

Illinois Department of Public Health  
Genetic and Metabolic Diseases Advisory Committee  
Minutes – October 29, 2009 Meeting  
Hilton Garden Inn – Springfield, IL

Members Present:

Joel Charrow, M.D., Committee Chair, Children's Memorial Hospital  
George Hoganson, M.D., University of Illinois Medical Center  
Susanna McColley, M.D., Children's Memorial Hospital  
Michael Schneider, M.D., Southern Illinois University School of Medicine  
John Ralston, M.D., Memorial Medical Center  
Lainie Friedman Ross, M.D., Ph.D., University of Chicago  
Margaret Westemeyer, M.S., CGC, University of Illinois College of Medicine in Peoria  
Barbara Goss, R.D., University of Illinois Medical Center  
Cathy Gray, R.N., University of Chicago  
Sean Clark, Parent

Other Attendees:

Pam Borchardt, Co-Executive Director, SIDS of Illinois  
Tess Rhodes, R.N., UIC, Division of Specialized Care for Children  
David Jinks, Ph.D, IDPH Newborn Screening Laboratory Director  
Tom Johnson, Chief, IDPH Division of Laboratories  
Mike Petros, IDPH Newborn Screening Laboratory Operations Manager  
Claudia Nash, IDPH Genetics/Newborn Screening Program Administrator  
Karen Burget, R.N., IDPH Genetics/Newborn Screening  
Barbara DeLuka, R.N., IDPH Genetics/Newborn Screening  
Nikki Woolverton, IDPH SIDS/Genetics/Newborn Screening  
IDPH Genetics/Newborn Screening Staff:  
Kathryn Seymore, Heather Gardner, Marti Williams, Marie Nagle, Debbie Box,  
Danielle Harrell, Doug Hart, Ben Layne

Dr. Charrow called the meeting to order at 10:05 a.m. Following introductions, the minutes of the April 30, 2009 meeting were approved.

Dr. David Jinks presented a summary of confirmed NBS disorders from June 2002 through July 2009. Over 1.14 million specimens were tested during this period, and 2,068 infants were diagnosed with some type of disorder. Dr. Jinks reviewed second tier testing for tyrosinemia type 1; and that the lab is now reporting low citrulline, which can indicate CPS or OTC deficiency. Most frequently diagnosed cases of fatty acid disorders are MCAD and SCAD, which are usually asymptomatic, and the most frequently diagnosed organic acid disorders are either 3MCC or IVA.

Barbara DeLuka provided a handout summarizing quality assurance measures for July through September 2009. There were 48,251 specimens submitted from 133 hospitals, and 75% of specimens arrived at the IDPH lab within three days of collection and 98% within five days. More than 100 hospitals use the IDPH provided UPS courier services for prompt shipping of newborn screening samples. There was a slight decline in the quality of specimens received during this period, which may be due to facilities collecting specimens on expired specimen cards or collecting specimens prior to 24 hours of age. The new specimen cards that were introduced in July request both time of birth and time of collection. If these fields are not completed and the newborn's age at time of collection cannot be calculated, specimens are tested, but are also reported as invalid and as collected prior to 24 hours of age.

Newborn screening process data for 2008 was reviewed, with the average time between birth and diagnosis at 26.5 days, slightly up from 2007. In 2008, 1.3% of 1583 presumptive positive cases were lost to follow-up prior to case closure or diagnosis, down from 1.9% in 2007. Out of the 335 diagnosed cases, 54 infants had an initial borderline test result, and 12 infants had an initial normal newborn screening test result. There was some discussion explaining the reasons for delays in some events included in the process data, especially for hemoglobin disorders. Many hematologists do not see children until two months of age, despite recommendations for prophylaxis by two months of age. Some primary care providers also are not referring to specialists on a timely basis. All detailed information on this process data is included in the packet.

Claudia Nash stated there are three follow-up positions that are vacant. Tom Johnson reported the approval of three positions to be posted for the lab. Claudia also informed the Committee that Illinois will be one of only nine states to receive an award for newborn screening from the March of Dimes on December 10, 2009.

Work is continuing on the Perkin Elmer newborn screening data system. They will be providing training to lab staff the week of November 2. Full rollout beta testing will occur in December, with full online functioning expected in April or May 2010. Claudia reported that Perkin Elmer will return to work with the follow-up program the week of November 16.

