

**Illinois Department of Public Health**  
**Genetic and Metabolic Disease Advisory Committee (GMDAC)**  
Minutes – November 15, 2012  
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  
Colleen Gibson, R.N., LaSalle County Health Department  
Susanna McColley, M.D., Lurie Children's Hospital  
Mary Niewinski, R.D., University of Illinois at Chicago  
Michael Schneider, M.D., Carle Foundation Hospital  
Jennifer Burton, M.S., University of Illinois at Peoria  
Cathy Wicklund, M.S., Northwestern University  
Timothy Geleske, M.D., Private Practice

**Members Not Present:**

Sheila Chalmers-Currin, Parent  
Cathy Gray, R.N., University of Chicago  
George Hoganson, M.D., University of Illinois at Chicago  
Praveen Kumar, M.D., Northwestern University Medical Center  
Karen Litwack, M.S.W., Center for Jewish Genetics  
Michael Msall, M.D., University of Chicago  
Alexis Thompson, M.D., Lurie Children's Hospital  
W. Patrick Zeller, M.D., Private Practice

**Other Attendees:**

Tess Rhodes, R.N., Division of Specialized Care for Children  
Sue Shafer, G.C., President, Genetic Task Force of Illinois  
Annemarie Valdez, SIDS of Illinois  
Brook Croke, G.C., University of Illinois College of Medicine at Peoria  
Parents: Marisol Alanis and Zacarias Aguilar, Natasha Schertz, Zina Berryhill, Amy Walsh

**IDPH Representatives:**

David Culp, Ph.D., Deputy Director  
Tom Schafer, Deputy Director  
Tom Johnson, Chief Division of Laboratories  
George Dizikes, Ph.D., Acting Chief Newborn Screening Laboratory  
Mike Petros, Dr. P.H., Newborn Screening Laboratory  
Elizabeth Paton, J.D., Legal Services

**Genetics/Newborn Screening Program:**

Claudia Nash, Program Administrator

Tracey Kreipe and Shannon Harrison, Nurse Consultants

Heather Shryock, Data Manager

Newborn Screening Staff: Carey Benedict, Debbie Box, Denise Kink, Israel Guerrero, Rhonda Hawkins, Jennifer Holloway, Jessica Miller, Agnes Beavers, Doug Hart,

Graduate Student Interns: Princess Johnson, Linda Robinson, Dennis Tiburzi

### **Introductions and Announcement of New Members**

The meeting was called to order at 10:30 a.m. Following general introductions, new members were welcomed to the committee: Mary Niewinski-registered dietician, Jennifer Burton-genetic counselor, Dr. Timothy Geleske-pediatrician and Dr. Ramsay Fuleihan-pediatric immunologist.

### **Retirements**

Dr. Mike Petros was acknowledged for his service to IDPH and the Committee pending his retirement at the end of December.

### **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. During 2013, only Colleen Gibson's term will expire with an option for renewal. Currently, two vacancies exist; pathologist and consumer representatives. Recommendations for new members to fill the openings were encouraged and may be submitted to Claudia.

### **Approval of Minutes - April 19, 2012 Meeting**

The minutes of the April 19, 2012 spring meeting were unanimously approved.

### **Subcommittee Reports**

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

A copy of the NSLS September 26, 2012 meeting minutes was provided to members, and Dr. Mike Petros summarized the meeting for the group. The lab will be using a new CF DNA mutation panel in the next 3-4 weeks which includes testing for up to 45 mutations. The benign I148t mutation will no longer be reported when this panel is implemented.

Approximately 19,300 samples have been analyzed thus far by tandem mass spectrometry (MS/MS) for amino acids and acylcarnitines using a non-derivatized sample preparation method, and cut-off values for MS/MS results will be adjusted as necessary.

Laboratory staff have been exploring how to proceed with screening infants older than 90 days of age, including adoptees and transfused infants, since current test cut-off levels are applicable only for newborns and not older infants. Dr. Petros queried other states regarding their practice of testing older infants through a national newborn screening ListServ, and found no consensus. The laboratory currently tests all samples received, but may consider discouraging submission of samples for the screening of children older than four months of age.

Review of galactosemia test data indicated 156 borderline abnormal specimens and 4 presumptive positives out of 10,000 samples tested during August and September 2012. Many newborn screening laboratories in other states also experience a high number of false positive galactosemia test results during warm months.

The laboratory reported an approximate turnaround time as three days for most specimens, from sample receipt to completion of testing. The lab processes approximately 14,200 newborn screening specimens per month.

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

Claudia Nash reported the NSES has not met since the last full committee meeting in 2011. During previous meetings, the NSES developed a formal submission process for review of suggested additions to the Illinois the NBS panel. Over the past few months, an Illinois parent of a child diagnosed with adrenoleukodystrophy (ALD) has contacted Claudia Nash about adding ALD to the Illinois panel. The parent was informed about the NSES expansion protocol and that the national Secretary's Advisory Committee on Heritable Disorders in Newborns and Children preliminarily considered ALD for inclusion in state screening panels, but is awaiting subsequent data to review before further discussion of this disorder. Additional review of ALD by the Secretary's Advisory Committee should be forthcoming in the next few months.

