

Illinois Department of Public Health
Genetic and Metabolic Disease Advisory Committee (GMDAC)
Minutes – October 2, 2014
William Tell Holiday, Springfield, IL

Members Present:

Joel Charrow, M.D., Chair, Lurie Children's Hospital
Ramsay Fuleihan, M.D., Lurie Children's Hospital
Timothy Geleske, M.D., Private Practice
Susanna McColley, M.D., Lurie Children's Hospital
Mary Niewinski, R.D., University of Illinois at Chicago
Alexis Thompson, M.D., Lurie Children's Hospital
Cindy Mitchell, R.N., St. John's Hospital
Amy Walsh, Parent
Cathy Wicklund, M.S. Northwestern University

Members Not Present:

Jennifer Burton, M.S., University of Illinois at Peoria
Sheila Chalmers-Currin, Parent
George Hoganson, M.D., University of Illinois at Chicago
Karen Litwack, M.S.W., Center for Jewish Genetics
Michael Msall, M.D., University of Chicago
Lainie Friedman Ross, M.D., PhD., University of Chicago
Michael Schneider, M.D., Carle Foundation Hospital
W. Patrick Zeller, M.D., Private Practice

Other Attendees:

Stephanie Gelb, Parent
Tess Rhodes, R.N., Division of Specialized Care for Children
Adrienne Savant, M.D., Lurie Children's Hospital
Denise Wilburn, Parent

IDPH Representatives:

David Culp, Ph.D., Deputy Director
Tom Schafer, Deputy Director
Tom Johnson, Chief Division of Laboratories
Newborn Screening Laboratory Staff
 George Dizikes, Ph.D., Chief Newborn Screening Laboratory
 Rong Shao, M.D.
 Hector Diaz
Genetics/Newborn Screening Program
 Shannon Harrison, Nurse Consultant
 Heather Shryock, Data Manager
 Nikki Woolverton, Grants Manager
 Ginger Mullin, Au.D., Newborn Hearing Screening Program Coordinator

Introductions and Announcement of New Members

The meeting was called to order at 10:05 a.m. with introductions.

Review of Committee Member Appointment Terms

Dr. Joel Charrow announced nine committee nominations are still pending. Tom Schafer stated the process is delayed statewide and has been taking approximately one year for state advisory committee member nominations to be approved. Tom Schafer thanked the committee members, whose term of appointment has expired, for their continued service to this committee.

Approval of Minutes - April 17, 2014 Meeting

Denise Wilburn stated she was unclear after reading the minutes about who was going to be charged for newborn screening results. Tom Schafer stated a \$25.00 fee would be charged to parents and/or individuals who request newborn screening results but not to medical providers. Dr. McColley motioned to approve the April 17, 2014 spring meeting minutes and Amy Walsh seconded the motion. Cathy Wicklund abstained. The April 17, 2014 spring meeting minutes were approved.

Subcommittee Reports

Newborn Screening and Laboratory Subcommittee

Dr. George Dizikes stated statewide screening for severe combined immune deficiency (SCID) began on June 9, 2014. Over 42,000 newborn screening samples have been tested. IDPH has identified one confirmed case of SCID, 2 DiGeorge cases and approximately 4 other immunodeficiency diagnoses since screening began. IDPH is referring children with positive screening results to 5 immunology centers. Dr. Dizikes stated he is going to contact the Massachusetts state laboratory to set up a teleconference to discuss their cutoff value for SCID. Massachusetts state laboratory has an assay most similar to IDPH's assay. The laboratory will also be changing from a derivatized method to a non-derivatized method after testing for LSDs begins, probably in 2015. The new non-derivatized method will allow succinylacetone testing on all samples, which should help identify possible cases of tyrosinemia type I.

Lysosomal Storage Diseases Subcommittee

Dr. Dizikes stated pilot testing will begin as soon as the software vendor Perkin Elmer can make the needed changes to the database. Until that time, lysosomal storage disorder (LSD) testing has been delayed.

Newborn Screening Cystic Fibrosis Collaborative

Dr. Susanna McColley reported the cystic fibrosis (CF) collaborative has been awarded a small grant from the Cystic Fibrosis Foundation to look at processes and help meet national goals. Dr. McColley stated the collaborative is still actively monitoring quantity not sufficient (QNS) rates for sweat testing at all CF centers. Comparative data between Illinois and the other states was presented by Dr. McColley for several different categories. Overall, Illinois is doing better than the national average. The CF Foundation grants will continue at a slightly higher level and focus on rapid and accurate diagnosis of disease.

