

ILLINOIS DEPARTMENT OF PUBLIC HEALTH
GENETIC AND METABOLIC DISEASES ADVISORY COMMITTEE

Minutes—April 10, 2008 Meeting
William Tell Inn—Countryside, IL

Members Present:

Susanna McColley, M.D., Children's Memorial Hospital, Substitute Chair
Debra Weese-Mayer, M.D., Rush University Medical Center
Kay Saving, M.D., St. Francis Hospital, Peoria
Sunetra Reddy, University of Chicago
Shelly Cummings, M.S., Genetic Counselor, University of Chicago
Anne Kozek, M.S., Dietician
Albert Olorvida, M.D., Family Practice Physician
Sheila Chalmers-Currin, Parent
Cathy Gray, R.N., University of Chicago
Karen Litwak, L.C.S.W., Chicago Center for Jewish Genetic Disorders
Michael Schneider, M.D., SIU School of Medicine
W. Patrick Zeller, M.D., Pediatric Endocrinologist
Carla Gillespie, R.N., Madison County Health Department

Nominees Present:

Praveen Kumar, M.D., Northwestern Memorial Hospital

Other Attendees:

Robert Vogt, Ph.D., Centers for Disease Control
Harry Hannon, Ph.D., Centers for Disease Control
Victor DeJesus, Ph.D., Centers for Disease Control
Morris Kletzel, M.D., Children's Memorial Hospital
Lainie Ross, M.D., University of Chicago
Ramsay Fuleihan, M.D., Children's Memorial Hospital
Michael Msall, Ph.D., University of Chicago
Tess Rhodes, B.S.N., DSCC
Lisa Dye, March of Dimes
Pam Borchardt, SIDS of Illinois
Bob Evanosky, Evanosky Foundation
John Adams, Newborn Screening Advocate, Toronto, Canada
Tom Johnson, IDPH Laboratories
David Jinks, Ph.D., IDPH Newborn Screening Laboratory
George Dziekes, Ph.D., IDPH Laboratories
Claudia Nash, M.S., IDPH Genetics/Newborn Screening Administrator
Karen Burget, R.N., M.P.A., IDPH Genetics/Newborn Screening
Barbara DeLuka, R.N., M.P.H., IDPH Genetics/Newborn Screening
Luna Okada, M.S., IDPH Genetics/Newborn Screening
Nikki Woolverton, M.S., IDPH Genetics/Newborn Screening

Kathryn Seymore, B.A., IDPH Genetics/Newborn Screening
Heather Gardner, IDPH Genetics/Newborn Screening

Due to absence of the Chair, Dr. Burton, the meeting was called to order at 10:10 by Dr. Susanna McColley. All attendees introduced themselves. The minutes of the October 4, 2007 meeting of the Committee were approved by vote.

The Chair reported that of the five conditions recommended to the HRSA Secretary's Advisory Committee for inclusion in the list of disorders on the "Uniform newborn screening panel", Pompe Disease and Severe Combined Immunodeficiency Disorder (SCID) have been approved by the screening subcommittee and forwarded to the external review group for in-depth study.

Dr. Jinks presented a summary of all confirmed cases of over one million infants screened since 2002. The combined frequency for all disorders is about 1/600 and 1/5,000 for MS/MS disorders. There have been two cases of cystic fibrosis (CF) diagnosed since testing began in March (1/7,500). The first case of argininemia has just recently been diagnosed. Minutes of the Laboratory Subcommittee are included in the meeting packet.

Barbara DeLuka presented a handout on newborn screening follow-up services from 2002-2007. The slight increase in lower level abnormal screens seen in 2007 is due to the new requirement for repeat testing on NICU infants. With regard to specimen submission, the percentage of specimens received by the lab in 3-5 days continues to increase. This is due to the UPS pickup and delivery service.

Dr. McColley reported on the CF pilot which began January 28. The IRT cutoff has been changed from 100 ng/ml to 150 ng/ml due to the number of false positives. In addition to referral to CF specialists, genetic counseling for the families of all babies with positive CF results is also recommended. The lab is using a cutoff of the top 4% to conduct the DNA mutation analysis. There will be missed cases that will be discovered later, since our testing only includes 44 mutations. She noted that there is no national registry for CF, however, the Cystic Fibrosis Foundation has kept a registry since 1966.

