Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee
Conference Call Meeting Minutes: December 15, 2010

Subcommittee Members Attending:
Dr. George Hoganson-University of Illinois at Chicago
Dr. Barbara Burton- Children's Memorial Hospital
Dr. Gopal Srinivasan- Mt. Sinai Hospital
Dr. Joel Charrow- Children's Memorial Hospital
Dr. Darrel Waggoner- University of Chicago
Sunetra Reddy- University of Chicago
Kristin Clemenz-Children's Memorial Hospital

Guests:
Bob Evanosky

IDPH Staff:

Dr. David Jinks	Mike Petros
Tom Johnson	Claudia Nash
Heather Shryock	Angela McCauley
Margie Nelson	

The meeting was called to order at 10 am and introductions were made. The minutes of the May 26, 2010 meeting were approved.

New Business

Staffing and Laboratory Resources
One newborn screening laboratory staff person has been reassigned to the lysosomal storage disease (LSD) pilot screening initiative. Laboratory supervisors continue to assist with processing the mail on a daily basis. Changes to galactosemia testing planned for early 2011 should free up 0.5 FTE .

Three positions have recently been filled in the follow up section; two replacing positions that have been vacant for two years, and one position to provide follow up for LSD screening. (Note: subsequent to this meeting other staffing changes have occurred: two follow up positions will be vacant by March, due to one employee retiring and one employee accepting a different job).

Data System/Reports
The Perkin Elmer data system is now being utilized by both the laboratory and follow up components of the Newborn Screening Program. Basically the system is functioning well with minor adjustments still being made. Laboratory staff in the quality assurance unit continue to review all of the reports prior to issuance of the report to the hospital since there have been some discrepancies noted between the first and second page of the new laboratory reports. Since the laboratory reports are now much more complex than those generated by the former data system, many hospital laboratories are not entering all the information provided by IDPH into their own hospital data system. Some hospitals are scanning the second page of the IDPH report into their data system, where it is retained as a pdf document. It is anticipated that the turn around time from receipt of specimens at IDPH to provision of report to the

submitting hospital may be decreased from the current TAT of 10 working days using the new data system once the new data system is fully operational.

Two other features of the Perkin Elmer system are not yet on line which can improve timeliness and accuracy of test result availability by hospitals. These are the use of HL7 data exchange which would allow electronic upload of maternal and birth information from hospitals into the IDPH data system, and download of test results from IDPH to the hospital medical record. Northwestern Memorial Hospital, the largest birthing hospital in Illinois (1,200 births/month), is engaged in collaborative efforts with IDPH to implement HL7 exchange of information, then other hospitals can also be approached to utilize this model. The Perkin Elmer system also has an alternative Internet application "eReports" to improve the exchange of information between IDPH and hospitals. With eReports hospital staff can enter maternal and birth information directly into the Perkin Elmer system, which would eliminate the need for IDPH staff to perform this function and hopefully eliminate data entry errors due to handwriting interpretation. The eReport feature also would allow hospitals and physicians to obtain newborn screening test results for their patients by password protected access to the Perkin Elmer data system.

Lysosomal Storage Disorders Pilot

Since November 1, 2010 when the LSD pilot began, approximately 1,500 samples from Northwestern Memorial and the University of Chicago have been screened for Pompe, Fabry and Gaucher disease. Screening for Krabbe and Niemann Pick should be implemented early in 2011 for newborns from these two hospitals. Effective June 1, 2011 the state mandate requires all Illinois newborns to be screened for all five above named LSDs. So far during this pilot screening for LSDs. One case of Fabry has been diagnosed. It appears that several of the newborns with a positive screen for Gaucher are infants with very low birth weight. Additional data will be available for review at the next meeting of this subcommittee.

Galactosemia Screening

A change in galactosemia testing will be implemented early in the 2011, where the galactose level and the GALT enzyme will be tested on each specimen using a dual channel system. GALT will be reported as either present, reduced or absent, and approximately 3,000-4,000 samples have been tested so far using this system in preparation to roll out this new method. (NOTE: since this conference call, the exact date of this change has been established to occur February 1, 2011). It was noted that a modification has been made to clarify laboratory reports where the total galactose level was reported as elevated on page 2, but on page 1 it was reported that the test for galactosemia was normal. Now this category of report states that the galactose elevation is not clinically significant for classic galactosemia.

NICU Protocol for Newborn Screening

Currently the Illinois Newborn Screening Administrative Rules require all infants in the neonatal intensive care unit (NICU) receive a second newborn screening test at two weeks of age or at discharge, whichever occurs first, since cases of primary hypothyroidism have been reported in the literature as being missed in premature infants. The Clinical Laboratory Standards Institute (CLSI) has recommended requiring a third test at 30 days of life and some states have instituted this practice. Some states also use birth weight cutoffs to determine the optimal time of testing. It was stated that approximately 2% of all NICU admissions are for babies <25 weeks gestation or <1,500 grams, with these infants at higher risk of having a false negative newborn screen. There was a discussion about the need for changing the Illinois requirement and it was determined that additional statewide data are needed regarding the gestational age and weight of newborns admitted to the NICU to adequately make a recommendation

regarding this issue. Newborn Screening staff will contact the IDPH Perinatal Advisory Committee to obtain these data.

Other Business

Dr. Hoganson reported that Dr. Jinks, the Chief of the IDPH Newborn Screening Laboratory, will be retiring effective December 31, 2010. Dr. Hoganson thanked Dr. Jinks for his professional leadership and accomplishments during his tenure and noted that significant changes in newborn screening in Illinois have occurred under his direction.

Another meeting of the subcommittee will be planned prior to the March 31, 2011 meeting of the full Genetic and Metabolic Diseases Advisory Committee. This subcommittee will plan on meeting quarterly in February, May, September and November, with meetings to alternate between morning and afternoon to accommodate the schedule of all subcommittee members.

The meeting was adjourned at 11 am.