

## **GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING**

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, August 25, 2017 at the Florida Department of Health Bureau of Public Laboratories, 1217 North Pearl Street, Jacksonville, Florida.

### **Call to Order:**

The meeting was called to order at 10:10 am EST by Paul Pitel, MD, Council Chairperson. Roll was taken and introductions were made.

### **Members Present:**

Paul Pitel, MD, Chairman, Hematologist  
David Auerbach, MD, Florida Hospital Association Representative  
Robert Fifer, PhD, Audiologist  
Dorothy Shulman, MD, University of South Florida (USF)  
Roberto Zori, MD, University of Florida (UF)  
Jeffrey Brosco, MD, Children's Medical Services (CMS) Deputy Secretary  
Gary Kleiner, MD, PhD, University of Miami (UM)  
Carina Blackmore, MS Vet Med, PhD, Department of Health, Delegate for the Surgeon General  
Cyril Blavo, DO, MPH & TM, Nova Southeastern University  
Lori Gephart, RN, Agency for Persons with Disabilities  
Heather Smith, Parent Consumer  
George Fox, Parent Consumer  
Brian Kirk, Newborn Screening/March of Dimes Representative

### **Guests:**

Gerold Schiebler, MD, CMS Consultant  
Jeanne Brunger, PerkinElmer, Inc.  
Larry Vroegindewey, PerkinElmer, Inc.  
Christina Vracar, AHCA  
Linh Hoang, PerkinElmer  
PJ Borandi, PerkinElmer  
Zhilin Lin, PerkinElmer  
Candice Brannen, Baebies  
Vamsee Pamula, Baebies  
Chanelle Garcia, UM Audiology  
Nathalie Chouery, UM Audiology  
Airi Matthews, UM Audiology  
Aimee Alilio, UM Audiology  
Mark Dant, National MPS Society  
Sharon Bowden, Pediatrix Medical Group  
Susan Weinger, Pediatrix Medical Group  
Karla Kiriako, Sanofi Genzyme  
Michael Wells, Sanofi Genzyme  
Helen Travers, Sanofi Genzyme

### **DOH Personnel Present:**

Cassandra Pasley, JD, BSN, Tallahassee  
Marcy Hajdukiewicz, MS, Tallahassee  
Dusty Edwards, BSN, RN, Tallahassee  
Jessica Meyer, MSW, Tallahassee

Bonnie Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville  
Patti Ryland, MT, BS, Bureau of Public Health Laboratories, Jacksonville  
Patty Parrish, Bureau of Public Health Laboratories, Jacksonville  
Gary Fields, Bureau of Public Health Laboratories, Jacksonville  
Ming Chan, PhD, Bureau of Public Health Laboratories, Jacksonville  
Sajani Mamallapalli, Bureau of Public Health Laboratories, Jacksonville  
Rachel Eastman, Contracts, Tallahassee  
Emily Reeves, BSN, RN, Nurse Follow-Up, Tallahassee  
Caitlin Roberts, Hearing Program, Tallahassee

**Attended Via Conference Call:**

Jennifer Moore, BSN, RN, Tallahassee  
Su Meter, BSN, RN, Tallahassee  
Sandra Crump, RN, Tallahassee  
Donna O'Steen, USF  
Emily Jones, March of Dimes  
Julia Jenkins, EveryLife Foundation

Dr. Paul Pitel introduced Dr. Jeff Brosco, CMS Deputy Secretary, new member of the Council and CMS representative; George Fox, new parent consumer for the Council; and Brian Kirk, March of Dimes representative for the Council.

**CMS Division Update**

Cassandra Pasley provided the Council an overview of changes resulting from the most recent legislative session update related to Child Protection Team (CPT), Newborn Screening (NBS), and Early Steps including changes in the requirements for CPT Medical Directors, authority overview of the Sexual Abuse Treatment Program, and the newly appointed CMS Forensic Interview Task Force.

Senate Bill 1124 provided mechanism for adding new disorders to Florida's newborn screening panel. The bill outlines timeframes for the GNSAC to consider addition the of disorders within one year of addition of a disorder to the Recommended Uniform Screening Panel (RUSP), then requires the program to begin screening within 18 months after the GNSAC recommends the disorder. Additionally, House Bill 1041 allows the NBS Program to release results to the newborn's parent or legal guardian, personal representative, or person designated by parent or legal guardian; added "detection" of disorders to language; and permitted four representatives from any of Florida's medical schools on the GNSAC.

