AGENDA

TUESDAY SEPTEMBER 21: GENETIC MODIFIER DISCOVERY

10:00 am – 10:10 am **Workshop Introduction and Acknowledgements**
Glen Nuckolls PhD, National Institutes of Health, NINDS

10:10 am – 10:15 am **Introduction to Genetic Modifier Discovery Session**
Trudy Mackay PhD, Clemson University

10:15 am - 11:15 am 15 min presentations plus Q&A

Genetic Modifiers of Huntington's Disease
James Gusella PhD, Harvard Medical School

Genome-wide In Vivo CNS Screening to Identify Genetic Modifiers of Mutant Huntingtin Toxicity
Myriam Heiman PhD, Massachusetts Institute of Technology

Strategies for Modifier Detection in Drosophila
Trudy Mackay PhD, Clemson University

11:15 am - 11:20 am **Break**

11:20 am - 12:00 pm **Virtual Poster Session 1**
Short, pre-recorded presentations selected from abstracts, live Q&A

Functional screening of lysosomal storage disorder genes identifies modifiers of neurodegeneration in synucleinopathy
Meigen Yu, Baylor College of Medicine

Parkinson's disease risk genes act in glia to control neuronal alpha-synuclein toxicity
Abby Olsen MD PhD, Brigham and Women's Hospital, Harvard Medical School

Targeting Sipa1l2 and other candidate modifiers in the Sox10/Egr2 co-expression network to treat CMT1A
George Murray, The Jackson Laboratory & The University of Maine

	Defining the molecular mechanism of GARS1-related Charcot-Marie-Tooth disease Sheila Marte, University of Michigan
	Systems-guided in vivo identification of causal mechanisms in Alzheimer's disease from human brain transcriptomic profiles Grant Mangleburg, Baylor College of Medicine
12:00 pm -12:15 pm	**Break**
12:15 pm - 1:15 pm	**Genetic Modifier Discovery Session (continued)** 15 min presentations plus Q&A
	A Decade Long Clinical-Genetics Effort Identifies SIPA1L2 as a Modifier Gene for the Common Peripheral Neuropathy Subtype CMT1A Stephan Züchner MD, PhD, University of Miami
	TMEM106B, A Key Protective Factor in the Brain Rosa Rademakers PhD, University of Antwerp
	Insights for Rett Syndrome from the Identification of Genetic Modifiers in Mecp2 Mice Monica Justice PhD, The Hospital for Sick Children (SickKids)
1:15 pm - 2:00 pm	**Open group discussion** *Moderated by Co-Chairs Monica Justice and Trudy MacKay*
2:00 pm - 3:00 pm	**"Happy Hour" for students, fellows and new investigator to meet with workshop presenters, Co-Chairs and NIH Program Directors (https://roseliassociates.zoomgov.com/s/1608146296)**
3:00 pm	**End of Day 1**

WEDNESDAY SEPTEMBER 22: MODIFIER CHARACTERIZATION AND MECHANISMS

10:00 am – 10:05 am	**Introduction to Modifier Characterization and Mechanisms Session** Huda Zoghbi MD, Baylor College of Medicine
10:05 am - 11:05 am	15 min presentations plus Q&A
	Genome Editing in Huntington's Disease Mice to Test Candidate Modifier Genes Vanessa Wheeler PhD, Massachusetts General Hospital, Harvard Medical School
	Regulatory Variants as Potential Modifiers of Coding Variant Penetrance Tuuli Lappalainen PhD, Columbia University
	An ALS Modifier Gene and a Potential Achilles' Heel for ALS Aaron Gitler PhD, Stanford University

11:05 am - 11:10 am **Break**

11:10 am - 12:15 pm **Virtual Poster Session 2**
Short, pre-recorded presentations selected from abstracts, live Q&A

Sequence variation upstream of a cytoplasmic tRNA modifies neurodegeneration induced by ribosome stalling
Michael J. Molumby PhD, University of California San Diego

Modifiers of neurodegenerative disease can act to extend lifespan and CNS healthspan in Drosophila melanogaster
Megan Mair, Baylor College of Medicine

Sequencing of phenotypic extremes to identify genetic modifiers of Rett Syndrome
Jonathan Merritt PhD, Vanderbilt University Medical Center

