AIM 2020: Leveraging Therapeutic Opportunity into Novel Treatment Paradigms
This conference is co-funded by the National Institute of Neurological Disorders and Stroke (NINDS) and National Center for Advancing Translational Sciences. All publications, posters, oral presentations at scientific meetings, seminars, and any other forum in which results of this co-funded research are presented must include a formal acknowledgement of the NINDS/other funding entity support, citing the NINDS grant number R13-NS115342.
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All AIM conference communications, materials and abstracts are confidential.

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**Wireless Internet Information**

The following is the wireless Internet connection information in the AIM meeting room:

- **Network Name** ........ Sheraton Meeting Room
- **Password** ............... ataxia

Please limit your connections to the Internet to one device only. When not using the Internet, please disconnect from the NAF WiFi Internet connection. More importantly, please focus your attention on the AIM presentations. Thank you.
March 3, 2020

Dear AIM 2020 Attendee,

On behalf of the Organizing Committee and my Co-Chair, Dr. Stefan Pulst, it is my great pleasure to welcome you to the 8th Ataxia Investigators Meeting (AIM), sponsored by the National Ataxia Foundation. **AIMs continue to bring ataxia investigators from around the world together to exchange ideas, and to move the field forward towards the ultimate goal of developing effective therapies to treat these devastating diseases.**

This year’s meeting will consist of presentations from leaders in the ataxia field and from internationally renowned physicians and scientists with expertise ranging from molecular biology to neuroscience to neurotherapeutics. We have assembled a diverse set of invited lecturers and Keynote speakers, including a Nobel Laureate and the Director of the NINDS, with selected lectures from Trainees and Junior Investigators. We will again emphasize interaction and discussion, with a format that will permit ample time for questions at the end of each talk. This year we will have **Open Discussion sessions** to facilitate exchange of ideas, after we have heard all presentations for a Theme. If you are a Trainee, we especially value your participation during the Q&A and open discussions. We will have two **Hot Chair sessions** that feature four-minute talks from Junior Investigators, who will outline their poster presentations in this longer time frame, as requested by attendees of the last AIM. New this year will be a **Shark Tank session**, during which a provocative proposal will be argued for, with probing questions put forth by a panel of “sharks,” all while questions and perspectives are solicited from audience attendees. We hope that the Shark Tank session will illustrate that creative, unorthodox ideas are always welcome and how we should be open-minded as a field. Last, but not least, please be sure to make the most out of numerous opportunities to interact with patients and their family members at various venues: 1) a **Poster Session for Patients and Families**, 2) the Thursday evening dinner with talks given by patients and family members, 3) the Annual Ataxia Conference/AIM Patient/Family Networking Session on Friday at the end of AIM and 4) **Birds of a Feather sessions** on Friday afternoon.

Certainly, the success of this meeting depends upon all of you, and I am grateful for the interest displayed in AIM 2020 by the ataxia research community, who together submitted a record number of abstracts (>120) for consideration. I genuinely hope that all of you leave this meeting inspired by the incredible advances taking place in the ataxia field and will establish productive collaborations that advance our efforts in basic research, clinical practice, and therapeutics.

*Albert La Spada, MD, PhD*

*AIM 2020 Chair*

P.S. – We kindly request that you complete the meeting survey, as we review all feedback and use it to make changes to the organization of the meeting (as we have done this year).
Meeting Schedule

8th Ataxia Investigators Meeting (AIM) 2020
“Leveraging Therapeutic Opportunity into Novel Treatment Paradigms”
— Sheraton Denver Downtown, Denver, CO —

TUESDAY, MARCH 3, 2020

3:00-6:30 PM .......... AIM Check-in and Poster Boards available to hang posters
5:00-6:30 PM .......... Welcome Reception & Opening Remarks
                  Albert La Spada, MD, PhD, Duke University
Evening ................... Networking Dinner On-Your-Own

WEDNESDAY, MARCH 4, 2020

7:30-8:30 AM .......... Breakfast

Theme 1 – Basic Cerebellar Function and Dysfunction

Session Chair: Marija Cvetanovic*, PhD, University of Minnesota

8:30-9:10 AM .......... Keynote: Sascha du Lac, PhD, Johns Hopkins
                  Cerebellar Modules

