

**Illinois Department of Public Health**  
**Genetic and Metabolic Disease Advisory Committee (GMDAC)**  
Minutes – April 18, 2013  
William Tell Holiday Inn, Countryside, IL

**Members Present:**

Joel Charrow, M.D., Chair, Lurie Children's Hospital  
Ramsay Fuleihan, M.D., Lurie Children's Hospital  
Michael Schneider, M.D., Carle Foundation Hospital  
Jennifer Burton, M.S., University of Illinois at Peoria  
Cathy Gray, R.N., University of Chicago  
George Hoganson, M.D., University of Illinois at Chicago  
Michael Msall, M.D., University of Chicago  
Alexis Thompson, M.D., Lurie Children's Hospital  
Amy Walsh, Parent

**Members Not Present:**

Lainie Friedman Ross, M.D., PhD., University of Chicago  
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  
Cathy Wicklund, M.S., Northwestern University  
Timothy Geleske, M.D., Private Practice  
Sheila Chalmers-Currin, Parent  
Karen Litwack, M.S.W., Center for Jewish Genetics  
W. Patrick Zeller, M.D., Private Practice

**Other Attendees:**

Barbara Burton, M.D., Lurie Children's Hospital  
Shelly Roat, Division of Specialized Care for Children  
Cindy Mitchell, RN, Perinatal Network Administrator, Springfield Network  
Parents: Denise Wilburn, Christina and Bryce LaBord

**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  
Jennifer Crew, Ph.D.,  
Khaja Basheeruddin, Ph.D.,  
Rong Shao, Ph.D.,  
Pat Kloppenburg,  
Claudia Nash, Genetics Section Administrator  
Tracey Kreipe and Shannon Harrison, Nurse Consultants

## **Introductions and Announcements**

The meeting was called to order at 10:45 a.m. Dr. Joel Charrow and Claudia Nash informed attendees that rainy weather and area flooding had prevented several members and interested individuals from attending, therefore a quorum was not achieved. Information was reviewed but no action was taken due to lack of a majority of members present. .

## **Approval of Minutes - November 15, 2012 Meeting**

The minutes of the November 15, 2012 fall meeting were included in the meeting folders. Formal discussion and approval was postponed until the fall 2013 meeting.

## **Committee Vacancies**

Three members are resigning due to retirement or new jobs. Cathy Gray, RN, Colleen Gibson, RN and Praveen Kumar, M.D were acknowledged for their service to the Committee and wished well by the meeting attendees. Cathy brought with her Cindy Mitchell, Springfield Perinatal Network Administrator, as a nominee for the hospital nurse appointment. Claudia Nash stated there were now a total of four (4) vacancies: Pathologist, neonatologist, hospital nurse and public health representative. Recommendations for new members to fill the openings were encouraged and may be submitted to Claudia.

## **Subcommittee Reports**

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

A copy of the NSLS February 27, 2013 meeting minutes was provided to members, and Dr. George Hoganson summarized the meeting for the group. The subcommittee continues to meet quarterly to review lab operations and reporting. The lab has been using a new CF DNA mutation panel and a few new mutations have been flagged as benign polymorphisms. Dr. Hoganson and the lab are concerned with causing unnecessary parental anxiety and referrals because of reporting these seemingly benign mutations. The subcommittee will consult with the CF Collaborative to further discuss this issue. The lab will participate in a regional CF quality improvement project with Dr. Phil Farrell of the University of Wisconsin, with many of the details still to be determined. Dr. George Dizikes stated the lab is considering dried blood spot storage and the many considerations which would be required including controlling conditions, equipment, staffing, and storage areas.

### **Lysosomal Storage Diseases Subcommittee (LSDS)**

Dr. Barbara Burton stated the subcommittee has not had any recent meetings but will convene May 17, 2013 at Lurie Children's Hospital. An agenda was provided and invitation extended to anyone who is interested in LSD implementation. Dr. Kathy Grange from Washington University in St. Louis will describe LSD screening that has been occurring in the state of Missouri.

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

Cathy Gray informed members that the Illinois CCHD Work Group has met for approximately eighteen months and accomplished its goals to establish a screening protocol, data collection tool, follow-up protocol, education for providers and parents, and to review additional

considerations. These resources have been distributed to all Illinois birthing hospitals, and many have already begun screening. The work group recommended that IDPH be supportive of Illinois CCHD Legislation and specifically Representative Gabel's House Bill (HB) 2661. Tom Schafer stated that Senator Steans is also supportive of CCHD and will endorse the bill in the senate and add some amended language that will "clean-up" some language currently in the Act. Copies of HB 2661 were distributed to meeting attendees per request. Dr. Hoganson offered additional changes to the bill as written. HB 2661 has been endorsed by American Heart Association, the March of Dimes and Illinois Hospital Association.

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

Tracey Kreipe 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. Recent discussion among Collaborative participants via monthly meetings and a listserv has been regarding the implementation of the new CF panel and reporting of benign mutations. Jennifer Burton informed attendees of a recent case involving F508C which is a non-disease causing mutation by itself but when paired with specific other mutations may have variable clinical consequences such as congenital bilateral absence of vas deferens (CBAVD) according to Dr. Susanna McColley. Tracey encouraged future discussion among IDPH Lab, NSLS and the Collaborative to address concerns about the above and to determine whether reporting some mutations may be unnecessary. Dr. McColley is also reapplying to the CF Foundation for continued funding on behalf of the CF Collaborative to examine health outcomes.

