Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: July 22, 2015

Subcommittee Members Attending
Barbara Burton, Chair, Katherine Keating, Lurie Children's Hospital
Jennifer Burton, University of Illinois College of Medicine at Peoria
David Dimmock, Medical College of Wisconsin
Dorothy Grange, Washington University School of Medicine
George Hoganson, Rich Dineen, University of Illinois Chicago
Linda Manwaring, St. Louis Children's Hospital
Vivian Pan, Advocate Lutheran General Children's Hospital
Tess Rhodes, DSCC
Michael Schneider, Carle Hospital-Champaign
Natasha Spencer, Consumer Representative
Darrel Waggoner, Maria Helgeson, University of Chicago

IDPH Staff
Khaja Basheeruddin, Jean Becker, David Culp, George Dizikes, Tom Johnson, Arthur Kohrman, Claudia Nash, Conny Moody, Heather Shryock

The meeting was called to order at 4:00 p.m. Dr. Barbara Burton asked if there were any questions about the aggregate data, with none, she commented that of the 40,690 total specimens tested, only 91 were abnormal or 0.2%.

IDPH Laboratory Status Report
Khaja Basheeruddin reported he expected the fifth instrument for LSD testing to be operational soon. The contract for Krabbe molecular testing was sent to the University of Illinois Health, and IDPH is still waiting for a response. Dr. Kohrman indicated he was aware of many positions needing to be filled at UIC which could slow this process.

Dr. Basheeruddin clarified three or more abnormal results on one specimen should be considered unsatisfactory/invalid. Dr. Kohrman asked if this could be affected by specimen transport time delay. Claudia Nash responded that the rationale might more be related to heat exposure versus transport delay. Dr. Kohrman asked that technical issues be reported to the centers via email communication between subcommittee calls.

Open Discussion
The issue of parental refusal of confirmatory molecular testing due to cost was raised. Genetic Counselor Keating indicated she has been working to get added coverage from Division of Specialized Care for Children (DSCC). Missouri indicated they work with the National Organization for Rare Diseases (NORD) to cover expenses that are not covered by insurance. Tess Rhodes, DSCC representative, explained that Pompe was the only LSD that met their agency's inclusion criteria since this disorder has cardiac involvement. Dr. Dimmock indicated that MPS 1 also has cardiac involvement. Dr. Kohrman voiced concern that a mechanism is not in place to cover costs to work-up the most critical disorders, MPS1 and Pompe.

It was stated that Gaucher and Fabry were not likely to need immediate treatment, and that enrollment into the Gaucher Registry provides molecular testing at no charge, and that there were ways to get molecular testing done free for Fabry. Dr. Burton has asked Ms. Keating to speak with Ms. Rhodes separately and disseminate information about available resources.

Consented Case Discussion

At this time, the specialists and genetic counselors discussed specific cases by institution. IDPH staff indicated they would be sending out notification to genetic counselors that the cutoff date will be one week prior to scheduled subcommittee calls for submission of data, and all consent forms and definitive diagnosis forms must be submitted to IDPH to be included on the spreadsheet of cases to be reviewed. For data purposes, the subcommittee agreed it was not necessary to track total parenteral nutrition (TPN) status. It is also not necessary to track cases on the spreadsheet that have a normal diagnostic work up.

To allow more time for data to be compiled, it was decided that the LSD Subcommittee would meet every other month. The next conference call is scheduled for **September 23rd at 4:00 p.m.** The meeting adjourned at 4:50 p.m.