Newborn Screening Hemoglobinopathy Collaborative

Dr. Alexis Thompson reported the hemoglobinopathy collaborative has been meeting bi-monthly by teleconference. The collaborative has been focusing on ways to improve the return of long-term follow up information from hematologists to IDPH. The long-term follow up form is currently being reviewed by collaborative specialists to make it less cumbersome in hopes of getting better compliance with completion and return. Another focus of the collaborative has been to assist patients with Medicaid in identifying a pediatric hematologist in their network. IDPH staff is drafting a list of the insurance carriers accepted at hematology center for pediatricians to use when determining Medicaid coverage for pediatric hematologists. Tess Rhodes with the Division of Specialized Care for Children

reminded the group that sickle cell is not medically eligible for the program. This is because DSCC does not have a category for hemoglobinopathies. DSCC operates under Administrative Rules that include the categories for medical eligibility. Any change to categories who require a formal process. Dr. Thompson concluded by saying the National Heart, Lung and Blood Institute is developing guidelines and educational materials for medical providers related to sickle cell disease.

Severe Combined Immune Deficiency Collaborative

Dr. Ramsay Fuleihan reported the severe combined immune deficiency (SCID) collaborative, which is comprised of 5 referral centers, has been meeting regularly every two to three months. Pilot testing began April 8, 2014 and statewide screening began on June 9, 2014. SCID data from June 9 through August 31, 2014 was provided to the group. The number of positive results has been greater in preterm infants as compared to full term infants. IDPH has received one confirmed case of SCID, 2 DiGeorge cases and approximately 4 other immunodeficiency diagnoses since screening began.

Amy Walsh asked if all centers are using the same diagnosis and treatment criteria for children referred through newborn screening. Dr. Fuleihan reported the diagnosis criteria being used by all five centers are similar, but the treatment protocols do vary slightly among the centers. Dr. Fuleihan commented that based on a treatment consortium at 25 centers, the outcome of a child is favorable if the child is transplanted before 100 days of life regardless of the treatment protocol used.

Amy Walsh reported the national Immune Deficiency Foundation is producing a video for parents of children who have positive SCID screening. The video has testimonials of other parents whose babies have been in a similar situation. When asked how the five centers have been handling this high volume of referrals, Dr. Fuleihan reported his center has been seeing these children within one week. Dr. Fuleihan added he believes the laboratory will eventually need to adjust the TREC cutoff values, but it is probably too early to do this now based on the amount of data we have.

Newborn Screening Expansion Subcommittee

Dr. Charrow reported no new activities or recent meetings for this group.

Newborn Metabolic Screening and Treatment Code Changes

Tom Johnson stated the laboratory code changes have been adopted. Changes include a specimen retention policy for negative and positive specimens and a clause stating NBS specimens may be used for quality control purposes. Tom Schafer stated the changes proposed by the Follow-up Program are still awaiting internal approval. These changes include a \$25 fee for releasing newborn screening results to parents/individuals and the addition of a section on critical congenital heart disease (CCHD) in order for the Program to establish protocols, data reporting and education guidelines. Tom Schafer stated he thought these changes will take another six months before being adopted.

Dr. David Culp stated there have been internal conversations regarding expanding the normal operating hours of the Newborn Screening Program to a six day work week. Both the laboratory and the follow-up program would work Monday through Saturday. Cathy Wicklund stated the Discretionary Advisory Committee for Heritable Disorders in Newborns and Children discussed laboratory weekend hours at their September meeting and also gave recommendations for turnaround times. She stated these recommendations should be posted on the Discretionary Advisory Committee website soon. Dr. Culp stated transitioning to a six day work week would not be easy due to staffing and union issues. He projected 25 laboratory staff members and two follow-up staff would be needed for Saturday coverage, so there would more than likely be a transitional period before full implementation of Saturday workdays could begin.

The group was asked how the laboratory would decide which conditions the laboratory will test and report on Saturdays. Dr. Charrow stated the committee will review the recommendations of the Discretionary Advisory Committee to make recommendations to the Illinois Newborn Screening Laboratory. Dr. Culp stated IDPH officials would then bring these recommendations to the Director, but this transition will likely not occur until fiscal year 2016.

Tom Schafer stated the Department is committed to operating six days a week, but there is increased cost involved. He stated there would likely be a newborn screening fee increase that would have to go through Joint Commission on Administrative Rules (JCAR), and that the department may have to operate at a loss for a while after the six day work week begins. Individuals asked if there was anything they could do in support of the six day work week. Dr. McColley stated that public input from parents and partner organizations is critical for issues such as these.

Update: Implementation of Expanded Testing for Lysosomal Storage

Dr. Dizikes stated there is a handout of the LSD validation data provided to members in their folder. Dr. Dizikes stated the laboratory participated in a specimen exchange with PerkinElmer Laboratory for the LSD validation process. He is anticipating starting the LSD pilot with four birthing hospitals in late October, then expanding to additional hospitals before screening statewide. The main concern at this time is screening for Krabbe since IDPH does not have a contract in place with the New York state laboratory to perform the DNA analysis for Illinois. Therefore, the laboratory will only be testing for five LSDs (Fabry, Gaucher, MPS I, Niemann-Pick and Pompe) once the pilot begins but will hopefully start Krabbe testing after January 1, 2015. Dr. Culp stated the laboratory is still trying to work out a contract with New York, but is also looking at the possibility of Mayo Clinic or a nationwide bid process for Krabbe testing. Tom Schafer stated once statewide LSD testing begins, the newborn screening fee will be increased by \$2.00 for MPS I testing.

