Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: March 11, 2015, 4:30 p.m.

Subcommittee Members Attending:

Dr. Barbara Burton, Lurie
Rich Dineen, UIC
Maria Helgeson, University of Chicago
Dr. George Hoganson, UIC
Katherine Keating, Lurie
Annie McRae, UIC
Tess Rhodes, DSCC
Dr. Michael Schneider, Carle
Dr. Zohra Shad, UIC
Dr. Darrel Waggoner, University of Chicago

IDPH Staff:

Rebecca Barnett
Dr. Khaja Basheeruddin
Jean Becker
Maria Crain
Dr. George Dizikes
Shannon Harrison
Dr. Arthur Kohrman
Tom Johnson
Conny Moody
Claudia Nash
Dr. Rong Shao
Heather Shryock

IDPH Report

Dr. Dizikes reported that to date approximately 6,800 samples from 8 hospitals have been tested for Pompe, Fabry, Gaucher, Niemann-Pick and MPS I. He indicated the lab continues to have problems with mass spectrometers staying operational without continued maintenance. Further expansion of testing to include samples from other hospitals will occur once Krabbe is added to the panel. Dr. Dizikes indicated that regarding Krabbe molecular testing, attempts to secure a contract with the New York state laboratory have failed, due to inconsistencies between IDPH and NY attorneys regarding the required contract language. A contractual arrangement is being finalized through the University of Illinois at Chicago to send samples to Mayo laboratory for this testing. Dr. Dizikes stated that Mayo has agreed to perform pyschosine and 30 kb deletion testing on all samples submitted, with a 1-2 day turnaround time, and will conduct sequencing for specimens as needed, with a proposed 7-10 day turnaround time. Subcommittee members questioned the clinical utility of psychosine testing since very little has been reported in the literature about psychosine as a predictor in newborns of the clinical expression of Krabbe disease. Committee members stated that the currently proposed plan for second tier Krabbe testing deviates from the original testing algorithm recommended by the LSD subcommittee.

Extensive discussion ensued as to when and how IDPH would report the results of the galactocerebrosidase or GALC enzyme from the IDPH newborn screen along with the pyschosine, 30 kb deletion and DNA sequencing results from Mayo. The specialists strongly reminded IDPH that the protocol was written so the state screening enzyme activity and DNA results from an outside lab would be reported at the same time, and that it would be harmful to report enzyme levels alone, which would cause newborns to be referred for diagnostic testing unnecessarily. Dr. Waggoner specifically stated that to do otherwise would be a direct conflict with the subcommittee's recommendation, and he indicated that screening enzyme results alone would not justify lumbar puncture, MRI and other diagnostic tests which may not be needed if DNA sequencing results indicate no cause for concern.

The subcommittee did agree if the psychosine level was exceptionally high or the testing for 30 kb deletion was homozygous, that should necessitate immediate reporting of these test results with the GALC enzyme level to the primary care physician, with recommendation to refer immediately to a specialist. Otherwise, the subcommittee members recommended holding all test results until the DNA sequencing is complete and at that time, report GALC level, psychosine, and sequencing results to the primary care provider with recommendations for follow-up.

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: March 11, 2015, 4:30 p.m.

It was also discussed at length that sequencing results must be reported to the primary care provider with an interpretation (disease causing, benign or unknown significance), and with follow-up recommendations, since the PCP cannot be expected to interpret these complex molecular results. The group also voiced concerns that documentation of the molecular results from Mayo should be included in the IDPH report (mailer) which is sent to hospitals, since the hospital could not incorporate results from a separate report in their data system.

NOTE: Hospitals will now need to be notified that additional changes will be required in their data systems from what they were originally instructed by IDPH on May 29, 2014, since the IDPH reporting protocol is being changed to include psychosine and 30 kb deletion testing.

Action Items:

- Jean Becker of the IDPH follow-up program will distribute to the subcommittee:
    - The proof of a paper that describes Mayo's psychosine assay (author-Matern)
    - The article about New York's psychosine concentration in dried blood spots from newborns that were identified via newborn screening to be at risk for Krabbe disease (author-Chuang)
    - Mayo's algorithm for processing samples for psychosine, 30kb deletion and DNA sequencing
- The subcommittee will further review these materials
- The LSD Subcommittee Chair, Dr. Burton, will provide Dr. Kohrman with the subcommittee's proposal for reporting Krabbe test results
- Dr. Kohrman will present the Director with the subcommittee's proposed protocol which will recommend:
    - Waiting to report enzyme activity, psychosine and DNA sequencing results, unless the following occurs:
    - Immediate reporting of enzyme, psychosine and 30kb deletion, when
        - Exceptionally high psychosine levels, or
        - Homozygous 30 kb deletion
- Dr. Dizikes will provide the subcommittee with the psychosine cutoff used by Mayo
- Dr. Dizikes will provide examples of how Mayo results will be reported on the IDPH report
- IGA with UIC will be further reviewed by IDPH staff and will include language that will eliminate avoidable time delays in transporting specimens.

The meeting adjourned 5:40 p.m.

The next scheduled call is April 22, 2015 at 4:00 PM

Call in number 888-494-4032
Access Code 1495450242#