

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

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, August 20, 2021, virtually through Go To Meeting platform.

### **Call to Order:**

The meeting was called to order at 9:30 am, EST, by Roberto Zori, MD, Council Chairperson. Roll was taken and introductions were made.

### **Members Present:**

Roberto Zori, MD, Chair, University of Florida (UF)  
Robert Fifer, PhD, Audiologist  
Dorothy Shulman, MD, University of South Florida (USF)  
Cynthia Gauger, MD (Hematologist)  
Gary Kleiner, MD, University of Miami (UM)  
Barry Byrne, MD, Practicing Pediatrician  
Desiree Schnoor, March of Dimes  
Lindsay Baker, Consumer  
Robert Karch, MD, Deputy Secretary of Children's Medical Services (CMS)  
George Fox, Consumer  
Carina Blackmore, MS Vet Med, PhD, Surgeon General Designee  
Jane Gibson, Ph.D., FACMG, University of Central Florida (UCF)  
Lindsay Khan, DO (Florida Hospital Association)

### **Guests:**

Alexa Bledsoe, UF  
Aliana Romero, AuD  
Amarillis Sanchez-Valle, MD, USF  
Anna McGuinness, UF  
Anton Graves  
Beena Vells  
Cheryl Garganta, MD, UF  
Colleen Lentz  
Dana Ferrell Birchfield  
Donna O'Steen, USF  
Eddy Varona  
Elizabeth Stoltz  
Gerold Schiebler, MD  
Jayson Marin, WPH  
Jean Polasky  
Jeanne Brunger, PerkinElmer, Inc.  
Judith Ranells, MD, USF  
Kristin Spence  
Larissa Vargas  
Lisa Elliott, Kansas NBS  
Marilee Lowry, Kansas NBS  
Marissa Couluris  
Mark Hudak, MD  
Meghan Kelley  
Melissa Racobaldo, USF  
Michelle Black, Kansas NBS

Michelle Mills, Kansas NBS  
Milvia Tellez, UM  
Myrtha Gregoire-Bottex, MD (UM)  
Paul Pitel, MD  
Rebecca Sieruga, AuD Extern  
Rebekah Hurd  
Sabah Bissainthe, Agency for Persons with Disabilities (APD)  
Stephanie Hacker, UM  
Willa Thorson, MD, UM  
Yanerys Colon

**DOH Personnel Present:**

Marcy R. Hajdukiewicz, MS, Tallahassee – CMS  
Dusty Edwards, BSN, RN, Tallahassee – CMS  
Kim Porter, Tallahassee – CMS  
Emily Reeves, BSN, RN, CPN, Tallahassee – CMS  
Bryan Wendel, Tallahassee – CMS  
Kristin Barnette, RN, Tallahassee – CMS  
Caitlin Roberts, Tallahassee – CMS  
Ken McCain, Tallahassee – CMS  
Lindsey Felt, Tallahassee – CMS  
Allison Westphal, RN, Tallahassee – CMS  
Linda Ebelini, BSN, RN, Tallahassee – CMS  
Makenzie Young, RN, Tallahassee – CMS  
Chantel Alfred, RN, Tallahassee – CMS  
Sandra Crump, BSN, RN, Tallahassee – CMS  
Stephane Crumbaker, RN, Tallahassee – CMS  
Jennifer Case, Tallahassee – CMS  
Jordon Reese, RN, Tallahassee – CMS  
Barbara Nolen, RN, Tallahassee – CMS  
Ericka Horne, Tallahassee - CMS  
Felicia Clowers, Tallahassee – CMS  
Peggy Waterman, Tallahassee – CMS  
Miranda Nerland, Tallahassee – CMS  
Natasha Rich, Tallahassee – CMS  
Chiquita Williams, Tallahassee – CMS  
Amie Bounds, Tallahassee – CMS  
Lisa Gidula, Tallahassee – CMS  
Bonnie Taffe, PhD, MPH, Bureau of Public Health Laboratories (BPHL), Jacksonville  
Patty Lewandowski, BPHL, Jacksonville  
Patti Ryland, BS MT, MPH, BPHL, Jacksonville  
Sajani Mamallapalli, BPHL, Jacksonville  
Reynaldo Abrea, BPHK, Jacksonville  
Elliott Bailey, BPHL, Jacksonville  
Ming Chang, BPHL, Jacksonville  
Najib Asgari, BPHL, Jacksonville  
Bijoy Anand, BPHL, Jacksonville  
Virginia Simmons, BPHL, Jacksonville  
Jennifer Martin, BPHL, Tallahassee

### **Housekeeping Items**

- Three new members of the GNSAC were introduced:
  - Dr. Cynthia Gauger, serving as a Hematologist representative.
  - Desiree Schnoor, representing the March of Dimes.
  - Lindsay Baker, serving as a consumer representative.

