The Genetics and Newborn Screening Advisory Council (GNSAC) Task Force meeting was held by telephone conference call on Tuesday, May 22, 2018.

Call to Order and Roll Call
Welcome/Introductions

The meeting was called to order at 3:04 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
Patricia Ryland, Administrator, Newborn Screening Laboratory, attended on behalf of Bonnie Taffe

Department of Health Personnel Present

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

REVIEW OF MINUTES

March 29, 2018 Genetics and Newborn Screening Advisory Council Task Force Meeting Minutes

Dr. Pitel called for concerns about the minutes. There were none. Dr. Brosco moved to approve the minutes. The motion passed unanimously.

PRESENTATION

Information from North Carolina regarding Mucopolysaccharidosis Type I (MPS I) and Pseudo-deficiencies, and Emily Reeves, BSN, RN

Ms. Reeves provided a briefing on North Carolina's screening procedures. Among other information, she shared that out of 60,000 screenings, 60 samples were sent for sequencing. After the starting to use the CLIR database 19 samples were sent for sequencing.

Discussion continued.
Ms. Edwards shared the following information about Illinois: Of 514,000 babies screened for MPS I, 313 positives were referred out, and of these 2 were true cases of MPS I, 96 were pseudo-deficiencies, 18 were unresolved or refusals, 13 were carriers, 148 were normal, 4 died, 30 are currently pending.

**OLD BUSINESS**

**Pompe Disease and Mucopolysaccharidosis Type I (MPS I)**

Discussion from the previous section continued, which included discussion on MPS I. Task Force members expressed a desire for additional information that would be helpful to their discussion. To assist with obtaining and providing the needed information, members were directed to submit questions to staff following this meeting. The findings will be placed on the agenda for the June meeting.

The meeting adjourned at 3:59 pm.