Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: June 26, 2014

Subcommittee Members Attending:

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

IDPH Staff:

Khaja Basheeruddin	Claudia Nash
Jean Becker	Tom Schafer
Maria Crain	Rong Shao
George Dizikes	Nitika Sharma

IDPH Report

Dr. Dizikes reported the lab was ready for the pilot to begin. The pilot was to begin at Northwestern, but they are experiencing problems with HL7 messaging and the LSD application. Since Northwestern's database will be down from June 30 – July 14, the pilot will now begin at Advocate Christ, Central DuPage, St. Francis Peoria and the University of Chicago. If everything is operational, Northwestern will be added in mid-July with statewide expansion in a month. Dr. Waggoner voiced concern that their institution was included in the pilot, and it was not an option. Dr. Ross indicated they should have had the opportunity to not be in the pilot phase and wait until statewide screening is ready. Dr. Dizikes explained all hospitals will be included since this was a legislated mandate; all hospitals statewide have received two notifications to prepare their data systems for the implementation and the rollout would be gradually phased-in. He indicated that the word "pilot" may not be the best terminology, but the testing validation has already been completed.

Dr. Waggoner informed the committee of a Hurler positive patient identified during the validation testing phase, and the mother is refusing follow-up testing due to cost. He felt this is an issue they will continue to face and that needed to be addressed. In this particular case, the patient has Blue Cross Blue Shield insurance that did not cover the confirming enzyme test, and the mother was refusing DNA and urine testing as they could not afford more bills. There was discussion about what insurance companies cover, and that financial responsibility will fall to the family for non-covered services. Dr. Ross pointed out that the family did not consent to newborn screening and since it is legislated, it is unfair to force necessary subsequent testing costs on the parents. With the implementation of LSDs, it was noted that all follow-up testing for many other disorders is covered in Illinois by DSCC, but only Pompe will be covered of the LSDs. Dr. Ross said this has placed the family in a very unfortunate position to not be able to afford knowing if their child has this disease. Claudia gave examples of diagnostic costs that parents feel are thrust upon them, and this has been, on occasion, an issue with other newborn screening tests.

Extensive discussion ensued around the positive versus negative impact of newborn screening. Dr. Waggoner proposed this should be studied, and asked Dr. Burton if she would consider not screening

should the negative impact outweigh the positive. Dr. Burton stated she felt patients have different experiences at Lurie than at the University of Chicago based on how the information is presented to them. Dr. Waggoner stated that was an unfair statement, and Dr. Ross added that Dr. Waggoner is fair and objective with his patients. Dr. Burton indicated she would be willing to participate in a study of the impact of LSD screening on families if it was an unbiased, objective study.

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, since it is now evident that the enzyme and molecular test results will not be provided at the same time as originally anticipated.

Dr. Waggoner, the author of the Illinois Krabbe follow-up protocol, reported the original protocol was based on the understanding that the NBS report would include both enzyme and mutation results at the same time. As it turns out, the state plan is to issue a report with all of the NBS results including the enzyme result for Krabbe, but not the DNA mutation analysis. Since these results will not be available for several days, a second independent report from the reference lab (Mayo or New York) is expected to be issued 5-6 after the enzyme report.

Dr. Waggoner feels it is important that the same protocol should be followed for each patient, at each institution, and the committee should decide whether the patient should be called in prior to receipt of the DNA results. Dr. Burton asked Dr. Waggoner to summarize the issue and Claudia will send to the entire committee for their comments.

Finally, the *Parent/Individual Consent and Authorization To Share Newborn Screening Data/Specimens* form has been approved by the IDPH legal staff and shared with the specialists. This consent will allow the LSD subcommittee to anonymously discuss individual cases. When a patient is referred to a specialist with an abnormal LSD screen, the specialist should ask the parent to provide consent to review the results with this group. There is also the option for parents to consent to sharing part of the leftover specimen with another lab as well. Once the form is signed, the specialist will need to return the forms to IDPH Follow-up Program, and information about this child will be added to the spreadsheet for discussion on the subcommittee calls.

Next call is scheduled for July 17, 2014 at 2:00 pm.

Meeting adjourned 2:45 p.m.