### **Critical Congenital Heart Disease (CCHD) Work Group**

Shannon Harrison informed members that the Illinois CCHD Work Group was formed in January 2012 as recommended by the full Genetic and Metabolic Diseases Advisory Committee. The work group has met nearly each month to review and develop a standardized screening protocol, data collection tool and educational materials. Illinois birthing hospitals were surveyed in August and over 72 hospitals reported they were currently screening or in the process of implementing screening for CCHD, 13 were not screening, and 40 hospitals did not respond to the survey. The work group is very close to finalizing a recommended pulse oximetry screening protocol for birthing hospitals to use on all newborns, and will issue their final report within the next two months. Legislation is needed in Illinois to mandate hospitals conduct CCHD screening of all newborns and will likely be proposed in the Spring 2013 Session. The Centers for Disease Control and Prevention have published educational materials for parents explaining pulse oximetry screening, which the IDPH CCHD work group is recommending to hospitals. The Children's National Medical Center in Washington DC developed a CCHD toolkit for hospitals to use for medical staff education and training. The work group reviewed a data collection tool from the state of Arizona and felt that it included all fields necessary for CCHD data collection. The next Illinois work group meeting is scheduled for December 5, 2012. IDPH staff attorney, Elizabeth Paton, stated the current NBS statute had been erroneously changed to indicate that all screening must occur through tandem mass spectrometry, so when the statute is amended to require CCHD screening, the existing newborn screening language should be modified to accurately reflect testing methods. The work group was informed that the Director's Office may be collaborating with advocacy organizations such as the American Heart Association and the March of Dimes to make CCHD a 2013 legislative priority.

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

Dr. McColley discussed the efforts of the Collaborative to review CF statewide screening and diagnostic data and to reduce the percentage of sweat tests with quantity not sufficient (QNS) results, which is nationally recommended to be no greater than 10 percent. The Collaborative is receiving a small amount of funding for the second year from the National Cystic Fibrosis Foundation (CFF) which has been used to conduct site visits to hospitals regarding their sweat test protocol, in an effort to troubleshoot and reduce QNS rates. During the past four quarters, data reviewed from all CF centers that serve Illinois, indicate a QNS rate of seven percent from 3-8 CF Centers, and an aggregate QNS rate of 11% from all centers. A call for applications for a new three year funding cycle will be released in the near future by the CFF, and the Illinois CF Collaborative intends to reapply for continued funding. Dr. Charrow inquired if the recent national chloride contamination of some types of sweat test kits should be a concern for Illinois. The Committee was informed that the test kits used by Illinois CF Centers were not a part of the contamination incident.

### **Remarks from IDPH Office of Director**

Dr. Kohrman introduced himself and explained his current responsibilities with IDPH, which include serving as a consultant to the Director and as chairman of the Institutional Review Board. Dr. Kohrman recently visited the IDPH Newborn Screening Laboratory and Follow up Program and is pleased with the overall functioning of the newborn screening process. Dr. Kohrman stated that the goal of Director Hasbrouck is to ensure proper supports are available with any proposed newborn screening expansion. He also stated that Dr. Hasbrouck considers the Advisory Committee to be the primary body to advise and guide IDPH through future expansion efforts. It is expected that the GMDAC will frequently use the recommendations of the Secretary's Advisory Committee to recommend new additions to the Illinois panel. Dr. Kohrman also stated that the Director's Office is committed to a new state public health lab facility, hopefully in the near future.

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

Dr. Dizikes reported that although IDPH does currently have statutory authority to perform screening for lysosomal storage disorders (LSDs) and severe combined immunodeficiency (SCID), screening has not yet been implemented due to delays in procuring the necessary testing equipment. Some pilot LSD testing has been completed for six of the seven mandated LSDs except MPS II. It is anticipated that another 2-3 months will be required to establish baseline and cutoff values, and the IDPH laboratory will conduct a comparison study with Perkin Elmer Genetics Laboratory to ensure consistent results. Regarding Krabbe testing, the New York Department of Public Health Laboratory has tentatively agreed to perform molecular studies on the estimated 70 presumptive positive cases per year for Illinois that are expected. A written agreement will be finalized.

The IDPH laboratory is also proceeding with preparations for SCID pilot testing. There was a question whether Perkin Elmer Genetics could be used on a contractual basis to begin SCID testing until all the necessary equipment is procured. Tom Johnson stated IDPH would still have to enter into a contract for services which frequently takes as long as the purchase of

equipment, and IDPH would need to build-up funds to pay for contractual testing services. Hospitals that will be included in the pilot have not yet been identified for either LSD or SCID testing.

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.

