Illinois Department of Public Health (IDPH)
Genetic and Metabolic Diseases Advisory Committee
Newborn Screening Laboratory Subcommittee
Minutes – April 1, 2009
2121 W. Taylor St., Rm. 139E
Chicago, Illinois

Subcommittee Members in Attendance
Dr. George Hoganson (Subcommittee Chair), University of Illinois at Chicago
Dr. Barbara Burton, Children's Memorial Hospital
Attendance by Audio Conference
Sunetra Reddy, University of Chicago
Dr. Gopal Srinivasan, Mt. Sinai Neonatology

IDPH Staff
Dr. David Jinks, Newborn Screening Laboratory
Mike Petros, Newborn Screening Laboratory
Attendance by Audio Conference
Barbara DeLuka, Genetics/Newborn Screening Program
Heather Gardner, Genetics/Newborn Screening Program
Claudia Nash, Genetics/Newborn Screening Program
Kate Seymore, Genetics/Newborn Screening Program

Call to order by Dr. Hoganson at 9:15 AM
Introductions
Minutes of November 5, 2008 meeting were approved.

IDPH Laboratory Report and Discussion
Dr. Jinks discussed laboratory results and confirmed cases for the period July 2002 through February 2009*. During this period 1,234,269 specimens were tested with a combined incidence of one diagnosed disorder for every 644 specimens. No unusual findings were noted. Since March 2008, when screening for cystic fibrosis was first implemented, 36 infants have been diagnosed with CF.
*See attached report "Confirmed Case Summary".

Dr. Jinks reported that the new laboratory database nears completion; lab instruments are currently downloading test data into the new system, and phase two testing is nearing completion. Testing of the new Follow-up Program data system has encountered delays and technical problems, and currently high level contractor discussions with the system sub-contractor are addressing these concerns. The follow-up data system is not ready for implementation and continued use of the mainframe laboratory system will be required for the interim.

Funding for development of LSD screening was discussed. Grant opportunities for IDPH laboratory participation with Advanced Liquid Logic Inc. in development of more efficient, less expensive micro-fluidic fluorometric assays for LSD screening may become

available in the near future. Screening assays for Fabry, Gaucher and Pompe have been demonstrated, and assays for Krabbe and Niemann-Pick disorders are under development by the company. If screening for Krabbe utilizing this type of fluorometric assay proves unfeasible, testing for this LSD disorder by tandem mass spectrometry (MS/MS) is still an option. The potential benefits of LSD screening with micro-fluidic fluorometric assay include fewer equipment and staff requirements and improved turn-around time for the testing, potentially making this testing system more cost effective. Dr. Jinks will remain in contact with CDC researchers, Dr. David Millington of Duke University and Advanced Liquid Logic representatives regarding this possible approach to LSD screening.

Discussion about the time line for LSD screening start-up recognized the need to begin pilot screening using anonymous samples by December 2010. Pilot testing for LSD's will be initiated at Northwestern Memorial and University of Chicago hospitals. Use of micro-fluidic fluorometric assays would eliminate the need for large increases in laboratory personnel, equipment and building renovations required by LSD screening methodologies utililizing MS/MS.

Personnel concerns were discussed; the laboratory has two vacant positions and Follow-up program has three vacancies, with one position currently posted for hiring.

Dr. Jinks informed members about recent developments in severe combined immune deficiency (SCID) testing. Baxter Healthcare Corporation, which is based in Illinois, has expressed some interest in assisting development of pilot newborn screening for SCID.

Members discussed the progress of the Health and Human Services Secretary's Advisory Committee with regard to review of disorders for addition to a uniform newborn screening panel. SCID was not added, a decision possibly related to failure of current pilots to detect a true SCID case, although the testing has detected cases of DiGeorge Syndrome. Pompe was also not yet approved, which may be related to the lack of published studies within the United States; only foreign pilot testing results are currently available. Krabbe will be further reviewed at the next Secretary's Advisory Committee meeting.

The Clinical Laboratory Standards Institute (CLSI) proposed guidelines for newborn screening in the NICU were discussed. Comparison of current Illinois NICU guidelines to the CLSI proposed ones, and those of surrounding states will continue, and Follow-up staff will report on the incidence of specimen collection after transfusion, a major concern for newborn screening in the NICU. No changes to the current guidelines were recommended by members at this time.

Other Discussion

There was discussion regarding detection of folate/cobalamin disorders and ornithine transcarbamylase disorder on MS/MS screening, which Dr. Jinks will research and address.

The next Subcommittee meeting is scheduled for July 22, 2009 from 9 to 11 AM.
The meeting was adjourned at 10:20 AM.

Addendum:
Newborn Screening Administrative Rule changes regarding LSD screening pilot testing and fee increases were approved by the State Board of Health and submitted to the Governor's Office for approval prior to being sent the Joint Committee on Administrative Rules (JCAR).

Minutes respectfully submitted by Barbara DeLuka 4/21/09