                  Cerebellar movement control

9:40-10:10 AM ...... Andreea Bostan*, PhD, University of Pittsburgh
                  Cerebellar connections with the cerebral cortex and the basal ganglia

10:10-10:30 AM ...... Break

10:30-10:45 AM ...... David Bushart*, PhD, University of Michigan
                  Region-specific dysfunction of an ion channel module in SCA1

10:45-11:00 AM ...... Paul Ranum*, PhD, The Children’s Hospital of Philadelphia
                  Spatiotemporally barcoded single-cell RNA-Seq enables characterization
                  of the murine cerebellum at fine resolution

11:00-11:15 AM ...... Naiara Akizu*, PhD, University of Pennsylvania
                  Uncovering pathogenic mechanisms of SNX14 associated cerebellar ataxia

11:15-11:30 AM ...... Mandi Gandelman*, PhD, University of Utah
                  Altered calcium signaling and STAU1-dependent ER stress in models of SCA2

11:30-12:00 PM ...... Theme 1 Open Discussion led by Marija Cvetanovic, PhD

12:00-1:00 PM ........ Lunch

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

WEDNESDAY, MARCH 4, 2020 (continued)

Theme 2 – Disease Modeling and Mechanisms
Session Chair: Sokol Todi, PhD, Wayne State University

1:00-1:40 PM .......... Keynote: Paul Taylor, MD, PhD, St. Jude’s Research Hospital
Bridging biophysics and neurology: the role of phase transitions in neurodegeneration

1:40-2:10 PM .......... Henry Houlden, MD, PhD, University College London
Genetics of late-onset ataxia

2:10-2:40 PM .......... Karl Herrup, PhD, University of Pittsburgh
Ataxia-Telangiectasia

2:40-3:10 PM .......... Theme 2 Open Discussion led by Sokol Todi, PhD

3:10-3:20 PM .......... Official Photo of Attendees

3:20-3:45 PM .......... Break

3:45-4:15 PM .......... Five “Hot Chair” Junior Poster Presenters
Owen Morgan*, BA, Johns Hopkins University
Motor and Cognitive Sequencing in Cerebellar Ataxia
Esther Becker*, PhD, University of Oxford
Human iPSC-derived cerebellar organoids recapitulate development of the cerebellum
Adam Avery*, PhD, Oakland University
Drug discovery for SCA, using novel fluorescence technology targeting β-III-spectrin
Larissa Nitschke*, BS, Baylor College of Medicine, Houston, TX
Modulating phosphorylation of ATXN1’s serine 776: a potential therapeutic approach for SCA1
David Szmulewicz, MD, Monash University, Australia
Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS

4:15-5:45 PM .......... Scientific Poster Session A with Wine and Cheese

6:00-6:15 PM .......... Depart for Outing to History Colorado Center

6:30-10:00 PM .......... Dinner Keynote: Nobel Laureate, Thomas Cech, PhD, University of Colorado
Thirty years after the Nobel:
Some thoughts on science, medicine and trinucleotide expansion
Meeting Schedule

THURSDAY, MARCH 5, 2020

7:30-8:30 AM .......... Breakfast

**Theme 3 – Neuroprotective Targets & Strategies**
Session Chair: Vikram Shakkottai, MD, PhD, University of Michigan

8:30-9:00 AM .......... Keynote: Walter Koroshetz, MD, NINDS Director
  NINDS strategy for funding from discovery to therapy

9:00-9:30 AM .......... Matthew Scaglione*, PhD, Duke University
  Novel chaperone systems for therapy

9:30-10:10 AM ........ Keynote: Viviana Gradinaru*, PhD, Caltech
  Vectors that cross the blood-brain barrier

10:10-10:35 AM ...... Theme 3 Open Discussion led by Vikram Shakkottai, MD, PhD

10:35-11:00 AM ...... Break

11:00-11:30 AM ...... Five “Hot Chair” Junior Poster Presenters
  Celeste Suart*, BHSc, McMaster University, Ontario, Canada
  *Ataxin-1 is signalled to DNA damage by ATM kinase
  Pan Li*, PhD, Johns Hopkins University School of Medicine
  *Knock-in mouse model of spinocerebellar ataxia type 12
  Young Woo Park*, PhD, University of Minnesota
  Measurements of cross-sectional differences and longitudinal changes with DTI in SCA1/2/3/6
  Leon Tejwani*, MS, Yale School of Medicine
  *Longitudinal single nucleus transcriptomic profiling of the SCA1 mouse cerebellum
  Jennifer Faber*, MD, German Center for Neurodegenerative Diseases
  *Data-driven model of dynamic biomarkers in SCA3

