Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee (NSLS)
Illinois Department of Public Health, Division of Laboratories
2121 W. Taylor St., Chicago, Illinois
Meeting and Conference Call Minutes: September 26, 2012

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

Other Participants:
Darryl Waggoner-University of Chicago

IDPH Staff:
George Dizikes, Mike Petros, Khaja Basheeruddin, John Nawrocki, Raj Singh, Bill Calvert, Matt Charles, Shunna Johnson, Glen Yoshimura, Hector Diaz, Rong Shao, Jennifer Crew, Claudia Nash, Tracey Kreipe, Barbara DeLuka, Heather Shryock, Jennifer Holloway, Israel Guerrero

The meeting was called to order at 9:18AM, followed by introductions. The minutes of the June 6, 2012 meeting were then approved.

New Business

Laboratory Report
Staffing and Laboratory Resources:
There have been no recent additions to Laboratory staffing and cross training of technologists in the various testing areas continues. The Newborn Screening Laboratory is undergoing re-organization and a listing of the laboratory supervisors and contact information are attached to the minutes. Dr. Petros announced he will be retiring at the end of the year, and Dr. Hoganson thanked him for his years of dedicated service to the newborn screening community.

Data System/Reports:
The Laboratory continues to work with the data system vendor on system improvements, and work continues with Northwestern Memorial IT, IDPH IT and the vendor to implement HL7 electronic data transfers, first in-bound sending of demographic data to IDPH to occur by November 1, 2012, which is to be followed by sending out-bound newborn screening results data back to the hospital to occur by January 1, 2013. Several other hospitals have expressed interest in implementing HL7 data transfers and IDPH staff are working with these hospitals.

Cystic Fibrosis (CF) Screening:
Staff training on use of the new CF DNA test panel (Hologic InPlex with 44 CF alleles) has been completed, and earlier this morning the CF Collaborative held a monthly conference call with CF specialists. During the call there was discussion about the I148T CF mutation that will be included in the new test panel. The national consensus among CF specialists is not to report this mutation since it is only associated with CF in the presence of a second allele that will not be included in the new CF panel. Jennifer Crew indicated the I148T mutation could be "blocked" and not reported when use of the new CF test panel begins. The CF specialists are aware of the differences in mutations between these two panels, and they will be notified when use of the new panel begins. Claudia has

provided information on the specific mutations in both the old and new panels to all the CF Centers that serve Illinois families. The information will also be made available to any hospital laboratories upon request.

There was also discussion about Dr. Phil Farrell's visit to the IDPH Chicago Lab and a proposed collaborative study of a newly developed 157 CF mutation panel in cooperation with Dr. Farrell and Dr. Mei Baker of Wisconsin. This proposed study would also include Minnesota, Indiana and Michigan State laboratories, and could potentially decrease the number of false positive screens for CF that require sweat testing. Dr. Suzanna McColley is also working with this proposed study group.

Severe Combined Immune Deficiency (SCID) Screening:
Dr. Crew reported that assay for SCID is working well, and the IDPH Molecular Laboratory is waiting for acquisition of the high volume extractors and PCR amplifiers necessary to implement the pilot and statewide SCID testing.

Lysosomal Storage Disease (LSD) Screening:
It was reported that LSD multiplex standards from the University of Washington laboratory may be superior to the previously available CDC standards, and laboratory staff are working with University of Washington on new internal standards for three enzymes and working to establish fixed or floating cut-off values for enzyme activity levels.

Procurement of equipment is ongoing, and CMS is moving forward with the process. No phase-in pilot will be initiated until all equipment needed for statewide testing is obtained. There was discussion about provisions for DNA testing through an outside laboratory to reduce the high false positive rate associated with Krabbe screening, and Dr. Dizikes indicated that the IDPH molecular lab may be capable of performing this testing without sending samples to New York or another external laboratory. There was discussion about requesting input from the ad hoc Lysosomal Storage Disease Subcommittee, of which Dr. Burton is the chair, on this issue.

Follow-up Program Report
Three full time office specialists were recently hired to fill Follow-up Program vacancies, and one additional position remains vacant. These new staff members are being trained to assist with reporting of NBS results and tracking of active follow-up cases.

