

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: December 18, 2014, 2:00 p.m.

Subcommittee Members Attending:

Lurie Children's Hospital	Barbara Burton, Chair
	Kitty Keating
Lutheran General	Brad Tinkle
University of Chicago	Lainie Friedman Ross
	Darrel Waggoner
University of Illinois Chicago at Peoria	Jennifer Burton
DSCC	Darcy Contri

IDPH Staff:

Rebecca Barnett	George Dizikes
Khaja Basheeruddin	Tom Johnson
Jean Becker	Claudia Nash
Marie Bukovic	Tom Schafer
Matt Charles	Rong Shao
	Heather Shryock

IDPH Report

Khaja reported that the pilot began November 3rd and the lab was testing for the following five LSDs: Fabry, Gaucher, MPS I, Niemann-Pick and Pompe at four hospitals (Advocate Christ, Central DuPage, OSF Peoria, and University of Chicago). Of the 1,317 samples test nineteen required a second confirmatory test. Of those, four have been reported as positives: 2 sample for Pompe, 1 sample for Pompe/Fabry, 1 sample for MPS 1.

Two genetic counselors indicated that families are overwhelmed with information at the initial specialist visit and have been unwilling to sign the Consent and Authorization to Share Newborn Screening Data/Specimens Form. It was stated that staff at the medical specialist office may need to request and reeducate on numerous attempts to get this consent signed, so that specific case information can be shared with the LSD committee.

The HL7 reporting issues are being resolved with Northwestern so the lab can start testing their samples for LSDS soon. To increase the volume being screened, the committee discussed adding other hospitals if the reporting issue with Northwestern was not resolved.

Dr. Waggoner indicated the IDPH lab report states the LSD test as "Pending". Khaja indicated that issue has been resolved to indicate "Profile Positive" or "Not Tested" if not a pilot hospital. However, if "Pending" does appear please contact the lab to have an amended report printed. Dr. Burton asked if Dr. Dizikes planned to submit data to the Newborn Screening Translational Research Network (NBSTRN) sponsored Pompe pilot project that involves New York, Georgia and Wisconsin. There is a bi-weekly conference call to discuss results and progress. Dr. Dizikes reported he does attend these conference calls and has reported Illinois' progress in establishing LSD testing in general and Pompe testing in particular. There is also an LSD section to the R4S database that Piero Rinaldo maintains at the Mayo Clinic. Dr. Dizikes also plans to provide data for this database as he has for MS/MS data.

The meeting adjourned 2:30 p.m.

Next scheduled call is January 22, 2015 at 2:00 PM
Call in number 888-494-4032
Access Code 1495450242#