

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: March 23, 2016

Subcommittee Members Attending

Barbara Burton, M.D., Chair, Jill Corkery, Lauren Whiteaker, Lurie Children's Hospital
Rich Dineen George Hoganson, M.D., Annie McRae, University of Illinois Chicago
Lainie Friedman Ross, M.D., Ph.D., University of Chicago
Tess Rhodes, DSCC
Natasha Spencer, Consumer Representative

IDPH Staff

Khaja Basheeruddin, Ph.D., Jean Becker, Matt Charles, Arthur Kohrman, M.D., Claudia Nash, Rong Shao, M.D., Heather Shryock

The meeting was called to order at 4:00 p.m. by Dr. Barbara Burton.

IDPH Lab Report

Dr. Basheeruddin reported the MPS II internal initial study was completed as a separate punch. The lab will be doing two punches as they move forward. He felt the data was reasonable. The actual validation studies have not been initiated yet. This will include doing at least 5000 specimens. Once Perkin Elmer updates the database to include completion of LIMS support, the projected timetable for implementation of screening will be after Neobase is completed in the lab.

Matt Charles indicated the Krabbe was an open procurement and the bid came down in mid-January. The bid included psychosine and DNA. Senior procurement and Legal staff are determining if bids are accepted. Implementation of Krabbe screening will be dependent on the procurement process.

Screening for adrenoleukodystrophy (ALD) is expected to be implemented in early 2017 and can be incorporated with the new version of the Neobase assay with acylcarnatine profile.

Claudia informed the subcommittee that staff from the Lab and Follow-Up Program have been participating on monthly calls with the Newborn Screening Translational Research Network (NBSTRN) regarding Pompe and MPS I screening.

Dr. Burton informed the subcommittee that she has reached out to State Senator Righter to seek support on health insurance coverage, but has not heard back from him. She has heard back from State Representative Laura Fine. Dr. Burton is seeking mandated coverage for any disorder included on newborn screening. Tess Rhodes, DSCC, indicated if molecular testing is denied by a managed care organization (MCO) to contact the Department of Healthcare and Family Services and they will contact the MCO directly. Contact Tess for help.

Aggregate data was discussed. Mutation A143+ should not be categorized at "Unresolved Undetermined", but as "Affected Late Onset". Also, do not classify pseudodeficiency as unresolved. Rich Dineen asked for guidelines so everyone reports the same. Dr. Burton will work on this and

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circulate it to the group. The positive predictive value spreadsheet was reviewed and the lab was asked if they were looking at things to reduce the false positives.

Consented Case Discussion

No discussion – Dr. Ross reminded the subcommittee that those cases with consent on file should be the only ones discussed.

Open Discussion

New York is piloting ALD. In Illinois, ALD has been legislated, and it is now on the Recommended Uniform Screening Panel. New York rates are in line with the estimates 1:15,000 in males and New York is detecting female carriers. ALD screening also picks up other peroxisomal disorders including Zellweger Syndrome. No follow-up protocols have been developed for ALD in Illinois, until the lab is prepared. The plan will be to refer to geneticists not neurologists.

Natasha Spencer, consumer representative, reported that Kentucky implemented Krabbe newborn screening recently. She said the University of Chicago diagnosed an 18 month old with Krabbe and the family had previously seen multiple physicians. Ms. Spencer is concerned that IDPH is aware of these family delays in diagnosis.

Dr. Burton invited the subcommittee to a face-to-face meeting on June 2nd which would include an update on Missouri LSD implementation, review of Illinois' data in detail, laboratory update, case presentations. Lunch and parking vouchers will be provided. The May 25 conference call is being cancelled.

Dr. Burton informed the subcommittee that the WORLD meeting was well received. There was tremendous interest and a lot of questions. Dr. Rong Shao presented at APHL on Fabry data. Specifically, the effects of birth weight on GLA alpha-galactosidase activity. Dr. Burton asked to review the data again at the June 2 meeting.

The next meeting will be face-to-face on **June 2nd at 10:00 a.m.** at the Lurie Children's Hospital, Room 11-160. The meeting adjourned at 4:51 p.m.

SAVE THE DATE
July 27 at 4:00 p.m.
Call-in Information:
Call-in #888-494-4032; Passcode 1495450242