



8th Ataxia Investigators Meeting (AIM) 2021 – Rescheduled
“Leveraging Therapeutic Opportunity into Novel Treatment Paradigms”
Virtual – Times are in Central Daylight Time

Due to unforeseen circumstances, this schedule is subject to change.

MONDAY, May 24, 2021

10:00-10:05 AM	NAF announcements, Andrew Rosen, NAF Executive Director
10:06-10:17 AM	Welcome to virtual meeting orientation, Chair, Albert La Spada, MD, PhD
<u>Theme 1 – Basic Cerebellar Function and Dysfunction</u>	
Session Chair: Marija Cvetanovic*, PhD, University of Minnesota	
10:20-11:00 AM	Keynote: Sascha du Lac, PhD, Johns Hopkins <i>Cerebellar Output Circuits</i>
11:03-11:30 AM	Sheng-Han Kuo*, MD, Columbia University <i>Cerebellar movement control</i>
11:30-11:40 AM	Break – Exhibit/Sponsor Hall and Lobby open for networking
11:40-12:07 PM	Andreea Bostan*, PhD, University of Pittsburgh <i>Cerebellar communications with cortex and the basal ganglia</i>
12:10-12:22 PM	David Bushart*, PhD, Ohio State University College of Medicine Region-specific dysfunction of an ion channel module in SCA1
12:25-12:37 PM	Ashley Robbins, PhD, The Children’s Hospital of Philadelphia <i>Spatiotemporally barcoded single-cell RNA-Seq enables characterization of the murine cerebellum at fine resolution</i>
12:40-12:52 PM	Naiara Akizu*, PhD, University of Pennsylvania <i>Uncovering pathogenic mechanisms of SNX14 associated cerebellar ataxia</i>
12:55-1:15 PM	Theme 1 Open Discussion led by Marija Cvetanovic, PhD
1:15-2:00 PM	Lunch – Exhibit/Sponsor Hall and Lobby open for networking
2:00-2:45 PM	Keynote: Nobel Laureate, Thomas Cech, PhD, University of Colorado, Boulder <i>The Magic of RNA: from CRISPR Gene Editing to mRNA Vaccines</i>
2:45-2:50 PM	Tech Break
<u>Theme 2 – Disease Modeling and Mechanisms</u>	
Session Chair: Sokol Todi, PhD, Wayne State University	
2:50-3:17 PM	Matthew Scaglione*, PhD, Duke University <i>Novel chaperone systems for therapy</i>
3:20-3:47 PM	Karl Herrup, PhD, University of Pittsburgh <i>Ataxia-Telangiectasia</i>
3:50-4:20 PM	Theme 2 Open Discussion led by Sokol Todi, PhD
Completion of Day 1	

TUESDAY, May 25, 2021

<u>Theme 3 – Neuroprotective Targets and Strategies</u>	
Session Chair: Vikram Shakkottai, MD, PhD, University of Texas Southwestern, Dallas	
10:00-10:40 AM	Keynote: Walter Koroshetz, MD, NINDS Director <i>NINDS strategy for funding from discovery to therapy</i>
10:43-11:10 AM	Henry Houlden, MD, PhD, University College London <i>Genetics of late-onset ataxia</i>

11:10-11:25 AM	Break – Exhibit/Sponsor Hall and Lobby open for networking
11:25-11:37 AM	Mandi Gandelman*, PhD, University of Utah, Salt Lake City <i>Altered calcium signaling and STAU1-dependent ER stress in models of SCA2</i>
11:40-11:52 AM	Monica Banez-Coronel*, PhD, University of Florida, Gainesville <i>CCG CGG interruptions found on highly penetrant SCA8 alleles increase RAN protein levels and cellular toxicity</i>
11:55 -12:07 PM	Pawel Switonski*, PhD, University of California, Irvine <i>Epigenetic Dysregulation Impairs High-Fidelity DNA Repair to Promote Cerebellar Degeneration in SCA7</i>
12:10-12:25 PM	Theme 3 Open Discussion led by Vikram Shakkottai, MD, PhD
12:25-12:30 PM	Tech Break
12:35-12:55 PM	“Hot Chair” Junior Poster Presenters <ol style="list-style-type: none"> Owen Morgan*, BA, Johns Hopkins University <i>Motor and Cognitive Sequencing in Cerebellar Ataxia</i> Esther Becker*, PhD, University of Oxford <i>Human iPSC-derived cerebellar organoids recapitulate development of the cerebellum</i> Adam Avery*, PhD, Oakland University <i>Drug discovery for SCA, using novel fluorescence technology targeting β-III-spectrin</i> Larissa Nitschke*, BS, Baylor College of Medicine, Houston, TX <i>Modulating phosphorylation of ATXN1's serine 776: a potential therapeutic approach for SCA1</i>
12:55-1:00 PM	Tech Break
1:00-2:00 PM	Scientific Poster Session A and Lunch-Exhibit/Sponsor Hall is open
2:00-2:40 PM	Keynote: Paul Taylor, MD, PhD, St. Jude's Research Hospital <i>Bridging biophysics and neurology: the role of phase transitions in neurodegeneration</i>
2:40-2:45 PM	Tech Break
2:45-3:10 PM	“Hot Chair” Junior Poster Presenters <ol style="list-style-type: none"> Leon Tejwani*, MS, Yale School of Medicine <i>Longitudinal single nucleus transcriptomic profiling of the SCA1 mouse cerebellum</i> Celeste Suart*, BHSc, McMaster University, Ontario, Canada <i>Ataxin-1 is signalled to DNA damage by ATM kinase</i> Young Woo Park*, PhD, University of Minnesota <i>Measurements of cross-sectional differences and longitudinal changes with DTI in SCA1/2/3/6</i> David Szmulewicz, MD, Monash University <i>Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS</i> Jennifer Faber*, MD, German Center for Neurodegenerative Diseases <i>Data-driven model of dynamic biomarkers in SCA3</i>
3:10-3:15 PM	Tech Break
3:15-4:30 PM	Moderated Breakout Session – Moderator: Liana Rosenthal, MD, PhD & Sokol Todi, PhD Divya Singhal, MD, FAAN <i>Diversity and Inclusion</i>
4:30 PM	Completion of Day 2

