-up Program regularly participate in regional and national work groups , including the Region 4 Genetics Collaborative and meeting sponsored by NewSTEPs and the Newborn Screening Translational Research Network. IDPH follow-up staff are also participating in a research study conducted by Emory University regarding outcomes of children with Duarte galacotosemia based on treatment differences.

Early Hearing Detection and Intervention Program
Information was included in the packet regarding the newborn hearing screening program. This program was integrated into the newborn metabolic screening program two years ago and is partially funded through a grant from the Centers for Disease Control and Prevention. Numerous quality improvement initiatives are being conducted by program staff.

Genetics Program Grantees
Nikki Woolverton provided a summary of the FY17 grant awards for the three types of grants IDPH distributes for each year for services, outreach and education. Grantees include 18 genetics centers, 8 pediatric hematology centers and 33 local health departments. Funding amounts will remain at FY16 levels. The Grant Accountability and Transparency Act is being implemented which aligns with federal requirements for state grants.

SIDS Program
Nikki Woolverton provided SIDS and other Sleep-Related Infant Death Data for 2014, the most current year for which data is available for. The number of SIDS deaths is decreasing while Undetermined deaths are increasing. The Cook County Medical Examiner is no longer using SIDS as a cause of death. This trend is occurring nationwide. The group was interested in learning more about the classification of infant deaths, and it was suggested Dr. Steven White with the Cook County Medical Examiner's office provide more information at the next meeting.

Reports from Partners:
Sickle Cell Disease Association of Illinois (SCDAI)
Staff from SCDAI were not present to report on their activities. Shannon Harrison did indicate that SCDAI is working closely with various hematologists to assist patients with getting enrolled in "Get Connected" through the Sickle Cell Disease Association of Illinois, which will provide them with more resources, and that SCDAI will be assisting numerous children attend camp SOAR in Michigan this summer. Dr. Hsu indicated that the Paul Newman foundation assists with transportation costs for families.

Center for Jewish Genetics

Jason Rothstein is the Director of the Center and as such will be replacing Karen Litwack as their ex-officio member on this committee, since Karen will be retiring. Jason provided an overview of the Center's DNA Day activities this past April. For the past several years the Center has received DNA Day funding from IDPH to promote the importance of knowing your family health history throughout the state by providing information to local public health agencies through printed materials and a webinar. This year's focus was on men's health, and various written materials were updated, including a brochure on newborn screening and on family health history and a new brochure was developed regarding men's health issues. The Family Health Record Keeper was also translated into Spanish, and made available to local public health agencies. Jason indicated that the Center looks forward to continuing the relationship with IDPH and continuing the DNA Day activities.

Genetic Task Force of Illinois (GTFI)

Christie Hoell, the President of the Genetic Task Force, reported that the Task Force is comprised of over 95 members and the next meeting will be held September 14 at the University of Chicago. GTFI conducts an annual educational symposium that is supported by IDPH. The next symposium will be held February 3, 2017 and will include current topics in clinical genetics. Other endeavors with which GTFI is involved, include a new one day workshop for prospective genetic counseling students that will be conducted this summer, and collaborative efforts with advocacy groups to develop educational materials on Down syndrome for families that can be provided by birth hospitals.

Sudden Infant Death Services (SIDS) of Illinois, Inc.

Staff from SIDS of IL were not present to report on their activities. Nikki Woolverton reported that historically IDPH has provided grant funding to this organization to support their bereavement support and educational efforts. For FY 16 this funding was not appropriated and it is not clear if funding will be appropriated for FY 17. IDPH continues to collaborate with this organization in many efforts.

Division of Specialized Care for Children (DSCC)

Tess Rhodes reported that DSCC has developed new information material for health care providers regarding care coordination and distributed these materials. DSCC continues to approve medical subspecialists for the IDPH Newborn Screening Program and covers diagnostic testing for many disorders included in the newborn screening panel.

Adjournment:

The meeting was adjourned at 1:30 PM. The date for the next meeting was not established. It is likely that a late fall 2016 meeting will be held by videoconference.