

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, August 18, 2023, virtually through Go To Meeting Platform.

Call to Order:

The meeting was called to order at 9:38 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)
Gary Kleiner, MD, PhD, University of Miami (UM)
George Fox, Consumer
Carina Blackmore, MS Vet Med, PhD, Surgeon General Designee
Lindsay Khan, DO, Florida Hospital Association
Jane Gibson, Ph.D., FACMG, University of Central Florida (UCF)
Cynthia Gauger, MD, Hematologist
Chrishonda Jenkins, MSN, RN, Agency of Persons with Disabilities

Guests:

Alexa Bledsoe, UF
Aliana Romero, AuD, UM
Amarillis Sanchez-Valle, MD, USF
Ami Mehta
Amy Jonasson
Andrea Sheilds
Anna Fowler, AuD Student, UM
Anna McGuinness, UF
Barbara Nolen
Cheryl Garganta, MD, UF
Corine Orr, UM
Daphne Applebee, UF Health Jax
Deanna Almanza
Deeanna Hebrank
Deborah Barbouth, MD, UM
Donna O'Steen, USF
Eileen Rivera
Jasmine Burton-Akright, UF Health Jax
Jeanne Brunger, PE
Jolan Walter, MD
Judith Ranells, MD, USF
Kansas Sampson
Krista Mantay
Kristin Spence, MHSc, RD, LD/N, USF
Lizbeth Mellin
Madison Lake
Majed Dasouki, MD
Marie-Claire Rowlinson
Matthew Ellinwood, CSO, National MPS Society

Melissa Lorquet, AuD Student, UM
Melissa Racobaldo, USF
Milvia Tellez, UM
Narendra Derreddy
Nicole Couch
Omer Abdul Hamid
Pamela Trapane, MD, UF Health Jax
Renee Bomar
Riley Carroll, UF Health Jax
Saadet Andrews, MD, University of Alberta
Sarah Goltsman, AuD Student, UM
Shelby Jackson, UF Health Jax
Stephanie Hacker, UM
Vania Mishkin, UM
Willa Thorson, MD, UM

DOH Personnel Present:

Marcy R. Hajdukiewicz, MS, Tallahassee-CMS
Andrea Gary, MS, RN, Tallahassee-CMS
Dusty Stern, 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
Kari Hope, Tallahassee-CMS
Ken McCain, Tallahassee-CMS
Lindsey Felt, Tallahassee-CMS
Chantel Alfred, RN, Tallahassee-CMS
Kirstie Gordon, RN, Tallahassee-CMS
Jessica Williams, RN, Tallahassee-CMS
Linda Ebelini, RN, Tallahassee-CMS
Amy Fritsch, RN, Tallahassee-CMS
Tori Davis, RN, Tallahassee-CMS
Philicia Corbett, Tallahassee-CMS
Felicia Williams, Tallahassee-CMS
Chiquita Williams, Tallahassee-CMS
Peggy Waterman, Tallahassee-CMS
Teryl Camus, AuD, Tallahassee-CMS
Miranda Nerland, Tallahassee-CMS
Natasha Rich, Tallahassee-CMS
Dajana Ujka, Tallahassee-CMS
Eric Tano, Tallahassee-CMS
Jennifer Givens, Tallahassee-CMS
Sandy Noel, Tallahassee-CMS
Susanne Crowe, MHA, Bureau of Public Health Laboratories (BPHL)-Jacksonville
Patti Ryland, BS MT, MPH, BPHL-Jacksonville
Sajani Mamallapalli, BPHL-Jacksonville
Bijoy Anand, BPHL-Jacksonville
Radley Remo, BPHL-Jacksonville
Najib Asgari, BPHL-Jacksonville

Reynaldo Abrea, BPHL-Jacksonville

Housekeeping Items

- Agenda format was reviewed.
- Criteria for submitting public comments was reviewed.

New Conditions: Guanidinoacetate Methyltransferase (GAMT) Deficiency

Dr. Saadet Andrews presented on GAMT deficiency.

- An overview of GAMT deficiency was provided.
- A review of current treatment options was provided.
- A review of newborn screening for GAMT deficiency was provided.
- A review of recommendations for GAMT deficiency newborn screening was provided.

Public Comments

- There were no public comments submitted.

Guanidinoacetate Methyltransferase Deficiency Consideration

- Consideration for adding GAMT deficiency to the NBS screening panel was conducted by the Council.
- Dr. Zori made a motion to add GAMT deficiency to the NBS screening panel at 10:29 AM.
 - The motion was seconded by Dr. Shulman.

----- The Council voted to add GAMT deficiency to the NBS screening panel at 10:30 AM.-----

Approval of Minutes from Previous Meeting

- Dr. Zori moved for the minutes to be accepted from the February 2023 meeting at 10:27 AM. The Council unanimously approved the minutes at 10:28 AM.

Children's Medical Services (CMS) Division Director Update

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

- An overview of legislative activities was provided.
- An overview of the Division related activities was provided.
- Andrea Gary, MS, RN, was introduced as the Division Director for CMS.

----- Dr. Zori called for a ten-minute break at 10:48 AM-----

----- The meeting reconvened at 11:03 AM-----

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.
- An overview of program accomplishments was provided.

- The NewSteps Report was reviewed with the Council.
- An overview of the NBS Annual Statistics was provided.
- An overview of the Critical Congenital Heart Disease (CCHD) Reports was provided.
- An overview of upcoming conditions was provided.

Newborn Hearing Screening (NBHS) Follow-Up Program Update

Mrs. Kim Porter provided the NBHS Follow-Up Program update to the Council.

- An overview of legislative activities was provided.
 - Congenital Cytomegalovirus (cCMV) implementation
 - Children’s Hearing Aid Program
- An overview of current program activities was provided.
- The NBHS data was reviewed with the Council.

----- Dr. Zori called for a fifteen-minute break at 12:28 PM-----

----- The meeting reconvened at 12:45 PM-----

Newborn Screening (NBS) Marketing and Quality Improvement (QI) Update

Mr. Bryan Wendel provided the NBS Marketing and QI update to the Council.

- An overview of the unit was provided.
- An overview of current priorities was provided.

Newborn Screening Laboratory Update

Ms. Susanne Crowe provided the Newborn Screening Laboratory update.

- An overview of current laboratory activities was provided.
- An overview of laboratory grant activities was provided.
- An update on QI activities was provided.
- An update on future goals was provided.

Public Comments

- Dr. Jolan Walter provided a public comment regarding Chronic Granulomatous Disease (CGD) screening.
- Mrs. Felicia Morton provided a public comment regarding CGD screening.
- Dr. Amarilis Sanchez-Valle provided a public comment regarding Hydroxocobalamin availability for patients with Cobalamin conditions.

New Discussions

- The upcoming timeline for the addition of GAMT deficiency to the NBS screening panel was discussed.
 - Florida statutes outlines, the NBS Program must implement screening within 18 months, February 2025.
- The next GNSAC Meeting will be held Friday, February 16, 2024.
 - This meeting will be held in-person at the Bureau of Public Health Laboratories-Jacksonville.

- Dr. Zori discussed desire to survey various service groups to evaluate how different aspects of the NBS Program are performing related to genetic services. The survey would be used to determine where improvements are needed.
 - Service groups would include such groups as clinicians and pediatricians.

The meeting adjourned at 2:08 PM.