

Illinois Department of Public Health
Lysosomal Storage Disease Subcommittee
Conference Call Minutes – January 22, 2015

Subcommittee Members Attending

Lurie Children's Hospital- Barbara Burton, Kitty Keating
University of Illinois at Chicago- Zohra Shad, Annie McRae
University of Illinois at Peoria- Jennifer Burton
The University of Chicago- Darrel Waggoner, Lainie Friedman Ross, Maria Helgeson
University of Illinois, Division of Specialized Care for Children-Tess Rhodes

IDPH Staff

George Dizikes	Tamra Simulick	Marie Bukovic
Rong Shao	Fran Balster	Rebecca Barnett
Arthur Kohrman	Claudia Nash	Maria Crain
Matt Charles	Heather Shryock	

The meeting was convened by the Chair, Dr. Barbara Burton, by conference call at 2:05 PM.

Overview of LSD Pilot

Dr. Dizikes provided an overview of the pilot screening activities to date. November 3, 2014, the IDPH laboratory began testing all newborn screening samples from four hospitals (Advocate Christ, OSF St. Francis in Peoria, the University of Chicago and Central DuPage) with a total of 2,848 specimens screened to date. Of those specimens, 1.4% (34) had enzyme levels below the cutoff on the initial screen, and 0.49% (14) had an enzyme level below the cutoff on the repeat screen of the same sample, therefore were reported to the primary care provider as abnormal for one or more LSDs.

Dr. Dizikes indicated that the laboratory is hoping to begin testing all samples from Northwestern in the near future as soon as database reporting issues are resolved. Newborn screening reports for Northwestern are currently issued through an electronic HL7 messaging system, and this system is being revised to incorporate the addition of LSD results.

Other hospitals are being considered for inclusion in the pilot as well; Edward Hospital, Evanston Northshore, St. Alexius and Advocate Lutheran General. Dr. Dizikes indicated these hospitals would be notified in the near future, and testing would not begin for another two weeks for newborns from these sites. He indicated the only other significant difficulty with LSD testing is keeping the tandem mass spectrometers operational. Currently only two out of four machines are functioning, making it difficult to consider proceeding with testing statewide.

Dr. Dizikes indicated that screening for Krabbe is not yet occurring due to delays in establishing a contract for molecular testing. He indicated that progress has been made in securing an intergovernmental agreement with the University of Illinois at Chicago to send specimens to Mayo Clinic, but testing cannot occur until these arrangements are finalized.

Dr. Burton asked for clarification regarding how a test result is designated as “borderline” rather than “positive”. Dr. Dizikes indicated that a borderline result occurs when the result falls between the screening cut off and the call-out cut off.

Review of Data

Current screening data were distributed to the committee and discussed. Fourteen abnormal test results have been identified for 10 newborns, with some samples having a low enzyme level for more than one LSD. Eight test results were reported as ‘positive’ with an enzyme level low enough to require referral to a specialist, 5 results were reported as ‘borderline’ which require a repeat newborn screen and 1 result was ‘invalid’, also requiring another newborn screening sample be collected. A total of 6 cases have been resolved as normal and further tests are pending for the remaining 4 newborns.

Dr. Burton indicated when a case is closed as normal, it would be helpful to know if that determination was made from a repeat normal newborn screen, or if diagnostic testing was done. This information will be added to the spreadsheet, according to the recommendation.

Dr. Burton also noted that most of the abnormal results identified so far have been with sick infants in the NICU, but the majority have been full term. Dr. Dizikes stated that much more data will be needed before it can be determined if this trend will continue, and cutoffs for NICU babies may need to be modified.

Other disorder-specific spreadsheets, previously developed, will be used in the future. The data presented were compiled in a summary sheet for this meeting to provide a condensed overview of the findings to date, since there are currently so few abnormal test results. The carrier status of molecular testing will also be included in future tracking spreadsheets.

Claudia Nash indicated that an error was regarding the normal cut-off level was listed for Fabry, which should be >18% instead of >31%.

Other Issues

Dr. Kohrman restated his support of advocating for an extended operation for newborn screening, since this continues to be a national issue to assure prompt reporting of abnormal results. Dr. Dizikes indicated that some additional laboratory staff had been requested to provide Saturday coverage, but there have been no updates regarding the status of those positions with the new administration.

Dr. Kohrman also provided some introductory information about the newly appointed IDPH Director, Dr. Nirev Shah.

Next Meeting

Meetings are generally scheduled for the third Thursday of each month, and the next meeting date will be Thursday, February 19, 2015 at 2 pm.