

RARE DISEASE DAY 2020

Precision Medicine Meets Rare Disease
The Family-Clinician-Scientist Partnership

THURSDAY, FEBRUARY 27, 2020

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

2:00–2:15 PM **Introduction** – Dr. Steven Walkley will discuss the importance of rare disease and introduce the Rose F. Kennedy IDDRC's personalized medicine initiative known as Operation IDD Gene Team, followed by today's program.

2:15–3:00 PM **The Forsman family** *(Alpha-mannosidosis)* – Dr. Walkley will describe the disorder known as alpha-mannosidosis and a therapy his lab pioneered for this disease back in the 1990's. He will then introduce Dr. Robert Marion who will interview Sarah Forsman and her parents, Danielle and Jeff, who will tell their story from diagnosis to treatment and beyond. Audience questions to follow.

3:00–3:45 PM **The Celiberti family** *(Gene: DYNC1H1)* – Dr. Melissa Wasserstein will introduce and interview Lindsay and Tom Celiberti and their son, Tyler, followed by Dr. Arne Gennerich providing an overview of the gene and the function of its protein in the brain and other tissues. Audience questions to follow.

3:45–4:40 PM **The Robl family** *(Gene: KDM5C)* – Dr. Julie Secombe will provide an overview of the KDM5C gene and its connection to X-linked intellectual disability. MSTP student Hayden Hatch will then interview Amy and Chris Robl and their daughters Gabby and Ashley. Audience questions to follow.

4:40–4:45 PM **Closing Remarks** by Dr. Walkley

5:00–6:00 PM **Main Street, in Forchheimer** – Please join us for a poster presentation on rare disease from labs and clinics at Einstein/Montefiore and for informal discussions with our visiting rare disease families, along with refreshments and music by the Einstein Jazz Ensemble.

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

Rare Disease Day®

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