Dr. McColley reported that all 50 states will provide newborn screening for cystic fibrosis by the end of December 2009. New CLSI guidelines for sweat testing will also be available in December 2009. In Illinois, genetic counseling occurs in 97% of cases, most frequently on the day of the sweat test. Premature infants account for a large number of unresolved cases with higher IRTs due to stress/asphyxia, and in low birth weight babies, obtaining an adequate amount of sweat is not always possible. Dr. McColley explained the new designation of "CF related metabolic syndrome," which includes various mutations that are thought to be benign or cause mild symptoms.

Nikki Woolverton reported on the SIDS/Infant Mortality Program and provided the Illinois Coroner/Medical Examiner Report Form. Under Illinois law, coroners and medical examiners must report any sudden and unexpected infant death to the Department within 72 hours, using this report form. The form has been adapted from the Centers for Disease Control and Prevention's (CDC) Sudden Unexpected Infant Death Investigation (SUIDI) Reporting Form. Ms. Woolverton also explained that the Department's Office of Vital Records has implemented an Electronic Death Registration System which allows physicians, coroners and funeral directors to electronically compile death information, therefore making the process much quicker. The SIDS Program may eventually have access to this system, which would enable staff to maintain up to date data and alleviate follow-up phone calls to coroners. October was SIDS Awareness Month, and as a way to reach underserved women, the IDPH contractor, SIDS of Illinois has provided community baby showers in an effort to educate parents on safe sleep, as well as other educational efforts.

The IDPH genetic grant awards had to be decreased for the first time since 1985 due to budget constraints. Karen Burget reported that grants to genetic centers were reduced by 12%, grants to local health departments which provide genetic referral or information in 60 Illinois counties, were reduced by 16%, and grants to 10 hematology centers were reduced by 21%. Two of the original 12 hematology grants were not funded. All funding to Illinois State Genetics Plan grantees was eliminated. Funding may be reconsidered in January for these programs and one hematology center. The annual Genetics Conference was also cancelled for next year. Claudia is working to restore the funding that has been lost, especially for the state plan grants.

Claudia Nash called attention to the Genetic Testing for Breast and Ovarian Cancer handouts. Flyers and materials will be posted on the IDPH web site and will be sent through various distribution lists as a way to provide better guidelines for testing and to promote genetic counseling. These materials were developed by IDPH and the CDC through collaboration with other Midwestern states in response to the direct to consumer marketing campaign by Myriad Genetics.

Dr. Jinks will attend a meeting at the CDC on November 15 to discuss lysosomal storage disorder screening. The changes to the Newborn Metabolic Screening and Treatment Code to add testing for five LSDs are in the Second Notice period, and are on the agenda for the JCAR meeting in mid-November. The rules include a newborn screening fee increase from \$59 to \$78. Dr. Ross commented that the proposed LSD testing is "research" and that she is troubled by the lack of informed consent for screening. She expressed her wish that the Committee take a stand on the continuation of this proposed screening. Dr. Charrow replied that he felt this was not necessary, since the Committee had already considered this issue. Claudia stated an opt-out provision could be discussed with IDPH Legal before the comment period ends if the Committee communicated with JCAR on this issue. Dr. Hoganson also pointed out that the

American College of Medical Genetics has a new certification which was not included in the Rules.

Dr. McColley presented information regarding a study completed by Dr. Ross, which assessed attitudes and beliefs of parents' understanding of CF newborn screening. This was one of the programs funded through the state plan grants. Phone surveys of parents of infants with positive newborn screening and negative sweat tests were conducted. Results found most parents were supportive of newborn screening for CF, and indicated 71% of parents have heard of CF, 70% understood carrier status, and 50% were not interested in carrier testing. There will be additional follow-up of these findings if funding is restored.

Dr. Charrow reported the LSD Subcommittee has drafted diagnostic protocols for each disorder, which were presented for review and revision to the Committee. These will be discussed further at their upcoming meeting. Dr. Schneider expressed interest in joining this Subcommittee.

Dr. Hoganson reported the Newborn Screening and Laboratory Subcommittee met in July. Minutes of this meeting are included in the packet.

Discussion of Development of IDPH Protocol for Newborn Screening Expansion was postponed. It may be appropriate to have a separate meeting of the Newborn Screening and Laboratory Subcommittee, with other individuals also invited to attend, to discuss this issue.

Since there was no new business, the meeting was adjourned at 2:10 p.m.