

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

The Genetics and Newborn Screening Advisory Council meeting was held on Friday, July 29<sup>th</sup>, 2016 at the Florida Department of Health Bureau of Laboratories, 1217 North Pearl Street, Jacksonville, Florida.

### **Call to Order:**

The meeting was called to order at 10:05 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, Practicing Pediatrician  
Robert Fifer, PhD, Audiologist - University of Miami (UM)  
Dorothy Shulman, MD, University of South Florida (USF)  
Roberto Zori, MD, University of Florida (UF)  
John Curran, MD, CMS Deputy Secretary  
Gary Kleiner, MD, PhD, University of Miami (UM)  
Carina Blackmore, MS Vet Med, PhD, Department of Health  
Cyril Blavo, DO, MPH & TM, Nova Southeastern University

### **Guests:**

Alfonso Mireles, MD, Early Hearing Detection Initiative Champion  
Gerold Schiebler, MD, CMS Consultant  
Mustafa Tekin, MD, Genetics, UM  
Amarillis Sanchez-Valle, MD, Genetics, USF  
Donna O'Steen, Genetics, USF  
Kristin Spence, Genetics, USF  
Larry Vroegindewey, PerkinElmer, Inc.  
Lindsay Raub, UF Genetics  
Stephanie Meyer, UM Genetics  
Jeanne Brunger, PerkinElmer, Inc.  
Lorrien Ragin-Dames, UM Genetics  
Rachel Mehringer, UM Audiology  
Brittany Cuevas, UM Audiology  
Allison Bechtel, MD, Nemours Jacksonville  
Vibhuti Agarwal, MD, Nemours Jacksonville  
Monique Johnson, RN, Agency for Health Care Administration (AHCA)  
George Fox, parent of child with Pompe

### **DOH Personnel Present:**

Cassandra Pasley, JD, BSN, Tallahassee  
Marcy Hajdukiewicz, MS, Tallahassee  
Dusty Edwards, BSN, RN, Tallahassee  
Pam Tempson, MSW, Tallahassee  
Bonnie Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville  
Patti Ryland, MT, BS, Bureau of Public Health Laboratories, Jacksonville  
Ming Chan, PhD, Bureau of Public Health Laboratories, Jacksonville  
Susanne Crowe, MHA, Bureau of Public Health Laboratories, Jacksonville  
Rachel Eastman, Tallahassee

**Attended Via Conference Call:**

Jennifer Moore, BSN, RN, Tallahassee  
Su Meter, BAN, RN, Tallahassee  
Emily Reeves, BSN, RN, Tallahassee

Dr. Pitel introduced Dr. Curran, CMS Deputy Secretary, as a new member of the council and the CMS representative and Dr. Kleiner was introduced as a new member of the Council and the representative from UM.

**2016 Legislative Update**

Cassandra Pasley provided the council updates on legislation related to x-linked adrenoleukodystrophy (ALD) screening and the Early steps Program, a meeting with the AHCA to address partnering on the billing process for upcoming newborn screening conditions and genetic testing, and Zika and how it relates to children with special health care needs.

The Department of Health is planning to partner with primary care physicians (PCPs) regarding possible ZIKA issues as well as OB/GYNs by reaching out to hospitals with information that provide direction and ensure there is a linkage of services with all programs available including Medicaid and Early Steps.

Dr. Blackmore provided a statewide update on Zika. Dr. Zori reported that the UF has received several requests for genetic testing regarding this virus. Dr. Curran discussed CMS' plans for management of children affected by perinatal Zika transmission. Information will be disseminated throughout the regions most affected.

The minutes from the February 19<sup>th</sup>, 2016 meeting were opened up for discussion and Dr. Pitel asked for approval. Dr. Zori motioned for approval, Dr. Auerbach seconded the motion, and all were in favor. The minutes were approved.

**Newborn Screening Laboratory Update**

Dr. Taffe addressed the council and provided an update on the Newborn Screening Laboratory. The laboratory is currently interviewing for the last two vacancies. Proficiency testing for both the first and second quarters were passed. Transit time and turnaround times have improved in the second quarter, in part due to the scanning of the NBS card.

A system upgrade has the potential for the fall from Perkin Elmer, which will aid the overall reporting mechanism for positives. The new version will assist with electronic ordering and electronic reporting project (ELO/ELR).

The laboratory has moved to the Luminex assay for Cystic Fibrosis (CF). Dr. Taffe reviewed the laboratory's process for the Hologic assay recall.

Dr. Taffe discussed the IRB approved lysosomal storage disorders (LSDs) project in coordination with PerkinElmer and UF. A technician begins on Monday and testing should begin for the 6 new lysosomal disorders. Mucopolysaccharidosis I (MPS-I) and Pompe are included in the 6 LSDs, which have been added to the RUSP. The study provides the laboratory the ability to begin testing for the disorders once FDA approval is achieved.

Dr. Zori addressed the group on behalf of the three newborn screening genetic referral centers with concerns relating to additional screening tests being cost-prohibitive and the need to address this now, as well as plan for how the diagnostic testing will be provided. The physicians have concerns as the genetic centers have not done this type of diagnostic testing prior to symptoms, which may create additional hurdles related to payment. Dr. Zori continued to express concern related to the unknown cost and timeframes associated with implementation and requested a workgroup be established to bring in experts from other states that have already addressed these issues. Dr. Taffe indicated the soonest a reliable test would be available to the program that could be used with the current instruments, prior to being mandated to do it, would be fall of 2017 or winter 2018.