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

Claudia Nash reported the NSES has not met since the last full committee meeting in 2011 and there have been no formal proposals for additional disorders to be added at this time, although there has been interest expressed by a parent for adrenoleukodystrophy to be considered.

### **Update: Timeline for Implementation of Expanded Testing – Lysosomal Storage Disorders (LSD) and Severe Combined Immunodeficiency (SCID)**

Dr. Khaja Basheruddin provided a presentation on progress made by the IDPH lab with regard to LSD testing, and stated that they anticipate full scale, statewide implementation by July 2014. Following the LSD presentation, Dr. Jennifer Crew explained the current status of SCID implementation and projected a similar full scale, statewide screening timeline by July 2014. Both individuals stated remaining hurdles for implementation include equipment procurement issues, finalizing validation studies, data collection and pilot testing.

Immunologists have been identified for SCID referrals, and the University of Chicago, Lurie Children's Hospital and St. Louis Children's Hospital are currently recognized as pediatric bone marrow transplant centers.

### **IDPH Report**

#### **Newborn Screening Laboratory**

Dr. George Dizikes reported that he has accepted the Newborn Screening Laboratory Section Chief position and will continue with some of his other duties as CLIA manager as well. A

total of 40,502 newborn screening specimens were tested between January–March 2013 with average turnaround time between 3.5-5.3 days depending on the assay. NeoBase MS/MS test validation for a non-derivatized sample preparation continues with approximately 19,000 specimens tested to date. Hector Diaz, lab supervisor, completed an analysis of 2012 calendar year galactosemia data and the impact of humidity on enzyme activity. He concluded that the increase in false positive results was directly related to humidity during warm summer months. Dr. Dizikes proposed the use of a desiccant when shipping specimens. Dr. Charrow asked if the lab would be able to submit aggregate data to the R4S, Region 4 database and Dr. Dizikes agreed that SCID and data regarding other disorders would be entered.

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

Tracey Kreipe reported staff continues to facilitate monthly meetings of the CF Collaborative, the quarterly Newborn Screening Laboratory Subcommittee, and as needed the Newborn Screening Expansion Subcommittee. In addition, staff participated in 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. A Hematology Collaborative will begin May 2013 with the purpose of providing a venue for discussion among credentialed Illinois pediatric hematologists and other medical professionals to improve follow-up coordination efforts with IDPH Newborn Screening and Genetics Program. Two Community Health Centers (CHCs), PCC Community Wellness and Friend Family Health Centers have designated individuals to be the NBS Coordinator for their institution. IDPH is actively seeking additional health centers to follow suit and designate a newborn screening coordinator which will better facilitate management of abnormal screening results, follow-up screenings, specialist referrals and education for parents and providers.

Shannon Harrison informed attendees that the Newborn Screening Follow up Program still has three staff vacancies. She also stated that the Critical Congenital Heart Disease Work Group has concluded its efforts and finalized their report which includes a recommended screening protocol and data collection tool. Shannon also reported that she is actively working with the Region 4 Genetics Collaborative and requested the Illinois Chapter of the American Academy of Pediatrics distribute a statewide survey to primary care pediatricians to assess their practice of collecting family health history.

### **Genetics Program Grantees**

For fiscal year 2014, Claudia Nash stated IDPH is providing funding to sixty-two 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). New legislation this year required IDPH to develop a formula for awarding grants to improve grantee accountability. Many

IDPH Programs experienced reductions in grant funding, however, NBS and Genetics was fortunate to keep most grantees at relatively level funding for the upcoming fiscal year.

### **SIDS Program**

Claudia Nash reported on the SIDS/Infant Mortality Program. The IDPH Center for Health Statistics most current data was provided from 2009, which indicates the number of SIDS related deaths had decreased, as had the birth rate. Accidental suffocation continues to be a significant cause of infant death. SIDS of Illinois is currently providing education regarding safe sleep and risk reduction across the state to daycare providers.

### **Educational Activities**

Shannon Harrison reported that she is sending monthly educational emails 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 are in development for Lynch Syndrome (consumer and provider versions) and congenital hypothyroidism. Changes to the Newborn Screening Practitioner's Manual to include information on LSDs and SCID have been completed. A new staff data system procedure manual and a policy and procedure manual for the entire Genetics Section is being developed. Denise Wilburn inquired about the galactosemia information provided by IDPH. Tracey Kreipe stated that a provider fact sheet and metabolic specialist list is sent to primary care providers, pediatricians and hospitals with all NBS reports that are borderline or positive for galactosemia. Denise and Amy Walsh suggested that IDPH consider development of a parent fact sheet that explains abnormal NBS results in general terms and a second parent fact sheet that is disorder specific.

### **Reports from Partners:**

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

Shannon Harrison stated DNA Day has become a month long event during April that is focused on family health history. On April 15, the Center in collaboration with IDPH and the Illinois Network for Education and Training (i-NET) provided a webinar on newborn screening, which included information on LSD and SCID for local health departments and other medical professionals. One credit hour of nursing continuing education was provided. The recorded DNA Day Webinar is at: <http://www.illinoisnetwork.org/programs/register.php?id=67>

#### **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.

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

Shelly Rhodes reported that DSCC is working to improve communication with families and providers using an array of technologies including an electronic care coordination information system and social media. In addition, DSCC is striving to move away from being just a “bill payer”.

### **Comments/Discussion:**

Tom Shafer asked that additional suggestions to change the language for the NBS Metabolic Screening Act be forwarded to Claudia Nash.

The meeting was adjourned at 2:05 PM.