

## The Walter J. Burdette Trainee Symposium on Human Genomics

### Program

#### 9:00am Opening remarks

Monkol Lek, Assistant Professor, Department of Genetics

#### 9:20am Opening Plenary

Dr. Eimear Kenny, Associate Professor and Director, Mount Sinai Center for Genomic Health

*10:10am Coffee break*

#### 10:30am Session 1

Sumantra Chatterjee, NYU School of Medicine

*Disrupted genetic and cellular networks in Hirschsprung disease*

José L. McFaline-Figueroa, University of Washington

*Massively multiplex chemical transcriptomics at single-cell resolution*

Yen-Chen (Anne) Feng, MGH and The Broad Institute

*Ultra-rare genetic variation in the epilepsies: a whole-exome sequencing study of 17,606 individuals*

Ida Surakka, University of Michigan

*Impact of rare loss-of-function mutations on population health*

*12:30pm Lunch served in foyer*

#### 1:30pm Session 2

Arthur Lee, Boston Children's Hospital, Broad Institute of MIT and Harvard

*Leveraging single-cell functional genomics towards non-coding variant interpretation in Mendelian regulatory disorders*

Gillian Belbin, Icahn School of Medicine at Mount Sinai

*Leveraging Fine-scale Structure to Explore Disease Risk in Health Systems*

Zeynep H. Coban Akdemir, Baylor College of Medicine

*Clan genomics and the population genetic architecture of disease: Long-sized AOH regions reveal IBD haplotypes that drive mutational burden at a locus*

*3:00pm Coffee break*

#### 3:20pm Session 3

Cigdem Sevim Bayrak, Icahn School of Medicine at Mount Sinai

*Computational evaluation of mutations and genes underlying human disease*

Ryan Collins, MGH, Harvard Medical School and The Broad Institute  
*gnomAD-SV: An open resource of structural variation for medical and population genetics*

Kymerleigh Pagel, The Johns Hopkins University  
*OpenCRAVAT: an open source collaborative platform for the annotation of human genetic variation*

**4:50pm Closing remarks**

Caroline Hendry, Scientific Director, Department of Genetics

*5pm depart*