Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee
Illinois Department of Public Health, Division of Laboratories
2121 W. Taylor St., Chicago, Illinois
Meeting and Conference Call Minutes: June 6, 2012

Subcommittee Members Attending:
George Hoganson-University of Illinois at Chicago – Chair
Barbara Burton, Lurie Children's Hospital
Gopal Srinivasan- Mt. Sinai Hospital
Sunetra Reddy-University of Chicago
Kristin Clemenz-Lurie Children's Memorial
W. Patrick Zeller-Pediatric Endocrinologist

Other Participants:
Pat McGleam-Pediatric Endocrinology nurse

IDPH Staff:
George Dizikes, Mike Petros, Khaja Basheeruddin, John Nawrocki, Raj Singh, Bill Calvert
Glen Yoshimura, Hector Diaz, Rong Shao, Shannon Harrison, Tracey Kreipe, Barbara DeLuka

The meeting was called to order at 1:05 PM, followed by introductions. The minutes of the February 22, 2012 meeting were approved.

New Business

Laboratory Report
Staffing and Laboratory Resources:
Laboratory staff was recently increased with two recently hired technologists currently in training to perform MS/MS testing. The Newborn Screening Laboratory is undergoing re-organization and candidates for the newborn section chief position are being sought. A request was made by Dr. Hoganson to provide to the Subcommittee members a listing of supervisors for the various Newborn Screening Laboratory testing sections.

Data System/Reports:
The Perkin Elmer laboratory data system now provides a two-page report for any specimens with abnormal or unsatisfactory results. Currently, there are 67 hospitals that have requested automated fax reporting of newborn screening (NBS) laboratory reports. IDPH laboratory and IT, the data system vendor and Northwestern Memorial IT and laboratory are working to implement electronic data exchange of birth record information, NBS orders, and test results. It was reported that the average turnaround time (time from receipt of NBS specimens to reporting of results) varies slightly each month and currently ranges from as low as 2.9 days for endocrine disorders to 7.7 days for CF two-tier molecular testing.

Cystic Fibrosis (CF) Screening:
While there is no shortage of supplies for CF DNA testing, the new DNA panel for CF screening will most likely be implemented in August 2012, following depletion of the DNA test kits currently in use.

Lysosomal Storage Disease (LSD) Screening:
In April the laboratory received a tandem mass spectrometer (MS/MS) instrument on loan, and will utilize the instrument in preparation for LSD testing. Laboratory staff have completed training and the instrument is optimized for testing. CDC has provided QC materials, and plans for LSD screening preparation include use of de-identified samples and dual testing in conjunction with Perkin-Elmer Genetics, a private lab that currently offers LSD screening. Six LSD disorders (Pompe, Fabry, Gaucher, Krabbe, Niemann-Pick, and MPS 1 - Hurler's) will be included in the dual testing project. A reagent for MPS2 (Hunter's) is not yet available. However, the University of Washington and Perkin-Elmer Genetics are working to develop one. The IDPH laboratory could possibly start a LSD pilot at one or two hospitals by late fall 2012. Following the pilot, statewide screening for LSD's is dependent upon procurement of additional tandem mass spectrometers and the necessary liquid handling equipment for sample preparation. The laboratory must be able to process upwards of 800 samples a day in order to implement statewide screening.

SCID Screening:
Dr. Nawrocki reported that IDPH Molecular Laboratory staff are ready for validation of SCID testing, but are waiting for acquisition of the initial robotic DNA extractors and high through-put PCR amplifiers necessary for pilot testing. These should start to become available during August 2012. State procurement issues have delayed equipment acquisition. Following the SCID pilot, four sets of this equipment will be necessary for statewide SCID screening, which is anticipated by July 2013.