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

Dusty Edwards provided an update for the Newborn Screening Follow-up Program beginning with staffing updates. Marcy Hajdukiewicz is the new Bureau Chief for Early Steps and Newborn Screening, Gerry Hicks has replaced Drew Richardson as the IT Coordinator for the Bureau, and Rachel Eastman is now the Contract Manager for Newborn Screening Referral Centers.

The three disorders discussed were Glycogen Storage Type II (Pompe), x-linked ALD, and MPS I. Testing for these disorders will be a significant change to the current testing methodology and the follow-up program. The program has reached out to other states, but there is limited information available as New York is the only state currently testing.

Dr. Zori requests for all three genetic referral centers be involved with the discussions with New York to address these issues, as previously done when the MS/MS expansion was implemented. The group discussed issues related to funding, treatments for the x-linked ALD, and the LSDs not being well defined, issues and the impact on the program related to the current disorders that have been added and the additional disorders that are likely to be added to the Recommended Uniform Screening Panel (RUSP), and the need to ensure the program is operating efficiently at the present time.

Dr. Pitel made a formal motion to 1) everyone work together for implementation for ALD, and 2) set up a formal process for how to add additional disorders and 3) analysis of the current operation of the program. Dr. Zori seconds the motion. Dr. Pitel stated an Ad Hoc Committee would be established to complete the tasks to include representatives from the 3 genetic centers, Dr. Shulman, the Laboratory, NBS Follow-up, and George Fox. All were in favor, and the motion was passed. A timeline was discussed to begin the Task Force prior to the next Advisory Council meeting in 6 months. Dr. Pitel will begin the process with a conference call to establish the task force.

Dusty Edwards addressed the CF Hologic assay recall and the steps the NBS Program has taken thus far during the recall. The next steps include a conference call with the contracted CF Referral Centers discuss a plan of action for those where the program was unable to complete a retest in the laboratory.

Dusty Edwards reviewed the NBS Follow-up statistics. Dissemination of results were presented and reviewed by the group. A significant increase in the number of partial unsatisfactory specimens was noted due to change in the way the assay is read by laboratory staff. There was recommendation by the group to look at the language of the partial unsatisfactory and clarify the language related to the reason for the requested repeat.

Dr. Auerbach questioned the NBS data spreadsheet information with regard to the number of CCHD cases and hearing loss cases. It was determined that the spreadsheet contained an error. The Program will work to create reliability/authenticity of NBS data statistics.

Dr. Shulman would like there to be a mechanism to capture the false negatives for the disorders screens. She expressed the information would be beneficial to use when adjusting cut-offs for disorders. The program would look into possible ways to obtain the information.

An update on the new specimen cards was provided. Some hospitals have received the latest version, which allow for the primary care physician, ordering physician, and National Provider Indicator number.

An update on the 64C-7 Rule development was provided. Dusty Edwards reviewed information related to the Rule Development Notice and the program is currently working on a creating a draft. Once available for dissemination, the council will be included in correspondence.

The NBS Data system and Vital Statistics has now been linked, thus creating the availability to compare contact information within our data system. This will expedite the process of calling out a referral to the referral centers and assist with finding families. The [www.FNSR.net](http://www.FNSR.net) website has continued to have increased in users. We are working on enhancements that will make it more PCP friendly.

### **Newborn Hearing Screening Update**

Pam Tempson provided the Newborn Hearing Screening Update and introduced the EHDI Pediatric Champion for hearing, Dr. Alphonso Mireles. A thorough update was provided on the status of the screened vs. not screened hearing data, eReports™, and the program's progress with pending cases. The program plan is to educate PCP's and audiologists to begin using eReports to submit hearing screening results to streamline result collection.

Drs. Zori and Auerbach requested additional information related to parental refusal. Pam Tempson explained that there may be two possible causes that the program has heard of, one relates to hospitals outsourcing the hearing screen causing an insurance coverage issue and the other relates to PCPs encouraging their patients to refuse the screen in the hospital in order to do the hearing screen in the office on the well check. Dr. Fifer stated that it would be good to determine the motivator for parent refused in order for the program to establish an approach for closing the gap.

Pam Tempson provided an update on the ELO/ELR process explaining it will dramatically improve the authenticity of the information received, reducing incorrect or missing data. Florida will be the first state to begin using ELO/ELR and other states are following our progress in hopes to establishing their own systems. Baycare Health Systems has offered to be pilot for this system and are close to being ready to go live. North Naples Hospital will be the second system to begin this testing, followed by Winnie Palmer which is the largest birthing hospital in the state.

## **New Items**

Dr. Pitel discussed the statutory language related to the Advisory Council member seats. The current language does not allow for representation for all disorders screened, which may be an issue as additional disorders are added. There was discussion on the need to look at the language in the future.

Dr. Auerbach discussed the occurrence of newborn screens he receives collected within one hour of life, with elevated TSH levels. Dr. Taffe informed the members a cutoff value would need to be valid relative to the time that was agreed upon. Dr. Shulman confirms that no result collected at less than 24 hours of age be considered for treating congenital hypothyroidism unless the TSH is greater than 150  $\mu\text{IU/ml}$ .

The meeting adjourned at 2:35 pm EST.