A role for L1CAM/SAX-7 and Erk signaling in fluid regulation and vulva development
Caroline Aragon, University of Minnesota

Gabra2 is a genetic modifier of Scn8a encephalopathy in the mouse
Wenxi Yu PhD, University of Michigan

Targeting Latent TGF-beta binding protein 4 (LTBP4) for the treatment of muscular dystrophy
Alexis R. Demonbreun PhD, Northwestern University

Genetic compensation at the AIS: Scn8a and Kcna1
Sophie Hill, University of Michigan

Using Drosophila melanogaster to identify loci modifying Coffin-Siris syndrome mutations
Rebecca MacPherson, Clemson University

Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling
Paul Marcogliese PhD, Baylor College of Medicine

12:15 pm - 12:30 pm **Break**

12:30 pm - 1:30 pm **Modifier Characterization and Mechanisms Session (continued)**
15 min presentations plus Q&A

Sphingolipids and Ceramides in the Pathogenesis of Lysosomal Storage Diseases and Parkinson's Disease
Hugo Bellen DVM PhD, Baylor College of Medicine

Transfer RNAs as Genetic Modifiers of Neurological Phenotypes
Susan Ackerman PhD, University of California, San Diego

Tau Translation Regulation by Paired Antisense Long Non-coding RNA Transcripts
Rohan De Silva DPhil, University College London

1:30 pm - 2:00 pm **Open group discussion**
Moderated by Co-Chairs Huda Zoghbi and Susan Ackerman

2:00 pm - 3:00 pm **"Happy Hour" for students, fellows and new investigator to meet with workshop presenters, Co-Chairs and NIH Program Directors (https://roseliassociates.zoomgov.com/s/1608146296)**

3:00 pm **End of Day 2**

THURSDAY SEPTEMBER 23: TRANSLATIONAL RESEARCH FROM GENETIC MODIFIERS

10:00 am – 10:05 am **Introduction to Translational Research Session**
Stephan Züchner, University of Miami

10:05 am - 11:25 am 15 min presentations plus Q&A

Personalized Medicine Using a Zebrafish Model for Dravet Syndrome
Scott Baraban PhD, University of California, San Francisco

Application of Genetics in Drug Discovery and Development
Aris Baras MD MBA, Regeneron

Identifying Genetic and Chemical Modifiers: Lessons from Congenital Myopathies
James Dowling MD PhD, The Hospital for Sick Children (SickKids)

Gaucher Disease "Complexity" in a "Simple" Mendelian Disorder
Ellen Sidransky MD, National Institutes of Health, NHGRI

11:25 am - 11:30 am **Break**

11:30 am - 12:15 pm **Research Resources for Modifier Research**
10 min presentations plus Q&A

Mouse Resources for Genetic Modifier Studies
Elissa Chesler PhD, The Jackson Laboratory

International HundredK+ Cohorts Consortium
Hakon Hakonarson MD, PhD, Children's Hospital Philadelphia

NHLBI Trans-Omics for Precision Medicine (TOPMed)
Albert Smith PhD, University of Michigan

12:15 pm - 12:30 pm **Break**

12:30 pm - 1:30 pm	**Translational Research Session (continued)** 15 min presentations plus Q&A *Fetal Hemoglobin: from Bedside to Bench to Bedside* Swee Lay Thein FRCP DSc FMedSci, National Institutes of Health, NHLBI *Prospects for Modifier Studies in Diverse Populations: Lessons from Africa* Neil A. Hanchard MD, DPhil FACMG, National Institutes of Health, NHGRI *Regional Effects of Modifiers: Lessons from Spinocerebellar Ataxia type1* Huda Zoghbi MD, Baylor College of Medicine
1:30 pm - 2:00 pm	**Open group discussion** *Moderated by Co-Chairs Huda Zoghbi and Stephan Zuchner*
2:00 pm - 3:00 pm	**"Happy Hour" for students, fellows and new investigator to meet with workshop presenters, Co-Chairs and NIH Program Directors (https://roseliassociates.zoomgov.com/s/1608146296)**
3:00 pm	**End of Day 3/Workshop**