Additional budget information was reviewed and the impact on the Division of CMS. Early Steps received 7 FTEs and additional funding, the NBS Hearing Program received 3 FTE, and 1 FTE was provided to the NBS Follow-up Program. The NBS Follow-up Program received funding for the implementation of x-linked Adrenoleukodystrophy (x-linked ALD).

Several Council members shared concerns regarding the possibility of not receiving current funding in the future. Cassandra Pasley clarified the funding was included in current program funds, but as with any legislative funding, could be line item vetoed in the future.

George Fox questioned funding for x-linked ALD and if additional funding would be requested for each new disorder added to the screening panel. Cassandra Pasley explained these funds are for x-linked ALD, in addition to future disorders added to Florida's panel.

Dr. Brosco suggested an independent review of new disorders up for consideration by the Council, possibly from a state currently screening for proposed disorders. Dr. Pitel requested state specific information for disorders considered by the Council.

### **Newborn Screening Laboratory Update**

Dr. Taffe addressed the Council and provided an update on staffing in the NBS Laboratory and the promotion of Sajani Mamallapalli. A quality assurance update provided to the Council regarding laboratory informing the laboratory has passed proficiency testing for the last two quarters. Penny Edwards, Dr. Garganta, and Dr. Shulman are assisting with cutoff evaluations in conjunction with the laboratory.

A data system upgrade is scheduled for January, with PerkinElmer, which will include updates for x-linked ALD testing. The laboratory is currently in the process of updating software for each disorder, which will include any necessary logic changes. The target date for Go Live of x-linked ALD is April 2018; however, this is subject to change. It will take at least 6 months to get instruments in the laboratory and begin the process of testing. The PerkinElmer kit for x-linked ALD testing is pending FDA approval.

ELO/ELR has gone live for all Baycare Hospitals. The laboratory is currently receiving 80 to 100 orders a day.

Dr. Taffe provided update regarding CDC cooperative agreement/grant to fund NextGen sequencing for SCID and lysosomal storage disorders (LSDs) project with PerkinElmer and UF. George Fox requested information regarding samples used for the project. Dr. Taffe confirmed that de-identified specimens are used.

Dr. Shulman questioned the 5-day turn-around time, for laboratory results, and where Florida rates compared to other large states. Per Dr. Taffe, Florida is on par with other large states. Dusty Edwards stated there is a report available, in the PerkinElmer data system, which assesses each facilities ability to submit satisfactory specimens (Unsatisfactory Report) and each facilities ability to submit specimens to the laboratory within 3 days (> 3 Day Report), but are currently not functioning properly. Aggregate information is available on the NBS website for the facilities. Continued education and training regarding proper collection and transit times of specimens is provided to all birth facilities for the purpose of maintaining program needs.

### **Newborn Screening Follow-Up Program Update**

Dusty Edwards provided an update for the NBS Follow-up Program. Dr. Pitel requested explanation as to how funding was divided among referral centers. Dusty Edwards provided an explanation to the Council regarding the process used to determine funding and the funding changes implemented.

The new timeline requirements for the NBS Program was reviewed to ensure members understood changes implemented with the statutory language change. Once a disorder has been approved by the GNSAC, the NBS Follow-up Program must begin testing within eighteen months. All funding received this legislative session is effective July 1<sup>st</sup>. Dr. Pitel remarked that the funding is not license to "sit on our hands," but to begin work appropriately. George Fox inquired about the process of adding disorders and if the GNSAC could decline addition of new

disorders to the Florida panel. Dusty Edwards informed the members on the process and clarified that the GNSAC may choose to make a recommendation or decline to add disorders.

NBS will be making changes to the Rule to allow the Follow-up Program to release NBS results to parents. Currently, the Follow-up Program directs parent requests to the laboratory.

Rule update (64C-7) became effective August 21, 2017. Includes pulse oximetry language and outlines responsibility inside and outside of a licensed birthing facility.

Dusty Edwards reviewed the NBS Program data with the Council and posed the question regarding what data is more beneficial for the Council. Heather Smith remarked that confirmed positive data is most useful. Several council members inquired about the “rule of 5’s” when presenting NBS data. Dr. Blackmore and Cassandra Pasley explained the “rule of 5’s” was used for privacy reasons. The Council agreed that false positives are a primary concern. Request was made to add page numbers to the data spreadsheets.

Dr. Kleiner indicated need for additional education to the SCID Referral Centers regarding SCID screening.

A review of the Medical Foods process is underway. The Long-Term Genetic Programs were provided funding to aid in maintenance of a medical foods pantry.

