

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: August 21, 2014, 3:00 p.m.

Subcommittee Members Attending:

Lurie Children's Hospital University of Chicago	Barbara Burton, Chair Lainie Friedman Ross Darrel Waggoner
University of Illinois at Chicago	Rich Dineen Zohra Shad
University of Illinois Chicago at Peoria DSCC	Jennifer Burton Tess Rhodes

IDPH Staff:

Khaja Basheeruddin	Shannon Harrison
Jean Becker	Tom Johnson
Marie Bukovic	Arthur Kohrman
Matt Charles	Claudia Nash
Maria Crain	Tom Schafer
George Dizikes	Rong Shao

IDPH Report

Dr. Basheeruddin reported that Perkin Elmer was continuing to make changes to the IDPH mailer to include LSD test results. He reported the lab did not consistently have instrumentation running that would accommodate the pilot or going statewide and suggested extending the August 25 start date to September 2. (UPDATE: Due to problems keeping lab instrumentation operational, software issues with the Perkin Elmer database necessary for the Follow-Up Program, and resolution for DNA mutation analysis the pilot is suspended until all issues have been fully resolved.)

The contract for DNA testing through New York state lab has been rejected by Health Research, Inc. (HRI), representing New York State Health Department (NYSHD) Wadsworth Center due to indemnification language. Elizabeth Paton, IDPH Legal Counsel, is working with the Department of Central Management Services to determine if the state contract requirements can be adapted in any way to accommodate this requirement. IDPH will use Mayo lab until the contract is finalized. Based on Illinois contract limits, the contract with Mayo is for a maximum of \$50,000, which may not cover a full year of DNA testing.

Dr. Burton reported that she was at a meeting with Dr. Michael Gelb last week and he said the MPS2 substrate should be available for purchase from Perkin Elmer. Dr. Basheeruddin indicated he had received the new substrate in March or April, but had only been able to complete one experiment as MPS2 needs to be assayed separately. He indicated difficulty in giving an accurate timeline until the lab had the 6-plex test online. They are also working to get additional MS/MS instrumentation.

With no further business, the committee discussed the recent Krabbe Consortium meeting attended by Dr. Joel Charrow and Dr. George Dizikes. Of interest was that Hunter's Hope

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Foundation may support psychosine testing at Mayo Clinic. Psychosine has been shown to accumulate in infants with Krabbe. New York State has diagnosed five Krabbe cases since screening began. Missouri will be doing Krabbe in-house, but only testing the enzyme and then referring to the specialist for diagnostic enzyme analysis followed by DNA sequencing if GALC activity is below the normal limit.

Dr. Dizikes indicated there was also discussion about stem cell transplantation as a last resort, and that it was not considered curative. However, transplants earlier in life seem to have better outcomes. Consequently, there is currently a trial underway at Duke looking at in utero transplants. Dr. Burton stated that you do not need a match for an in utero transplant because the fetus is immune incompetent and a parent could be used as a bone marrow donor.

The meeting adjourned 3:18 p.m.

Special call set for September 9, 2014 at 2:00 PM
Next scheduled call is September 18, 2014 at 2:00 PM

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