

**Illinois Department of Public Health
Genetic and Metabolic Diseases Advisory Committee
Meeting - October 23, 2019
Crowne Plaza Chicago SW Burr Ridge**

Members Present:

Celia Anguiano, Parent
Carrie Balian, EHDI Parent
Barbara Burton, M.D., Lurie Children's Hospital
Glynnis Cailteux, R.N., Kankakee County Health Department
Julie Fleischer, M.D., SIU School of Medicine
Lauren Hitchens, A.P.N., Lurie Children's Hospital
Rachel Katz, L.S.W., Lurie Children's Hospital
Janine Yasmin Khan, M.D., Lurie Children's Hospital
Mary Kreiter, M.D., Lurie Children's Hospital
Beth Leeth, M.S., Lurie Children's Hospital
Melanie Makhija, M.D., Lurie Children's Hospital
Shannon O'Brien, R.D., University of Illinois Medical Center
Erin Paquette, M.D., J.D., Lurie Children's Hospital
Adrienne Savant, M.D., Lurie Children's Hospital

Other Attendees:

Amy LaFine, Kankakee County Health Department
Aisha Ahmed, Lurie Children's Hospital
Sibyl Cox, SIU School of Medicine

IDPH Representatives:

Laura Ashbaugh, Newborn Screening Follow-up Nursing Supervisor
Rebecca Barnett, Grants Coordinator
Shannon Harrison Newborn Screening Follow-up Nursing Supervisor
Chrissy Knepler, Newborn Screening Follow-up Nursing Supervisor
Ginger Mullin, Newborn Hearing Screen Program Coordinator
Pam Smith, Newborn Screening Program Manager

Welcome and Introductions

At 1033 a.m., Dr Burton welcomed members and guests.

Vote on Bylaws

A motion to approve bylaws by Dr Paquette. None opposed, all in favor. Bylaws were approved.

Approval of May 21, 2019 Meeting Minutes

One correction with member present was note. No other corrections were noted. Carrie Balian gave motion to approve. No members opposed, all in favor. Meeting minutes were approved.

Committee Member Status

Pam Smith stated all members have been approved and there are no pending committee memberships. There is one vacancy on committee. Members discussed filling with a hearing community member, and an audiologist was suggested. Rich Dineen agreed. Ginger Mullin stated she will try to identify a pediatric

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audiologist for the committee. All in favor, no one opposed. Pam Smith will also take recommendations from committee members for an audiologist.

IDPH Reports

A. Newborn Screening Laboratory

**Laboratory staff was not in attendance to give updates on their agenda items.*

1. Less than 24-hour sample collection: Shannon Harrison stated that the Program will no longer be reporting abnormal test results for disorders that are not valid if collected at < 24 hours of age. The Perkin Elmer data vendor is currently working on these data system changes. Biotinidase deficiency, galactosemia, hemoglobinopathies, and severe combined immune deficiency will be the disorders reported if collected at <24 hours of age.
2. ALD screening: Laura Ashbaugh updated the committee on the status of ALD screening. There have been no issues with screening or referrals. IDPH has only identified one female carrier through screening.
3. Projected SMA implementation timeline: Shannon Harrison stated IDPH will begin screening by July 1, 2020, and is currently forming a workgroup to discuss follow-up protocols, a specialist list and reporting algorithm. During the validation phase, the IDPH laboratory did identify one positive case. The specimen was sent to Wisconsin State Health Department laboratory, and a diagnosis is pending at an Illinois referral center.
4. NBS fee increase: Pam Smith stated the IDPH newborn screening fee increased to \$128 per specimen on 1/1/2019 for the implementation of spinal muscular atrophy. There is a legislative proposal for the IDPH director to allow the Department to change fee via rules than the act because it is a much easier mechanism when a fee increase is needed.
5. GAMT legislation: On May 10, GAMT screening legislation was referred to committee assignments and went to a specific committee. No new movement has occurred since that date.
6. Legislative Rules: The follow-up staff wrote several changes to the NBS rules, and it has been passed to the laboratory. Matt Charles is now adding a clause to make it so that a fee change can be made in the rules and not in the act. Dr. Burton asked if there was a change to the rules about releasing NBS specimens to physicians. Pam Smith stated it was added to the rules' changes, but it will still have to go through the entire rules process before this can happen.

B. Newborn Screening Follow-Up Program

1. SMA designated consultants: Shannon Harrison stated the SMA workgroup will be discussing whether genetics or neurology will be the designated SMA consultants for IDPH. Dr. Burton stated a neuromuscular specialist at Lurie needs to be on workgroup. She also voiced concerns about the IDPH screening not picking up carriers, not picking up all positive babies, that we would be missing a fraction of babies, and that we need to get neuromuscular physicians involved right away.
2. Saturday operations: Pam Smith stated that the IDPH Laboratory and Follow-up Program have staff that are working a regular Tuesday-Saturday schedule. Follow-up has one registered nurse and three clerical office specialists working Tuesday-Saturday. The laboratory is not reporting on Saturdays at this time unless they find critical value, but the plan will be to report the same time-sensitive disorders that are currently reported on holidays.
3. UPS courier shipping: Shannon Harrison stated that their birthing hospitals, several federally qualified health care centers, and one Amish community have all been set up with UPS

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CampusShip services. IDPH is continuing to pay for all UPS charges for those centers that wish to use UPS courier services to ship their specimens to the IDPH Laboratory.

