Illinois Department of Public Health (IDPH)
Genetic and Metabolic Diseases Advisory Committee
Newborn Screening Laboratory Subcommittee
Minutes – November 5, 2008
2121 W. Taylor St., Rm. 139E
Chicago, Illinois

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

IDPH Staff
Dr. David Jinks, Newborn Screening Laboratory
Tom Johnson, IDPH Office of Health Protection
Mike Petros, Newborn Screening Laboratory
Attendance by Audio Conference
Barbara DeLuka, Genetics/Newborn Screening Program
Heather Gardner, Genetics/Newborn Screening Program
Kate Seymore, Genetics/Newborn Screening Program

Call to order by Dr. Hoganson at 9:00 AM
Introductions
Minutes of August 27, 2008 meeting were reviewed and approved.

IDPH Laboratory Report and Discussion
Dr. Jinks discussed laboratory results and confirmed cases for the period July 2002 through September 2008*. During this period 1,160,813 specimens were tested with a combined incidence of one diagnosed disorder for every 647 specimens. No unusual findings were noted. Since the implementation of screening for Bart's hemoglobin in July, four cases of alpha thalassemia have been diagnosed. Since cystic fibrosis screening was started in March, 25 infants have been diagnosed with CF. Galactosemia screening now includes Beutler assay of all specimens from babies NPO at the time of specimen collection, which has increased the number of specimens with reduced GALT enzyme activity requiring follow-up services. *See attached report "Confirmed Case Summary".

Dr. Jinks reported on progress with the laboratory database, and beta testing of the system is expected to begin later this month. The new data system will eventually allow hospitals direct access to test results and provide automated downloading of demographic data through HL-7 compatible systems.

There was discussion about funding for resources needed to develop LSD screening tests and laboratory protocols. CDC is still working on test development, and a private

corporation has recently published research papers on LSD screening. Dr. Jinks will remain in contact with CDC researchers.

Dr. Hoganson indicated the March meeting of the American College of Medical Genetics plans topics including, ethics, informed consent, and use of residual dried blood samples. A session on the addition of lysosomal storage disease screening to the Illinois newborn screening test panel is also planned.

There was discussion about the Genetic and Metabolic Diseases Advisory Committee (GMDAC) meeting, and future plans for the Newborn Screening Laboratory Subcommittee. The members were informed of the GMDAC Advisory Committee recommendation that responsibility for development of any proposed guidelines and/or criteria for the addition of new disorders to the newborn metabolic screening panel should be delegated to this Subcommittee.

Other Discussion

Members asked questions regarding the IDPH requirements for use of residual dried blood spots. Concerns regarding use of samples including, follow-up for the patient if an abnormality is detected and the need for informed consent, must be addressed in any request applications. Current IDPH policy requires disposal of residual samples after three months. To date, no residual samples have been released for research. Formal applications must include an IRB approval and must be submitted to the IDPH Data Research and Release Committee (DRRC). DRRC approval may include review by the IDPH institutional review board. Key concerns of the DRRC include whether the samples are identified or de-identified. The application must address specimen data and demographics, provide a case for use and indicate a public health research impact. Following review by the DRRC, a memo of agreement between the researcher and IDPH are signed that indicate the specifics of the transfer of material and address publication rights. Any IDPH concerns about use of the specimens or the proposal must be addressed by the researcher. Subcommittee members recognized the potential public health research benefits for use of de-identified residual blood samples.

There was discussion regarding developments in testing for severe combined immune deficiency disorders (SCID). Meetings with CDC and Dr. Morris Kletzel regarding SCID testing are anticipated in November and December.

Dr. Hoganson asked members if the current schedule of alternating morning to afternoon meetings every three months was agreeable to the group, and there were no objections. Dr. Hoganson will contact Dr. Charrow for advice regarding the addition of new members to the Subcommittee, and to discuss future considerations for the Subcommittee, including review of residual specimen use and development of a protocol for consideration of new diseases to be added to the newborn screening panel.

The meeting was adjourned at 10:15 AM.