Emily Reeves presented plan for implementing x-linked ALD. The NBS Program goal for implementation is to begin screening by the end of the fiscal year, June 30, 2018. NBS Follow-up will be conducting monthly calls with Genetics Centers to discuss implementation and protocols. The Genetic Centers would like to have panel discussions with states currently screening for x-linked ALD. States currently screening for x-linked ALD includes: New York, Connecticut, California, Minnesota, and Pennsylvania. Dr. Brosco inquired as to what information is provided by national groups. NewSteps is a national newborn screening resource that works with states to provide data and implementation assistance and the program will be using this resource.

Dr. Pitel requested there be a discussion on the meeting minutes from February 3, 2017. Dr. Shulman made a motion to accept the draft minutes, pointing out a typo on the last page, 2<sup>nd</sup> paragraph, 5<sup>th</sup> word; and was seconded by Dr. Zori and all were in favor.

### **Baebies Presentation**

Candace Brannen provided a presentation on the Seeker Platform, with an emphasis on Pompe and MPS I.

### **PerkinElmer**

PJ Borandi provided a presentation on behalf of PE regarding their screening platform for x-linked ALD and LSDs.

### **Discussion on MPS and Pompe**

Mark Dant, with the National MPS Society, stated there are have not been any known issues with treatment reimbursement for MPS I.

The Council identified the need for a vote on MPS I and Pompe as both conditions have been added to the RUSP. Multiple concerns were discussed, regarding the voting timeframe and ensuring information is available. The Council voiced other concerns regarding Pompe and MPS I screening including lack of specialists in Florida and Genetic Center staffing.

Dr. Fifer proposed use of telemedicine in rural areas where it can be difficult for families to travel. Dr. Zori shared his experience with telehealth as a means of reaching those in outlying rural areas. Dr. Zori voiced concerns regarding confirmatory testing for false positives, as some expenses are not reimbursable. Christina Vracar from the Agency for Health Care Administration stated some expenses are reimbursable. Christina Vracar would like to be notified of issues referral centers encounter to achieve resolution and will follow up.

The Council requests Pompe and MPS I information from states that have implemented screening. Dr. Byrne will serve as lead for the Pompe discussion at the next meeting.

### **Newborn Hearing Screening Follow-Up Program Update**

Jessica Meyer provided an update for the NBS Hearing Program, including program staff. Grant updates are as follows: The NBS Hearing Program received the HRSA and EHDI grants. As a grant requirement, an EHDI-specific Advisory Committee will be developed. Twenty-five percent of this committee will be comprised of parents or family members of infants/children who are deaf or hard of hearing, and deaf or hard of hearing individuals.

September is the kick-off for the CDC grant regarding information that they would like to have reported back to them. In 2015, the State of Florida had 224,273 babies born with 96.8% screened and 2.5% not screened for hearing.

An update was provided regarding parent refusals. Data was collected for births which occurred between July 2016 and July 2017. Primary reasons for refusal include:

- Cost/insurance concerns;
- Primary care physician recommendation for refusal in the hospital, and subsequent hearing follow-up occurring at their desk.

### **Zika Screening Protocol**

Dr. Fifer provided information from Dr. Gonzalez regarding Zika screening protocols. Dr. Zori indicated Dr. Rasmussen has extensive knowledge of this topic and offered to contact her to provide additional information to the Council. An update was provided regarding Florida specific Zika screening in Florida.

### **NICU Collection Procedures**

Dr. Auerbach voiced concerns regarding false positives. Information was provided regarding a false-positive study conducted at Winnie Palmer Hospital. Amino acid changes were the largest grouping of false-positives. To test this theory, Total Parental Nutrition (TPN) was discontinued a few hours prior to specimen collection. Positive outcomes related to this study could lead to practice changes. A proposal was made not to collect specimens within the first 24 hours of life, but instead, collect first specimen on day 2 of life. This topic is to be added to the next Agenda.

### **National MPS Society**

Mark Dant, President and CEO of the MPS Society, gave his family's and other families' stories. Ryan, Mark Dant's son, was diagnosed with MPS I in 1991 at the age 3. The Ryan Foundation was created to raise awareness and disease research for MPS I.

Dr. Gerold Schiebler suggests that there should a survey conducted regarding protein foods and believes cost of medical foods should be paid for by insurance agents. Dr. Zori informed the members that UF treats 120 PKU patients. Dr. Pitel suggests having something ready for the next legislative session.

Dr. Schiebler would like to add Dr. Howell from the University of Miami to have him participate in these discussions. Dr. Pitel would like to have that done.

The meeting adjourned at 2:35 pm EST.