

ILLINOIS DEPARTMENT OF PUBLIC HEALTH  
GENETIC AND METABOLIC DISEASES ADVISORY COMMITTEE  
Subcommittee on Lysosomal Storage Disorders  
Minutes—September 9, 2008, 1:00 p.m.

This meeting was held by conference call, and was called to order at 1:05 p.m. by Dr. Burton, Subcommittee Chair.

Members Present:

Dr. Barbara Burton, Children's Memorial Hospital  
Dr. Darrel Waggoner, University of Chicago  
Dr. Lainie Friedman Ross, University of Chicago  
Dr. Kay Saving, St. Francis Medical Center, Peoria  
Dr. George Hoganson, University of Illinois (joined meeting briefly)  
Dr. David Jinks, IDPH Newborn Screening Laboratory  
Mike Petros, IDPH Newborn Screening Laboratory  
Claudia Nash, Genetics/Newborn Screening Follow up Program Administrator  
Barbara DeLuka, Genetics/Newborn Screening Program  
Kate Seymore, Genetics/Newborn Screening Program  
Heather Gardner, Genetics/Newborn Screening Program

Dr. Burton presented an overview of the proposed language changes to the Metabolic Screening and Treatment Code (Administrative Rules for Newborn Screening). There was discussion of revisions to Section 661.35 of the Administrative Code, which pertains to Designation of Consultants. It was agreed that the proposed new language in the section being added should require additional criteria for evaluation of infants with a positive screen for Krabbe disease, than would be required for the other four LSDs. Infants with a positive screen for Pompe, Fabry, Gaucher and Niemann-Pick should be referred only to designated specialists (clinical biochemical geneticists or clinical geneticists) who can demonstrate extensive experience in the diagnosis and treatment of patients with LSDs, have a team of pediatric specialists including cardiology, and the capacity to provide enzyme replacement infusion therapies.

Since it is necessary to perform stem cell transplant by 30 days of life for newborns affected with neonatal onset Krabbe, the Committee members decided that initial referral of infants with a positive screen for Krabbe should be made only to specialists (clinical biochemical geneticists or clinical geneticists) that have a team in place which includes a pediatric neurologist(s) with the capability to perform stem cell transplants, to expedite the clinical evaluation process. Currently Children's Memorial Hospital and the University of Chicago are the sites in Illinois that meet these criteria. (Note: after this meeting, Claudia Nash contacted Dr. Kathy Grange at St. Louis Children's Hospital of Washington University, who indicated their site is also currently performing transplants.)

The Committee members discussed the need for molecular testing at the IDPH lab. Dr. Jinks indicated that the laboratory administration is currently supportive of adding this

piece to the screening protocol for Krabbe. Dr. Jinks also indicated that he would further modify Section 661.30, Interpretation of Results to make the language not so specific or restrictive with regard to testing methodology. Claudia Nash indicated that the final fee increase is being reviewed by IDPH to include the gene sequencing costs, and that all changes to the Administrative Rules language should be finalized and submitted to IDPH Legal staff for preliminary review within the next 2-3 weeks.

Dr. Burton provided protocols and data from New York state about their experience with newborn screening for Krabbe. She also provided information about the current clinical evaluation protocol in use in New York from Dr. Patricia Duffner, pediatric neurologist at the Women and Children's Hospital in Buffalo, NY. Subcommittee members discussed the data obtained from the NY screening program, which reflect a small false positive rate. A small group of infants will require continued follow up since they are considered at 'moderate' risk for developing the disease. The need for developing standard evaluation and monitoring protocols for all Illinois newborns identified through screening was discussed and will be considered by future meetings of this group, and others, including pediatric neurologists and members of the transplant teams.

The meeting adjourned at 1:50 pm.