

Illinois Department of Public Health  
Genetic and Metabolic Disease Advisory Committee  
Minutes—February 7, 2011  
Teleconference Call

Members Present:

Dr. Joel Charrow, Chair, Children's Memorial Hospital  
Dr. Darrel Waggoner, University of Chicago  
Dr. Susanna McColley, Children's Memorial Hospital  
Michael Schneider, M.D., SIU School of Medicine  
Lainie Freidman-Ross, M.D., Ph.D., University of Chicago Hospital  
Colleen Gibson, R.N., LaSalle County Health Department  
Alexis Thompson, M.D., Children's Memorial Hospital  
George Hoganson, M.D., University of Illinois at Chicago Hospital  
Patrick Zeller, M.D., Pediatric Endocrinologist-Wheaton  
Karen Litwack, Chicago Center for Jewish Genetic Disorders  
Barb Goss, University of Illinois at Chicago Hospital  
Sean Clark, Parent

Other Attendees:

Dr. Barbara Burton- Children's Memorial Hospital  
Bob Evanosky, the Evanosky Foundation  
Dr. Ramsay Fuleihan, Children's Memorial Hospital  
Gerri Clark and Tess Rhodes, Division of Specialized Care for Children  
Amy Walsh, Parent  
Dr. Naga Chadaram, Advanced Liquid Logic

IDPH Newborn Screening Lab

Dr. David Culp, Ph.D., IDPH Deputy Director  
Tom Johnson, Division Chief IDPH Laboratories  
Mike Petros, Newborn Screening Laboratory  
Dr. Rong Shao, Newborn Screening Laboratory

IDPH Newborn Screening Follow up Program:

Tom Schafer, IDPH Deputy Director  
Claudia Nash, Barbara DeLuka, Nikki Woolverton, Margie Nelson, Angela McCauley

Introduction

The meeting was called to order at 10:35 a.m. Roll call was taken and a quorum of committee members was achieved. The chair, Dr. Charrow, provided some background information, stating that he recently received a letter from Dr. Damon Arnold, the Director of IDPH, asking the committee to consider the addition of severe combined immunodeficiency (SCID) to the Illinois newborn screening panel. Dr. Charrow indicated that legislation is pending in Illinois to require the addition of SCID, and that a separate legislative initiative is also being considered for the

addition of MPS I and II to the Illinois panel. Dr. Charrow also stated that several months ago the Department of Health and Human Services Secretary's Advisory Committee on Heritable Disorders in Newborns and Children (SACHDNC) recommended the addition of SCID to all state newborn screening programs. MPS I/II have not yet been proposed to the SACHDNC for review, however, enzyme replacement therapy and stem cell transplant are available treatments for these conditions to improve the quality of life, with earlier treatment being most effective.

#### Expansion Protocol

Dr. Waggoner, as Chair of the Newborn Screening Expansion Subcommittee, indicated that this subcommittee has drafted a protocol for adding new disorders to the Illinois panel. Dr. Waggoner described this proposed process as having two means for considering disorders to be added; 1- disorders recommended by the SACHDNC can be fast tracked for a vote by the full committee, and 2- for any disorder not approved by SACHDNC, various criteria have been established that must be addressed, including:

- consumer involvement in decision to mandate screening;
- availability of an accurate screening test;
- disorder is treatable and requires early treatment;
- significant, life challenging risk of morbidity if disorder is untreated;
- reasonable price of screening test;
- significant prevalence of disorder;
- resources needed by the IDPH Genetics Program to ensure adequate follow up;
- resources and access to treatment and counseling;
- positive health benefits must outweigh risks and burdens;
- existence of mechanisms for regular review of scientific and medical rationale; and
- results of population based studies of the specific proposed test.

Any individual can bring forth a disorder for consideration by the Expansion Subcommittee but must address all these criteria in their proposal. A vote regarding acceptance of this draft proposal will be taken at the upcoming meeting of the Expansion Subcommittee on February 16.

#### Open Discussion-SCID

The Immune Deficiency Foundation representative indicated they have educational materials developed for parents which they will provide to IDPH. Various committee members indicated that adding SCID to the Illinois panel should be fairly straightforward since SCID has been approved by the SACHDNC. A motion was made and seconded to add SCID to the Illinois newborn screening panel and was passed unanimously by committee members at this meeting.

#### Open Discussion-MPS I/II

Various members expressed concern about voting on the addition of MPS I/II at this time, without further review by the Expansion Subcommittee, since MPS I/II have not yet been considered by the SACHDNC. It was also stated that the committee should not make a decision to react to legislation, but should follow the proposed process outlined by the Expansion Subcommittee for consideration of disorders not yet approved by SACHDNC. Some committee members expressed concerns regarding adding new tests to the state panel without more preliminary studies and data, and cautioned that there is widespread controversy now regarding

screening for Krabbe disease, which Illinois is mandated to perform on every baby effective June 1, 2011. It was also stated that in New York, the only state currently screening for Krabbe, adequate follow up has not been completed to evaluate the impact of newborn screening on all families with a positive screen, which requires long range monitoring of the infant. It was mentioned that implementation of new tests without adequate review and data can yield unexpected results as with the recent pilot screening for Gaucher disease in Illinois, where higher levels of positive screening results have been noted in very low birth weight newborns, and the hospital where these newborns are patients must now bear the cost of diagnostic testing. However, it was pointed out that there are medical benefits of newborn screening and early treatment for MPS I/II and that testing for MPS I/II would be easy ad-ons for the IDPH lab with minimal additional cost since the equipment vendor for the current lysosomal storage disorder tests (Advanced Liquid Logic) already has the reagents for MPS I/II and the IDPH lab staff are currently performing other tests using this assay. Proponents of MPS screening also expressed concerns regarding delays in considering MPS I/II since the protocol for expansion has not yet been finalized and accepted by the full committee, and stated that consideration of MPS I/II would likely be delayed until the fall meeting of the full advisory committee.

A motion was made and seconded to table consideration of MPS I/II at this time until further review can be conducted by the Expansion Subcommittee, and this motion was passed unanimously by Committee members at this meeting.

#### Conclusion

The Expansion Subcommittee will meet February 16 to review and vote on accepting the proposed protocol for adding new disorders to the Illinois panel. Information regarding MPS I/II will be compiled according to the proposed criteria outlined in the expansion protocol and will be presented to the Expansion Subcommittee prior to the March 31 meeting of the full Genetic and Metabolic Diseases Advisory Committee. It is anticipated that the Expansion Subcommittee will summarize the criteria for considering MPS I/II and the full committee will vote on adding these disorders to the Illinois panel at the March 31, 2011 meeting.

The meeting was adjourned at 11:10 a.m.