Review of Data

Heather Shryock stated several data handouts are provided in the folder, including the turnaround times from receipt of specimen by the IDPH laboratory to reporting of results; a table of SCID positive screenings from June 9-August 31, 2014; the LSD validation data; and compiled data from 2012, 2013 and a portion of the 2014 positive screens and diagnosed cases. Dr. Charrow commented that Lurie Children's Hospital had seen fewer cases of SCAD and IBCD in the past couple of years and the data provided confirms that fewer of these cases have been reported. Dr. Rong Shao stated the laboratory reviews all confirmed cases and compares their values to modify laboratory cutoffs based on data from the follow-up program.

IDPH Report

Newborn Screening Follow-Up Program

Shannon Harrison reported the follow-up program is fully staffed except for the education coordinator position. The Newborn Hearing Screening Program officially joined the metabolic program in May 2014. An organizational chart is provided to reflect these recent changes. The Newborn Screening Program is in the process of transitioning over to a new website that will be broken down into the different program sections. The website should be more user friendly so medical professionals and parents can navigate through the material easier. The newborn screening records release consent forms will also be available online.

Shannon Harrison also reported the Newborn Screening Program is in the process of merging data in the electronic birth certificate from the Illinois Vital Records System (IVRS) into the PerkinElmer

(Newborn Metabolic Screening) and HiTrack (Newborn Hearing Screening) data systems. This interface with IVRS will populate information into a module for metabolic, hearing and CCHD screening. All three vendors are still collaborating but are hopeful the program will have something to test by the end of the year. This interface will enable IDPH to track each birth to assure screening has occurred and will provide more accurate and complete information as well as save hospital and IDPH time with data entry.

Early Hearing Detection and Intervention Program

Dr. Ginger Mullin stated the Early Hearing Detection and Intervention Program joined the Newborn Metabolic Screening Program in May. This merging of teams will facilitate sharing of resources and intervening with the same population of babies in a timely manner. Newborn hearing data were presented for 2013, and Ginger stated ever since hospitals were mandated to screen infants after birth, Illinois hospitals have met the national benchmark in regards to percentage of babies screened after birth.

Genetics Program Grantees

Nikki Woolverton provided the group with a summary of the five types of grants IDPH distributes each year for services, outreach and education. Grantees includes 15 hospitals, 34 local health departments in 72 counties, eight pediatric hematology centers and two general revenue appropriations to the University of Illinois at Chicago and the SIDS of Illinois. She added IDPH was able to give an additional grant to Advocate Illinois Masonic Hospital and Warren County Health Department this year.

SIDS Program

Nikki Woolverton stated October is Sudden Infant Death Syndrome awareness month, and the IDPH SIDS Program has been participating in various activities to promote SIDS awareness and infant safe sleep practices. The SIDS Program is also involved with a three way state agency collaboration to hold press conferences in Springfield and Chicago to discuss infant safe sleep practices and SIDS risk reduction.

Educational Activities

Shannon Harrison reported the newborn screening program staff have been giving several presentations around the state to medical professionals and the perinatal network regarding the addition of SCID and LSDs to the newborn screening panel. She stated fact sheets for SCID and LSDs are available on the newborn screening website.

Reports from Partners:

Sickle Cell Disease Association of Illinois (SCDAI)

Staff from SCDAI were not present to report on their activities.

Center for Jewish Genetics

Staff from the Center for Jewish Genetics were not present to report on their activities.

Genetic Task Force of Illinois (GTFI)

Staff from GTFI were not present to report on their activities, but Cathy Wicklund stated the licensure bill for genetic counselors has been renewed.

Sudden Infant Death Services (SIDS) of Illinois, Inc.

Nikki Woolverton reported staff from SIDS of Illinois were not able to be present but sent a report. In FY15, SIDS of Illinois has trained over 5,000 health care providers, parents, social workers and nurses in grief and bereavement support for families. They have given 65 cribs to families in need through their CRIB program. In October, they received a Proclamation stating October is Infant Safe Sleep Month from Mayor Rahm Emanuel of Chicago.

Division of Specialized Care for Children (DSCC)

Tess Rhodes reported the DSCC has new brochures available with their new office locations listed. The organization is working on a new care coordination system entitled "Efforts to Outcomes." This system will help determine what issues families need assistance with, such as travel, referrals, etc. Tess Rhodes also reiterated the DSCC will help with travel, and serves children up to age of 21. If providers are unsure if a child will qualify for DSCC services, the provider should refer the child anyway because many of these children have co-morbid conditions. She also stated DSCC is exempt, so if a child is enrolled in DSCC, the child is waived from managed care organization programs. New DSCC forms can also be found on their website. Of the new disorders being added to the IDPH screening panel, Tess Rhodes stated the DSCC will only cover treatment for Pompe disease because this particular LSD affects the heart muscle.

Adjournment:

The meeting was adjourned at 1:36 PM. The next meeting will be scheduled for April 2015 in Springfield.