

Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee
Illinois Department of Public Health, Division of Laboratories
2121 W. Taylor St., Chicago, Illinois
Meeting and Conference Call Minutes: January 29, 2014

Subcommittee Members Attending:

George Hoganson-University of Illinois at Chicago – Chair
Barbara Burton-Lurie Children’s Hospital
Denise Lonigro-Advocate Christ Hospital
Kristin Clemenz-Lurie Children’s Memorial
W. Patrick Zeller- Endocrinologist- Private Practice

IDPH Staff:

George Dizikes, Arthur Kohrman, Bill Calvert, Khaja Basheeruddin, Rong Shao, Raj Singh, Jennifer Crew,
Claudia Nash, Shannon Harrison, Jean Becker, Heather Shryock, Nitika Sharma, Linda Robinson

The meeting was called to order at 1:00 PM, followed by introductions. The minutes were approved for the meeting held September 18, 2013.

Old Business

There were no items for discussion.

New Business

Laboratory Report

Updates:

Dr. George Dizikes indicated the new testing area is complete; however they are having difficulty maintaining a stable room temperature. Therefore, Neobase testing has been stopped until a new room with a stable room temperature can be set up. Lysosomal storage disorder (LSD) procedures for tandem mass spectrometers are running as expected. The laboratory is enrolled in a CDC proficiency testing (PT) program and has identified all positive LSD specimens they have tested. The laboratory does not yet have firm cut off values established for the LSDs, but over 5,000 specimens have been tested. The severe combined immune deficiency (SCID) staff have also enrolled in the CDC PT program, and they have tested several thousand specimens. All molecular staff are now cross trained to do SCID testing. Their testing room is complete and functional.

Data System/Reports:

Dr. Dizikes indicated Perkin Elmer database changes for SCID testing are expected to be done by April, but there is still an additional piece of the SCID database that has to be developed for follow-up activities. Perkin Elmer is behind schedule for database changes for LSD testing. Additionally, Perkin Elmer has to roll out a database upgrade before LSD testing can begin. Dr. Dizikes also indicated that an emergency preparedness plan with Perkin Elmer is being developed so there can be a transfer of result data directly into the IDPH data system should Illinois ever need Perkin Elmer to take over our testing temporarily.

Pilot Study:

Dr. Dizikes indicated the SCID pilot is expected to begin April 1, 2014, on all samples submitted from both Northwestern Memorial Hospital and Advocate Christ Hospital. The LSD pilot is expected to begin May 1, 2014, for only Northwestern Memorial Hospital samples. Letters will soon be sent to both hospitals, as well as to all Illinois birthing facilities, stating what new tests will be added to the NBS panel and what data field changes are required so that facilities have ample time to upgrade their data systems.

Follow-up Program Report

Updates:

Claudia Nash voiced concerns about how the Perkin Elmer software upgrade may affect the result flow, especially during the time we will be adding new tests. Laboratory staff stated this upgrade is needed in order to report LSDs. Claudia Nash stated the follow up program will send a letter to all birthing facilities informing them of the new tests being added and the fee increase of \$2 per specimen effective July 1, 2014. Three office specialist positions are posted for new follow up staff. One nursing supervisor position is posted as well.

Review of Data:

Claudia Nash stated the follow up staff has received inquiries from various medical specialists concerned about the increase in the number of positive newborn screening results that are being reported and various other issues, summarized below:

- 1.) Congenital Adrenal Hyperplasia (CAH) – Dr. Ghai, a neonatologist with Illinois Masonic Hospital, had called the follow up program with concerns regarding the increase of positive screening results for CAH, especially with babies in the neonatal intensive care unit (NICU). Dr. Ghai voiced he felt that the Department was putting the burden on physicians with this high number of positive screens. IDPH follow up program compiled CAH data for Illinois Masonic Hospital from 2011-2013 and statewide. After review by this subcommittee, it was felt that the number of positive screens was not higher than expected. Although no diagnosed cases were identified at Illinois Masonic Hospital, elsewhere some diagnosed cases had initial borderline screens, so it seems unwise to raise the cut off. The follow up program agreed to send a summary of the data and meeting discussion to Dr. Ghai.
- 2.) Hypothyroidism (HYP) – The data for this disorder was consistent with prior years.
- 3.) MS/MS – Follow up program indicated there has not been a significant change in the number of diagnosed cases for these disorders; however, there has been a 33% increase in number of positive screening samples. Rong Shao reported the MS/MS laboratory is periodically updating their test cut-off algorithm according to their testing results and confirmed case feedback from follow-up program and neonatal specialists. After comparing positive screening result numbers from 2012-2013, there was more than a 30% increase in the number of positive screening results in 2013 as compared to 2012. The main increase was in the fatty acid oxidation disorder (FAO) category, especially in these 3 disorders:
 - a. CACT/CPT II (Acylcarnitine Translocase Deficiency/Carnitine Palmitoyltransferase II Deficiency)
 - b. MCAD (Medium Chain Acyl-CoA Dehydrogenase Deficiency)
 - c. VLCAD (Very Long chain Acyl-CoA Dehydrogenase Deficiency)

The MS/MS laboratory is fine tuning their cut-off values by using more secondary markers in their reporting algorithms. Dr. Hoganson instructed the laboratory staff to review the data in the upcoming month to determine the reason for the increase number of positive tests and provide a report to this committee. Additional detailed data will be circulated by email to members of this subcommittee regarding the increase in MS/MS positives by disorder type to identify the reason for the increased trend in 2013, and the lab staff will review cut-off levels.

- 4.) Hemoglobinopathies (HGB) – There has not been a change in the number of positive test reports, however, there has been a marked increase in the number of “low fetal to adult” (LFA) hemoglobin results. Data from 2012 to 2013 were reviewed for non-NICU, >38 weeks gestational age infants. In 2012, there were 148 such newborns with LFA reported compared to 253 in 2013. Hector Diaz reported the LFA criteria is based on the percentages between Fetal (F) and Adult (A) hemoglobin. Samples are reported LFA whenever “A” hemoglobin is greater than 20%, the sample indicates transfusion or NICU status and the date of birth is less than 2 months. Follow up staff will provide

laboratory with a list of LFA cases, and laboratory staff will review the reported LFA cases from 2013 to look at the relative percentages of hemoglobin "F" and "A".

- 5.) Cystic Fibrosis (CF) – Follow up program reported Dr. Gopal Srinivasan wanted clarification of the cutoff value for IRT levels. Laboratory staff confirmed any IRT >170 ng/mL would have DNA analysis. The second concern raised was the increase in the number of positive reports in 2013, which the laboratory said is probably due to the change in the DNA panel. The CF Collaborative had recommended that the lab block the reporting of F508C and the IDPH laboratory staff confirmed their SOP was recently approved to eliminate reporting of this mutation. The laboratory staff requested the follow up staff to ask the CF Collaborative for their recommendation on whether or not to remove the mutation D1270N from the panel for similar reasons. Jean Becker will discuss this issue with the CF Collaborative at their next meeting.

Meeting was adjourned at 2:20 PM.