4. Grant funding: Rebecca Barnett stated FY19 grants were very late, but they are all closed out now. (See handouts for details)
 - a. Genetic counseling grants - \$1.55 million to centers
 - b. Sickle Cell grants - \$258,000 to eight centers
 - c. Genetics education was changed from a grant to a contract. Local health departments screen clients with Family Health History Questionnaire. They screened over 2,000 clients from April-June 2019.
 - d. FY2020 grants are back on track to be executed in a timely manner.
 - e. A new FY2020 Schaaf Yang Research Grant was created to conduct research for this fiscal year only. IDPH had to develop the grant setup from the beginning. The grant should be posted in E-grants today for one month. IDPH will need an expert level of reviewers with genetic background and requested committee volunteers to review applications. The timeline is in handouts. This competitive grant up to \$300,000 for this year only. The money came from general review funds. A legislator proposed it and passed legislation. The grant has a very short timeline and has to awarded to someone in Illinois, although it can be for clinical trials. Dr. Savant suggested to build a registry, but Dr. Fleischer stated there is already a Schaaf Yang registry. Dr. Fleischer stated she will email Dr Schaaf for suggestions.
 - i. Volunteers: Doctors Fleischer, Paquette, Makhija, Khan. Rich Dineen suggested we reach out to Dr. Mary Kreiter because the treatment is endocrinology; Dr. Burton will do it if need be.

C. Early Hearing Detection and Intervention Program

Ginger Mullin reported he Illinois EHDI Program is an unfunded mandate with shared goals by three state agencies: Illinois Department of Public Health, University of Illinois at Chicago – Division of Specialized Care for Children (DSCC), Department of Human Services, Bureau of Early Intervention. The DSCC receives funding through the Health Resource Service Administration specific to the EHDI program. DSCC is currently writing for funding which would cover April 1, 2020 through March 31, 2024. IDPH received funding through the Centers for Disease Control and Prevention

IDPH EHDI major accomplishments include continued data integration with birth certificates (IVRS), continued weekly data exchange with Medicaid at HFS to help locate families and physicians for unresolved infant records, finishing transition to HiTRACK 5 which allows for greater versatility, reporting and security. Current efforts are CDC EHDI cooperative agreement and PHHS block grant funding annual reports and an annual data submission required by December 16th (3 months early).

IDPH EHDI Data Highlights include screening rates continue at greater than 98% of all births in medical care facilities. Illinois' preliminary Loss to Follow-up at time of diagnosis (LTF) for 2018 shows a continuous decrease.

- 2017 was 30% based on the CDC algorithm
- 2018 will go in at 25%
- All time low since the 2013 birth cohort peak of 78%

Preliminary 2018 data shows at best a stable rate of enrollment into Early Intervention Services compared to last year; challenges remain with engaging families in the voluntary state program

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Early Hearing Detection and Intervention Program (EHDI)

Ginger Mullin reported on the EHDI program which works with UIC-DSCC and Part C Early Intervention to achieve the national goals of screening by 1 month; diagnosis by 3 months and intervention by 6 months for hearing screening and follow-up. Recent work focuses to finish transition to HiTRACK 5.3 by all birth facilities, refining the data integration with birth certificates and maximizing the data exchange with Healthcare and Family Services. EHDI is fully grant funded. The annual reports for both the CDC EHDI cooperative agreement and CDC Public Health and Human Services grant are being finalized for submission. The annual CDC data report for the 2018 birth cohort will be submitted by December 16, 2019.

Preliminary Programmatic data was shared revealing: statewide greater than 98% of infants are screened with an overall refer rate of 3.2%; and the loss-to follow-up for the 2018 birth cohort is anticipated to be 25% down from 30% for the 2017 birth cohort and down from 78% for the 2013 birth cohort. Enrollment in Part C Early Intervention data shows a preliminary rate of approximately 50% which is stable to lower than previous years.

II. Other Reports

- A. Newborn Screening and Laboratory Subcommittee-no report given.
- B. Newborn Screening Cystic Fibrosis Collaborative-no recent activity.
- C. Newborn Screening Hemoglobinopathy Collaborative-Shannon Harrison stated the collaborative continues to meet bi-monthly to discuss national trends, research projects, and educational efforts.
- D. Severe Combined Immune Deficiency Newborn Screening Collaborative-Shannon Harrison stated there has been no recent activity, but she will be sending out future meeting dates so the IDPH Laboratory can discuss a change in the reporting and a cutoff value change.
- E. Newborn Screening Expansion Subcommittee-no recent activity.

III. Reports from Partners

- A. Norton & Elaine Sarnoff Center for Jewish Genetics-no report given.
- B. Sickle Cell Disease Association of Illinois-no report given.
- C. Illinois Society of Genetic Professionals-no report given.
- D. Sudden Infant Death Services of Illinois, Inc.-no report given.
- E. Division of Specialized Care for Children-Tess Rhodes not available. A video was played.

IV. Other New Business

- A. Next meeting Date-discussed the possibility of a virtual meeting in six months.
- B. Emergency contact information was handed out to members.

V. Meeting was adjourned at 1:55 p.m.