

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: September 17, 2014

Subcommittee Members Attending:

George Hoganson-University of Illinois at Chicago – Chair
Denise Lonigro-Advocate Christ Hospital
W. Patrick Zeller- Endocrinologist- Private Practice

IDPH Staff:

Tom Johnson, Tom Schafer, George Dizikes, Khaja Basheeruddin, Raj Singh, Claudia Nash, Shannon Harrison, Jean Becker, Heather Shryock, Marie Bukovic, Nitika Sharma

The meeting was called to order at 9:05 PM, followed by introductions. The minutes were approved for the meeting held January 29, 2014.

Old Business

There were no items for discussion.

New Business

SCID Testing- Three Month Update

Shannon Harrison of the follow-up program gave a report on the outcome of Illinois SCID testing since it was implemented statewide June 9, 2014. A SCID/Immunology Collaborative has been formed consisting of pediatric immunologists from each of the five SCID specialty centers, as well as staff from the IDPH newborn screening laboratory and follow-up program, and has been meeting bi-monthly. The five SCID diagnostic centers are Lurie Children's Hospital, Advocate Christ Hospital, the University of Chicago, all located in Illinois and Cardinal Glennon Children's Hospital and St. Louis Children's Hospital, both located in St. Louis, MO. Data were reviewed that showed out of 42,400 samples tested in the first three months, 175 preterm and 72 term newborns tested positive, and one term infant has been diagnosed with X-linked SCID. The current normal cutoff is less than 300 TRECs/microliter. Illinois staff will contact the Massachusetts newborn screening program, which has been testing for SCID much longer than Illinois, to compare data and cutoffs, since the testing methodology used by Massachusetts is similar to that used in Illinois. Due to the few number of SCID diagnostic centers, Dr. Hoganson questioned if it is acceptable for local physicians to consult with specialists who recommend and review results of specific diagnostic tests, with the local ordering physician then signing off on cases based on another physician's review without seeing the child in person. Shannon indicated this manner of case closure is being accepted by IDPH since it is not always feasible for families to travel long distances for a face to face diagnostic consultation.

Lysosomal Storage Disorders

Jean Becker of the follow-up program reported that the target date for the start of the LSD pilot is now October 13, 2014 for five LSDs, Pompe, Gaucher, Fabry, MPS I and Niemann-Pick. The target date for the Krabbe testing pilot will be January 2015. Causes of this delay include difficulty getting a contract in place for the Krabbe molecular testing, continued problems with sufficient mass spectrometers to be properly working and errors with the follow-up side of the database that the software vendor is correcting. Currently, the database vendor, Perkin Elmer, is making modifications to the reports to eliminate any reference to Krabbe, from the newborn screening final report, until the molecular testing contract issues are resolved and testing for Krabbe can begin.

The value of having molecular results for Krabbe as part of the newborn screening test was discussed by Dr. Hoganson who stressed this will eliminate the need to refer some newborns with low enzyme levels for further evaluation, and will also help the specialists determine how best to proceed with truly affected newborns. Dr. Hoganson stated that a second tier testing method for MPS I (enzyme followed by molecular testing) would also be beneficial since a high number of MPS I pseudodeficiency cases will likely be identified, based on the screening experience in Missouri. Molecular testing should be included as part of the newborn screen since it would also negate the need to refer the vast majority of these infants to a specialist for further diagnostic evaluation.

Data from the March 18-April 29, 2014 validation testing phase for LSDs was reviewed which indicated 42 of 15,000 specimens tested were identified as 'positive' by IDPH and sent to Perkin Elmer, Mayo and or New York state lab. Of these, only 15 were validated as abnormal by these outside laboratories and then reported by the IDPH follow-up staff to the primary care physician. Of these 15 newborns referred, one was diagnosed with Gaucher and one with Niemann-Pick; seven newborns were classified with a pseudodeficiency, and three as carriers, one case is still pending and the remaining two were normal.

