

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting Minutes: July 27, 2016

Subcommittee Members

Barbara Burton, MD, Chair
Rich Dineen, Genetic Counselor, UIC
Linda Manwaring, St. Louis Children's Hospital
Maria-Reneé Plona, Genetic Counselor, UIC
Michael Schneider, MD, Carle Hospital Champaign
Zohra Shad, MD, UIC
Natasha Spencer, Consumer Representative
Darrel Waggoner, MD, University of Chicago

IDPH Staff

Khaja Basheeruddin, PhD; Matt Charles, Claudia Nash, Heather Shryock

Dr. Basheeruddin asked if there were any questions regarding the data summary. There was no discussion about the Summary Data or Consented Case Data.

There was a correction to the June 2nd minutes. "It was proposed that samples from infants weighing less than 2000 **not 200** 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." This change will be made to the official minutes.

Lab Update

The lab reported they would be changing the MPS I cutoff within the week. MPS II has not started testing in a live environment. There has been no change to Krabbe molecular testing contract progress. Dr. Burton spoke with Dr. Dieter Matern from Mayo who thought it was finalized. Matt Charles was not aware of this.

Illinois state newborn data and Washington University pilot study data for LSD screening using de-identified specimens were discussed. Dr. Basheeruddin highlighted the differences in studies including LSD method/chemistries and population demographics, as well as number of specimens screened.

The lab continues to evaluate positive screenings and current cutoff values for alpha-galactosidase (GLA) enzyme activity for Fabry in which the lab reviewed data for 60,000-70,000 samples and saw an effect of birth weight <2000 grams on the enzyme. The lab will continue to monitor this data. In addition, the lab is reviewing cut off values for alpha-glucosidase (GAA) for Pompe and beta-glucocerebrosidase (ABG) for Gaucher. The lab feels this will reduce false positive rates with confidence.

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Dr. Burton requested a summary of first year data for June 1, 2015 – May 31, 2016. Heather Shryock will prepare this summary.

Dr. Schneider was concerned about the ethics of diagnosing adult onset disease in newborn cases. Dr. Burton indicated that the Secretary's Advisory Committee was not concerned with late onset for Krabbe or MPS I, and that the range of onset can be from 12 – 30 years of age.

The meeting adjourned at 4:31 p.m.

The next conference call is scheduled for **September 28 at 4:00 p.m.**

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

SAVE THE DATE
Future Meeting Dates
November 23