Illinois Department of Public Health (IDPH)
Genetic and Metabolic Diseases Advisory Committee
Newborn Screening Laboratory Subcommittee
Minutes – February 20, 2008
2121 W. Taylor St., Rm. 139E
Chicago, Illinois Dept. of Public Health

Subcommittee Members in Attendance
Dr. George Hoganson (Subcommittee Chair), University of Illinois at Chicago
Kristin Culp-Clementz, Children's Memorial Hospital
Audio Conference Attendance
Dr. Gopal Srinivasan, Mt. Sinai Medical Center
Sunetra Reddy, University of Chicago
Dr. Patrick Zeller, Pediatric Endocrinologist
Guest
Dr. Alexis Thompson, Children's Memorial Hospital
IDPH Staff
Dr. David Jinks, Newborn Screening Laboratory
Mike Petros, Newborn Screening Laboratory
Tom Johnson, Office of Health Protection
Claudia Nash, Genetics/Newborn Screening Program
Kate Seymore, Genetics/Newborn Screening Program
Barbara DeLuka, Genetics/Newborn Screening Program
Heather Gardner, Genetics/Newborn Screening Program

Call to order and introductions by Dr. Hoganson at 1:20 PM
Minutes of October 31, 2007 meeting were reviewed and approved.

IDPH Laboratory Report and Discussion
Dr. Jinks presented an analysis of all confirmed newborn screening disorders for the period July 2002 through January 2008. During this period 1,032,980 specimens were tested, 1,556 confirmed newborn screening disorders were reported, and just recently the first diagnosed case of argininase deficiency was confirmed.

Congenital hypothyroidism - Congenital hypothyroidism - The incidence of confirmed congenital hypothyroidism cases was discussed, and it was noted that during the period from 2003 through mid-February 2008, approximately one third of all confirmed transient and primary hypothyroidism cases were diagnosed following an initial borderline screening test in which the T4 level was within normal limits (8-23ug/dL) and the TSH level was less than 54 uIU/mL, but above the low-end cutoff. IDPH uses the TSH level as the primary screen for hypothyroidism, followed by T4 levels on all samples in the top 4% of TSH levels for the day's run of samples. Dr. Zeller offered to review TSH and T4 laboratory data for the Committee.
Note: Data was sent to Dr. Zeller and Dr. Hoganson on Feb. 21st, see attached summary.

Cystic fibrosis - CF phase-in screening was initiated January 28, 2008 for specimens received from 14 hospitals. Between January 28 and February 5, 16 positive screening

results were reported with these newborns referred for sweat testing; positive screens included one case with 2 CFTR mutations, one case with 1 mutation and 13 cases with an elevated IRT and no mutations. The timeline for the IRT and CFTR mutation testing is less than four business days after specimen receipt. Over 2,000 positive CF screens are anticipated each year with the current IRT cut-off of 100mg/mL as the fail safe for positive screens with no CFTR mutations detected.

Note: Subsequent to the meeting CF screening was instituted on March 3, 2008 for all samples received. Following review of data collected during the CF phase-in period, the fail safe IRT cut-off value for specimens with no CFTR mutations detected was increased to 150 mg/mL. It is anticipated that this revision will decrease the number of false positive CF screens, and be more in line with the CF screening protocols instituted by other state newborn screening programs.

Quality Assurance – Barbara DeLuka provided statewide data on specimen submission for review. Ninety percent of specimens are collected at 24-48 hours of age. Specimens collected after 48 hours of age appear to be retests, although the current data system does not allow for matching of specimens to confirm this. There was discussion about age adjusted cut-off values.

Lysosomal Storage Diseases – The sample preparation process required for Krabbe pilot testing in New York State was discussed. Dr. Jinks has been in contact with Joe Orsini in New York State Health Department and the Centers for Disease Control and Prevention (CDC) in preparation for future pilot lysosomal storage disease (LSD) screening. Mike Petros has been contacted by CMS approved contractors concerning the Newborn Screening Laboratory's projected needs for lab space, electrical, ventilation, plumbing and other mechanical requirements. Although no specific time frame to initiate the pilot screening is yet available, laboratory staff are reviewing the current resources and needs for space, equipment and staffing. It was noted by the group that no other state is pilot screening for more than one LSD, although New York is considering adding pilot screening for an additional LSD. CDC has established a manual assay, however, the sample preparation process needed for an automated high – throughput screening operation has not yet been developed.

Projected staffing needs for adding screening for five LSD disorders include six or more additional laboratory staff. Staff needs are high due to the intensive sample preparation and manual sample extraction requirements. Five additional sample plates for each specimen are required for each additional LSD. Five additional MS/MS instruments and five liquid handlers must also be purchased to allow universal screening for five LSD's using currently available experimental testing methodologies. Dr. Jinks indicated that purchase of one MS/MS instrument and one liquid handling system may be possible in the fall of 2008; if adequate laboratory space can be acquired, testing development for five LSD's could begin following acquisition of this equipment.

The Subcommittee discussed increasing the screening fee to finance additional equipment and staffing needs, and the necessary timelines for Administrative Rule changes allowing the fee increase. The general consensus was that the efforts to amend the Administrative Rule should begin in July 2008, with intent to increase the fee during the fall of 2009. It was acknowledged by the group that amendments to the Phenylketonuria Testing Act

stipulate LSD screening implementation in 2010, but must depend on the availability of a reliable methodology for high volume screening.

Alpha thalassemia – Mike Petros provided information on the incidence of hemoglobin screening results indicating possible alpha thalassemia; those samples with greater than 25% Bart's hemoglobin. From February to December 2007, three to four samples per month were indicative of possible alpha thalassemia. If possible alpha thalassemia cases are reported, new letters will be needed to report these results to primary care providers. Dr. Hoganson and the Subcommittee members present made a recommendation that the IDPH laboratory should begin reporting possible alpha thalassemia results as soon as possible. Claudia Nash indicated that new report letters and modifications to the database are needed; it is anticipated that these changes should be completed in one month. The newborn screening follow-up staff will develop report letters and a fact sheet for primary care providers. Kristin Culp-Clementz will assist staff with the fact sheet and letters.

No additional business was presented and the meeting was adjourned at 2:40 PM. The next scheduled meeting is set for May 21, 2008 from 9AM to 11AM.

Minutes prepared by Claudia Nash and Barbara DeLuka 3/6/08