

GENETICS AND NEWBORN SCREENING ADVISORY COUNCIL MEETING

The Genetics and Newborn Screening Advisory Council (GNSAC) meeting was held Friday, August 24, 2018, 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 9:04 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)
Jeffrey Brosco, MD, Children's Medical Services (CMS) Deputy Secretary
Carina Blackmore, MS Vet Med, PhD, Department of Health, Delegate for the Surgeon General
Heather Smith, Parent Consumer
George Fox, Parent Consumer
Brian Kirk, Newborn Screening/March of Dimes Representative
Barry Byrne, MD, Practicing Pediatrician
David Auerbach, Florida Hospital Representative
Gary Kleiner, University of Miami

Guests:

Jeanne Brunger, PerkinElmer, Inc.
Larry Vroegindewey, PerkinElmer, Inc.
Angela Darnell, PerkinElmer
Sharon Bowden, Pediatrix Medical Group
Susan Weinger, Pediatrix Medical Group
Mario Estevez (Project Alive)
Julia Jenkins (EveryLife Foundation)
Cheryl Garganta, UF
Anna McGuinness
Alexa Bledsoe
Amy Jonasson
Amy VanMeter
Evan O'Connell
Cassandra Pio
Jolan Walter (USF SCID)
Pamela Thompson
Robert Mons (Congressman Ron DeSantis office)
London Rotundo (Senator Rubio)
Donna Fogle
Melanie Block
Vanessa O'Connell
Christina Frigo
Richard Finkel (SMA)
Susan Weinger (Pediatrix)
Sharon Bowden (Pediatrix)
Joy Sherrick (Biogen)

Bridget Diaz
Lisa McLaine
Audra Butler
Avram Joseph (MPS Superhero Foundation)
Monica Anaya
Amanda Camp
Corrine Campbell Romano
Karen Maurins

DOH Personnel Present:

Cassandra G. Pasley, JD, BSN, Tallahassee – CMS
Kim Tendrich, JD – DOH/CMS
Dusty Edwards, BSN, RN, Tallahassee – CMS
Jessica Meyer, Tallahassee – CMS
Patty Lewandowski, Bureau of Public Health Laboratories, Jacksonville
Bonnie Taffe, PhD, MPH, Bureau of Public Health Laboratories, Jacksonville
Patti Ryland, MT, BS, Bureau of Public Health Laboratories, Jacksonville
Patty Parrish, Bureau of Public Health Laboratories, Jacksonville
Ming Chan, PhD, Bureau of Public Health Laboratories, Jacksonville
Sajani Mamallapalli, Bureau of Public Health Laboratories, Jacksonville
Lori Bair, Bureau of Public Health Laboratories, Jacksonville
Melissa Rodriguez, Bureau of Public Health Laboratories, Jacksonville
Elliott Bailey, Bureau of Public Health Laboratories, Jacksonville
Sherry Ray, Bureau of Public Health Laboratories, Jacksonville
Ken McCain, Tallahassee – CMS
Erik Kirk, Tallahassee – CMS
Allison Westphal, BSN, RN, Tallahassee – CMS
Rachel Eastman, Contracts, Tallahassee – CMS
Emily Reeves, BSN, RN, Tallahassee – CMS
Charlene Willoughby, Tallahassee – CMS

Attended Via Conference Call:

Jennifer Moore, BSN, RN, Tallahassee
Su Meter, BSN, RN, Tallahassee
Sandra Crump, RN, Tallahassee
Lori Gephart, APD Representative
Emily Jones, March of Dimes
Matt Brackett, AHCA
Lori Gephart (Member of GNSAC)
Donna O'Steen (USF Genetics)
Amarillis Sanchez-Valle (USF Genetics)
Kristin Spence (USF Genetics)

Dr. Pitel thanked Dr. Auerbach for his continued participation on the Council as it is his last meeting and he will be retiring.

CMS Division Update

Cassandra Pasley gave the update for the Division of Children's Medical Services.

