

Illinois Department of Public Health (IDPH)
Genetic and Metabolic Diseases Advisory Committee
Newborn Screening Laboratory Subcommittee
Minutes – May 21, 2008
2121 W. Taylor St., Rm. 139E
Chicago, Illinois

Subcommittee Members in Attendance

Dr. George Hoganson (Subcommittee Chair), University of Illinois at Chicago
Kristin Culp-Clementz, Children's Memorial Hospital

Audio Conference Attendance

Sunetra Reddy, University of Chicago
Dr. Patrick Zeller, Pediatric Endocrinologist

IDPH Staff

Dr. David Jinks, Newborn Screening Laboratory
Mike Petros, Newborn Screening Laboratory
Barbara DeLuka, Genetics/Newborn Screening Program
Heather Gardner, Genetics/Newborn Screening Program

IDPH Audio Conference Attendance

Claudia Nash, Genetics/Newborn Screening Program
Kate Seymore, Genetics/Newborn Screening Program

Call to order by Dr. Hoganson at 9:20 AM

Introductions

Minutes of February 20, 2008 meeting were reviewed and approved.

IDPH Laboratory Report and Discussion

Dr. Jinks presented an analysis of all confirmed disorders for the period July 2002 through April 2008. During this period 1,078,905 specimens were tested, and 1645 confirmed newborn screening disorders were reported, with a combined incidence of 1 disorder for every 657 specimens. The incidence of MS/MS disorders was 1 in 4,611 specimens, and for cystic fibrosis was 1 in every 5,800 specimens. The confirmed disorder total also includes galactosemia carriers, and this policy may be revised in the future. Although few states with birthrates equal to Illinois have performed MS/MS screening for as many years, these incidence rates are similar to those of other states with comparable racial and ethnicity mixes.

With regard to alpha thalassemia reporting, both the laboratory and follow-up program are prepared to begin reporting positive results on samples with 25% or greater Bart's hemoglobin. These results indicate possible alpha thalassemia, hemoglobin H disease, and three to four positive screens requiring follow-up services will be expected each week.

Cystic fibrosis (CF) screening is progressing and to date six babies with two CFTR mutations have been identified and subsequently diagnosed with CF*. The current IRT cut-off is 150 ng/mL, although Dr. Jinks indicated other states utilize a cut-off value of 170 ng/mL. The IRT cutoff may be revised as more data on confirmed cases becomes available.

* As of 5/22/08, a total of seven confirmed cases of CF have been reported, six with two CFTR mutations detected and an additional case with one CFTR mutation detected by screening.

Increased incidence of congenital hypothyroidism cases was again discussed. Dr. Zeller indicated a world-wide increased incidence has been reported, and the Illinois increased incidence may be associated with increases in Hispanic and Asian birthrates in Illinois, as these populations tend to have a higher incidence of hypothyroidism. Mike Petros reported that TSH cut-off value has not been changed over the past few years, and that of diagnosed hypothyroidism cases, 18.5% were detected on a second screen; 39.8% of confirmed cases had specimens with a normal T4 value and were detected by elevated TSH values. This does not reflect a change from 2007 findings.

Dr. Jinks reported on the GMDAC Advisory Committee meeting held April 10. Speakers from CDC provided information about LSD testing related to plans for IDPH pilot testing of sample preparation and testing for five LSD's and development of methods applicable to high-put through requirements of newborn screening laboratories, including liquid handling systems suitable for this type of testing. Grant funding may be offered to state programs to assist in developing pilot studies for combined assays. Dr. Morris Kletzel of Children's Memorial also presented information about a new, reliable and inexpensive method of screening for several types of severe combined immune deficiency (SCID) disorders focusing on assays for the IL7 protein. Currently, there is work in Wisconsin to develop a different testing method detecting T cell DNA receptor excision circle (TREC) copy number associated with SCID. Treatment of this fatal disorder is stem cell transplantation, which is highly successful when performed early. Dr. Kletzel is working towards on an informed consent pilot study with collection of newborn blood samples at Northwestern Memorial Hospital.

Dr. Jinks reported that the laboratory will acquire one liquid handling system and a new MS/MS instrument in order to begin development of mandated testing for five LSD disorders. This testing will require five additional plates for each specimen, one for each of the five LSD disorders. No other state is currently doing pilot testing for more than one LSD disorder. The newborn screening fee will be increased through Administrative Rule change to fund purchase of laboratory equipment and necessary renovations and to cover staffing needs, and Claudia Nash indicated this process will start after July 2008, with a possible fee increase in late 2009. The proposed fee increase was based on earlier estimates of expenses

for implementation of LSD screening was estimated to be around \$12, making the total fee \$71, which is well in line with fees charged by other state newborn screening programs.

There discussion by the chairman and other members about disposal of residual newborn screening specimens. After two months all normal specimens are decontaminated and disposed of. There were questions from the chairman and members about the Data Release Committee review process and legal considerations regarding use of residual specimens for research, and the members asked that representatives from IDPH Legal and the DRC attend the next scheduled Subcommittee meeting on August 20, 1-3 PM.

In conclusion, the members found it reasonable for the IDPH Laboratory to investigate the feasibility of newborn screening testing for SCID, and requested that input from Legal Services and the DRC regarding this issue be provided prior to the next scheduled meeting. One member questioned the effects of recent genetic discrimination legislation on newborn screening, but it was concluded that reporting of abnormal screening results is in the best interests of the newborn, and should not be affected by this legislation.

The meeting was adjourned at 11:00 AM.

Minutes prepared by Barbara DeLuka 5/23/08