

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: July 17, 2014

Subcommittee Members Attending:

Lurie Children's Hospital	Barbara Burton, Chair
University of Chicago	Lainie Friedman Ross
	Darrel Waggoner
University of Illinois Chicago at Peoria	Jennifer Burton

IDPH Staff:

Khaja Basheeruddin	George Dizikes
Jean Becker	Tom Johnson
Marie Bukovic	Rong Shao
Maria Crain	

IDPH Report

Dr. Dizikes reported that Perkin Elmer was finishing configuration of the IDPH revised mailer to include LSD test results and projected August 4th for the pilot to begin. (UPDATE: 8/25/14 new projected date.) Northwestern HL7 message test environment is being updated, and this hospital will be added to the four hospitals included in the pilot once it is operational. The contract for DNA testing through New York state lab has been submitted for their review. IDPH will use Mayo lab until the contract is finalized.

Dr. Waggoner raised the issue regarding the appropriateness of the current Illinois diagnostic follow-up protocol for patients with an abnormal newborn screen for Krabbe. The proposed NBS plan would not include reporting both enzyme and mutation results at the same time. The DNA mutation analysis will not be part of the first tier screening. The medical specialists were surveyed and Dr. Burton reported the consensus was to wait until DNA results were received before reporting enzyme levels. Dr. Grange had previously told Dr. Burton that Missouri screening results will be reported after only enzyme assay, and they will discontinue DNA testing, leaving this to be performed by the consultants at the time of evaluation of the screen positive infants. Burton felt if DNA results are received within a couple of days, then it would be acceptable to wait and report both together. If there are significant delays in receiving the DNA results, the protocol will need to be revised.

After enzyme results are finalized, the IDPH plan is to initially report the enzyme level, and the report will indicate to the primary care physician and specialist that DNA is pending. This should generate the referral to the specialist who will know that DNA testing is in process. Once DNA results are received, the lab will send an amended report with a copy of the DNA result and recommendations from Mayo or New York. The amended report will indicate if the DNA lab recommends or does not recommend follow-up. Dr. Dizikes said Mayo is willing to discuss follow-up recommendations with specialists. Dr. Waggoner indicated based on this plan there is no need to change the protocol.

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Dr. Ross questioned why Missouri switched from New York doing both enzyme and DNA testing. Dr. Burton stated it was for financial reasons, and Missouri always intended to do enzyme testing internally. Once Missouri begins enzyme testing in their state lab, the specialist handling the referral will request DNA testing. Neither Illinois nor Missouri have plans to conduct DNA testing at their state NBS lab.

Next call is scheduled for, August 21, 2014 at 2:00 PM

Meeting adjourned.