The Genetics and Newborn Screening Advisory Council (GNSAC) Task Force meeting was held by telephone conference call on Thursday, March 29, 2018.

Call to Order and Roll Call
Welcome/Introductions

The meeting was called to order at 1:03 pm EST by the chair, Dr. Paul Pitel. Those who identified themselves on the conference call are captured below.

GNSAC Task Force Members Present

Paul Pitel, MD, Chair, Hematologist
Carina Blackmore, MS Vet Med, PhD, Director, Division of Disease Control and Health Protection
Jeffrey Brosco, MD, PhD, Deputy Secretary, Children’s Medical Services
Barry Byrne, MD, Practicing Pediatrician, Cardiologist
George Fox, Parent Consumer
R. Rodney Howell, MD, Clinical Biochemical Genetics-Medical Genetics
Emily Reeves, BSN, RN, Newborn Screening Nurse Consultant
Bonita G Taffe, PhD, MPH, Director of Chemistry and Newborn Screening
Roberto Zori, MD, University of Florida Clinical Geneticist

Department of Health Personnel Present

Cassandra G. Pasley, BSN, JD
Kimberly Tendrich, CMS Senior Attorney
Dusty Edwards, BSN, RN, Director of Newborn Screening Follow-up Program
Ivy Shivers, Government Operations Consultant III

Dr. Pitel thanked and welcomed everyone to the first GNSAC Task Force meeting. He explained that the task force arose from a discussion at the most recent advisory council meeting regarding Pompe disease and Mucopolysaccharidosis Type I (MPS I).

Clarification was provided that a consulting firm had not been engaged at the time of the meeting on this topic. Dr. Pitel then directed attention to the presentation by Ms. Tendrich.

Kimberly Tendrich - Addendum to the Agenda Overview Presentation of Excerpts from the Sunshine Law, Chapter 286, Florida Statutes; Public Officers and Employees: General Provisions, Chapter 112, Florida Statutes; The Public Records Law, Chapter 119, Florida Statutes, and Form 8A

Ms. Tendrich summarized the laws regarding meeting in the sunshine; the duties of public officers, specifically relating to recusal; and public records.
Emily Reeves, BSN, RN - Overview Presentation Inventory of State Screening for Pompe and Mucopolysaccharidosis Type I (MPS I)

Ms. Reeves provided an overview of the material provided to the members for this meeting.

Nationwide statistics show that 11 states currently screen for Pompe disease, three of which have been screening since 2015. They are Illinois, Missouri and New York. Ten states screen for MPS I.

Ms. Reeves provided additional statistical information for the members’ consideration. Upon inquiry, Ms. Reeves volunteered to have office staff send members the source of the statistical information being provided.

R. Rodney Howell, MD - Presentation on Pompe

Differentiate between Infantile Onset Pompe Disease, Late Onset Pompe disease, and Pseudo-Deficiency in Pompe Disease

Dr. Howell provided various details about Pompe disease. He states that Pompe is a spectrum disease and that IOPD and LOPD can be differentiated by genetic analysis. Further, IOPD and LOPD can be differentiated from pseudo-deficiencies.

Task Force discussion included symptoms in babies as well as adults, testing at different times depending on onset of symptoms, treatment availability, genetic sequencing in relation to determining pseudo-deficiencies and avoiding false positives, second screenings, long term treatment, and the Muscular Dystrophy Association as a resource.

There was a follow-up discussion about laboratory processes in screening. Discussion included the possibility of testing approaches available, laboratory reporting, referrals, and turnaround times related to specimens sent for gene sequencing.

There was an inquiry on the status of a bill sponsored by Senator Aaron Bean related to hematopoietic stem cell transplants. Ms. Pasley will research and provide an update during the next Task Force meeting.

Ms. Edwards explained that the ability to differentiate between Pompe disease and pseudo-deficiencies will decrease the volume of referrals that require follow-up.

Dr. Byrne shared that Florida had not reported any cases of IOPD in the past three years.

Ms. Pasley requested that Dr. Byrne prepare a document for the advisory council’s review, on the full treatment of this disease. Dr. Byrne agreed to prepare a paragraph to present to the council at its August 2018 meeting.

Funding for screening was briefly discussed, with mention of a member project sponsored during the 2018 legislative session by Senator Lauren Book for funding associated with Pompe disease.

R. Rodney Howell, MD - Presentation on Mucopolysaccharidosis Type I (MPS I)

Differentiate between Pathogenic Variants and Pseudo-Deficiencies in MPS I

Dr. Howell shared information about MPS I, including symptoms and agreed to further discussion.
Based on the discussion, Ms. Reeves volunteered to obtain information from North Carolina on working with Collaborative Laboratory Integrated Reports to decrease pseudo-deficiencies in MPS I. Dr. Taffe will contact the North Carolina laboratory regarding reporting MPS I mutations. Ms. Edwards will follow-up with the Newborn Screening Translational Research Network on obtaining a list of known alleles for MPS I.

**Future Meetings**

The members chose to cancel the scheduled April meeting, and Ms. Shivers will reschedule the May meeting based upon members' availability.

The meeting adjourned at 2:59 pm.