

Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee
Minutes—March 31, 2011
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
Alexis Thompson, 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
George Hoganson, M.D., University of Illinois at Chicago
Cathy Wicklund, M.S., Northwestern University
Praveen Kumar, M.D., Northwestern Memorial Hospital
Colleen Gibson, R.N., LaSalle County Health Department
Michael Msall, M.D., University of Chicago

Other Attendees:

Barbara Burton, M.D., Children's Memorial Hospital
Ramsay Fuleihan, M.D. Children's Memorial Hospital
Pam Borchardt, SIDS of Illinois
Tess Rhodes, R.N., Division of Specialized Care for Children
Megan Hinton, President, Genetics Task Force of Illinois
Luna Okada, St. Alexius Medical Center
Amy Walsh, Parent
Zena Berryhill, Parent
Mike Petros, IDPH Newborn Screening Laboratory
Rong Shao, M.D., IDPH Newborn Screening Laboratory
George Dizikes, Ph.D., IDPH Laboratories
David Culp, Ph.D., IDPH Deputy Director
Tom Schafer, IDPH Deputy Director
IDPH Newborn Screening Follow up Program:
Claudia Nash, Heather Gardner, Nikki Woolverton, Ben Layne, Margie Nelson

Members Not Present:

Sheila Chalmers Currin
Sean Clark
Barb Goss
Karen Litwack
Albert Olorvida, M.D.
John Ralston, M.D.
Maggie Westermeyer
W. Patrick Zeller, M.D.

Introductions/Comments

The meeting was called to order at 10:10 a.m. Following introductions, the minutes of the September 30, 2010 and February 7, 2011 meetings were approved. Claudia Nash reported the term of seven committee members expired in February, but they will be reappointed since they have completed only one term.

Dr. Charrow announced that two committee members, Dr. Thompson and Cathy Wicklund have both recently been appointed to the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.

A paper was included in the handouts, entitled "Limitations of Universal Newborn Hearing Screening in Early Identification of Pediatric Cochlear Implant Candidates" that was published in the March 2011 issue of the *Archives of Otolaryngology-Head and Neck Surgery*. This paper presents information regarding newborn hearing screening in Illinois and will be discussed further at the next meeting with a representative from the IDPH newborn hearing screening program.

Dr. Burton addressed a publication also included in the handouts regarding screening for Critical Congenital Heart Disease (CCHD), which the Secretary's Advisory Committee on Heritable Disorders added in September 2010 to the list of disorders recommended for inclusion in each state newborn screening panel. Committee members discussed how screening could be implemented in Illinois. This issue will be referred to the Newborn Screening Expansion Subcommittee for further discussion.

SCID Screening Update

Staff reported that IDPH is in the process of amending the newborn screening administrative rules to add testing for Severe Combined Immunodeficiency (SCID). It is anticipated that the administrative rule change could be approved by January 2012 and that the proposed fee will likely increase by \$10 from \$78 to \$88, which is well within the range for what other states charge for newborn screening. Legislation, Senate Bill 1761, has also been introduced to add SCID to the Illinois newborn screening panel. (*Update: SB 1761, which also mandates the addition of newborn screening for MPS I and II passed both houses and is currently awaiting the Governor's signature*).

Laboratory staff indicated that IDPH will use the TREC method of testing, which is the same method as other states screening for SCID. It is important to note that DSCC will not cover treatment for SCID as this disorder does not meet their eligibility criteria. Committee members discussed the need for a coordinated approach in implementing screening and follow up for SCID.

Newborn Screening Laboratory Subcommittee

The last meeting of this subcommittee was held December 15, 2010. The IDPH laboratory is now testing galactose and GALT enzyme levels on all newborn screening samples.

A report by the Clinical Laboratory Standards Institute has recommended a third newborn screening sample be collected at 30 days of life to increase the detection of hypothyroidism. Currently, the Illinois newborn screening administrative rules require all infants in the NICU receive repeat testing at two weeks of age or upon discharge. The American Academy of Pediatrics Committee on the Fetus and Newborns is considering the recommendation, and this IDPH Subcommittee is gathering additional data on NICU admissions. A change in the existing policy in Illinois would require a revision in the IDPH newborn screening administrative rules.

