SUNDAY, JANUARY 24
Arrival and Registration

MONDAY, JANUARY 25
Welcome and Keynote Address
Ricardo E. Dolmetsch†, Novartis Institutes for BioMedical Research, USA
What Tools Do We Need to Develop Effective Treatments?

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

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 Jacqueumont, University of Montreal, Canada
Mapping the Effect of CNVs Genome-Wide on Cognition, Brain Structure and Functional Connectivity
Short Talks Chosen from Abstracts

Poster Session 1

TUESDAY, JANUARY 26
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 Chahrour, University of Texas Southwestern Medical Center, USA
Neurodevelopmental Disorders: From Genetics to Mechanisms
Short Talks Chosen from Abstracts

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

Leveraging Animal Models for the study of RGD
Jill L. Silverman, University of California, Davis, USA
Mouse Behavior
Jens Hjerling Leffler, Karolinska Institute, Sweden
Molecular Cell Type Diversity and Why It Matters for Disease

Nicholas Katsanis, Northwestern University Feinberg School of Medicine, USA
Leveraging Zebrafish Models
Short Talk(s) 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
Stephan Sanders, University of California, San Francisco, USA
Phenotypes, Biology, and Therapeutic Strategies for SCN2A
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
Paul Wang, Simons Foundation, USA
Talk Title to be Announced
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
Tara Dutka, NIMH, National Institutes of Health, USA
NIMH Interest in Rare Genetic Disorders
Short Talks Chosen from Abstracts

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

THURSDAY, JANUARY 28
Departure

* Session Chair † Invited but not yet accepted     Program current as of June 20, 2020. Program subject to change. Meal formats are based on meeting venue. For the most up-to-date details, visit https://www.keystonesymposia.org.