

Illinois Department of Public Health
Lysosomal Storage Disorders Subcommittee
Illinois Department of Public Health
Meeting and Conference Call Minutes: January 27, 2016

Subcommittee Members Attending

Kelly Bontempo, Advocate Lutheran General Children's Hospital
Shantel Buhl, Vivian Pan, Advocate Lutheran
Jill Corkery, Lauren Whitaker, Lurie Children's Hospital
David Dimmock, M.D., Medical College of Wisconsin
Dorothy Grange, M.D., Washington University
George Hoganson, M.D., Zohra Shad, M.D., University of Illinois Chicago
Tess Rhodes, DSCC
Natasha Spencer, Consumer Representative
Darrel Waggoner, M.D., University of Chicago

IDPH Staff

Khaja Basheeruddin, Ph.D., Jean Becker, Matt Charles, Maria Crain, Shannon Harrison, Arthur Kohrman, M.D.,
Claudia Nash, Conny Moody, Rong Shao, M.D., Heather Shryock, Rich Zimmerman

The meeting was called to order at 4:00 p.m. by Dr. Darrel Waggoner.

IDPH Lab Report

Dr. Basheeruddin gave the lab report indicating the IDPH lab is currently screening for 5 LSDs using a 5-plex assay and has analyzed approximately 157,000 specimens, including 15,000 during the Pilot Phase. During the Validation Phase, approximately 15,000 samples were analyzed. Total diagnosed cases: Fabry – 4; Gaucher – 2; MPS I – 1; Niemann-Pick – 1; Pompe – 7. Overall, the LSD rate is close to 1.1:10,000 specimens. He reported the LSD assay is coming along nicely and four MS/MS instruments are working. The lab is not experiencing any problems with turnaround time.

Matt Charles indicated the Krabbe procurement has been posted on the CMS website and expires next week. It is an open bid process. CMS will procure the contract for IDPH. The procurement includes timeliness and cost requirements. Matt indicated that Wadsworth, Mayo and the University of Illinois were aware of the posting. Dr. Waggoner indicated the committee would like to be involved in determining the quality of the vendor, and Dr. Kohrman agreed with him that someone from the committee should have the opportunity to review the proposals. Dr. Waggoner was most concerned with quality of molecular testing. Deputy Director Moody agreed it made sense to have someone on the committee review the bids.

Dr. Basheeruddin gave an update on MPS II. The lab recently used the substrate received from Perkin Elmer to perform multiplex analysis for MPS II with the five other enzymes and it seems to be working nicely. The lab is initiating validation studies.

Consented Case Discussion

Dr. Waggoner led the discussion to review aggregate data and cases where consent to discuss has been obtained.

Open Discussion

Deputy Director Conny Moody provided an update on health insurance coverage. She indicated that newborn children have to be provided coverage if dependent coverage is offered under the plan. Under the law, all accident and health insurance policies and HMO certificates must provide benefits to a newborn child immediately from the moment of birth. The newborn must be added to the policy regardless of illness, injury, congenital defects, birth abnormalities or premature birth and benefits must be paid for those conditions to the extent of the policy provisions. The Department of Insurance website provides information on mandated benefits

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for health insurance providers in Illinois. Often referred to as “body parts legislation”, the benefits coverage may be specific to a certain disease or type of procedure (e.g., breast exams, mental health services, autism, etc.) Illinois law only requires coverage to the extent it is provided in the policy. The newborn is not entitled to benefits, which are not contained in the policy. Therefore, if the insurance policy excludes a certain type of procedure, the company is within its rights to deny those claims for the baby. It appears from speaking to a Department of Insurance call center representative, the type of testing necessary for confirmatory testing and treatment of an LSD would have to be statutorily mandated for all health insurance providers to be mandated to provide coverage. Dr. Dimmock interjected that if a plan was doing business in Illinois it must meet the requirements for Illinois plans. A copy of the newborn laws (215 ILCS 5/356c for insurance companies or 215 ILCS 125/4-8 for HMOs) and the HIPAA law (215 ILCS 97/35) may be obtained from http://insurance.illinois.gov/Rules/laws_regs.asp.

Dr. Waggoner concluded that explanation did not help the child much and that shouldn't those parties who mandate the screening be asked to add coverage for diagnostic testing. He suggested working through the March of Dimes to initiate legislation. Dr. Kohrman indicated that some of these newborns should be eligible if adequate advocacy efforts, even in current system, through Ever Thrive, a maternal and child health advocacy group. Dr. Waggoner stressed that it is an extremely burdensome process to get approval for testing, and Dr. Hoganson added that timing does not allow for us to wait for approvals. The parents want to have the testing, but cannot afford the bills.

Dr. Waggoner proposed the committee begin to move to advocate for changes in required insurance coverage. Dr. Kohrman and Deputy Director Moody discussed the best way to do that either through legislation or an advocacy group.

In discussing the data, it was decided that a category of “unresolved, phenotyped undetermined/VUS” be added. This led to a discussion about review of clinical diagnostic protocols again, and it was suggested that Dr. Burton reassemble the original groups to review the original disorder protocols.

Dr. Waggoner also mentioned the usefulness of national database repositories and indicated the IBEMC pays \$100 for each case entered. The Inborn Errors of Metabolism Information System (IBEM-IS) is a web-based registry system designed to monitor health outcomes for people who have inborn errors of metabolism. The IBEM-IS allows the medical community to understand more about the natural history and outcomes for rare inborn errors of metabolism, develop treatment protocols, and support clinical research. IRB approval is required for all data collection. <https://www.ibem-is.org/Professionals.aspx>

The next conference call is scheduled for **March 23rd at 4:00 p.m.** The meeting adjourned at 4:50 p.m.

Call-in Information:

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

SAVE THE DATE
Face-to-Face Meeting
Lurie Children's Hospital
June 2, 2016
10:00 a.m. - 2:00 p.m.