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

- Dr. Zori moved for the minutes to be accepted from the February 2021 meeting at 9:38 AM. The Council unanimously approved the minutes at 9:39 AM.

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

Mrs. Marcy Hajdukiewicz provided a review of ongoing and upcoming activities within CMS.

- Bills impacting NBS during the 2021 legislative session:
  - HB 833/SB 1140 - Unlawful Use of DNA
  - SB 1770/HB 1233 - Genetic Counseling
  - SB 362/HB 407 - Pediatric Cardiac Care

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

Mrs. Kim Porter provided the NBS Hearing Screening Program update to the Council.

- Hearing staff continues to train and enroll audiologists in the electronic reporting system.
  - Enhancements to the system are currently in development.
- Collaboration with the School for Deaf and Blind and the EHDI Program has led to an increase in families across the state receiving service and support.
  - A new three-year contract was executed in June 2021.
- eReports Diagnostic Module Usability Evaluation sent out to users in August 2021.
  - System changes/updates to integrate user responses.
- The NBS Hearing data was reviewed with the Council.

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

Mrs. Emily Reeves provided the NBS Follow-up Program update to the Council.

- An overview of program staff was provided.
- Current program priorities were reviewed which include:
  - Virtual Hospital Technical Assistance
  - NewSTEPS Quality Indicator Report
  - Update the NBS Guidelines
- The NBS statistics were reviewed with the Council, including statewide Unsatisfactory data.
- An overview of outreach activities was provided.
- An overview of the CCHD Reports was provided.
- An overview of the Funding Allocation Project was provided.
  - Contracted with Mercer Health & Benefits, LLC
- COVID-19 and the impacts on the NBS Program were reviewed.

## **Special Review of the Minimum Newborn/Long-Term Genetic Guidelines**

- Dr. Shulman motion made at 10:36 AM to adopt the guidelines. The Council unanimously approved the motion at 10:37 AM.

## **CCHD Discussion**

Dr. Mark Hudak initiated discussion regarding Critical Congenital Heart Disease. Dr. Hudak's discussion points were addressed by both the Council and NBS Program representatives.

----- Dr. Zori called for a ten-minute break at 11:40 AM.-----

----- The meeting reconvened at 11:50 AM.-----

## **Newborn Screening New Conditions Update**

Mrs. Kristin Barnette provided an update on new conditions that have been added to the Newborn Screening test panel.

- Reviewed updates to testing processes and protocols based on available data.
- Reviewed x-linked adrenoleukodystrophy (X-ALD) data/statistics.
- Review of Pompe and Mucopolysaccharidosis Type I (MPS I) data/statistics.
- Review of SMA data/statistics.

## **Newborn Screening Laboratory Update**

Mrs. Patricia Ryland gave presentation on Next Gen Sequencing for CF

- Video shown prior to presentation.

Dr. Bonnie Taffe provided the Newborn Screening Laboratory update.

- Provided an overview of current laboratory Quality Improvement activities.
- Provided an overview of current grant opportunities.

## **New Discussions**

- Dr. Zori proposed an overview of the NBS Program in its entirety to evaluate productivity and identify gaps for improvement purposes.
  - The formation of a small group to oversee this process was proposed.
- Motion made by Dr. Zori at 1:03 PM to form a Task Force Group to review resources available to complete a review of the NBS Program. Dr. Shulman seconded the motion. The Council unanimously approved the motion to create the Task Force group at 1:05 PM.

## **Public Comment**

Dr. Zori called for public comment at 1:08 PM.

- No submitted public comments.

The meeting adjourned at 1:10 PM.