MS/MS Testing:
Dr. Shao informed the Subcommittee that following a review of MS/MS reporting, all abnormal results indicative of possible VLCAD will be considered positive screens, and referral to a metabolic specialist for evaluation and diagnostic testing will be recommended in these cases. This protocol is being implemented due to recognition of the rapid normalization of the primary VLCAD analyte (C14:1) as newborns obtain adequate intake of breast milk or formula in the days and weeks following birth. Dr. Shao is working with the laboratory data system vendor to produce reports that include all of the MS/MS analytes for any MS/MS abnormal screens. These more informative reports will be available upon request by the physicians. Dr. Hoganson requested screening result data for recently confirmed MS/MS and galactosemia cases and he and Dr. Burton offered the assistance of the metabolic specialists in on-going review of MS/MS results data.

Follow-up Program
Due to staff acceptance of promotional opportunities with IDPH, there are now three vacant positions in the Follow-up Program. Hiring for these positions has been approved.

> Note: Following this meeting these three open positions were posted to be filled, and one additional staff member has resigned to accept a promotion in another IDPH program.

Follow-up staff have been participating in monthly national Web casts on the progress of individual state efforts to implement SCID screening. Staff will also be working with the data system vendor to produce necessary abnormal reports for SCID and LSD's.

Critical Congenital Heart Disease (CCHD):
The CCHD workgroup has had four meetings to date (January, March, April, and May), and has identified several goals the workgroup would like to accomplish:

- To coordinate a statewide comprehensive program that would require screening and reporting of all results to IDPH
- To develop a standardized protocol for screening and follow-up
- To provide families and health care professionals with education and training

- To ensure that an adequate system is in place to provide diagnostic testing and treatment statewide
- To possibly link SpO2 results into an existing data base system for reporting
- To explore billing and reimbursement issues

The work group is developing a protocol to screen all babies born in Illinois for CCHD, and will follow the protocol developed by Secretary's Advisory Committee on Heritable Disorders in Newborns and Children. Also, there will be guidelines in place for hospitals to follow for failed screens.

Region 4 Endocrine Project:

The Region 4 Collaborative Short-term Follow-up work group will be surveying parents and the pediatric endocrinologists caring for children born in 2007 who were diagnosed with congenital hypothyroidism (CH). A data use agreement will be executed between IDPH and the Michigan Public Health Institute (MPHI) in accordance with the HRSA funded multi-state Region 4 Endocrine Project. Dr. Zeller, who is currently chair of an Illinois pediatric endocrinology professional group offered to inform fellow pediatric endocrinologists in advance of survey distribution.

Note: On June 21 the Data Use Agreement was executed by Dr. Hasbrouck, Director of IDPH, and on June 28 this survey project was approved by Dr. Kohrman, Chair of the IDPH Institutional Review Board. The purposes of these surveys are to:

- Determine how pediatric endocrinologists manage long term follow-up for children diagnosed with CH
- Identify current practices for educating parents about CH and long term follow-up

A similar study performed in Michigan found a number of children diagnosed with CH whose treatment for CH was stopped by parents without the benefit of follow-up testing or monitoring for normal thyroid functioning. De-identified data will be analyzed by MPHI and results provided to the seven states (IL, IN, MN, MI, WI, KY, and OH) participating in the study.

Dr. Hoganson also requested that data on CH cases diagnosed in pre-term, low birth weight newborns following the recent IDPH recommendation for a routine third newborn screen at day 28 of life in this population be made available to the Subcommittee members.

Hemoglobinopathies:

There was discussion about the validity of hemoglobinopathy screening when the initial screening sample is collected prior to transfusion and/or at 24-48 hours of age in very pre-term babies. Members were informed by IDPH laboratory staff that with the current high performance liquid chromatography (HPLC) used for this screening, HPLC is capable of detecting as little as 1.0% of adult or normal hemoglobin A and 1.0% of fetal, hemoglobin F in dried blood spots. Dr. Hoganson suggested this issue be discussed further with Dr. Alexis Thompson and other pediatric hematologists; and hemoglobin screening results of confirmed sickle cell disease and other hemoglobinopathy cases be provided to the members and pediatric hematologists.

The meeting was adjourned at 1:45PM. The next meeting is set for September 26, 2012 from 9-11AM.

Respectfully submitted,
Barbara DeLuka

Approved - 9/26/12