

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, August 23, 2024, virtually through GoTo Meeting Platform.

Call to Order:

The meeting was called to order at 9:33 AM, EST, by Roberto Zori, MD, Council Chairperson. Roll was taken, quorum established, 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)
George Fox, Consumer
Jane Gibson, PhD, FACMG, University of Central Florida (UCF)
Chrishonda Jenkins, MSN, RN, Agency of Persons with Disabilities
Tenesha Avent, March of Dimes
Barry Byrne, MD
Gary Kleiner, MD, PhD, University of Miami (UM)

Guests:

Alexander Davis
Amarilis Sanchez-Valle, MD, USF
Amy Jonasson, UF
Andrea Shields, USF
Anna Fowler, AuD Student, UM
Anna Grace Lewis
Anna McGuinness, UF
Cheryl Garganta, MD, UF
Christina Samper, AdventHealth
Daphne Applebee, UF Health Jax
Deanna Almanza, USF
Eileen Rivera
Heather Stalker, UF
Holden Duchon
Jasmine Burton-Akright, UF Health Jax
Jeanne Brunger, Revvity
Jesus Llaguno, USF
Kansas Sampson (Greenwell), UF
Lizbeth Mellin, UF Health Jax
Madeleine Franchi
Mary Lawlor-Barry
Megan Boothe
Meghan Kelley
Natalie Shallow
Nicole Couch
Pamela Trapane, MD, UF Health Jax
Rachel McCaffrey (Obi)
Renee Bomar
Riley Carroll, UF Health Jax

Ryan Brown-Ezell, GC
Shelby Jackson, UF Health Jax
Stephanie Hacker, UM
Vania Mishkin, UM
Yemila Lowry, RDN

DOH Personnel Present:

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
Sandra Crump, RN, Tallahassee-CMS
Gail Simpson, RN, Tallahassee-CMS
Karli Clark, RN, Tallahassee-CMS
Jessica Williams, RN, Tallahassee-CMS
Amy Fritsch, RN, Tallahassee-CMS
Tori Davis, RN, Tallahassee-CMS
Linda Ebelini, Tallahassee-CMS
Promise Santos, Tallahassee-CMS
Peggy Waterman, Tallahassee-CMS
Teryl Camus, AuD, Tallahassee-CMS
Miranda Nerland, Tallahassee-CMS
Natasha Rich, Tallahassee-CMS
Sharon Lundberg, Tallahassee-CMS
Gowshitha Vavilthota, Tallahassee-CMS
Jennifer Givens, Tallahassee-CMS
Celica Coleman, Tallahassee-CMS
Sandy Noel, Tallahassee-CMS
Michelle Dawson, Tallahassee-CMS
Daisy King, Tallahassee-CMS
Marie-Claire Rowlinson, Bureau of Public Health Laboratories (BPHL) -Jacksonville
Susanne Crowe, MHA, BPHL -Jacksonville
Patti Ryland, BS MT, MPH, BPHL-Jacksonville
Iruvanti Sunitha, BPHL-Jacksonville
Bijoy Anand, BPHL-Jacksonville
Najib Asgari, BPHL-Jacksonville
Sajani Mamallapalli, BPHL-Jacksonville
Xihui Newman, BPHL-Jacksonville
Yi Chen, BPHL-Jacksonville

Approval of Minutes from Previous Meeting

- Dr. Dorothy Shulman moved for the minutes to be accepted from the February 2024 meeting at 9:35 AM. Dr. Jane Gibson seconded the motion; the Council unanimously approved the minutes at 9:35 AM.

Housekeeping Items

- Dr. Roberto Zori announced the Housekeeping Items for the meeting at 9:36 AM.
 - Criteria for submitting public comments was reviewed.
 - Announced Dr. Robert Fifer's retirement.
 - Announced Lindsey Baker's (Parent Representative) end of term.
 - Announced the next GNSAC meeting date, February 21, 2025.
 - Purpose of the GNSAC meeting was reviewed.

GNSAC By-Laws

Mrs. Emily Reeves provided an overview of the proposed GNSAC By-laws at 9:39 AM.

- Dr. Dorothy Shulman motioned that the updated by-laws be accepted.
- Dr. Gary Kleiner seconded the motion.
- Council Members unanimously voted to accept the GNSAC By-Laws at 9:42 AM.

At 9:42 AM, Mrs. Reeves introduced Dr. Barry Byrne and his presentations on Krabbe Disease and Duchenne Muscular Dystrophy (DMD).

Overview of Infantile Krabbe Disease

Dr. Barry Byrne presented on Infantile Krabbe disease at 9:44 AM.

- An overview of Krabbe disease was provided, which included:
 - Signs and symptoms
 - Treatment
 - Infantile Krabbe disease prognosis (if untreated)
 - Long-term outcomes following umbilical cord blood transplant
 - Late Infantile Krabbe disease onset
 - Long-term outcomes following hematopoietic stem cell transplantation in late Infantile Krabbe disease

Krabbe Disease - Council Member Questions:

- Dr. Zori questioned the screening method efficacy for Krabbe disease.
- Dr. Zori requested a more detailed presentation on Krabbe disease screening method efficacy.

- Mrs. Reeves announced the vote to add Krabbe disease to Florida's screening panel would take place at the February 2025 GNSAC meeting, a more detailed presentation will be provided at that time.
- Dr. Zori requested the presentation and materials on Krabbe disease be provided prior to the meeting for council member review.