Nikki Woolverton explained the basics of the IDPH SIDS Program. IDPH contracts with SIDS of Illinois to provide support to families, and professional education programs. Coroners and Medical Examiners are required to report all sudden infant deaths to the program, who will then contact the local public health nurse and send a packet of materials and letter to the family. There has been a dramatic decrease in SIDS since 1993, from 298 cases to 88 last year. Pam Borchardt of SIDS of Illinois noted that they provide services to all families who experience a sudden infant death, whether SIDS or another cause. They also provide a connection to other families. There was some discussion of the Child Death Review Team, and the need to mandate that certain samples be collected for metabolic analysis during autopsies.

The IDPH Genetics Grant program has been in existence since 1984. Karen Burget explained that currently there is a total of \$2.5 million awarded to 68 grantees statewide; clinical genetic centers, sickle cell centers and local public health departments. Fourteen centers recently added CF counseling services. Grants to local health departments for genetic services cover 90 of the 102 counties. Also, there were over 500 attendees at the annual genetic nursing conference this past February.

Luna Okada, genetic counselor for IDPH, has been working with staff of the IDPH chronic disease prevention programs to help them include language about genomics into their state plans as they are being prepared. She reported that the State Genetics Plan was completed last year, with seven focus areas being described. Grant funding has been awarded for implementation of five of these areas; Professional Education, Billing/Reimbursement, Public Education, Reducing Barriers to Access, and Ethical, Legal and Social Issues. Updates on these are included in the meeting packet.

Claudia Nash reported that three staff have been added to the newborn screening program, with the addition of CF to the testing panel. She also noted that the Sickle Cell Disease Association of Illinois (SCDAI) is receiving a federal grant to partner with IDPH for newborn screening follow-up and education. SCDAI has also received a grant from HRSA for the Illinois Sickle Cell Action Network. This program aims to improve access to care for sickle cell families statewide, using the medical home concept. A training module has been developed for physicians, especially those working in Federally Qualified Health Centers.

Dr. Jinks reported that the lab is ready to start reporting Bart's hemoglobin for alpha thalassemia. He expects to see two to three positives per week. Kate Seymore is working with staff of Children's Memorial to finalize the reporting letter and fact sheet. Another lab concern is that 17% of confirmed cases of hypothyroidism last year were initially reported as borderline on the first test. They are willing to explore changing the cutoff values.

The afternoon speakers were Dr. Harry Hannon, Dr. Robert Vogt and Dr. Victor DeJesus from the Centers for Disease Control in Atlanta. They were presenting information related to Illinois' proposed testing for five Lysosomal Storage Diseases (LSDs). Dr. Hannon provided a history of quality assurance for newborn screening and the Newborn Screening Translation Research Initiative (NSTRI) for LSDs and Severe Combined Immunodeficiency Disorder (SCID). Dr. Vogt explained the CDC Foundation, which was created to expedite funding for any state newborn screening program to test for new disorders. As stated by Dr. DeJesus, Genzyme will provide the necessary reagents, four of which have been approved by the FDA. Quality control materials will be developed by the CDC and tested by other labs. They are currently trying to collect samples from existing patients. Claudia interjected that an Administrative Rule change will be necessary prior to implementation of this expanded screening.

Dr. DeJesus continued by explaining the CDC's role in providing protocols, reagents and training. They are working on several pilot studies to find the best way to combine

assays. They expect a very small false positive rate for this testing. Diagnostic testing will be done by labs that specialize in these disorders.

Tom Johnson presented a handout with the proposed timeline for the IDPH lab to begin testing for LSDs by December of 2010. There are a number of steps involved-- to change the Administrative Rule to increase the newborn screening fee; to initiate development of methods to test for each of the LSDs; to relocate everyone in the laboratory building to obtain and renovate the necessary space; to purchase the initial equipment; to establish positions and begin to hire the necessary staff, 7 positions projected. The Rule change itself will take about one year.

Dr. Morris Kletzel gave a presentation on a pilot being initiated at Children's Memorial to screen for SCID. This screening is inexpensive, and treatment can be provided, by means of stem cell transplantation. They have diagnosed four babies recently, at 1-5 months of age. They are working to develop a stem cell therapy that does not destroy the immune system. Twenty-three SCID patients have been transplanted during 2000-2007. This is an outpatient treatment, with quick recovery. Children's also plans to provide outreach and education activities in the future.

Claudia asked for nominations for the nine committee seats being vacated in the fall. A nomination form is included in the packet. The next meeting will be Thursday, September 18, in Springfield.

The meeting was adjourned at 2:20 p.m.

Minutes prepared by Kate Seymore.