Kim Tendrich addressed the Council on their responsibilities regarding public meetings, public records requests, and recusing oneself during a vote. The Council is subject to Sunshine

Law/public records requests. A meeting is defined as any discussion (by any method, i.e., email, phone, text, mail, or in person) between two or more members of the Council.

- All meetings must be publicly noticed at least 7 days prior to the meeting. The NBS Program must provide documents that already exist. The NBS Program is under no obligation to create documents due to a public records request.
- Handwritten notes made by Council members are not included in public records documentation.
- Members may not talk to each other about potential issues that may be brought before the Council without being publicly noticed. Members may not communicate through DOH employees.
- Members can be asked to review documents prior to the meeting but may not discuss the document prior to the meeting.

Approval of Minutes from Previous Meeting

Dr. Pitel asked for the minutes to be accepted. Dr. Shulman moved to accept the minutes from the February meeting. Dr. Brosco seconded her motion at 9:45 am.

Newborn Hearing Screening Follow-Up Program Update

Jessica Meyer provided the Newborn Hearing Screening Program update. Information regarding staffing was provided, as well as information concerning the new Early Hearing Detection and Intervention Advisory Committee. The first committee meeting was held in April 2018.

Newborn Screening Follow-Up Program Update

Dusty Edwards provided an update to the Council.

- The NBS statistics were reviewed with the Council. Dr. Brosco inquired about the Thyroid data and suggested tracking the data to monitor trends.
- Ms. Edwards elaborated on the NBS integration of Severe Combined Immunodeficiency (SCID) with FloridaShots. The Council discussed integrating the NBS report into FloridaShots. Ms. Edwards stated the issue with providing the NBS report through FloridaShots is the accessibility requirements are different for each system.
- A Memorandum of Agreement was signed with NewSTEPS on August 1, 2018, which will allow Florida to submit data into a national data repository.
- Discussed the NBS Laboratory will be using NextGen Sequencing as a 3rd tier test for Cystic Fibrosis.
- An update was provided regarding medical foods and formula for Phenylketonuria (PKU). Council members would like to identify barriers and develop provisions for affected families.
- PKU Coordinator is being hired in NBS. This position will collaborate with the Public Health Pharmacy and other entities to maximize the resources available and how these families can receive, and improve, these benefits.

Dr. Jolan Walter gave a presentation regarding the genetic landscape and diagnostic approach for SCID.

Public Comment (11:20 am) – Below is a list of parents/advocates that provided public comment in support for adding MPS I and Pompe Disease:

- Donna Fogle
- Mario Estevez
- Melanie Block
- Avram Joseph
- Monica Anaya
- Julia Jenkins
- Christina Frego
- Vanessa O’Connell
- Jennifer Miller Smith
- Amanda Camp
- Andrea Butler
- Cassandra Pio

New Conditions Update

Mrs. Emily Reeves provided an update on X-ALD. X-ALD protocol, which included a diagnostic algorithm and protocol following diagnosis, was presented to the Council. A vote by the Council was requested to adopt the X-ALD Protocol. Dr. Brosco motioned to accept the X-ALD Protocol and was seconded by Dr. Auerbach at 1:04 pm.

Ms. Edwards provided the Council with information obtained through the Genetics and Newborn Screening Task Force regarding MPS I and Pompe Disease.

Committee votes on Pompe Disease – Dr. Pitel made a motion to add Pompe Disease to Florida’s screening panel at 2:11 pm. George Fox seconded the motion. The motion passed unanimously.

Committee votes on MPS I – Dr. Fifer made a motion to add MPS I to Florida’s screening panel at 2:12 pm. Dr. Kleiner seconded the motion. The motion passed unanimously.

Dr. Finkel gave a presentation to the Council regarding Spinal Muscular Atrophy (SMA), detailing statistics on the occurrence of SMA in the US and Florida as well as the advances in medical technology. The “Framework for Adding Newborn Screening Conditions in Florida” will be used to create a review of SMA and will be presented to the Council at the next meeting.

The meeting adjourned at 2:57 pm.