Krabbe Public Comments

Mrs. Reeves opened the floor for public comment regarding the presentation on Krabbe disease at 10:02 AM.

- Dr. Sanchez-Valle provided her experience as a geneticist when having to care for infants with Krabbe disease.

Overview of Duchenne Muscular Dystrophy (DMD)

Dr. Byrne presented on DMD at 10:06 AM.

- An overview of DMD was provided, which included:
 - Benefits of screening
 - Signs and symptoms
 - Confirmatory testing options
 - Genetic testing
 - Gene therapy treatment options

Duchenne Muscular Dystrophy (DMD): Council Member Questions

- Dr. Zori questioned if DMD screening was added to the federal Recommended Uniform Screening Panel.
- Mrs. Reeves informed the Council that the vote to add DMD to the RUSP was postponed until the winter Advisory Committee on Heritable Disorders in Newborns and Children meeting.
- George Fox provided comment on the early diagnosis and treatment to assist in extending lives for newborn screening conditions, even if the treatment is not perfect.
- Dr. Zori requested a more detailed presentation on DMD with informative handouts on the screening efficacy at an upcoming council meeting.
- Dr. Kleiner informed the council that next generation sequencing is needed for DMD testing. He then questioned if the laboratory staff could predict any impediments for testing that council members should be discussing.
- Dr. Zori commented that the state is closer to Krabbe disease approval than DMD and assessment on the readiness to implement screening and lab preparedness at next meeting.

Duchenne Muscular Dystrophy (DMD) Public Comments

Mrs. Reeves opened the floor for public comment regarding the presentation on DMD at 10:50 AM.

- Dr. Sanchez-Valle provided insight that not every condition needs to be screened during the Newborn Screening test. There are other tests that can be done later in life that won't exhaust the newborn screening system.

----- Dr. Zori called for a ten-minute break at 10:55 AM -----

----- The meeting reconvened at 11:05 AM -----

Newborn Screening (NBS) Follow-up Program Update

Mrs. Reeves provided the NBS Follow-up Program update to the Council at 11:05 AM

- An overview of program staff and staffing changes was provided.
 - New staff for Regional Perinatal Intensive Care Center and Sickle Cell Registry positions.
- NBS Educational Opportunities included:
 - Association of Women's Health, Obstetric and Neonatal Nursing Conference
 - Association of Public Health Laboratories NBS Symposium
- Opportunity to partner on the Co-Propel Grant was discussed.
- A review of the NBS annual statistics.
- An overview of the Critical Congenital Heart Disease (CCHD) reports was provided.
- An overview of new conditions was provided.
 - Guanidinoacetate methyltransferase (GAMT) deficiency and Mucopolysaccharidosis Type II (MPS II)
- An overview of the upcoming conditions was provided.
 - Krabbe disease, DMD, and Metachromatic Leukodystrophy (MLD)

Council members, George Fox and Dr. Zori stated interest in having an ethics discussion during the next GNSAC meeting.

Newborn Hearing Screening (NBHS) Follow-up Program Update

Mrs. Kimberly Porter provided the Newborn Hearing Screening (NBHS) Program update to the Council at 11:45 AM.

- An overview of the program was provided.
- An update on legislative activities was provided.
 - Congenital Cytomegalovirus (cCMV) screening and expansion implementation
 - Children's Hearing Aid Program (CHAP)
 - Senate Bill 1528 Section 4 Uniform Hearing Screening and reporting requirements.
- The Health Resources and Services Administration grant requirements were discussed to include the grant goals and activities.
- An overview of the program's Centers for Disease Control and Prevention grant requirements was provided.
- NBHS performance data was reviewed with the Council.

----- Dr. Zori called for a thirty-minute lunch break at 12:27 PM-----

----- The meeting reconvened at 1:03 PM-----

Marketing and Process Improvement Update

Mr. Bryan Wendel provided the Continuous Quality Improvement (CQI) Follow-up update at 1:03 PM.

- An overview of unit priorities was provided.
 - Marketing initiatives
 - Quality improvement activities.
- A summary of the NBS focus group and survey was provided.
 - Goals of the NBS campaign
 - Accomplishments achieved
 - Key survey themes
 - Key focus group themes
 - Recommendations for future campaigns
 - NBS response to FSU focus group and survey
- The NBS annual statistics were discussed.

Newborn Screening Laboratory Update

Ms. Susanne Crowe provided the Newborn Screening Laboratory update at 1:30 PM.

- An overview of MPS II and GAMT implementation activities was provided.
- An update on current ordering and reporting systems, including electronic ordering and reporting, was provided.
- A summary of laboratory grant activities was provided.
 - State Newborn Screening Interoperability Implementation Project
 - APHL Informatics Messaging Services (AIMS) Project
 - CDC NBS Center of Excellence
- An update on quality assurance projects was provided.

Public Comments

Mrs. Reeves opened the floor for public comments at 1:56 PM.

- No comments from the public.

New Discussions

Mrs. Reeves opened the floor for new discussions at 1:57 PM.

- Mrs. Emily Reeves requested council members monitor email for upcoming documents regarding Krabbe disease and DMD.
- The next GNSAC Meeting will be held Friday, February 21, 2025
 - This meeting will be held in person, meeting invite to be sent at the conclusion of today's meeting.
- Final farewell and best wishes to Dr. Fifer and Dr. Fifer's parting remarks.

----- George Fox motioned to adjourn, Tenesha Avent seconds the motion -----

----- The meeting adjourned at 2:03 PM -----