

Illinois Department of Public Health  
Genetic and Metabolic Disease Advisory Committee  
Minutes—April 30, 2009  
William Tell Holiday Inn, Countryside, IL

Members Present:

Joel Charrow, M.D., Chair, Children's Memorial Hospital  
Darrel Waggoner, M.D., University of Chicago  
Susanna McColley, M.D., Children's Memorial Hospital  
W. Patrick Zeller, M.D., Pediatric Endocrinologist  
Karen Litwack, L.C.S.W., Chicago Center for Jewish Genetic Disorders  
Cathy Gray, R.N., University of Chicago  
Carla Gillespie, R.N., Madison County Health Department

Other Attendees:

Barbara Burton, M.D., Children's Memorial Hospital  
Alan Horwitz, M.D., University of Illinois at Chicago  
Alexis Thompson, M.D., Children's Memorial Hospital  
Jose Arrom, UIC Midwest Latino Center  
Pam Borchardt, SIDS of Illinois  
Cathy Wicklund, Genetic Counselor  
Rich Dineen, Genetic Counselor  
Sean Clark, Parent  
Lauren Pena, M.D.  
Gerri Clark, R.N., DSCC  
David Jinks, Ph.D., IDPH Newborn Screening Laboratory  
Mike Petros, IDPH Newborn Screening Laboratory  
George Dziekes, Ph.D., IDPH Laboratories  
Claudia Nash, IDPH Newborn Screening Program Administrator  
Karen Burget, R.N., IDPH Genetics/Newborn Screening Program  
Barbara DeLuka, R.N., IDPH Genetics/Newborn Screening Program  
Kathryn Seymore, IDPH Genetics/Newborn Screening Program

The meeting was called to order by Dr. Charrow at 9:40 a.m. The minutes of the September 18 meeting were approved. Claudia explained that this would be a short meeting because of today's Region 4 site visit to Illinois, which will start immediately after this meeting, at 10:30 a.m. There will be reports throughout the day, a working lunch, and a meeting for families only at 3:00 p.m.

Claudia also reported that there are now 10 vacancies on this committee, with 9 nominations waiting at the Governor's office. There are forms in the meeting packet to nominate a hospital representative.

Dr. Jinks provided a handout showing the abnormal results for 1.2 million newborn screenings, including MS/MS and the legacy disorders. These results continue to be about as expected for the various disorders. So far, only one case of Hemoglobin H Disease (Bart's) has been diagnosed since testing began last summer.

The newborn screening program currently has three vacant follow-up staff positions. Claudia reported that she just received a grant announcement from the CDC for a sickle cell surveillance project, and will probably submit an application. The first year of testing for cystic fibrosis has gone well, with genetic counseling being provided in most centers at the time of the sweat test. IDPH is conducting further review of these procedures. Budget issues continue to be a problem, with \$500,000 recently swept from the Metabolic Screening Fund. There have been delays with development of the follow-up portion of the newborn screening data system, due to problems with the subcontractor. The primary vendor, Perkin Elmer, has terminated the subcontractor, and will finish the project with their staff.

Dr. Charrow proposed that this Committee draft a letter addressing the budget issues and fund sweep to the Governor and IDPH Director.

There was no Laboratory Subcommittee report. Minutes of the last meeting of that subcommittee are included in the meeting packet.

Dr. Jinks reported that he has applied for a grant for an alternative testing method for lysosomal storage disorders (LSDs). He hopes to start a pilot for 3 of the 5 disorders in the fall with samples from Northwestern/Prentice and the University of Chicago. This method is less expensive than MS/MS, requires fewer staff to run, and has a shorter turn-around time.

The LSD rules have been approved by the State Board of Health and were sent to the Governor's Office April 3. The six month pilot is scheduled to begin in November, 2010.

There was some discussion of the need to make plans for follow-up diagnostic testing for LSDs, therefore, a meeting of the ad hoc LSD subcommittee will be scheduled in the upcoming weeks. Protocols could be based on the New York program, at least for Krabbe. There was a question if DNA testing to be done by the state lab was included in the proposed NBS fee increase.

The meeting was adjourned at 10:40 a.m.

Minutes prepared by K. Seymore