WEDNESDAY, May 26, 2021

Theme 4 – Biomarker Discovery and Clinical Disease Phenotyping	
Session Chair: Joel Gottesfeld, PhD, The Scripps Research Institute	
10:00-10:40 AM	Keynote: Viviana Gradinaru*, PhD, Caltech <i>Vectors that cross the blood-brain barrier</i>
10:43-11:10 AM	Leonard Petrucelli, PhD, Mayo Clinic, Jacksonville, FL <i>CSF biomarkers</i>
11:10-11:25 AM	Break – Exhibit/Sponsor Hall and Lobby open for networking
11:25-12:05 PM	Keynote: Alice Chen-Plotkin, MD, University of Pennsylvania <i>Lessons from biomarker work in PD</i>
12:08-12:20 PM	Theme 4 Open Discussion Led by Joel Gottesfeld, PhD
12:20-12:25 PM	Tech Break
12:25-1:15 PM	“Shark Tank”: <i>Single therapies for patients with DIFFERENT forms of ataxia!</i> Presenters – Vikram Shakkottai, MD, PhD, and Sokol Todi, PhD Sharks – Harry Orr, PhD and Joel Gottesfeld, PhD and Susan Perlman, MD
1:15-2:20 PM	Scientific Poster Session B and Lunch – Exhibitor/Sponsor Hall is open
2:20-2:50 PM	Gilles Guillemin, PhD, OM, MacQuarie, Australia <i>TRP metabolism, kynurenine</i>
2:53-3:20 PM	Matthis Synofzik, MD <i>Update on Ataxia Global Initiative (AGI)</i>
3:20-3:25 PM	Tech Break
3:25-4:30 PM	Break-out Session II Puneet Opal, MD, PhD, Northwestern University – <i>Biomarkers</i> Henry Paulson, MD, PhD, University of Michigan – <i>Mentorship</i> Marija Cvetanovic, PhD, University of Minnesota and Janghoo Lim, PhD, Yale <i>Single Cell RNA Sequencing</i>
4:30 PM	Completion of Day 3

THURSDAY, May 27, 2021

9:00-9:55 AM	<i>SARA/Clinical Ataxia rating scales training session</i> Jennifer Faber, MD and Heike Jacobi, MD
9:00-10:00 AM	Exhibitor/Sponsor Hall is open
Theme 5 – Clinical Trial Organization	
Session Chair: Liana Rosenthal*, MD, PhD, Johns Hopkins School of Medicine	
10:00-10:35 AM	Sophie Tezenas du Montcel*, MD, PhD, Sorbonne University, Paris <i>Trial Design for Rare Diseases</i> <i>Conversion of individuals at risk for SCA types 1, 2, 3, and 6 to manifest ataxia</i>
10:38-11:23 AM	Keynote: Holly Kordasiewicz, PhD, Ionis Pharmaceuticals <i>ASO therapy development</i>
11:23-11:48 AM	Break – Exhibit/Sponsor Hall and Lobby open for networking
11:51-12:04 PM	Robert Wessells*, PhD, Wayne State School of Medicine <i>Chronic Exercise slows disease progression in Drosophila models of SCA</i>
12:07-12:20 PM	Phyllis Faust, MD, PhD, Columbia University <i>A pathologic approach in a spectrum of cerebellar degenerative diseases</i>
12:20-12:40 PM	Theme 5 Open Discussion Led by Liana Rosenthal, MD, PhD
12:40-12:45 PM	Tech Break

	<u>Late Breaking Abstracts</u> Session Chair – Al La Spada, MD, PhD University of California Irvine
12:45-1:00 PM	Jayashree Chandrasekaran, MS, University of Minnesota <i>Multimodal MRI detects premanifest and early brain alterations in SCA1 and SCA3</i>
1:03-1:18 PM	Megan Keiser, PhD, Children’s Hospital of Philadelphia <i>Translating Gene Therapy for Spinocerebellar Ataxia Type 1: Optimization of efficacious therapeutic vectors in nonhuman primates</i>
1:21-1:36 PM	David Lynch, MD, PhD, Children’s Hospital of Philadelphia <i>Efficacy of Omaveloxolone in Patients with Friedreich's Ataxia: Baseline-Controlled Study</i>
1:39-1:54 PM	Xiang-Dong Fu, PhD, University of California San Diego <i>Making New Neurons To Replenish Lost Ones In Neurodegenerative Disease</i>
1:57-2:17 PM	Giulia Coarelli, MD, Hopital de la Salpetriere, Paris, France <i>Plasma neurofilament light chain predicts cerebellar atrophy and clinical progression in spinocerebellar ataxia</i>
2:20-2:40 PM	Al La Spada, MD, PhD <i>Wrap-up of the 2020-21 Conference, AIM farewell and Closing Remarks</i>
2:40 PM	AIM 2020-21 Adjourned

**Junior Investigators may include post docs, professors, and graduate students.*