Illinois Department of Public Health
Genetics and Metabolic Diseases Advisory Committee
Newborn Screening Laboratory Subcommittee
Conference Call/Meeting Minutes: September 28, 2011

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

IDPH Staff:

George Dizikes	Claudia Nash	Tom Johnson	Shunna Johnson
Mike Petros	Barbara DeLuka	Margie Nelson	Dennis Tiburzi
Rong Shao	Heather Shryock	Angela McCauley	Laura Harris
John Nawrocki	Khaja Basheeruddin	Matt Charles	
Jennifer Crew	William Calvert		

The meeting was called to order at 9:05 AM, and introductions were made. The minutes of the May 18, 2011 meeting were approved.

New Business

Laboratory Staffing and Resources

Four clinical laboratory technologist I positions were posted and interviews were begun, but there is uncertainty about when actual hiring will be approved and the positions will be filled.

Data System/Reports

The Perkin Elmer laboratory data system continues to be adapted to meet the needs of hospitals and physicians, and in the near future the second page of the laboratory mailer will be printed whenever the results are abnormal or the specimen is declared unsatisfactory. Currently, only the one page mailer is routinely printed, but the second page with actual test values is available upon request. Both pages are provided routinely for hospitals that request the test values. Physicians may also request the complete report. Automated faxing of the laboratory reports is also now available for hospitals that request this service.

Preparations with Northwestern Memorial Prentice Women's Hospital to initiate HL7 demographic and test result data transfers between the hospital and the IDPH Newborn Screening Laboratory continue. Several other hospitals have expressed interest in developing the capacity for HL7 data transfers with IDPH lab, and are in communication with the system vendor and IDPH information technology staff.

Follow up program is also utilizing the new data system for all reporting of abnormal results and follow-up services, with exception of metabolic formula orders, which continue to be processed through the old NOMAD data system.

DNA Testing and Cystic Fibrosis (CF)

Dr. Nawrocki discussed the progress in securing new, more automated DNA testing equipment to reduce bench preparation time of samples by around 15-20 hours per week. This equipment will also be necessary to implement recently legislated addition of statewide screening for severe combined immune deficiency (SCID). There will also be a change in the cystic fibrosis DNA test panel due to State procurement and contract requirements. The CF specialists have been notified of changes in some of the alleles in the proposed new panel, and have not voiced any concerns about this necessary change. There has been a net gain in the total number of alleles included in the proposed Inplex DNA system, and implementation is expected this winter.

Galactosemia Screening

Dr. Petros provided data (see attached) on the galactosemia screening results, and changes to the reporting algorithm. While the introduction of the two channel process for testing both total galactose and galactose1-phosphate uridyltransferase (GALT) enzyme activity on all samples appears to be working well, initially there were concerns about the high number of borderline abnormal galactosemia screens (samples with normal total galactose and reduced GALT enzyme activity) detected and reported. In June during a conference call with several of the metabolic specialists and the Newborn Screening Laboratory and Follow-up staff, a new reporting algorithm was announced and discussed. The Newborn Screening Laboratory will continue to monitor galactosemia screening results and confirmed cases on a monthly basis, however, the new reporting algorithm appears to have greatly reduced borderline abnormal galactosemia screens from the high levels noted in May and early June 2011. A question was raised about the possibility of IDPH Molecular laboratory providing DNA testing for galactosemia, but this does not appear feasible at the present time. Other NBS laboratories have made similar observations during the warm weather months with regards to GALT enzyme testing.

Lysosomal Storage Disorders (LSD) Pilot

Dr. Dizikes reported that Dr. Basheeruddin and Dr. Shao have worked with CDC and received training on the tandem mass spectrometry method of screening for lysosomal storage diseases. CDC currently has controls for Krabbe, Gaucher, Pompe, Fabry, Niemann-Pick and MPS 1 (Hurler's disease), but at the present time, there is no substrate available for MPS 2 (Hunter's disease).

The next preparation step will be MS/MS testing for LSD's at IDPH and would most likely include testing of de-identified samples. Internal validation of the testing may be problematic, and without validation IDPH reporting of results on identified samples may conflict with Clinical Laboratory Improvement Amendment (CLIA) standards and practices. The need for informed consent and/or IRB approval for the LSD phase-in pilot was discussed in detail by the members.

