Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee (GMDAC)
Minutes—April 19, 2012
Holiday Inn, Countryside, IL

Members Present:
Joel Charrow, M.D., Chair, Children's Memorial Hospital
George Hoganson, M.D., University of Illinois at Chicago
Praveen Kumar, M.D., Northwestern Memorial Hospital
Susanna McColley, M.D., Children's Memorial Hospital
Michael Schneider, M.D., Carle Foundation Hospital
Alexis Thompson, M.D., Children's Memorial Hospital
W. Patrick Zeller, M.D., Private Practice
Barb Goss, R.D., University of Illinois at Chicago
Colleen Gibson, R.N., LaSalle County Health Department
Cathy Gray, R.N., University of Chicago
Karen Litwack, M.S.W., Chicago Center for Jewish Genetic Disorders
Margaret Westemeyer, M.S., University of Illinois at Peoria
Cathy Wicklund, M.S., Northwestern University

GMDAC Members Not Present:
Sheila Chalmers-Currin, Parent
Sean Clark, Parent
Lainie Friedman Ross, M.D., PhD., University of Chicago
Michael Msall, M.D., University of Chicago
Albert Olorvida, M.D., Private Practice
John Ralston, M.D., Private Practice
Darrel Waggoner, M.D., University of Chicago

Other Attendees:
Zina Berryhill, Parent
Elizabeth Berry-Kravis, M.D., PhD., Rush University Medical Center
Barbara Burton, M.D., Children's Memorial Hospital
Ramsay Fuleihan, M.D., Children's Memorial Hospital
Alexa Hart, M.S.
Tess Rhodes, R.N., Division of Specialized Care for Children (DSCC)
Annemarie Valdez, SIDS of Illinois
Amy Walsh, Parent

IDPH Representatives:
Craig Conover, M.D., Medical Director
David Culp, Ph.D., Deputy Director
Tom Schafer, Deputy Director
Tom Johnson, Division Chief of Laboratories

George Dizikes, Ph.D., Acting Chief Newborn Screening Laboratory
John Nawrocki, PhD, Molecular Laboratory
Mike Petros, Dr. P.H., Newborn Screening Laboratory
Elizabeth Paton, J.D., Legal Services
Maureen McBride, Division of Laboratories

Genetics/Newborn Screening Program:
Claudia Nash, Program Administrator
Tracey Kreipe, Nurse Consultant
Shannon Harrison, Nurse Consultant
Barbara DeLuka, Nurse Consultant
Heather Shryock, Data Manager
Margie Nelson, Intern University of Illinois, Springfield
Dennis Tiburzi, Intern University of Illinois, Springfield

Introductions and Comments:
The meeting was called to order at 10:10 a.m. Following introductions, the minutes of the October 27, 2011 fall meeting were approved.

Bylaws – Review of Changes:
Claudia Nash discussed appointments and term limits, and announced that three members' terms will end this year, Barb Goss, Dr. Waggoner, and Dr. Olorvida. Drs. Waggoner and Olorvida have reached the membership term limit. Recommendations for new members to fill the openings for a dietician, geneticist, and primary care physician were encouraged and may be submitted to Claudia.

Subcommittee Reports:
Newborn Screening and Laboratory Subcommittee (NSLS)
A copy of the NSLS March 8, 2012 meeting minutes were provided to members, and Dr. Hoganson summarized the meeting for the group. The Newborn Screening Laboratory is evaluating a new non-derivatized sample preparation for tandem mass spectrometry (MS/MS) screening, and cut-off values for MS/MS results will be adjusted as necessary. The CF Collaborative has been informed that a new cystic fibrosis DNA mutation panel will be implemented later this year. MS/MS technology will be implemented for lysosomal storage disorders (LSD) screening. The need for institutional review board (IRB) review and approval for the LSD pilot was discussed due to concerns expressed at prior Advisory Committee meetings by some members, who felt LSD testing was research, requiring informed consent. Statewide screening for severe combined immune deficiency (SCID) will require high throughput PCR testing on a multiplex system, and IDPH is working to resolve equipment procurement issues, although pilot testing of a limited number of samples is feasible using currently available equipment. Based on guidelines from the Clinical Laboratories Standards Institute, NSLS members voted to send a recommendation for collection of a routine third newborn screening sample at day 28 of life for babies born at less than 34 weeks and/or weighing less than 2000 grams to all prenatal hospitals. A copy of the letter and the algorithm were provided to members.

Newborn Screening Expansion Subcommittee (NSES)

There have been no requests for review of disorders to be added to the newborn screening test panel, and no additional meetings of the NSES since the October 3, 2011 meeting.

Critical Congenital Heart Disease (CCHD) Work Group

Dr. Kumar informed members that the Illinois CCHD Work Group is focusing on identifying the newborn population to be included in the CCHD screening (well baby/NICU baby), how the screening should be performed, education and training, follow-up for positive screens, data collection, billing and hospital reimbursement, and the need to identify false negative cases. Dr. Kumar noted that pulse oximetry protocols vary among the states that have initiated the screening, but that a CCHD work group appointed by the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children has recommended screening for all babies born at $\geq$ 35 weeks gestation, and has established a protocol for the screening that the Illinois CCHD work group will encourage hospitals to adopt. There were questions from members about data collection of screening results and tracking false negative cases, and comments about the association between the genetic disorder, DiGeorge syndrome, which has immune deficiency complications. SCID screening may also detect DiGeorge cases and the CCHD often associated with this disorder. The Newborn Screening Program currently relies on the medical specialists for information about false negative cases in which babies have normal screening results, but are subsequently diagnosed with a newborn screening disorder, which would also occur with CCHD.