Laboratory Report

Dr. George Dizikes discussed various equipment issues. He indicated that Neobase testing is currently on hold and the mass spectrometers that will be used will need to be reinstalled. Neobase testing using a non-derivatized method, will not occur until early in 2015 at the earliest, and will include succinylacetone screening on all specimens, which will reduce the number of false positives for tyrosinemia. The high pressure pumps on the mass spectrometers to be used for LSD testing are malfunctioning due to leaky seals. The thermocyclers used in SCID testing are not durable and may need to be replaced. Perkin Elmer is developing a SCID testing kit and a six-plex assay for LSDs, which is a simpler assay than IDPH will be using. Reagents needed for the current testing method for the endocrine disorders will be exhausted by March 2015. A contract with Perkin Elmer to obtain a new single instrument to test for hypothyroidism, congenital adrenal hyperplasia, galactosemia and biotinidase deficiency is in process, and needs to be signed by the end of 2014 to assure the ability to continue endocrine testing by March 2015 when the reagents will be depleted for the current test method.

Follow-up Program Report

Claudia Nash reported that the newborn hearing screening program was integrated into the newborn metabolic screening program in May, with the addition of four staff and two graduate students. This program receives some funding from the Centers for Disease Control and Prevention and works closely with two other state agencies, the Division of Specialized Care for Children of the University of Illinois and the Early Intervention Program of the Department of Human Services. Procedures in the two newborn screening programs will be reviewed and revised as feasible, to assure consistency for birth hospitals and medical providers.

Claudia stated the follow-up program is currently fully staffed in preparation for LSD testing to begin. A major development that is underway is an interface with the Illinois Vital Records System to allow for import of data into the newborn screening databases (metabolic and hearing) that is collected for the birth certificate. This will provide much more complete and accurate data and will reduce duplicate data entry for birth hospital staff.

Review of Data

Turn- Around Time

Dr. Dizikes reviewed data regarding turn-around time for July-August 2014. These data indicate that the average time from collection to IDPH lab receipt is 1.5 days; from receipt until abnormal tests are reported is 3.0-3.5 days and from receipt to report of NORMAL results is 7.5 days.

Screening/Diagnosed Case Data: 2012-2014

Data for each disorder category was reviewed for calendar years 2012, 2013 and 2014 to date. Incidence for positive screens has remained stable for most disorders across these three years, with the following exceptions.

MS/MS: After examining positive screening result numbers from 2012-2013, there was a 30% increase in the number of positive screening results in 2013 as compared to 2012, while the number of diagnosed cases remained stable. The primary increase was in the fatty acid oxidation disorder category, especially in these 3 disorders:

- CACT/CPT II (Acylcarnitine Translocase Deficiency/Carnitine Palmitoyltransferase II Deficiency)
- MCAD (Medium Chain Acyl-CoA Dehydrogenase Deficiency)
- VLCAD (Very Long chain Acyl-CoA Dehydrogenase Deficiency)

The laboratory reviewed normal cutoff values and 2014 data will be evaluated when the year is complete.

Galactosemia: The number of abnormal screening tests was extremely high in 2012, (837) but returned to a typical level in 2013 (169).

Diagnosed Case Data: Diagnosed cases remained stable across this period of time for all disorders. Dr. Hoganson remarked the number of diagnosed newborns with sickle cell disease seems lower in 2013, but follow-up staff indicated that there are still some unresolved cases from 2013, since some pediatric hematologists will not confirm a diagnosis until one year of age.

Other Issues

Weekend Hours-Dr. Dizikes mentioned that IDPH is exploring the possibility of a six day operation.

Blood collection cards- Bill Calvert mentioned that the laboratory recently received an order of 300,000 newborn screening collection cards and another order of 200,000 cards will arrive soon, so the supply concerns are alleviated.

Dr. Hoganson mentioned that the full Genetic and Metabolic Diseases Advisory Committee will meet October 2, 2014.

Another meeting date for the Newborn Screening and Laboratory Subcommittee has not yet been determined.

The meeting adjourned at 9:50 a.m.