

**Illinois Department of Public Health**  
**Genetic and Metabolic Disease Advisory Committee (GMDAC)**  
Minutes – October 31, 2013  
William Tell Holiday Inn- Countryside

**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  
Susanna McColley, M.D., Lurie Children's Hospital  
Mary Niewinski, R.D., University of Illinois at Chicago  
Jennifer Burton, M.S., University of Illinois at Peoria  
Cathy Wicklund, M.S., Northwestern University  
Timothy Geleske, M.D., Private Practice  
George Hoganson, M.D., University of Illinois at Chicago  
Karen Litwack, M.S.W., Center for Jewish Genetics  
Cindy Mitchell, R.N., St. John's Hospital  
Alexis Thompson, M.D., Lurie Children's Hospital  
Michael Msall, M.D., University of Chicago

**Members Not Present:**

Amy Walsh, Parent  
Michael Schneider, M.D., Carle Foundation Hospital  
Sheila Chalmers-Currin, Parent  
W. Patrick Zeller, M.D., Private Practice

**Other Attendees:**

Tess Rhodes, R.N., Division of Specialized Care for Children  
Annemarie Valdez, SIDS of Illinois  
Mike Petros, Dr. P.H. - Former IDPH Newborn Screening Laboratory Staff  
Parents: Zina and Randy Berryhill

**IDPH Representatives:**

David Culp, Ph.D., Deputy Director  
Tom Schafer, Deputy Director  
Tom Johnson, Chief Division of Laboratories  
Matt Charles, Division of Laboratories  
Patricia Kloppenburg, Division of Laboratories  
Newborn Screening Laboratory Staff  
    George Dizikes, Ph.D., Chief  
    Rong Shao, M.D.  
    Khaja Basheeruddin, Ph.D.  
    Jennifer Crew, Ph.D.

### **Genetics/Newborn Screening Follow-Up Program:**

Claudia Nash, Program Administrator  
Jean Becker, Nurse Consultant  
Heather Shryock, Data Manager  
Nikki Woolverton, Grants Manager  
Linda Robinson, Graduate Intern

### **Introductions**

The meeting was called to order at 10:15 a.m. and introductions were made for members and others present.

### **Review of Committee Member Appointment Terms**

Claudia Nash reviewed the committee structure and reminded attendees that member appointments are for a three year term with eligibility for one reappointment. Currently, two vacancies exist; pathologist and local public health agency representative. In June 2014, the term of appointment for six members will expire, and new members are being sought. Recommendations for new members to fill the openings were encouraged and may be submitted to Claudia.

### **Approval of Minutes**

The minutes of the April 18, 2013 spring meeting and the November 15, 2012 meeting were unanimously approved.

### **Subcommittee Reports**

#### **Newborn Screening and Laboratory Subcommittee (NSLS)**

A copy of the NSLS February 27, 2013 and September 18, 2013 meeting minutes was provided to members, and Dr. Hoganson summarized the meetings for the group. Dr. Hoganson noted that the number of positive galactosemia reports was significantly less during the summer of 2013 than in the previous year. The status of IDPH laboratory developments with regard to LSD and SCID testing and changes in MS/MS testing was discussed. The importance of developing an electronic interface between IDPH and the birth hospitals and specialists was reiterated.

#### **Lysosomal Storage Disorders Subcommittee**

The minutes of the last subcommittee meeting held October 17, 2013 were reviewed. Diagnostic protocols for each disorder included in LSD screening have been developed and approved by members of this group, which will be implemented to assure consistency among centers with regard to diagnostic follow-up and testing. Claudia Nash reported this subcommittee will meet next on December 19, 2013 and will discuss methods of data collection and sharing for all newborns with a positive LSD screening test. The group has obtained and reviewed data from Missouri, since LSD newborn screening has begun in that state.

#### **Cystic Fibrosis (CF) Collaborative**

Dr. McColley indicated that the Collaborative continues to meet regularly, holding two meetings nearly each month, with representation from most of the fifteen CF centers that serve

Illinois patients. The last meetings were held September 19 and 25. A sweat test checklist was developed for labs to utilize in an effort to improve the rate of satisfactory sweat tests. The collaborative recommended that the IDPH lab discontinue reporting the F508C mutation. IDPH will not be able to participate in a research study at the University of Wisconsin regarding next generation sequencing, since currently there is no authority for IDPH to share samples with another laboratory. The sensitivity of the current mutation testing panel was discussed with Dr. McColley indicating nationally there is a 95-97% sensitivity rate.

### **Newborn Screening Hemoglobinopathy Collaborative**

Dr. Thompson reported that this collaborative has been meeting every 2-3 months with participation from several of the pediatric hematologists. Minutes of the last meeting held August 26, 2013, were distributed. Long term follow-up information collected by IDPH is being reviewed and the collection form may be modified to improve compliance with reporting. IDPH staff will review more data and report further to the collaborative. Requests to IDPH for older newborn screening records to document trait status to comply with the NCAA policy of requiring sickle cell status for college athletes was discussed. The impact this practice is having on state newborn screening programs is being reviewed by the Secretary's Discretionary Advisory Committee on Heritable Disorders in Newborns and Children, which will provide further guidance to states. This is an ongoing topic of discussion by SDACHDNC and American Society of Hematology. When newborn screening results are provided to consumers for this purpose, many states include a disclaimer that this was not the intentional use for newborn screening, and many states also include educational information about the implications of trait status. The Centers for Disease Control and Prevention are revising their educational information on sickle cell trait, which should be available in the next few months.