11:30-1:00 PM ........ Scientific Poster Session B

1:00-2:00 PM .......... Reception Luncheon with Divya Singhal, MD, FAAN, University of Oklahoma
  Diversity and Inclusion

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

THURSDAY, MARCH 5, 2020 (continued)

Theme 4 – Biomarker Discovery & Clinical Disease Phenotyping
Session Chair: Joel Gottesfeld, PhD, The Scripps Research Institute

2:00-2:40 PM .......... Keynote: Alice Chen-Plotkin, MD, University of Pennsylvania
Lessons from biomarker work in PD

2:40-3:10 PM .......... Gilles Guillemin, PhD, OM, MacQuarie, Australia
TRP metabolism, kynurenine

3:10-3:40 PM .......... Leonard Petrucelli, PhD, Mayo Clinic, Jacksonville
CSF biomarkers

3:40-4:00 PM .......... Break

4:00-4:15 PM .......... Monica Banez-Coronel*, PhD, University of Florida
CCG CGG interruptions found on highly penetrant SCA8 alleles increase RAN protein levels and cellular toxicity

4:15-4:30 PM .......... Pawel Switonski*, PhD, Duke University School of Medicine
Increased DNA damage activates PARP1 to deplete cerebellar neurons of NAD+ and thereby promote SCA7 disease pathogenesis

4:30-4:45 PM .......... Robert Wessells*, PhD, Wayne State School of Medicine
Chronic Exercise slows disease progression in Drosophila models of SCA

4:45-5:00 PM .......... Theme 4 Open Discussion Led by Joel Gottesfeld, PhD

5:00-5:30 PM .......... “Shark Tank”: Single therapies for patients with DIFFERENT forms of ataxia!

5:30-6:45 PM .......... Poster Session for Patients and Families
Posters must be removed after this session

7:00-9:00 PM .......... Dinner Banquet: Keynote: Patient-Family Perspective

Please Note: The AIM Meeting Room changes to Governors Square 15 on Friday morning
Meeting Schedule

FRIDAY, MARCH 6, 2020

Please Note: The AIM Meeting Room changes to Governors Square 15

7:30-8:30 AM .......... Breakfast

Theme 5 – Clinical Trial Organization
Session Chair: Liana Rosenthal*, MD, PhD, Johns Hopkins School of Medicine

8:30-9:10 AM .......... Keynote: Holly Kordasiewicz, PhD, Ionis Pharmaceuticals
   ASO therapy development

9:10-9:40 AM .......... Sophie Tezenas Du Montcel*, MD, PhD, Sorbonne University, Paris
   Trial Design for Rare Diseases

9:40-9:55 AM .......... Heike Jacobi*, MD, German Center for Neurodegenerative Diseases, Bonn
   Conversion of individuals at risk for SCA types 1, 2, 3, and 6 to manifest ataxia

9:55-10:10 AM ........ Phyllis Faust, MD, PhD, Columbia University
   A pathologomic approach in a spectrum of cerebellar degenerative diseases

10:10-10:20 AM ...... Thomas Klockgether, MD, PhD, DZNA
   Update SCA Global

10:20-10:35 AM ...... Theme 5 Open Discussion led by Liana Rosenthal, MD, PhD

10:35-10:55 AM ...... Wrap-up of entire AIM by organizing chairs:
   Al La Spada, MD, PhD & Stefan Pulst, MD

10:55-11:00 AM ...... Closing Remarks – Stefan Pulst, MD, University of Utah

11:05-11:15 AM ...... AIM Attendees move to AAC General Sessions

11:15-11:45 AM ...... NAF Annual Ataxia Conference/AIM Patient/Family Networking Session

AIM Adjourns

2:00-4:30 PM .......... “Birds of a Feather” Sessions at the Annual Ataxia Conference
   Junior Investigators are encouraged to attend these small groups.
   See page 9 for meeting rooms for these groups.

