# Illinois Department of Public Health
# Genetic and Metabolic Disease Advisory Committee (GMDAC)
Minutes—April 17, 2014
Hilton Garden Inn, Springfield, IL

**Members Present:**
Joel Charrow, M.D., Chair, Lurie Children's Hospital
Lainie Friedman Ross, M.D., PhD., University of Chicago
Ramsay Fuleihan, M.D., Lurie Children's Hospital
Timothy Geleske, M.D., Private Practice
George Hoganson, M.D., University of Illinois at Chicago
Alexis Thompson, M.D., Lurie Children's Hospital
W. Patrick Zeller, M.D., Private Practice
Jennifer Burton, M.S., University of Illinois at Peoria
Cindy Mitchell, R.N., St. John's Hospital
Sheila Chalmers-Currin, Parent
Amy Walsh, Parent

**Members Not Present**:
Karen Litwack, M.S.W., Center for Jewish Genetics
Susanna McColley, M.D., Lurie Children's Hospital
Michael Msall, M.D., University of Chicago
Mary Niewinski, R.D., University of Illinois at Chicago
Michael Schneider, M.D., Carle Foundation Hospital
Cathy Wicklund, M.S. Northwestern University

**Other Attendees:**
Tess Rhodes, R.N., Division of Specialized Care for Children
Brook Croke, M.S., University of Illinois College of Medicine at Peoria
Julie Fleisher, M.D., Washington University
Zina Berryhill, Parent
Emily Smith, Ph.D., Molecular Geneticist

**IDPH Representatives:**
David Culp, Ph.D., Deputy Director
Tom Schafer, Deputy Director
Tom Johnson, Chief Division of Laboratories
Newborn Screening Laboratory Staff
    George Dizikes, Ph.D., Chief Newborn Screening Laboratory
    Rong Shao, M.D.
    Khaja Basheeruddin, Ph.D
Elizabeth Paton, J.D., Legal Services

**Genetics/Newborn Screening Program:**
Claudia Nash, Program Administrator
Jean Becker and Shannon Harrison, Nurse Consultants
Heather Shryock, Data Manager

Nikki Woolverton, Grants Manager
Newborn Screening Staff: Denise Kink, Jennifer Holloway, Jessica Miller, Doug Hart, Derek Williamson, Belinda Szerletich, and Amber Lear
Graduate Student Interns: Nitika Sharma and Jesse Andrews

## Introductions and Announcement of New Members

The meeting was called to order at 10:10 a.m. with introductions.

## Review of Committee Member Appointment Terms

Claudia Nash announced the term for six members will expire in June. She reviewed the committee structure and reminded attendees that member appointments are for a three year term with eligibility for one reappointment.

## Term Expirations

Tom Schafer thanked the six committee members, whose term of appointment has expired, for their service and expertise with guiding the committee on behalf of the newborn screening program, the IDPH Director, and the state of Illinois.

## Approval of Minutes – April 17, 2014 Meeting

The minutes of the April 17, 2014 spring meeting were unanimously approved.

## Subcommittee Reports

### Newborn Screening and Laboratory Subcommittee

A copy of the NSLS January 29, 2014 meeting minutes was provided to members, and Dr. George Dizikes summarized the meeting for the group. Dr. Dizikes reviewed the Perkin Elmer database changes and developments since the previous meeting.

### Lysosomal Storage Diseases Subcommittee

Claudia Nash spoke on behalf of subcommittee chair Dr. Burton. Claudia reported the LSD committee has met to discuss preparations for testing. The committee has developed diagnostic protocols for each of the six LSD disorders, and each were reviewed and given final approval. In Missouri, positive LSD cases are shared and reviewed among the Missouri specialists. The LSD committee is interested in organizing a data collection approach similar to Missouri. After consulting with the IDPH legal staff, it has been advised that the sharing of records among Illinois specialists would require consent. The committee has drafted a consent form that will be utilized by specialists to obtain consent from families/guardians.

Claudia also addressed concerns regarding how the LSD specialist list was comprised. Claudia stated the administrative rules outlines the credentials medical specialists must have in order to be an IDPH consultant. A copy of the LSD list specialist list was also provided to members.

