

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

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, September 27, 2019, 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 9:32 am, EST, by Paul Pitel, MD, Council Chairperson. Roll was taken and introductions were made.

### **Members Present:**

Paul Pitel, MD, Chairman, Hematologist  
Robert Fifer, PhD, Audiologist  
Dorothy Shulman, MD, University of South Florida (USF)  
Roberto Zori, MD, University of Florida (UF)  
Carina Blackmore, MS Vet Med, PhD, Department of Health, Delegate for the Surgeon General  
Gary Kleiner, MD, University of Miami  
Mark Weatherly, MD, Practicing Pediatrician

### **Guests:**

Larry Vroegindewey, PerkinElmer, Inc.  
Jesse Bottcher, AHCA-Medicaid  
Willa Thorson, MD  
Cheryl Garganta, MD  
Sharon Bowden, Mednax  
Courtney Trewick, Mednax  
Nancy Passerini  
Aliana Romero  
Allison Lukomski  
Gerold Schiebler, MD, CMS Consultant  
Mark Hudak, MD

### **DOH Personnel Present:**

Cassandra G. Pasley, JD, BSN, Tallahassee – CMS  
Marcy R. Hajdukiewicz, MS, Tallahassee – CMS  
Dusty Edwards, BSN, RN, Tallahassee – CMS  
Jessica Meyer, MSW, Tallahassee – CMS  
Emily Reeves, BSN, RN, Tallahassee – CMS  
Lindsey Felt, Tallahassee – CMS  
Bryan Wendel, Tallahassee – CMS  
Allison Westphal, Tallahassee – CMS  
Robert Brooks, Tallahassee – CMS  
Erik Kirk, Tallahassee – CMS  
Patty Lewandowski, Bureau of Public Health Laboratories, Tallahassee  
Bonnie Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville  
Patti Ryland, BS MT, MPH Bureau of Public Health Laboratories, Jacksonville  
Sajani Mamallapalli, Bureau of Public Health Laboratories, Jacksonville  
Elliott Bailey, Bureau of Public Health Laboratories, Jacksonville  
Rey Abrea, BS MT, MD Bureau of Public Health Laboratories, Jacksonville

### **Attended Via Conference Call:**

Barry Byrne, MD, Practicing Pediatrician (Member of GNSAC)  
Lori Gephart, APD Representative (Member of GNSAC)  
Lindsay Kahn, Florida Hospital Association (Member of GNSAC)  
Amarillis Sanchez-Valle, MD University of South Florida  
Kristin Spence, USF Genetics  
Bobbie Jean Armstrong, RN, CMS  
Sandra Crump, RN, CMS  
Susan Meter, RN, CMS  
Kirstie Gordon, RN, CMS  
Kristin Barnette, RN, CMS  
Linda Deterding, RN, CMS

### **Children's Medical Services (CMS) Division Director Update**

Mrs. Cassandra Pasley provided a CMS legislative overview.

- Telehealth
- The NBS Program will provide a Critical Congenital Heart Disease (CCHD) report to Agency for Health Care Administration (AHCA) quarterly. The first report will be sent October 31, 2019.
- PKU formula
- Funds provided to increase genetic services in Northeast Florida.

### **Newborn Hearing Screening (NBS) Follow-Up Program Update**

Mrs. Jessica Meyer provided the Newborn Hearing Screening Program update.

- An update was provided regarding grant funding for the Hearing Program.
- The NBS Hearing data was reviewed with the Council.

### **Newborn Screening (NBS) Follow-Up Program Update**

Ms. Dusty Edwards provided an NBS follow-Up Program update to the Council.

- The NBS statistics were reviewed with the Council, including review of the statewide Unsatisfactory Rate.
- Discussed the COOP Plan with the Council and additional information was requested to be presented during the next meeting.
- Updates to the Phenylketonuria (PKU) process were reviewed, including the addition of a PKU Nurse Coordinator to the Program.
- New Florida Newborn Screening Results website launched July 9, 2019.

### **Newborn Screening Laboratory Update**

Dr. Bonnie Taffe provided the Newborn Screening Laboratory update.

- New testing kits for mass spectrometry went live June 24, 2019. These new kits include X-ALD testing.
- Information regarding renovations was provided, which is in second phase.
- An update regarding the laboratory's readiness for new disorders; Pompe, Mucopolysaccharidosis Type I (MPS I), and Spinal Muscular Atrophy (SMA) was provided.

### **New GNSAC Appointment**

Dr. Pitel announce Lindsay Kahn's appointment to the Council at 11:40 am.

### **Florida's Newborn Screening Genetic Service Delivery Report**

Mrs. Emily Reeves, Mr. Bryan Wendel, and Mrs. Allison Westphal presented an overview of the report, at the request of the legislature, regarding genetic services in Florida.

- Three Task Forces were established by the GNSAC Chair: Mapping of Existing Services, Physician Expertise for Genetic Services, and Telehealth.
- A summary of findings and recommendations were presented to the Council.

### **Council Comments made during presentation:**

- Dr. Zori discussed difficulty seeing patients in the panhandle due to CMS offices closing.
- Dr. Pitel stated there is a need for a fourth genetics center; however, location needs are unclear.
- The Council discussed recommendations regarding telehealth and reimbursements to aid the referral centers.
- Dr. Blackmore proposed working with neighboring states to create a potential center since the whole region is underserved.
- Ms. Edwards recommended to have a call in October to discuss the final comments from the study with the Council. Dr. Pitel called for a meeting.
- The Council recognized the need for additional centers; however, the problem is how many centers are needed to best serve families.

### **The Future of Florida NBS**

Dr. Paul Pitel discussed current challenges the NBS Program is facing and the future of newborn screening.

Dr. Pitel recommended a second study should be completed based on the NBS Program Study conducted in 2002. Dr. Zori makes the motion at 1:41 pm and Dr. Fifer seconded the motion.

### **Approval of Minutes from Previous Meeting**

Dr. Shulman moved for the minutes to be accepted from the August meeting. Dr. Weatherly seconded the motion at 1:45 pm.

### **Public Comment**

Dr. Pitel called for public comment at 1:46 pm.

- Dr. Gerold Schiebler commented regarding reimbursement deficiencies and telehealth.
- Dr. Mark Hudak was recognized by Dr. Pitel to discuss CCHD screening.
- Mrs. Pasley requested Dr. Hudak be added to February's agenda to discuss CCHD.

The meeting adjourned at 2:09 pm.