*Junior Investigators may include post docs, professors and graduate students
NAF’s Ataxia Research Grant Program for FY 2021

The National Ataxia Foundation is committed to funding the best science relevant to hereditary and sporadic types of ataxia in both basic and translational research. NAF invites research applications from domestic and international non-profit and for-profit institutions. Non-U.S. citizens are eligible to apply for a research grant from NAF. More information is available on the NAF website: www.ataxia.org.

**Seed Money Research Grant**

For new and innovative studies that are relevant to the cause, pathogenesis, or treatment of the hereditary or sporadic ataxias. Offered primarily as “seed monies” to assist investigators in the early or pilot phase of their studies and as additional support for ongoing investigations upon demonstration of need. One-year seed money awards granted up to $15,000 but promising proposals up to $30,000 will be considered for projects deserving special consideration. Applicants must be faculty members. Letter of intent due September 21, 2020. Application due October 19, 2020.

**Young Investigator Award**

The Young Investigator Award was created to encourage young clinical and scientific investigators to pursue a career in the field of Ataxia research. It is NAF’s hope that Ataxia research will be invigorated by the work of young, talented individuals supported by this award. Award amount is up to $35,000 for one year. Applicants must have an MD or PhD degree. Applicants must also have an appointment as a junior faculty member, senior post-doc or clinical fellow. Letter of intent due September 28, 2020. Application due October 26, 2020.

**Post-Doc Fellowship Award**

Post-doctoral fellowship awards are to serve as a bridge from post-doctoral positions to junior faculty positions. Applicants should have shown a commitment to research in the field of Ataxia. Award amount up to $35,000 for one year. Applicants should have completed at least one year of post-doctoral training, but not more than two at the time of application. Letter of intent due October 5, 2020. Application due November 2, 2020.

**Pioneer SCA3/MJD Translational Research Award**

The Pioneer SCA3/MJD Translational Research Award is for a research project that will facilitate the development of treatments for Spinocerebellar Ataxia Type 3/Machado Joseph Disease. The total grant funding is $100,000 for one year for one grant. Applicants must be established ataxia researchers. Letter of intent due October 5, 2020. Application due November 2, 2020.

For more information on NAF’s research funding program, contact Kelsey Trace at kelsey@ataxia.org.
Partnering with Ataxia Families

The location of the 2020 Ataxia Investigators Meeting dovetails with the 63rd Annual Ataxia Conference of the National Ataxia Foundation: “Partnering for Progress The Time is Now!” The hope is to maximize the impact of this meeting for scientists and patients alike by providing opportunities for meaningful interactions between researchers, patients, family members and caregivers. There are four dedicated opportunities to interact with persons affected by ataxia and their family members and caregivers to which you are invited to participate.

Patient and Family Poster Session:
On Thursday, March 5, from 5:30–6:45 p.m. there is a dedicated poster session for patients and families. During this session we ask that all Junior poster presenters be available at their posters so that patients and families can meet you and learn more about your ataxia research efforts. Please bring a chair to your poster so that you may sit down when speaking with a person who uses a wheelchair.

Thursday AIM Dinner:
On Thursday, March 5, at 7:00 p.m. at the AIM dinner, speakers from the ataxia community will share their stories and challenges of living with ataxia. From the surveys taken after prior AIMs, many attendees stated that this was the best part of the meeting. Don’t miss this!

Friday at 11:15-11:45 a.m. AIM Patient/Family Networking Session (NEW THIS YEAR)
Please sign up at the AIM registration table to be a part of this session. We need 80 researchers to attend.

Read what the attendees at the Family conference are looking forward to: “When is the last time you sat down with an Ataxia scientist? You’ll get that chance here! Ataxia researchers will come in and join your tables for an informal chat session. Dr. La Spada will kick off the session with a brief overview of takeaways from the Ataxia Investigators Meeting that will have just concluded. Then you’ll have the opportunity to ask questions and get to know an Ataxia scientist at your table.”

“Birds of a Feather” Small Group Sessions:
After the AIM 2020 has adjourned, patients and family members will meet in facilitated small groups to learn of the latest research, meet and share openly with others who have the same type of ataxia. You are very welcome to attend any session that would be of particular interest to you.

