Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee
Conference Call Meeting Minutes: February 22, 2012

Subcommittee Members Attending:
George Hoganson-University of Illinois at Chicago – Chair
Gopal Srinivasan- Mt. Sinai Hospital
Sunetra Reddy- University of Chicago
Kristin Clemenz-Children's Memorial
Other Attendees:
Joel Charrow, Genetics and Metabolic Diseases Advisory Committee(GMDAC) Chair-Children's Memorial Hospital

IDPH Staff:
George Dizikes, Claudia Nash, Tom Johnson, Bill Calvert
Barbara DeLuka, Matt Charles, Shunna Johnson, Maureen Mc Bride
Rong Shao, Heather Shryock, Shannon Harrison
Khaja Basheeruddin, Raj Singh, Mike Petros

The meeting was called to order at 9:07 AM, followed by introductions. The minutes of the September 28, 2011 meeting were approved.

New Business

Laboratory Report

Staffing and Laboratory Resources

Laboratory staff was recently increased with one internal transfer within Division of Laboratories, and two new hires. New staff will be trained in MS/MS testing, along with the routine training rotations through other laboratory sections.

Data System/Reports

The Perkin Elmer laboratory data system remains a work in progress as system improvements continue. Currently, laboratory staff are utilizing new QA report options. The Division of Laboratories now issues two-page specimen reports with reference ranges for all samples with abnormal and unsatisfactory results. A second "reference range" page may also be requested for any "normal" report upon request by the physician or submitter. Sixty hospitals have requested and are receiving direct faxing of laboratory reports, and this process appears to be working well. There was a question about adding the hospital lab accession number to the data system specimen record, and including this hospital identifier on all printed newborn screening reports, a request that Matt Charles offered to address.

Galactosemia Screening

The galactosemia algorithm was revised February 10, 2011, and following discussion with the metabolic specialists and review of test findings, analytic cutoffs for the "borderline" category was revised on July 23, 2011. Ongoing review of the galactosemia screening results from September 2011 found 15-20 positive screens per each 10,000 samples tested. The revised algorithm appears to be working well. Dr. Hoganson and Dr. Charrow reported that several galactosemia carriers have been identified with the revised algorithm, as would be expected for galactosemia newborn screening.

Cystic Fibrosis (CF) Screening

The new DNA panel for CF screening has not yet been implemented. IDPH Fiscal is working on a new acquisition process that has delayed purchase o f the new kits. Purchase of the new CF kits is anticipated by June 2012, and there is no current shortage of supplies. The new CF kits have been utilized by many public health laboratories and the new panel has been reviewed by the CF Collaborative Workgroup, which felt this panel to be acceptable for CF screening. There are some differences in the mutations between the two panels and a listing of CF mutations for both panels is available to hospitals and physicians.

Lysosomal Storage Disease (LSD) Screening

Khaja Basheeruddin reported lab staff are optimizing the MS/MS screening methodology for six of the LSD's, and should receive the seventh LSD reagent in the very near future. Reagents are available for Pompe, Gaucher, Fabry, Niemann Pick, Krabbe, and MPS 1, and the MPS2 reagent is expected soon. Procurement of equipment has become increasingly problematic. A loaner instrument may become available for verification testing of the 6 plus 1 multiplex LSD testing. Verification of any IDPH testing of identified samples would be arranged with Perkin-Elmer Genetics, a private company that currently provides LSD testing to the public market. IDPH has two contracts with Perkin-Elmer Genetics, one for verification studies of LSD testing and another for severe combined immune deficiency (SCID) screening. In addition, plans include procurement of five additional MS/MS instruments to replace older units.
It was also reported that preliminary information about LSD screening was presented to the IDPH Institutional Review Board last week. The IRB has yet to determine if LSD testing would be considered as research, or if informed consent would be needed.

SCID Screening

Screening for SCID will include PCR testing for TREC utilizing a multiplex system. A pilot using low through-put PCR instruments may be necessary, as procurement of the high through-put equipment needed for statewide SCID screening may not be procured until 2013. Plans for piloting and statewide SCID screening could be delayed until 2013, as the need to delay statewide screening following the pilot could not be avoided due to the volume of samples to be tested with the current equipment.
Dr. Fuleihan, the pediatric immunologist at Children's Memorial has offered to provide flow cytometry testing for any babies with positive screens for SCID, once pilot and statewide testing is implemented.

MS/MS Testing

There was brief discussion about plans to implement non-derivatized sample preparation for MS/MS screening. Current trials of the non-derivatized method show screening results may vary from those obtained with the derivatized preparation method, and reference ranges may need to be adjusted accordingly. The benefits of changing MS/MS sample preparation are being reviewed by IDPH, and it was acknowledged that CDC is working to provide proficiency testing for those labs using non-derivatized samples. Dr. Hoganson and Dr. Charrow agreed that if the testing is validated for accuracy and normal reference ranges are provided, the change to the non-derivatized method would not be cause for concern for the metabolic specialists.

Follow-up Program

Several new follow-up positions have been filled, with one additional new hire anticipated. The data system programmers will be on site in Springfield this week to work with Follow-up staff on several system development issues.

Critical Congenital Heart Disease

The Critical Congenital Heart Disease Workgroup held an introductory conference call and a second call is planned for early March, and thereafter every other month. Dr. Praveen Kumar is the workgroup chair. It appears that several perinatal hospitals have implemented this screening, and Follow-up staff are requesting CCHD screening protocols to share with the Workgroup. Although a change to the Newborn Screening law is necessary prior to mandating CCHD screening, members suggested that interested hospitals could be encouraged to start the screening prior to these legal changes, as this may not occur before 2013.

New requirements for Newborn Screening in the NICU

Members were reminded that the Clinical Laboratory Standards Institute (CLSI) has recommended serial screening (multiple routine sample collections) of babies born at less than 34 weeks gestation and/or birth weight of less than 2000 grams. CLSI guidelines recommend that in addition to the routine second sample collection now required by IDPH at day 14 for all babies admitted to the NICU, that a third routine sample be collected from pre-term/low birth weight babies at day 28 of life or prior to NICU discharge whichever is first.

Although the GMDAC did not vote to make a recommendation for this additional sample collection requirement, there were no objections at the October meeting. The Newborn Screening Laboratory Subcommittee members agreed that following the CLSI guidelines was advisable, and would be beneficial in detecting additional cases of hypothyroidism, as well as helpful in resolving abnormal earlier screenings due to the effects of total parenteral nutrition. Although third sample collection cannot be required without an Administrative Rule change, a recommendation through the perinatal network and directly to the hospitals could be made, much as is proposed for pulse oximetry screening for CCHD.

Other Discussion

There was discussion about the validity of hemoglobinopathy screening when the initial screening sample is collected prior to transfusion or at 24-48 hours of age in very pre-term babies. Dr. Hoganson suggested that this issue be discussed further with Dr. Alexis Thompson and other pediatric hematologists.

The meeting was adjourned at 10:05. The next meeting is set for June 6, 2012 from 1-3PM.

Respectfully submitted,
Barbara DeLuka

Approved - June 6, 2012