RARE DISEASE DAY 2018

Living with a Rare Disease – The Family-Clinician-Scientist Partnership

WEDNESDAY, FEBRUARY 28, 2018

Price Center/Block Research Pavilion • LeFrak Auditorium

2:00 – 2:10 PM Introductory Remarks by Dr. Steven Walkley

MEET THE FAMILIES

2:10 – 2:35 PM The Stearns (PPM1D) – Dr. Robert Marion will introduce and interview the family; Dr. Herbert Lachman will provide a brief discussion of the gene, protein and an approach to the investigation of possible treatments.

2:35 – 3:00 PM The Foglios (SLC17A5; Salla Disease) – Dr. Melissa Wasserstein will introduce and interview the parents; Dr. Kostantin Dobrenis will provide a brief discussion of the gene and protein and possible therapy approaches.

3:00 – 3:25 PM The Flanagans (HNRNPH2) – Dr. Marion will introduce and interview the parents.

3:25 – 3:50 PM The Appells (Hermansky-Pudlak Syndrome; HPS1, AP3B1, HPS3, HPS4, HPSS, HPS6, DTNBP1 and BLOC1S3) – Dr. Marion will introduce Donna Appell, R.N., executive director and founder of the Hermansky-Pudlak Syndrome Network Inc., and her daughter, Ashley.

INTRODUCING - OPERATION IDD GENE TEAM

3:50 – 4:00 PM Drs. Wasserstein and Walkley will briefly describe ongoing efforts by the Rose F. Kennedy IDDRC and the department of pediatrics to determine the genetic cause of IDDs in patients seen in Montefiore clinics, and to advance understanding of these conditions through an innovative new program known as Operation IDD Gene Team. The relationship of this effort to the recently funded program known as NYCKidseq will also be provided.

4:00 – 5:30 PM Main Street, in Forchheimer – Join us for an interlude of informal discussions with the families over refreshments, a singing performance by Alena Galan, and music by the Einstein Jazz Ensemble.

Sponsored by the Rose F. Kennedy Intellectual and Developmental Disabilities Research Center.

http://einstein.yu.edu/centers/iddrc/