Tom Johnson informed the committee that the IDPH newborn screening laboratory has drafted a continuity of operations retainer contract with Perkin Elmer Genetics Laboratory. IDPH had explored the possibility of utilizing neighboring states, but unfortunately none could handle the volume of Illinois samples in the event of an emergency shut down of the IDPH laboratory.

Dr. Ross verbalized her ongoing concerns regarding the failure to require parental consent for the expanded testing for lysosomal storage disorders, which she indicated would be expressed in forthcoming publications.

### **IDPH Report**

#### **Newborn Screening Laboratory – Section Chief Update**

Dr. George Dizikes reported that he remains the acting Newborn Screening Laboratory Section Chief, but has continued to work with CMS on various recruitment strategies. Further suggestions regarding recruitment methods or concerns from qualified individuals should be addressed to Dr. Dizikes.

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

Tracey Kreipe introduced three new follow-up staff and two graduate student interns from the Newborn Screening Follow up Program. There remain two vacancies for cystic fibrosis and galactosemia/biotinidase follow-up. A Northwestern Genetic Counseling Student has also been assigned to work on a part time basis with the follow up program.

Staff continue to conduct monthly meetings of the CF Collaborative and CCHD Workgroup, the quarterly Newborn Screening Laboratory Subcommittee, and the Newborn Screening Expansion Subcommittee. In addition, staff participated on monthly national conference calls regarding SCID implementation, and hemoglobinopathy screening, as well as in Region 4 Genetics Collaborative workgroups regarding long-term follow-up and endocrine disorders. Representatives from IDPH Lab, contact hospitals, specialists, and parents attended the Region 4 annual meeting held in September in Michigan.

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.

Preparations are also underway for a staff business continuity exercise.

The Newborn Screening Laboratory Subcommittee, laboratory and follow up program staff are reviewing cut-off values regarding the high volume of galactosemia abnormal results that occurred during the warm summer months, to better prepare for the summer of 2013.

### **SIDS Program**

Nikki Woolverton reported on the SIDS/Infant Mortality Program and IDPH Genetics grants. Under Illinois law, coroners and medical examiners must report any sudden and unexpected infant death to IDPH within 72 hours. Following coroner notification, the staff contacts a local public health nurse and requests a visit to the family to inform parents of community support services. IDPH also sends letters of condolence to families and provides information about bereavement resources. Approximately 200 infant death reports are received annually, including those for possible SIDS, overlay and entrapment or other causes of sudden, unexpected infant deaths.

### **Genetics Program Grantees**

For fiscal year 2013, 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. (SIDS of Illinois).

### **Educational Activities**

Shannon Harrison provided the following summary: 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 have been completed. A new Data System procedure manual for staff and a Policy and Procedure manual for the entire Genetics Section is being developed.

### **Secretary's Advisory Committee on Heritable Disorders in Newborns and Children**

Various documents from the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children were provided and discussed, including: 1) Summary of NBS panel expansion; 2) Condition Review / Matrix Document; 3) Minutes from the May 2012 Meeting and the 2012 Annual Report; 4) Summary document of HHS Secretary's recent recommendations; 5) Retention and use of dried blood spots. Cathy Wicklund, who serves on

the Secretary's Advisory Committee, stated there was an evidence based review underway to consider the addition of Pompe disease to the recommended uniform newborn screening panel, and that a final report would be released in January 2013.

Dr. Ross stated that screening studies to date for Pompe have been considered research and required parental consent. Dr. Kohrman indicated that it would be helpful to have model statute language regarding expansion for states to adopt. Cathy concluded that the current Matrix document is solely addressing state readiness from the laboratory perspective but in time the Committee would also consider statutory, procurement, and political implications.

### **Reports from Partners:**

#### **Center for Jewish Genetics**

DNA Day has become a month long event during April, that is focused on family health history. Recently the Center received federal grant funding from Baby's First Test, to conduct an outreach program used to educate local health departments and high schools.

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

No report-representative was not present.

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

Sue Shafer reported a recent needs assessment survey was completed with a 43% response rate regarding future directions of the Task Force. As a result of the survey, an outreach committee will attempt to recruit other professionals to participate in the Task Force, which has historically been comprised of medical geneticists and genetic counselors. Sue also informed the committee that the Illinois Genetic Counseling Licensure Act expires every ten years and is due to expire January 1, 2015. The approaching expiration of the Act provides an opportunity for amendments, if needed. The annual Genetics Symposium will be held at Northwestern Memorial Hospital February 8, 2013, to provide current information for health care professionals regarding developments in the field of medical genetics.

#### **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 DSCC is working to improve communication with families and providers using an array of technologies and social media. In addition, DSCC is interested in providing more care coordination and moving away from just being a "bill payer".

### **Comments/Discussion:**

Claudia Nash reminded members of the required annual Open Meetings Act and Ethics trainings, and the need to print and submit a certificate of completion as soon as possible.

The meeting was adjourned at 1:38 PM.