Follow-up staff have been participating in monthly national Web casts on the progress of individual states 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):
There have not been any additional CCHD workgroup meetings since the February NSLS meeting. Shannon Harrison at the NBS Follow-up Program is working to establish a standing monthly meeting date and working with workgroup members to finalize a CCHD protocol that will be sent to all Illinois birthing hospitals. A recent survey of the perinatal network administrators found that:

- 52 hospitals are currently screening for CCHD
- Over 20 hospitals are in the process of implementing CCHD screening
- 13 hospitals are not providing the screening
- 29 hospitals have not yet reported on their screening status

Changes to the NBS legislation to give IDPH authority to require CCHD screening of all Illinois newborns is being proposed, and once enacted, changes to the NBS Administrative Rule will be needed. Although CCHD screening will probably not be mandated before early 2014, as stated above, many hospitals have already implemented the screening, or have plans to do so. Shannon is also working to compile CCHD consumer and professional educational materials.

Region 4 Genetics Collaborative Endocrine Project:
With approval of the newly formed IDPH IRB Board, in late August surveys were sent to the parents of ninety-four Illinois children who were born in 2007 and diagnosed with congenital hypothyroidism. Surveys were also sent to the physicians who are caring these children. This mirror designed study of parents' understanding about their child's condition and physicians' care practices includes children born in seven Region 4 states. In late October the Illinois de-identified surveys will be sent to the Michigan Public Health Institute for data entry and analysis. So far, of the ninety-four parent surveys sent, eighteen have been returned completed and twenty-four were returned unclaimed or undeliverable. Of the ninety-four physician surveys, thirty have been returned completed.

Other Discussion

Tandem Mass Spectrometry (MS/MS) Testing:
The number of out of range MS/MS newborn screens for possible very long chain acyl-CoA-dehydrogenase deficiency (VLCAD) has lead to more newborns being referred for diagnostic testing and increased false positive rates for this disorder. However, this does not appear problematic and this protocol change has alleviated concerns about delays in obtaining repeat specimens for borderline initial screens resulting in false negative repeat NBS results due to normalization of the long chain acylcarnitines in the first week(s) of life.

There was continued discussion about creating age adjusted cut-offs for MS/MS testing of older infants, especially those over 3 months of age. While the number of older infants whose specimens are received for screening is relatively small, there is concern that a disclaimer may need to be added to these specimen reports. Dr. Petros will seek information through the National Newborn Screening and Genetics Resource Center list serv as to how other states laboratories address this issue to assist in resolving this matter.

Hemoglobinopathies:
Discussion of this topic will be deferred until the next NSLS meeting when Dr. Alexis Thompson, a pediatric hematologist is available, and to allow more time to collect data on the issue of hemoglobinopathy screening and the possible effects of birth at early gestational ages.

Galactosemia Screening:
Due to the high volume of NBS specimens with reduced galactose-1-phosphate uridyltransferase activity tested during the summer months, data is being collected on the outcomes of these abnormal screens and possible factors that can be addressed to limit the number of false positive cases. Discussion about this issue was deferred until data can be collected for review.

Adrenoleukodystrophy:
Members were informed that a request from the parent of a child with adrenoleukodystrophy (ALD) has been received asking that this condition be considered for addition to the Illinois NBS test panel. This parent provided literature about possible testing and treatments for ALD. Members noted that ALD has been presented to the Secretary's Advisory Committee on Heritable Diseases in Newborns and Children (SACHDNC) for review for addition to the uniform panel of newborn screening disorders for all states. The SACHDNC requires rigorous review and documentation that is completed by professionals from many medical and scientific fields. With the formation of the Newborn Screening Expansion Subcommittee (NSES), of which Dr. Waggoner is the Chair, as a standing Genetics and Metabolic Diseases Advisory Committee (GMDAC) subcommittee, a protocol is in place for addressing requests for additions to the Illinois NBS test panel. The NSES protocol places heavy emphasis on SACHDNC decisions and recommendations, although requests for review and related materials may also be submitted to the NSES.

This final scheduled meeting of 2012 was adjourned at 10:10AM. Meetings for 2013 will be scheduled and members will be notified of proposed dates before year end.
Respectfully submitted, Barbara DeLuka