### **Newborn Screening Expansion Subcommittee (NSES)**

Claudia Nash reported the NSES has not met since the last full committee meeting, and there have been no requests for additional disorders to be reviewed by this group.

### **Critical Congenital Heart Disease (CCHD) Newborn Screening Update**

Public Act 98-0440 was signed into law by the Governor August 16, 2013 which mandated Illinois birth hospitals to screen all newborns for critical congenital heart disease using pulse oximetry, effective immediately. The IDPH Director, Dr. Hasbrouck, has determined that at this time, IDPH will not collect any screening data, but will simply provide guidance to hospitals and serve as a resource for information. The newborn screening administrative rules are currently being amended to address the addition of CCHD to the Illinois screening panel. In addition, other changes are being made to the administrative code to address newborn screening sample retention, and data sharing. These amendments to the code should be finalized in the summer of 2014.

### **Update - Timeline for Implementation of Expanded Testing**

Dr. Dizikes reported that work is proceeding with testing of de-identified samples for both LSDs and SCID, and a sample exchange with Perkin Elmer Genetics laboratory is being planned during the validation phase of testing. It is anticipated that a pilot testing period for

SCID will begin in the Spring of 2014 with samples from two birth hospitals, and will begin a month or so later for LSDs with samples from one hospital. Following the conclusion of the pilot testing period, statewide testing will be implemented, no later than July 1, 2014 for both SCID and LSD screening. Testing for MPS II is not yet ready, and will not be implemented at that time. Changes to the Perkin Elmer database are being completed to accommodate the new testing, as well as changes to the hard copy laboratory report, which will be increased to three pages. Hospital IT programs will be notified of these upcoming additions to the newborn screening panel, so they can make necessary modifications to their database prior to July 2014.

## **IDPH Report**

### **Newborn Screening Laboratory**

Dr. Dizikes also stated that work is continuing regarding implementation of an electronic data transfer interface between the IDPH newborn screening laboratory and Northwestern Memorial Hospital, which has the highest number of births in the state. This HL7 interface would create an electronic transfer of patient information to IDPH directly from the medical record and would transfer newborn screening test results from IDPH to the hospital medical record. Once this process is successfully in place at Northwestern, it will be implemented at other birth hospitals, which should reduce data entry time and errors for hospital and IDPH staff.

Dr. Dizikes reported that work is proceeding with moving to a non-derivatized method of testing for amino acids and acylcarnitines, but this won't be implemented until testing for LSDs and SCID is operational. This method will also include succinylacetone testing for each specimen, but will no longer detect malonic aciduria. New GSP testing instruments will be installed for T4, TSH and 17-hydroxy progesterone testing, which ultimately will also be used for total galactose, GALT and biotinidase assays.

### **Newborn Screening Follow-Up Program**

Claudia Nash introduced Jean Becker, a nurse recently hired with the follow-up program. Staff have been working to provide more detailed feedback to hospitals regarding timeliness of specimen submission, with many hospitals now requesting monthly data. Staff have developed educational materials for SCID and LSDs and are making necessary changes to the IDPH Web site and hard copy educational materials. Staff continue to conduct monthly or periodic meetings of the CF Collaborative, Hemoglobinopathy Collaborative, LSD Subcommittee, and Newborn Screening Laboratory Subcommittee. In addition, staff participate in monthly national conference calls regarding CCHD screening, SCID implementation, and hemoglobinopathy screening, as well as in Region 4 Genetics Collaborative workgroups regarding long-term follow-up and endocrine disorders. With continued requests for data being received, further efforts are being made to explore improved methods of extracting data from the Perkin Elmer newborn screening data system.

## **SIDS Program**

Nikki Woolverton reported that along with the Sudden Infant Death Services of Illinois, Inc. IDPH is developing webinars regarding SIDS risk reduction and bereavement issues, that can be used by staff at local health departments, child care centers and hospitals.

## **Genetics Program Grantees**

Grants for fiscal year 2014 are continuing, with the first quarter completed. Nikki Woolverton stated IDPH is providing funding to sixty-one agencies. Grantees include seventeen hospital-based medical genetics programs, ten pediatric hematology programs, and thirty-four local health departments. IDPH also provides funding for 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.

## **Educational Activities**

Monthly educational emails are being sent to all Illinois birthing hospitals regarding newborn screening and to local public health departments regarding various topics related to genetics, newborn screening, and SIDS. Current newborn screening fact sheets have been updated and are on the IDPH website. Practitioner fact sheets for SCID and LSDs have been finalized. Changes to the Newborn Screening Practitioner's Manual to include information on LSDs and SCID are being completed. A new Data System procedure manual for staff and a Policy and Procedure manual for the entire Genetics Section is being developed.

## **Reports from Partners:**

### **Center for Jewish Genetics**

Karen Litwack reported on DNA Day activities conducted in April. IDPH provides funding to the Center which has developed various materials and programs during that are focused on family health history. For 2014, it is anticipated that the focus will be integrating genetics into nursing practice, and a webinar will be conducted for nurses and other health care providers.

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

No report-representative was not present.

### **Genetic Task Force of Illinois**

No report-representative was not present.

### **Sudden Infant Death Services (SIDS) of Illinois**

No report- representative was not present for the duration of the meeting.

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

Tess Rhodes reported that of all the LSDs to be included in screening, DSCC will cover only Pompe. However, DSCC can provide care coordination for any newborn diagnosed through screening.

**Adjournment:** The meeting was adjourned at 2:05 PM.