

KEYSTONE SYMPOSIA

on Molecular and Cellular Biology

Neuropsychiatric and Neurodevelopmental Disorders: Harnessing Rare Variants (A3)

January 24-27, 2021 • Embassy Suites by Hilton Boulder • Boulder, CO, USA

Scientific Organizers: Jennifer G. Mulle, Audrey Thurm, Christa L. Martin and Carrie E. Bearden

Supported by the Directors' Fund

Scholarship Deadline: October 7, 2020 / Abstract Deadline: October 21, 2020 / Discounted Registration Deadline: November 23, 2020

SUNDAY, JANUARY 24

Arrival and Registration

MONDAY, JANUARY 25

Welcome and Keynote Address

Speaker to be Announced

CNV Disorders

Raquel E. Gur, University of Pennsylvania, USA
22q11.2 Deletion and/or 22q11.2 Duplication

Jennifer G. Mulle, Emory University School of Medicine, USA
3q29 Deletion and 3q29 Duplication

Wendy K. Chung, Columbia University, USA
Deep Dive into 16p11.2 Deletion and Duplication

Short Talks Chosen from Abstracts

Roundtable Discussion 1: Different RGDs (Rare Genetic Disorders) and the Differences/Similarities Between Them

Single Gene Disorders

Speaker to be Announced

Stephan Sanders, University of California, San Francisco, USA
Phenotypes, Biology, and Therapeutic Strategies for SCN2A

Speaker to be Announced

Short Talks Chosen from Abstracts

Poster Session 1

TUESDAY, JANUARY 26

How Do We Measure Phenotypes? How Do We Harmonize across Studies?

Christa L. Martin, Geisinger Health, USA
Population Genomic Screening for Brain Disorders

Audrey Thurm, NIMH, National Institutes of Health, USA
Harmonization of Phenotypes

Elise Robinson, Harvard T.H. Chan School of Public Health and Broad Institute, USA
The NeuroDev Study: Phenotyping in African Populations

Sébastien Jacquemont, University of Montreal, Canada
Mapping the Effect of CNVs Genome-Wide on Cognition, Brain Structure and Functional Connectivity

Short Talks Chosen from Abstracts

Roundtable Discussion 2: What the Outstanding Questions are and What Tools are Needed to Answer Them?

Mechanistic Studies of RGDs

Lucy R. Osborne, University of Toronto, Canada
Mechanistic Studies of 7q11.23 Copy Number Variation

Thomas Bourgeron, Institut Pasteur, France
Functional Impact of Autism-Associated Mutations

Maria Chahrouh, University of Texas Southwestern Medical Center, USA

Neurodevelopmental Disorders: From Genetics to Mechanisms

Short Talks Chosen from Abstracts

Poster Session 2

WEDNESDAY, JANUARY 27

Convergent Biology and Translation to Idiopathic Disease

Carrie E. Bearden, University of California Los Angeles, USA
Talk Title to be Announced

Nenad Sestan, Yale School of Medicine, USA
Tools to Identify Convergent Biology

Armin Raznahan, NIMH, National Institutes of Health, USA
Genetics First Approaches and Childhood Onset Neuropsychiatric Disease

Daniel H. Geschwind†, University of California, Los Angeles, USA
Convergent Biology

Short Talks Chosen from Abstracts

How Do We Get to Treatment?

Randall L. Carpenter, Rett Syndrome Research Trust, USA
When Clinical Trials Fail: A Post-Mortem

Ricardo E. Dolmetsch†, Novartis Institutes for BioMedical Research, USA
What Tools do we need to Develop Effective Treatments?

Mustafa Sahin, Boston Children's Hospital, USA
Clinical Trials and TSC

Short Talks Chosen from Abstracts

Panel Discussion: How Do RGD Fit within the NIH Mission?

Anne R. Pariser, NCATS NIH, USA
Novel Approaches for Accelerating Rare Diseases Therapeutics Development

Melissa A. Parisi, NICHD, National Institutes of Health, USA
Overlapping Psychiatric and Developmental Disorders: NIH Approaches to a Continuity of Phenotypes

Linda Brady†, NIMH, National Institutes of Health, USA
Talk Title to be Announced

Short Talks Chosen from Abstracts

Meeting Wrap-Up: Outcomes and Future Directions (Organizers)

THURSDAY, JANUARY 28

Departure