Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting Minutes: June 2, 2016

Subcommittee Members Attending
Barbara Burton, MD; Zohra Shad, MD; Brad Tinkle, MD; Mindy Li, MD; Rich Dineen, MS; Jill Corkery, MS; Tess Rhodes, DSCC; Andrea Atherton, MS; Erika Vucko, APN; Shanna Widera, APN; Lauren Whiteaker, APN; Maren Schmiester, Joe Schneider, Shelley Kinzer-Corley, Sheela Shrestha, Jon Solamillo, Carolyn Ries, Vicki Sanders, MS; Rachel Katz, MSW; Soo Shim, MSW; Marie-Renee Plona, MS; Andrea Paras, MS; Katherine Kim, MS

IDPH Staff
Khaja Basheeruddin, PhD; Matt Charles, Rong Shao, MD

Guests
Dawn Peck, MS, University of Missouri

The meeting was held at Ann & Robert H. Lurie Children's Hospital of Chicago and convened at 10:05 AM by Dr. Barbara Burton, Subcommittee Chair. Dr. Burton reviewed the cumulative data on LSD newborn screening circulated by email to subcommittee members by the Springfield follow-up staff. Subsequently, Khaja Basheeruddin, PhD presented an update from the Newborn Screening laboratory. Dr. Basheeruddin reported on efforts in the laboratory to put into place a new assay for IDUA using lactone as an inhibitor of beta-glucuronidase which has iduronidase-like activity. It is anticipated that use of this new assay should help reduce the false positive rate for MPS1. He also shared data on GLA levels in low birth weight and term infants as a function of chronologic age. He pointed out that low birth weight infants have higher GLA levels than term infants although these gradually return to levels comparable to term infants by about one month of age. It was proposed that samples from infants weighing less than 2000 grams that are obtained before 14 days of age, and have a GLA level below the batch median, be reported as invalid with a repeat sample requested after 14 days of age. Dr. Tinkle suggested that the lab pull data on low birth weight infants who have had sequential samples analyzed to assess the effectiveness of this strategy before actually implementing it. Dr. Basheeruddin indicated that he would look into this.

Dr. Basheeruddin reported on the status of Krabbe newborn screening. He indicated that the major hold has been in getting a contract finalized with Mayo for the follow-up psychosine and DNA testing. He also pointed out that some modifications of the laboratory information management system (LIMS) will be required before implementation. He presented a rough timeline indicating how long each step in implementation might take. Some discussion followed regarding the excessive delays in getting this initiated. Matt Charles stated that he thought that the terms of the contract had been almost finalized by the lawyers and stated that he understood that initiating Krabbe and MPS II screening is a high priority. It was asked if some of the steps outlined by Dr. Basheeruddin on his timeline could not be done in parallel. Matt Charles stated that this was occurring. Dr. Basheeruddin presented some information on the MPS II assay and data on 90+ samples thus far analyzed. He stated that the assay appears to be working very well although a full scale validation will be required with a much larger number of samples as well as staff training. There was inquiry as to why this has not yet occurred. Dr. Basheeruddin reported that there have been times when some of the MSMS machines have been down so that no machine was available for test development.

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Dawn Peck from the University of Missouri presented Missouri's experience to date with LSD newborn screening. They are using the digital microfluidic method to screen for Pompe, Gaucher, Fabry , MPS1 and Krabbe diseases. The advisory committee in Missouri has recommended inclusion of MPSII as well but this cannot yet be done using the digital microfluidic method. The incidence of Pompe, Fabry, and Gaucher disease and MPS1 is similar in Missouri to that observed in Illinois. For Krabbe disease, the lab does both enzyme assay and 30 kb deletion testing. A referral to the consultant is made when at least one copy of the 30 kb deletion is detected or when the enzyme activity is very low. No cases of infantile Krabbe disease have thus far been detected in Missouri. Ms. Peck reported that this approach has significantly reduced the number of referrals to diagnostic centers for Krabbe disease.

A series of cases of interesting or complex cases were presented by Jill Corkery, Lauren Whiteaker and Rich Dineen. There was a lively discussion of these.

At the end of the meeting, action items were discussed. It was decided that the committee will continue with bimonthly teleconferences, with the next one at the end of July. Dr. Tinkle suggested that more frequent calls may be needed once Krabbe and MPSII screening begins. Dr. Tinkle also recommended that all consultants use the same diagnostic laboratories for follow-up of infants with a positive screen for LSD's. Some discussion ensued, and it was concluded that this would be ideal but is probably not possible in every case. Dr. Burton asked if the group felt it was still necessary to consent every family for possible discussion of the specifics of their case since we no longer go through most cases. She suggested that consents be obtained only from the parents of those infants that the physicians specifically want to discuss with the group. No decision was made on this point. The issue of trying to get legislation requiring insurance coverage of the follow-up testing necessary to confirm a diagnosis of an LSD was again raised. Dr. Burton reported that she had contacted both Dale Righter and Laura Fine about this. No response was received from Senator Righter. Representative Fine responded and said she would look into it. Dr. Burton indicated that she would follow-up with both of them. Tess Rhodes suggested some other possible contacts. Dr. Burton indicated that she would follow-up after the meeting with a memo to the committee regarding the definition of some of the categories used in the summary data so that patients can be classified in the same way.

The next conference call is scheduled for **July 27th at 4:00 p.m.**

Call-in Information:
Call-in #888-494-4032; Passcode 1495450242

SAVE THE DATE
Future Meeting Dates
September 28
November 23