### Newborn Screening Cystic Fibrosis Collaborative

Jean Becker reported on behalf of Collaborative Chair Dr. McColley. Dr. McColley made a recommendation to the IDPH Laboratory to suppress two CFTR sequence variations, I148T and F508C, on the newborn screening panel for cystic fibrosis. These variations were well characterized and clearly are not disease causing so are no longer being reported.

The data as submitted by IDPH reflects an overall improvement by centers to be below the goal of <10% Quantity Not Sufficient rate for sweat tests performed. The statewide rate has remained <10% for the past two quarters. This improvement can be attributed to the implementation of the sweat test check list previously approved and recommended by this group.

Of concern are changes to the federal SMART Act (Save Medicaid Access and Resources Together). The State of Illinois is implementing the SMART Act which will impose limitations on prescription drugs for more than four prescriptions in a thirty day period unless prior approval is received for all prescriptions in excess of the four prescription limit. While advocates are trying to get the Act overturned, Dr. McColley suggested patients be proactive and request an override, as sixty percent of such requests are approved for adults. This is an annual process and if the number of medications change from the original exemption, the appeal must be submitted again. Antibiotics are excluded without prior authorization, but most CF children are on more than four medications.

## Newborn Screening Hemoglobinopathy Collaborative

Shannon Harrison reported the Hemoglobinopathy Collaborative has been meeting bi-monthly. Members have been reviewing data to determine why there is a large number of patients who are lost to follow up after diagnosis. The group has also reviewed long term follow up letters that are sent to medical specialists yearly, after a child is diagnosed, to ensure the information IDPH is collecting is relevant to the care of the child. The group has given guidance to IDPH for updating the treatment section of the letter to make it more useful.

## Newborn Screening Expansion Subcommittee

Claudia Nash reported no new activities or recent meetings for this group.

## Critical Congenital Heart Disease (CCHD) Work Group

Shannon Harrison reported since Public Act 98-0440 was signed by Governor Pat Quinn last August, which mandated Illinois birthing hospitals to screen all newborns for CCHD using pulse oximetry before hospital discharge, perinatal administrators have reported that all their respective hospitals are screening. Although there has been no new activity since the bill was signed and IDPH is still not collecting any screening data, the NBS follow up program is providing education and guidance to hospitals and medical practitioners. The NBS website has been updated to include the CCHD screening algorithm, the recommended screening guidelines and educational materials.

Cindy Mitchell, Perinatal Administrator for Springfield, IL, stated hospitals have asked where they should document the screening results. It was discussed that other states are collecting this screening data on the newborn screening card and in the birth certificate. Tom Schafer stated we are in discussions with the Illinois Vital Records System (IVRS) and their vendor to add birth related data to the electronic birth certificate. This project is expected to be added to the IVRS task list in the next year. IDPH has asked the IVRS vendor if there is a possibility to add CCHD information to this project.

## Newborn Metabolic Screening and Treatment Code Changes

Tom Schafer also stated information regarding CCHDis being included in the rules change to the Newborn Metabolic Screening and Treatment Code. These changes have been somewhat delayed due to other changes being made to the rules. Tom stated IDPH has added a clause stating a fee will be charged for releasing newborn metabolic screening results to parents/individuals. A provision was also added for IDPH to share information with other public health authorities after a parent's consent is given. Two draft consents are currently being reviewed by IDPH legal staff for releasing test results.

### Newborn Screening Release of Information

Shannon Harrison stated the Newborn Screening Program has developed two draft consents that are currently awaiting review by the IDPH legal staff. The first consent is for medical providers to request newborn screening results for patients under their care. The second consent is for an individual or parent who is requesting a newborn screening result for their own records. This second consent will require a notary public to ensure the identity of the person requesting the record. This consent will be required for individuals seeking newborn screening results to comply with the NCAA requirements for participation in college sports.

### Update: Implementation of Expanded Testing for Lysosomal Storage Disorders and Severe Combined Immunodeficiency

Dr. Dizikes reported the pilot for Severe Combined Immune Deficiency (SCID) began Tuesday April 8, 2014. Northwestern Memorial Hospital and Advocate Christ Hospital serve as the two hospitals participating in the pilot, with a combined rate of 150 specimens daily. In a month, Dr. Dizikes expects to implement statewide testing. The test examines DNA for a characteristic target that is present in maturing T-cells. Doctor Dizikes also reports Lysosomal Storage Disorders (LSD) are in the test validation phase. A database update will take place on May 18, 2014. Pilot testing is scheduled for May 19, 2014. After a month of testing, statewide screening will be implemented.