Cystic Fibrosis (CF) Collaborative

Dr. McColley discussed the Collaborative efforts to review CF screening and diagnostic data and to reduce the percentage of sweat tests with quantity not sufficient (QNS) results. Dr. Le Grys from the CF Foundation (CFF) is providing site visits to labs offering newborn sweat testing in an effort to improve sweat testing and reduce the number of QNS results. The CLSI has also published guidelines for sweat testing of newborns and infants. Dr. McColley mentioned a national effort by the CFF to track late diagnosed CF cases and those not identified through newborn screening. Ninety percent of all CF patients receive care and treatment through CF Centers.

Newborn Screening Laboratory Report:

Lysosomal Storage Disease Implementation Update

Dr. Dizikes reported that a new MS/MS instrument has been received that will allow pilot training of staff for LSD pilot testing of de-identified samples by October 2012. The plan is to pilot test with de-identified samples prior to testing identified samples. This will allow time to resolve the IRB review of the research/informed consent issues regarding LSD screening.

Severe Combined Immune Deficiency Implementation Update

Dr. Nawrocki reported that the validation testing for SCID screening has been optimized. The lab now has one high throughput DNA extractor and is participating in a CDC quality control program that will help determine the best cut-off values for SCID screening. A SCID pilot could be started in 2012, but statewide screening will require additional instruments due the

high number of samples that must be tested on a daily basis. Four to seven additional instruments will be needed to initiate statewide SCID screening.

The Newborn Screening Laboratory has been able to hire three new employees who are currently being trained on MS/MS testing, and will be working with Perkin Elmer for LSD test training. The Life Cycle data system is still being developed to meet laboratory needs, and auto-faxing of laboratory reports to 63 perinatal hospitals was recently implemented. Turn-around-time for newborn screening testing is less than 4.5 days for most disorders and 3 days for endocrine disorders.

Newborn Screening Follow-Up Program:

Claudia Nash introduced two new nursing staff members for the Program, and reported that due to several vacancies there is a forty percent reduction in the follow-up staffing level. Fact sheets on newborn screening disorders have been updated on the IDPH Web site, and members asked about Web site linkages with American College of Medical Genetics and other professional organizations offering newborn screening information for health care professionals and parents. This will be addressed by Program staff.

Process data on confirmed cases, abnormal screen cases and false negative cases is being compiled and evaluated for calendar years 2010 and 2011. Quality assurance reports regarding newborn screening specimen submission by perinatal hospitals was provided to the members. There was discussion about the Perinatal Network efforts to track and monitor hospitals performance and to encourage improvements in submission timeliness.

Currently, follow-up staff are participating in monthly national SCID conference calls. Staff will work to establish collaborative groups of pediatric hematologists and pediatric endocrinologists, similar to the current CF Collaborative group.

Genetic Grantee Update:

Genetics grants for fiscal year 2013 will continue to be provided to medical genetics centers and to local health departments to support genetic services for children and adults, and to pediatric hematology centers to assist in providing services for individuals with sickle cell diseases, most likely at the current funding level.

Reports from Partners:

Sudden Infant Death Services (SIDS) of Illinois

Annamarie Valdez expressed gratitude for continued IDPH grant funding. She announced that 126 perinatal hospitals will now be required to distribute "Safe Sleep" and SIDS information to all new parents. The organization continues to provide "SIDS" trainings to daycare centers and offers baby cribs to parents in need of safe sleep items.

Division of Specialized Care for Children

Tess Rhodes provided DSCC folders with new information about services, and stressed that any questions about services should be directed to her or the local DSCC sites provided in the brochures. She informed members that family travel assistance for treatment or evaluations

may be provided by DSCC, and that DSCC also provides medical case management for children with special health care needs, even though their medical conditions may not technically be covered by the agency.

Chicago Center for Jewish Genetic Disorders (CCJGD)
Karen Litwack reported that DNA Day activities for fiscal year 2012 will again include promoting awareness of genetics issues and education about genetics. A "Baby's First Test" grant from the Genetic Alliance has enabled the CCJGD to provide information about newborn screening in both English and Spanish. The "Baby's First Test" brochures are also available to local health departments and efforts are underway to provide the brochures through OB/GYN physician offices. The "Genetics 101" CD's will also be made available to local health departments and a new Spanish version will also be offered. The CCJGD is also working with public health department nurses to survey parents who receive genetics presentations to assess their understanding of the materials presented.

Sickle Cell Disease Association (SCDAI)
No representative was present.

Guest Speakers:
Dr. Berry-Kravis presented "Fragile X Newborn Screening: A Pilot Study". Rush University Medical Center is participating in this NIH grant funded, IRB approved, informed consent study offering newborn screening for fragile X using a separate dried blood spot sample. The fragile X sample is collected following collection of the IDPH newborn screening sample from babies whose mothers consent to the testing.

Alexa Hart presented her master's thesis, "Illinois Public Opinion of the Storage and Use of Newborn Screening Samples".

Comments/Discussion:
Dr. Thompson and Cathy Wicklund, who serve on the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children discussed the processes involved in review of new disorders proposed for addition to the Secretary's uniform newborn screening test panel.

Elizabeth Paton, IDPH Legal Counsel, reminded members of the IDPH required annual Open Meetings Act and Ethics trainings, and the need to print and send a certificate of completion to Claudia Nash as soon as possible.

There were questions about the possibility of participation in GMDAC meetings by teleconference and this possibility was affirmed, although logistically teleconference locations may prove difficult, and communication and discussion by teleconference can also be difficult.

The meeting was adjourned at 2:00 PM.