RARE DISEASE DAY 2019

Rare Disease Meets Personalized Medicine

WEDNESDAY, FEBRUARY 27, 2019

Price Center/Block Research Pavilion • LeFrak Auditorium
Forchheimer Building • Main Street

3:00–3:15 PM  **Introduction** – Dr. Steven Walkley will introduce the Rose F. Kennedy IDDRC initiative known as Operation IDD Gene Team followed by today’s program.

3:15–3:55 PM  **The DeGoris family** (*Lowe Syndrome*) – Drs. Herb Lachman and Solomon Moshe will introduce and interview the family, Corina DeGoris and her son Matt, followed by Dr. Lachman and graduate student, Jesse Barnes, providing a discussion of the syndrome and the studies being carried out in the Lachman lab.

3:55–4:35 PM  **The Manaster family** (*Gene: CACNA1A*) – Dr. Robert Marion will introduce and interview Lisa Manaster and her daughter, Emily, followed by Dr. Kamran Khodakhah and postdoctoral fellow, Dr. Heather Snell, providing an overview of the gene and the function of its protein in the brain.

4:35–5:15 PM  **The Rhew family** (*Gene: ANKS1B*) – Dr. Pamela Counts will introduce and interview April and Emeline Rhew, followed by Dr. Bryen Jordan and MSTP student, Abigail Carbonell, providing an overview of the gene and the function of its protein in the brain.

5:15–6:00 PM  **Closing Remarks** by Drs. Walkley, Marion and Wasserstein

5:15–6:00 PM  **Main Street, in Forchheimer** – Join us for an interlude of informal discussions with the families over refreshments, a viewing of an art exhibit on rare disease by Beyond the Diagnosis and music performed by the Einstein Jazz Ensemble.

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

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