Ask. Listen. Learn.
Saturday, June 29 – Location: Terrace Room
8:30 a.m. to 10:00 a.m.

The UMDF is pleased to offer this special workshop session for patients and families to join in informal discussions with investigators and coordinators to Ask, Listen and Learn about various studies that are near and dear to their interests. Note: This is for informational purposes only and it is a “pilot” year for the session.

2019 Studies/Trials Participating

1. **Phase III Trial of DCA in PDC Deficiency** - Peter Stacpoole, PhD, MD and Tracie Kurtz, University of Florida, Gainesville, FL

2. **Metabolic Consequences of Primary Mitochondrial Disease**
   Marni Falk, MD* and Elizabeth McCormick, CGC*
   Development of an O2 nanosensor in Mitochondrial Myopathy
   Zarazuela Zolkipli Cunningham, MD*; Colleen Muraresku, CGC*; and Laura MacMullen, CRC*
   Development of a Mitochondrial Myopathy Rating Scale
   Zarazuela Zolkipli Cunningham, MD*; Jean Flickinger, PT, PCS*; Colleen Muraresku, CGC*; and Laura MacMullen, CRC*
   Prevalence of Past Exposures in the Mitochondrial Myopathy Patient Population
   Zarazuela Zolkipli Cunningham, MD*; Jean Flickinger, PT, PCS*; and/or Laura MacMullen, CRC*
   * Mitochondrial Medicine Frontier Program (MMFP); Center for Epigenomic and Mitochondrial Medicine (CMEM), Children’s Hospital of Philadelphia.

3. **The NIH MINI Study- Metabolism, Infection and Immunity in Mitochondrial Disease**
   Peter McGuire, MS, MBBCh; Eliza Gordon Lipkin, MD; and Shannon Kruk, RN, National Institutes of Health, National Human Genome Research Institute, Bethesda, MD

4. **North American Mitochondrial Disease Consortium (NAMDC) Registry and Biorepository** - Xiomara Q. Rosales, MD*
   Treatment of TK2 Deficiency With Thymidine and Deoxycytidine - Michio Hirano, MD* and Kris Englestad, MS, CGC*
   An Open-Label Study of Continuation Treatment With Combination Pyrimidine Nucleosides in Patients With TK2 - Michio Hirano, MD* and Kris Englestad, MS, CGC* Natural History Study - Mitochondrial Disease - Kris Engelstad, MS, CGC*
Mitochondrial Stress Brain Imaging and Epigenetic - Xiomara Q. Rosales, MD*, Marissa Cross*, and Kris Engelstad, MS, CGC*

m.3243A>G Biomarker Study - Michio Hirano, MD* and Kris Englestad, MS, CGC*

*Columbia University Medical Center, New York, NY

5. Anesthesia in Patients with Mitochondrial Disease - Maria Matuszczak, MD; Rahmat Adejumo, MBBS, MPH and Wafa Quilkhán, MBBS, UT Mitochondrial Clinic at The McGovern Medical School, Houston, TX

6. Pre-clinical Study Using Skin Biopsies from MELAS Patients to Test a Novel Pharmacological Therapeutic Strategy - Andrea Gropman, MD and Anne Chiaramello, PhD, Children’s National Medical Center, George Washington University School of Medicine, Washington, DC

7. Treatment of Nitric Oxide Deficiency with L-citrulline in MELAS: A Phase 1 Dose-Finding and Safety Study - Fernando Scaglia, MD and May Ali, Baylor College of Medicine, Houston, Texas

8. MMPower-3 Trial Update: A Phase 3, Randomized, Double-Blind, Placebo-Controlled Trial of Elamipretide in Patients With Primary Mitochondrial Myopathy - Mary Kay Koenig, MD, UT Mitochondrial Clinic at The McGovern Medical School, Houston, TX

9. A Phase Ia/Ib, SAD and MAD Study of of KL1333 in Healthy Subjects and Patients With Primary Mitochondrial Disease

Magnus Hansson, MD, PhD and/or Matilda Hugerth, NeuroVive Pharmaceutical, Lund, Sweden

10. A Phase I/II, Open Label, Single Dose Clinical Study to Evaluate the Safety and Therapeutic Effects of Transplantation of MNV-BM-BLD (Autologous cd34+ Cells Enriched With Blood Derived Mitochondria) in Pediatric Patients With Pearson Syndrome – Noa Sher, PhD, Minovia Therapeutics LTD, Tirat Hacarmel, Israel