Below is the listing of sessions meeting on Friday, March 6 from 2:00-4:30 p.m. and their locations:
• SCA 1 ..................................................... Governor’s Square 9
• SCA 2 ..................................................... Plaza Court 6
• SCA 3 ..................................................... Governor’s Square 14
• SCA 6 ..................................................... Governor’s Square 11
• SCA 5, 7, & 8 ........................................ Plaza Court 5
• All other SCAs, EA & DRPLA ............... Governor’s Square 12
• Friedreich Ataxia ................................. Governor’s Square 16
• AOA and other Recessives ...................... Governor’s Square 17
Scientific Poster Sessions

Poster Numbering
The poster number is followed by the theme which is followed by the presenting author.

Themes
1. Basic cerebellar function and dysfunction
2. Disease modeling and mechanisms
3. Neuroprotective targets and strategies
4. Biomarker discovery and clinical disease phenotyping
5. Clinical Trial Organization

Session Times
• Wednesday Scientific Poster Session is from 4:15-5:45 PM on Wednesday, March 4
• Thursday Scientific Poster Session is from 11:30-1:00 PM on Thursday, March 5
• Thursday Poster Session for Patients and Families is from 5:30-6:45 PM on Thursday, March 5

Wednesday Scientific Poster Sessions (4:15-5:45 PM)

2_Theme 1_Wed ................................... ‡Amokrane, Nadia .... Columbia University Medical Center
Impulsivity in Patients with Cerebellar Ataxia: a case-control study

5_Theme 3_Wed ..................................... *Avery, Adam .............. Oakland University
Drug discovery for spinocerebellar ataxia, using novel fluorescence technology targeting β-III-spectrin

7_Theme 2_Wed ................................. Bannister, Roger A. ...... University of Maryland, Baltimore
Zebrafish as a model system for the study of severe CaV2.1 channelopathies characterized by ataxia, cerebellar atrophy and global developmental delay

8_Theme 3_Wed .................................... *Becker, Esther ............... University of Oxford
Human iPSC-derived cerebellar organoids recapitulate development of the cerebellum

10_Theme 3_Wed ................................. Bushart, David ................. University of Michigan
Identification of appropriate potassium channel targets to improve neuronal function and motor impairment in Spinocerebellar Ataxia type 1

12_Theme 5_Wed ................................. Chen, Helen ................. Massachusetts General Hospital
The genetic counselor: Increasing recognition of their critical role in ataxia centers in the era of genomic medicine
Scientific Poster Sessions

Wednesday Scientific Poster Sessions (continued)

14_Theme 3_Wed ......................... Chen, Yong Hong ...... Children's Hospital of Philadelphia
Novel adeno-associated viral vectors for spinocerebellar ataxia therapies

18_Theme 4_Wed ............................... Corben, Louise Anne ... Murdoch Childrens Research Institute
Patient reported perspective of a new measure of upper limb function in Friedreich ataxia.

20_Theme 3_Wed ............................ Costa, Maria do Carmo ..................... University of Michigan
Exploring the efficacy of aripiprazole-related compounds to reduce cellular levels of ATXN3

22_Theme 2_Wed ............................. Denha, Sarah ............................ Oakland University
Unique N-terminus of β-III-spectrin modulates toxicity of a SCA5 mutation

24_Theme 3_Wed .............................. ‡Duarte, Sónia ......................... Center for Neuroscience and Cell Biology: CNC
MicroRNA profiling in Spinocerebellar ataxia type-3: On the development of a miRNA-based therapy

26_Theme 2_Wed ............................ Edamakanti, Chandrakanth Reddy .... Northwestern University
Circuit level disruption in Spinocerebellar ataxia type 1 as a plausible therapeutic target

28_Theme 1_Wed ............................. Elsayed, Liena ............................ University of Khartoum, Sudan
Ataxic neurodegenerative disorders in Sudan: when whole exome sequencing is combined to high consanguinity and old genome

30_Theme 3_Wed .............................. Fagan, Kelly ............................. University of Pennsylvania
CRISPR-Cas9 gene editing strategies to treat spinocerebellar ataxia type 1

32_Theme 1_Wed ............................. Figueroa, Karla ......................... University of Utah
Are We Related? Exploration of Haplotype and Ancestry Research

34_Theme 4_Wed .............................. Gupta, Anoopum ............... Massachusetts General Hospital
Detection of Oculomotor Dysmetria in Ataxia from iPhone Video of the Horizontal Saccade Task