## <u>IDPH Report</u>

### Newborn Screening Laboratory

Tom Johnson reported the laboratory has experienced three procurement issues which they are addressing. The issues include securing contracts for newborn screening collection cards, reagents for cystic fibrosis mutation analysis reagents and reagents for sickle cell testing.

### Newborn Screening Follow-Up Program

Claudia Nash reported the newborn metabolic screening program is being reorganized and will be integrating the newborn hearing screening program into existing operations effective May 1, 2014. This integration of programs should provide improved customer service for hospitals and health care providers. Claudia also reported that due to the article written in the *Milwaukee Journal Sentinel* regarding the timeliness of newborn screening submission, there has been an increased awareness among Illinois hospitals regarding the importance of timely testing. IDPH has received an increased number of telephone calls from hospitals inquiring about their individual hospital reports, and has been posting the hospital quarterly reports on the IDPH newborn screening website.

### Genetics Program Grantees

For fiscal year 2015, Nikki Woolverton reported IDPH is providing funding to sixty-one agencies for services related to genetic counseling, sickle cell disease and local health departments for case follow-up and referrals. Two additional grants are direct appropriations from general revenue funds; the Comprehensive Sickle Cell Clinical Care Program through the University of Illinois at Chicago Sickle Cell Center and Project Safe Sleep Education and Outreach through Sudden Infant Death Services of Illinois, Inc. (SIDS of Illinois). This year IDPH has transitioned to an Electronic Grants Administration and Management System (EGrAMS) which will allow all applications and supporting documents to be available online.

### SIDS Program

Nikki Woolverton provided SIDS and Infant Mortality data for 2010, the most current year for which this information is available. The number of SIDS deaths had decreased, however accidental suffocation continues to be a significant cause of infant death. IDPH and SIDS of Illinois, Inc. provide education regarding infant safe sleep and SIDS risk reduction to health professionals and to communities. Several full day workshops have been held this past year for professionals who provide education on infant safe sleep and SIDS risk reduction strategies, as well as professionals who provide grief and bereavement services to families who have experienced an infant death. Strategies on communicating with families following an infant death and the most current American Academy of Pediatrics recommendations were reviewed.

### Educational Activities

Shannon Harrison reported IDPH is in the process of updating the newborn screening website to include information on CCHD, newborn hearing, LSDs and SCID. IDPH staff are also working on restructuring the entire website to make it easier to navigate through the various topics. Also, two education sheets on hypercholesterolemia and cardiomyopathies have been drafted and are awaiting revisions.

## Reports from Partners:

### Center for Jewish Genetics

Karen Litwack was not present to give a report, however, the 2014 DNA Day Report was provided to the group for review.

### Sickle Cell Disease Association of Illinois (SCDAI)

Staff from SCDAI were not present.

### Genetic Task Force of Illinois (GTFI)

Staff from GTFI were not present.

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

SIDS of Illinois, Inc., implements Project Safe Sleep, in which they work with local health departments and/or social workers in determining families who currently do not have a safe sleep environment for their infant. Through an application process, SIDS of Illinois, Inc. provides families with a portable crib. The project is going strong, however, the need for cribs is greater than the number available. Nancy Maruyama, RN, with SIDS of Illinois, Inc. is serving as co-chair for the Department of Human Services Collaborative Improvement & Innovation Network (COIIN) to reduce infant mortality. The COIIN Collaborative aims to reduce infant mortality and improve birth outcomes through best practices.

### Division of Specialized Care for Children (DSCC)

Tess Rhodes reported the DSCC is experiencing a transition from hard copy files to electronic case records. DSCC has a new website which is user friendly and is geared towards medical providers. It will allow providers to complete applications to become a DSCC provider and to locate forms for newborn metabolic and hearing screening. Tess stated Pompe disease will be the only LSD disorder covered by the DSCC since this disorder has cardiac involvement. Pompe is also the only disorder being recommended by the Discretionary Advisory Committee on Heritable Disorders in Newborns and Children to be added to the uniform screening panel.

## Adjournment:

The meeting was adjourned at 2:04 PM. The next meeting will be scheduled for September or October 2014.