

Illinois Department of Public Health  
Genetic and Metabolic Disease Advisory Committee  
Minutes—April 29, 2010  
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  
Michael Schneider, M.D., SIU School of Medicine  
George Hoganson, M.D., University of Illinois at Chicago  
Praveen Kumar, M.D., Northwestern University Medical School  
Lainie Freidman-Ross, M.D., Ph.D., University of Chicago Hospital  
Barbara Goss, R.D., University of Illinois at Chicago  
Sean Clark, Parent  
Margaret Westemeyer, M.S., Northwestern University Hospital  
Alexis Thompson, M.D., Children's Memorial Hospital

Other Attendees:

Barbara Burton, M.D., Children's Memorial Hospital  
Pam Borchardt, SIDS of Illinois  
Tess Rhodes, R.N., Division of Specialized Care for Children  
Talana Hughes, Sickle Cell Disease Association of Illinois  
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  
Barbara DeLuka, R.N., IDPH Genetics/Newborn Screening Program  
Kathryn Seymore, IDPH Genetics/Newborn Screening Program  
Brooke Croke, IDPH intern, Genetic Counseling Student

The meeting was called to order at 10:15 a.m. Following introductions, the minutes of the October 29, 2009 meeting were approved.

Laboratory Subcommittee

Dr. Hoganson reported on the Newborn Screening and Laboratory subcommittee meeting that was held on January 6, 2010. The minutes are included in the meeting packet. Items discussed at that meeting were the 2 cases of low citrulline that were detected and change in the cystic fibrosis molecular testing kits. The subcommittee also discussed changing the cut-offs for

galactosemia to detect more cases of GALK and GALE. There was discussion about the 14-day repeat newborns screens for babies in NICU, and whether to continue this or to change to 28 or 30 days, as is done in some states. More information will be collected from other states for further discussion of this at the next subcommittee meeting. It was suggested that there be a separate ad hoc committee formed to discuss creating a formal system for adding disorders to the newborn screening panel, such as SCID, and a policy for the retention of blood cards for future use.

### LSD Subcommittee

The metabolic specialists have developed follow-up protocols for the five lysosomal storage disorders that will be added in November. This group will discuss data collection at the next meeting in October before the pilot starts in November. The state of Missouri is preparing to test for the same 5 LSDs, and New Mexico and New York are expected to follow. Informational materials for these disorders are being developed by a committee of the NIH.

### LSD Screening Update

Dr. Jinks reported that the delivery of the instrument from Advanced Liquid Logic (ALL) had been delayed until next month because of issues with a formal agreement between IDPH and ALL. The IDPH lab will start a pilot project with three of the disorders. Also, it has been decided that DNA screening for Krabbe will be done outside the state laboratory.

### Cystic Fibrosis Update

Dr. McColley gave a report on several aspects of cystic fibrosis testing, including a project conducted by a genetic counseling student to determine the knowledge of parents about cystic fibrosis and the appropriate time for provision of genetic counseling for parents about the disease. At this time, there are no known missed cases of CF during the first two years of newborn screening. The false positive rate is about as expected. It was also explained that DSCC policy for coverage of CF care is that annual follow-up must be done at an accredited CF center.

### IDPH Newborn Screening Laboratory

It was noted that there have been a significant number of confirmed cases with borderline initial newborn screening results. There were two cases of tyrosinemia with found medically that had negative initial NBS results. There were two cases of organic acid disorders an MMA and a 2MBCD with initial normal screening results, which are to be investigated, and a large number of 'unknown variant galactosemias'. It was suggested that the program change their diagnosis information forms which are sent to specialists, in order to get more precise diagnostic information and perhaps improve compliance with completing these forms. Also, it is assumed that there have been more than the three known hemoglobin H confirmed cases, probably because primary care providers are not following up with DNA testing on these babies.

With regard to the Perkin-Elmer database, the lab is entering some samples into both the current and new databases. The lab intends to go live with the Perkin-Elmer system July 1, with the follow-up staff ready by August 1. Ultimately, electronic exchange of data with hospitals/providers and IDPH will be possible.

#### Newborn Screening Follow-up

Information was presented regarding the timing of laboratory receipt of specimens from hospitals. Ninety-eight percent of specimens are now received within 5 days of collection. There was a question as to whether 100% of newborns are tested. This is not known, however, there are only 5 known babies born in 2009 that were not tested.

#### SIDS Program

A handout was provided that shows a dramatic decrease in SIDS deaths over the years. It was also reported that legislation was just passed for all hospitals to provide SIDS education to new parents and/or caregivers.

#### Genetics Grants

Claudia gave a brief overview of the grant program. Karen Burget, who is in charge of about 60 of the grants, is going to be retiring at the end of the year. The genetics conference was cancelled for this year, however, genetics staff organized two school health conferences for nurses, which were well received. They also provided information to genetic counselors and health departments about family health history in conjunction with DNA day. The level of funding for FY2011 is not known, but it is expected that funding for the State Genetics Plan grants will not be renewed. Kate Seymore, who takes care of the sickle cell grants, will also be retiring this year.

Further discussion included the following:

Genetics Program/NBS Follow-up Program will be moving in June, and will also begin validation testing the of the Perkin-Elmer system in June. They also need to update fact sheets, practitioner's manual, brochures to include the LSDs. There are also still two staff vacancies.

There is one vacancy on the Genetics and Metabolic Screening Advisory Committee. Nominees must undergo a background check prior to being appointed.

The Sickle Cell Disease Association of Illinois is continuing its work with Federally Qualified Health Centers for adults with sickle cell disease to promote the medical home concept. They also still receive referrals for all newborns with trait from IDPH.

A new Director has been appointed at DSCC.

The meeting was adjourned at 1:50 p.m. The next meeting will be September 30 in Springfield.

Minutes prepared by Kate Seymore