Illinois Department of Public Health
Newborn Screening Laboratory Subcommittee
Illinois Department of Public Health, Division of Laboratories
2121 W. Taylor St., Chicago, Illinois
Meeting Minutes: March 4, 2015

Subcommittee Members Attending:
George Hoganson-University of Illinois at Chicago – Chair
Denise Lonigro-Advocate Christ Hospital
Gopal Srinivasan-Mt. Sinai Hospital
Ramsay Fuleihan- Lurie Children's Hospital
Kristin Clemenz-Lurie Children' Hospital

IDPH Staff:
George Dizikes, Matt Charles, Arthur Kohrman, Tom Johnson, Claudia Nash, Shannon Harrison, Jean Becker, Heather Shryock

The meeting was called to order at 9:08 AM, followed by introductions.

Old Business
The minutes were approved for the meeting held December 3, 2014. There was no other old business for discussion.

New Business
SCID Testing- Six Month Update
Data were reviewed for the 8 month period of statewide SCID testing and for the pilot test period. During this time, over 104,000 samples were screened, and there were 529 abnormal tests in preterm infants and 285 in term infants. A total of 9 newborns were diagnosed with an immunodeficiency since screening began: 1 X-liked SCID, 1 SCID variant, 1 untyped SCID, 5 newborns with DiGeorge syndrome and 1 with trisomy 21. At the last meeting of the SCID Collaborative, based on the Illinois screening data and discussions with the Massachusetts newborn screening program, the pediatric immunologists from all five Illinois immunology centers recommended that IDPH lower the cutoff from 300 TRECs to 250 TRECs. The laboratory staff are now expressing concern about lowering the cutoff, but will review this issue further internally.

Lysosomal Storage Disorders- Review of Pilot Test Implementation
The LSD pilot began November 3, 2014, with newborns from four hospitals being tested for five LSDs; Pompe, Gaucher, Fabry, MPS I and Niemann-Pick. Additional hospitals have been added to the pilot, and currently all specimens from 8 hospitals, including Prentice Women's Hospital, are included. To date, approximately 4,700 samples have been tested with 14 newborns having an abnormal finding on screening, and no cases diagnosed.

Regarding Krabbe molecular testing, attempts to secure a contract with the New York state laboratory have failed, due to inconsistencies between IDPH and NY attorneys regarding the required contract language. A contractual arrangement is being finalized through the University of Illinois at Chicago to send samples to Mayo laboratory for this testing. Dr. Dizikes stated that Mayo has agreed to perform pyschosine and 30kb deletion testing on all samples submitted, with a 24 hour turn-around time, and will conduct sequencing for specimens as needed, with a proposed 7 day turn-around time. Dr.

Hoganson stated, that since this deviates from the protocol originally approved by the LSD subcommittee, this change should be reviewed and approved at the next meeting of the LSD subcommittee, as well as the timing for how and when results should be reported by IDPH. It is anticipated that Krabbe will be added to the pilot prior to further expansion of testing specimens for LSDs from other hospitals.

Review of 2013/2014 Data

Abnormal screening data and diagnosed case data were compared for 2013 and 2014. During 2014, there were approximately 100 more samples with a positive amino acid screening result and 155 more with an abnormal fatty acid oxidation result than in 2013, although there were 32% fewer diagnosed cases in these categories in 2014. Other categories were comparable for the two years.

Some pediatricians and specialists have questioned why IDPH is reporting test results as "presumptive positive" for some amino, fatty acid and organic acid analytes that are in the normal range (all of these cases have been normal with further diagnostic testing). Dr. Dizikes indicated other ratios are abnormal in these cases, but no explanation has been provided on the report to physicians. Dr. Dizikes also indicated there are other cutoffs used for older babies. Dr. Hoganson requested that an explanatory note be added to these reports to lessen the confusion for physicians. Dr. Hoganson and Dr. Kohrman also requested that IDPH compare screening/diagnosed case data with that in other states and to the Illinois birth rate.

Information regarding newborn screening testing turn-around time was reviewed for December 2014 to February 2015, which indicates a mean time of 1.6 days from specimen collection to lab receipt, 4.1 days from receipt to report of abnormal results and 9.4 days from specimen receipt to report of normal results.

Adrenoleukodystrophy

Claudia Nash indicated that legislation has been introduced to add X-linked adrenoleukodystrophy (ALD) to the Illinois newborn screening panel. IDPH staff have gathered information from New York, the only state screening for ALD, and the IDPH Director, Dr. Shah, has been discussing this issue with the legislative sponsor. Further information about ALD newborn screening was provided at the February 2015 meeting of the HHS Discretionary Advisory Committee for Heritable Disorders in Newborns and Children, which may vote on the inclusion of ALD later this summer.

Timeliness Issues

Recommendations were made at the February 2015 meeting of the HHS Discretionary Advisory Committee for Heritable Disorders in Newborns and Children, regarding five areas related to newborn screening timeliness. State newborn screening programs are being encouraged to address these areas, with a goal of 95% compliance by 2017. They are:

1- All initial specimens should be collected by 48 hours of life
2- All specimens should be received in lab within 24 hours of collection
3- Results of all time critical tests should be reported by the 5th day of life
4- Results of all presumptive positive results should be reported by the 7th day of life
5- All tests should be completed by the 7th day of life

Newborn screening staff will track these metrics on a quarterly basis beginning with a baseline of the fourth quarter of calendar year 2014.

IT System Developments

Birth Related Data System
Progress is being made toward developing an interface between the newborn screening data system and the vital records (birth certificate) data system. This will allow for direct population of information from the birth record into the newborn screening system, which will yield more accurate and more complete information, as well as document that a newborn screening specimen has been received for each birth. Testing is planned for the summer of 2015 at the two birth hospitals in Springfield.

Direct Access to Newborn Screening Results by Provider (eReports)
A feature in the newborn screening data system is being developed which would allow hospitals and physicians to look up and print newborn screening results for their patients. It is anticipated this feature may be available by late summer 2015.

The meeting adjourned at 10:05 a.m.