It was felt that the University of Chicago and Northwestern Memorial Prentice Womens' Hospital would again be excellent sites for the LSD phase-in pilot, as specialists at these institutions would be accessible for referrals of newborns with positive LSD screens, and in the case of newborns diagnosed with Krabbe, stem-cell transplantation would also be available. There were questions about how abnormal screens for LSD would be reported, if informed consent for the phase-in pilot was obtained. Concerns about the need for full-time staff at these hospitals to meet informed consent requirements were voiced, as well as, concerns about the logistics of tracking samples and signed consent forms for any samples to be tested for LSD's. It was also discussed that these hospitals will require internal IRB review approval, prior to involvement in the phase-in process. Development of a reliable timeline for implementation of the LSD screening is difficult until the questions about the need for informed consent and institutional IRB approval for both the hospital and IDPH are resolved. It was decided that while consent for phase-in

pilot testing and IRB's have not previously been an issue for newborn screening, additional discussion of these issues at the Genetic and Metabolic Diseases Advisory Committee (GMDAC) will be necessary.

SCID Newborn Screening

The Administrative Rule (Section 661.30, I,3) for the addition of SCID, MPS 1 and MPS2 is currently in the second posting for comments period. Dr. Nawrocki indicated testing for SCID will provide quantification of TRECs, and lab staff have worked with materials available for internal controls and TREC quantification using five confirmed SCID blood samples from New York State. The lab will start to test CDC performance evaluation samples. Division of Laboratories is working with State Central Management Services to obtain the contracts for the high through-put DNA testing equipment needed for SCID screening. With this equipment the lab can process 384 samples in 2 hours time. The SCID pilot plan is to start with 4,000 identified samples which will be sent to another state that has successfully implemented SCID testing; Wisconsin is a likely candidate. SCID screening will also be performed by IDPH, however, the test results will be reported as determined by the other state lab. Both laboratory and follow-up staff continue to participate in monthly SCID Webinars provided by the Association of Public Health Laboratories.

Follow-up Report

Two additional public health specialist positions have been filled, and the new staff in training for follow-up services. Program staff are now working to verify demographic information on specimen cards with IDPH Vital Records data in an effort to provide physicians with more accurate and reliable contact information for families of babies with abnormal screens. They are also working more closely with the Department of Children and Family Services to assist in locating foster families and case managers for follow-up care, and with the IDPH birth defects registry to learn of children who may be diagnosed with newborn screening disorders that were not previously reported to the program.

NICU Newborn Screening

This discussion was based on the Clinical Laboratory Standards Institute (CLSI) national guidelines for newborn screening in the NICU. Data on the number of congenital hypothyroidism cases identified in low birth babies with normal initial newborn screens and the number of NICU admissions was previously provided (see attached). Additional information from the perinatal community on this topic was requested, but none was received. Dr. Zeller, a pediatric endocrinologist, and members agreed to provide a recommendation to the GMDAC to require collection of a routine third newborn screening sample from newborns with a birth weight less than 2,000 grams at 28 days of age, or prior to discharge from the NICU. This new recommendation will comply with CLSI guidelines. There were no objections voiced against this decision, although changes to the Administrative Rule for Newborn Screening will be necessary prior to any implementation of the new requirement.

Additional Business

Recently five late diagnosed cases of congenital adrenal hyperplasia were reported to the Follow up program and available information about these cases was discussed. All cases had normal initial newborn screens for 17-OHP levels, and it was acknowledged that all cases were non-salt wasting CAH, diagnosed clinically at ages from 4 months to 5 years. Efforts to encourage additional pediatric endocrinologists to join this Subcommittee will be made at an upcoming meeting of the Illinois Chapter of the Pediatric Endocrinologist Society. Follow-up program may also be participating in a Region 4 Genetic s Collaborative project that surveys physicians on treatment of congenital hypothyroidism and thyroid challenge testing at age 3 years. Efforts will be made to encourage closer connections with the

pediatric endocrinologists as their knowledge and expertise would be very beneficial to the Newborn Screening Program and this subcommittee.

The meeting was adjourned at 10:10 AM. The next meeting will be scheduled in early 2012.

Note: Information that Perkin Elmer may provide testing for six LSD disorders through their reference laboratory was received and e-mailed the members on October 4, 2011.

Respectfully submitted,
Barbara DeLuka
10/13/11