Newborn Screening Expansion Subcommittee

The last meeting of this subcommittee was held March 9, 2011. The draft proposal for addition of new tests to the Illinois newborn screening panel was reviewed in detail. After discussion, the full Committee voted to approve the proposed protocol with the following four changes: 1) in addition to the core members, additional subcommittee members can be added as needed to review specific disorders; 2) the Newborn Screening Expansion Subcommittee will be added as a permanent standing subcommittee of the Genetic and Metabolic Diseases Advisory Committee, with the bylaws to be amended to reflect this addition; 3) the language in the draft proposal will be changed to remove the requirement that any new test be approved by the FDA and state that the screening tests must be validated by standard laboratory procedures; and 4) proposals for additions to the Illinois screening panel will be submitted to the Chair of the Advisory Committee, who will assign the proposal to this subcommittee for consideration, and notify members of the parent committee. The draft protocol will be revised to reflect these changes and will then be sent to all committee members. It was also stated that since there now exists a standardized, formalized process for making changes to the Illinois newborn screening panel, this process should be the preferred manner utilized for making changes, as opposed to legislation.

LSD Ad hoc Subcommittee

Dr. Burton reported on the pilot screening for LSDs which began November 1, 2010. There have been 5,266 infants screened. One infant screened positive for Pompe and was later classified as normal. There were seven positive screens for Fabry, and three were confirmed. There have been several false positives for Gaucher, mostly restricted to low birthweight, NICU infants. Only four were positive in non-NICU babies. Review of further clinical data related to babies with a positive screen for Gaucher was suggested to determine if other factors related to feeding or antibiotics could be affecting the screening results. Dr. Ross indicated the need for informed consent since further research is being suggested to explain screening findings. The next meeting of this subcommittee will be held in early May.

Cystic Fibrosis Collaborative

Dr. McColley summarized the handout given regarding the CF Newborn Screening Collaborative for April-December 2010. The overall inadequate sweat test rate is 12%, which is somewhat above the recommended national standard of 10%. Collaborative members are sharing best practices among centers to assure timely sweat testing, provision of genetic counseling to all families and to reduce QNS rates. The Illinois Collaborative is also submitting an application to the national Cystic Fibrosis Foundation for Quality Improvement grant funding.

IDPH Newborn Screening Laboratory

Mike Petros provided an update from the newborn screening lab, and abnormal screening data were presented for the period of July 2002 – June 2010. Dr. Rong Shao is responsible for implementing the methodology for LSD testing. Testing for Krabbe and Niemann-Pick have been delayed due to research issues with the vendor, Applied Liquid Logic (ALL), but they anticipate possibly being ready to add these two disorders in May 2011. The current testing format used by ALL is a 10 channel plate, however they will be changing to a 40 channel plate in the future. Additional laboratory space and testing instruments are needed to increase the capacity for LSD testing. As of February 1, 2011, the IDPH Lab initiated testing all samples for total galactose and transferase, which seems to be going well so far.

Newborn Screening Follow-Up Program

Three vacant positions have been filled recently in the follow up program, and these staff are being trained. The Perkin Elmer database is now being used by the follow up staff and the reports to physicians have been changed to a much clearer format. The administrative rule language changes to add testing for severe combined immunodeficiency have been drafted for internal review.

SIDS Program

IDPH is providing a grant to SIDS of Illinois, Inc. to assist in offering bereavement services to families and training activities for health care professionals. The Co-Executive Director of SIDS of IL, Pam Borchardt, reported that legislation requiring Illinois birthing hospitals to provide all families of newborns with information about safe sleep practices is now in effect and SIDS of IL has developed a program to provide hospitals with this information. Other legislation is pending which will amend the Child Care Act of 1969 to require all licensed child care facility employees that care for newborns and infants to complete training every three years on the nature of sudden unexpected infant death, sudden infant death syndrome and the safe sleep recommendations of the American Academy of Pediatrics.

IDPH Genetics Grants

The FY 2011 grant applications have been processed and were reviewed by a designated grant review committee. The genetic counseling and sickle cell grants were restored to FY 2009 funding levels, after a decrease last year, and all grant funding will be retroactive to July 1, 2010 even though there has been a delay getting the contracts in place. Funding is also being provided to the Chicago Center for Jewish Genetic Disorders to coordinate statewide educational activities for DNA Day in April. Activities this year include activities and presentations by genetic counselors in health departments, colleges and high schools in many Illinois communities.

Newborn Screening for Krabbe Disease – the New York Experience, Patti Duffner, M.D.

Dr. Duffner gave a presentation about screening for Krabbe Disease in New York, the only state currently screening for Krabbe.

Adjournment

The meeting was adjourned at 2:40 p.m. The next meeting will be held October 27, 2011 in Springfield.