Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: May 5, 2014

Subcommittee Members Attending:

Lurie Children's Hospital	Barbara Burton, Chair
St. Louis Children's Hospital (Wash U)	Kathy Grange
University of Chicago	Amy Knight Johnson
	Lainie Friedman Ross
	Darrel Waggoner
University of Illinois Chicago	Rich Dineen
University of Illinois Chicago at Peoria	Jennifer Burton
	Brook Croke
	Jen Tarpinian

IDPH Staff:
Jean Becker
Dave Culp
Shannon Harrison
Tom Johnson
Arthur Kohrman
Claudia Nash
Tom Schafer
Nikki Woolverton

Background
The meeting was called to order at 3:00 PM by Dr. Burton who indicated this was a special meeting called to discuss LSD reporting. Subcommittee members verbalized concern with abnormal test reporting during the validation phase as this was not expected to occur prior to the pilot phase beginning. Dr. Ross voiced concern that no data system is currently in place to track who was tested and who was not tested.

IDPH Report
Deputy Director Schafer acknowledged that concerns of the subcommittee members had been carefully reviewed and he apologized for any miscommunication. He indicated that no additional samples would be sent to Perkin Elmer Genetics for validation and that enough samples have been tested to complete the validation phase by the IDPH lab. Perkin Elmer Genetics results that were sent previously to primary care providers with handwritten names and contact information will be reissued to primary care physicians and specialists with a cover letter explaining the results had been previously reported.

Deputy Director Culp echoed that concerns of the subcommittee were taken seriously. He indicated that the Department is waiting for Perkin Elmer to finalize database updates before the pilot testing phase of just Northwestern samples can begin, which is now estimated to start June 9. Dr. Waggoner asked if the follow-up staff would follow the LSD abnormals already reported during the validation phase, and Claudia Nash indicated they would be followed the same as any other abnormal test. There are ten abnormal test results that have been referred to specialists since April 10, with three specimens currently in process at Perkin Elmer Genetics and eight specimens pending DNA at the New York laboratory. It was reported that the IDPH laboratory could only identify those screenings sent to Perkin Elmer for validation since all newborns were not tested during the validation phase.

The specialists discussed in general terms the patients that had been referred to them so far. Several patients still have diagnostic work ups pending.

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Krabbe Reporting

Deputy Director Schafer reported that New York was reluctant to do second tier screening without additional indemnification language in an agreement with IDPH. The Wadsworth Center of New York State Public Health lab is believed to be the only laboratory doing second tier DNA testing on dried blood spots and are conducting this test currently for their own specimens. Dr. Waggoner was concerned that if second tier testing cannot be arranged for Illinois, then the protocol developed is not being followed and Krabbe testing should be suspended. Dr. Burton indicated other laboratories can do second tier testing and this issue should be investigated further. Dr. Culp indicated that a workable agreement with New York was being explored and if no agreement can be reached, consideration of a private laboratory who could meet turnaround time would be reviewed.

Next call is scheduled for May 15, 2014 at 2:00 pm.

Meeting adjourned 3:30 p.m.

ADDENDUM: Since this meeting, Dr. Dizikes has clarified that although many specimens have been tested by IDPH during the validation, only those sent to Perkin Elmer Genetics should actually be reported since only those test results have been "validated". Tests conducted only by IDPH that were determined to be normal, should not be reported, since those results were not validated by Perkin Elmer. The lab is now in the process of obtaining all normal reports from Perkin Elmer which will also be reported to the primary care physicians and documented in the IDPH database along with the abnormal specimen results.