

Illinois Department of Public Health  
Genetic and Metabolic Disease Advisory Committee  
Minutes—September 30, 2010  
Hilton Garden Inn, Springfield, 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  
Michael Schneider, M.D., SIU School of Medicine  
Lainie Freidman-Ross, M.D., Ph.D., University of Chicago Hospital  
Cathy Gray, R.N., University of Chicago  
Margaret Westemeyer, M.S., University of Illinois at Peoria  
Cathy Wicklund, M.S., Northwestern University Hospital  
Colleen Gibson, R.N., LaSalle County Health Department  
Sheila Chalmers-Currin, Parent

Other Attendees:

Pam Borchardt, SIDS of Illinois  
Tess Rhodes, R.N., Division of Specialized Care for Children  
Amy Walsh, Parent  
David Culp, Ph.D., IDPH Deputy Director  
Tom Schafer, IDPH Deputy Director  
Tom Johnson, Division Chief IDPH Laboratories

IDPH Newborn Screening Program:

Claudia Nash, Barbara DeLuka, Karen Burget, Kathryn Seymore, Nikki Woolverton, Heather Gardner, Marie Nagle, Debbie Box, Marti Williams, Denise Kink, Doug Hart, Ben Layne, Margie Nelson, Angela McCauley

The meeting was called to order at 10:30 a.m. Following introductions, the minutes of the April 29, 2010 meeting were approved.

Newborn Screening Laboratory Subcommittee

Barb DeLuka reported on the meeting that was held May 26. The minutes are included in the meeting packet. There was discussion of certain diagnosed cases, the change in CF test kits, and the change in the alpha thalassemia testing method. Also, regarding newborn screening for babies in the NICU, the repeat testing on day 14 of life may be changed to day 28. This would require a change in the IDPH Administrative Rules.

Newborn Screening Expansion Subcommittee

Dr. Ross reported on the first meeting of this subcommittee. There are several ways to approach expanding the newborn screening panel. These include the recommendations of the Secretary's Advisory Committee, exploring evidence-based research information, and looking at what is being discussed in other states. This Subcommittee would accept addition interested members and will select a chair at the next meeting.

#### Other Discussion Regarding Expansion

Following the report of the Newborn Screening Expansion Subcommittee, there was discussion by many Committee members about changes to the testing panel including also periodically reviewing the existing panel to determine if, over time, any condition should be removed from the screening panel. Dr. Ross also suggested that perhaps the Expansion Subcommittee should reconsider some of the proposed LSDs to be included, and add or remove certain disorders, based on more recent data that is available.

#### Cystic Fibrosis NBS Collaborative

This group consists of the 15 CF centers in Illinois. Dr. McColley explained the current protocol for follow-up of positive newborn screening results for CF. Genetic counseling is provided to 100% of families at 11 of the sites. The QNS percentage is 9% overall, which is acceptable by national standards. The goals of the Collaborative are to overcome barriers to genetic counseling and to timely sweat testing of infants, thereby improving the quality of services to families.

#### Lysosomal Storage Disorder Subcommittee

Dr. Charrow summarized a report on this subcommittee prepared by Dr. Burton, which is included in the meeting packet. The Illinois pilot screening program for the LSDs is to begin November 1 at two hospitals. Missouri, New Mexico and possibly New York are preparing to add LSDs to their newborn screening panels. Illinois laboratory staff currently are being setting up testing using Advanced Liquid Logic instrumentation for two of the disorders. The American College of Medical Genetics is developing ACT sheets for the LSDs, which will be available on the Internet. The Illinois lab will not be doing DNA sequencing for Krabbe; this will be done by the New York state laboratory, which has been testing for Krabbe disease for 4 years. There was some discussion about the findings from the New York state Krabbe screening program.

#### Newborn Screening Follow-Up Program

Barb DeLuka provided data on confirmed and presumptive positive cases for calendar year 2009 (included in packet). The packet also includes samples of the new reports that are generated by the Perkin Elmer data system. The specimen submission report shows that 77% are received by the lab within 3 days, and 98% within 5 days, with 99.8% of specimens being of satisfactory quality.

#### SIDS Program

Nikki Woolverton reported on training activities being conducted for families and professionals, in conjunction with SIDS of Illinois.

## IDPH Genetics Grants

The FY 2011 grant applications are being processed and will be reviewed by a designated committee. The genetic counseling and sickle cell grants were restored to their FY 2009 funding levels, after a decrease last year, and all grant reimbursements will be retroactive to July 1.

## Newborn Screening Program Update

Claudia Nash briefly summarized current activities of the Genetics/Newborn Screening Program, as follows: looking at a different method of formula distribution; providing NBS information for new mothers online, rather than printing brochures; validating the new data system, which is to go live November 1; reporting all new confirmed cases from newborn screening to the IDPH birth defects registry; continued collaboration with Region 4, the Sickle Cell Disease Association of Illinois, the Illinois Sickle Cell Action Network and the Chicago Center for Jewish Genetic Disorders. Ms. Nash attended a conference on LSDs in Kansas City September 29, at which Dr. Duffner from New York was the keynote speaker. Dr. Duffner has agreed to speak at the next meeting of the Illinois Genetics Advisory Committee March 31, 2011.

The Secretary's Advisory Committee on Heritable Disorders recently added Severe Combined Immunodeficiency Disorder (SCID) and Critical Cyanotic Congenital Heart Disease (CCCHD) to the list of disorders recommended to be included in each state newborn screening panel.

Amy Walsh, parent of a child with SCID, told the story of her son, who is 10 years old. He is now doing well after a bone marrow transplant from his brother.

There is still a vacant position on the Committee that needs to be filled.

It was suggested that this Committee give consideration to the issue of dried blood spot storage and usage. This has become a nationwide issue, as these could be used for research purposes. The Newborn Screening Laboratory Subcommittee will address this issue and Cathy Wicklund offered to have a genetic counseling student summarize how this is being addressed in other states.

Dr. Charrow acknowledged the upcoming retirement of IDPH staff Karen Burget and Kate Seymore.

The meeting was adjourned at 1:55 p.m.