36_Theme 2_Wed .............................. ‡Handler, Hillary ....................... University of Minnesota
Role of non-cerebellar nuclear localization in SCA1 pathology

38_Theme 1_Wed ............................. Hilger, Allison ........................ Northwestern University
Impaired auditory feedback control for pitch production in speech in ataxic dysarthria

*“Hot Chair” Poster Presenter  ‡Travel Stipend Recipient
Scientific Poster Sessions

Wednesday Scientific Poster Sessions (continued)

40_Theme 4_Wed .................. Huryn, Larissa ................................. NEI/NIH
Ophthalmic Manifestations of Spinocerebellar Ataxia Type 7

42_Theme 1_Wed .................. Jacoby, Brigitte .............. Massachusetts General Hospital
Sensitivity and specificity of the Cerebellar Cognitive Affective Syndrome (CCAS) scale versus MMSE and MoCA for detection and characterization of cognitive deficits in cerebellar versus other neurocognitive disorders

44_Theme 2_Wed .................. Ke, Michael ..................... KPU Bio-Innovation Lab
Whole Exome Sequencing to Identify Novel Mutations in Two Ataxia Families

45_Theme 2_Wed .................. Keiser, Megan S. ........ Children's Hospital of Philadelphia
Combinatorial Gene Therapy for Spinocerebellar Ataxia Type 1

46_Theme 2_Wed .................. Keiser, Megan S. ........ Children's Hospital of Philadelphia
Combined transgene and intron-derived miRNA therapy for the treatment of SCA1

48_Theme 4_Wed .................. Knudson, Karin .............. Massachusetts General Hospital
Estimation of ataxia severity and disease classification from wearable sensor recordings with machine learning

50_Theme 1_Wed .................. Lin, Chi-Ying ...... Columbia University Medical Center
The Contribution of The Cerebellum to Cognitive Function in the Disease Process of Dementia

52_Theme 2_Wed .................. Luttik, Kimberly .................... Yale University
Differential effects of enhanced Wnt-β-catenin signaling in Purkinje cells and glial cells in SCA1

54_Theme 2_Wed .................. ‡Maciel, Patricia .......................... University of Minho
Genetic modifiers of Spinocerebellar ataxia 3/ Machado-Joseph disease (SCA3/MJD)

56_Theme 2_Wed .................. Maciel, Patricia .................... University of Minho
Suppression of proteotoxicity by serotonin signaling

57_Theme 5_Wed .................. MacMore, Jason .............. Massachusetts General Hospital
Patient Reported Outcome Measure for Ataxia (PROM-Ataxia). Further development with test-retest reliability and external validation

59_Theme 3_Wed .................. Macpherson, Chelsea Erin ...... Teachers College, Columbia University
Engage-Ataxia: Development of a physical activity coaching intervention in individuals newly diagnosed with ataxia
Scientific Poster Sessions

Wednesday Scientific Poster Sessions (continued)

61_Theme 5_Wed ................................. Millar, Jennifer .................. Johns Hopkins Medicine
Patients with cerebellar ataxia reporting gaze instability demonstrate abnormal oculomotor patterns during passive head rotation

62_Theme 1_Wed ............................... *Morgan, Owen ..................... Johns Hopkins Univ.
Motor and Cognitive Sequencing in Cerebellar Ataxia

64_Theme 2_Wed ................................. Napierala, Jill ................ University of Alabama at Birmingham
Upregulation of Mitochondrial Aldehyde Dehydrogenase Activity Inhibits Lipid Peroxidation in Friedreich’s Ataxia Cells

66_Theme 2_Wed ............................... *Nitschke, Larissa ............. Baylor College of Medicine
Modulating phosphorylation of ATXN1’s serine 776 residue: a potential therapeutic approach for Spinocerebellar Ataxia Type 1

68_Theme 4_Wed ................................. ‡Pane, Chiara ............... Federico II University of Naples
The Upper Limb Cardiopulmonary Exercise Test in Friedrich Ataxia Patients

70_Theme 2_Wed ................................. Paul, Sharan ..................... University of Utah
Decreased miR-217 derepresses Staufen2 and regulates autophagy in Spinocerebellar ataxia type 2 (SCA2)

72_Theme 1_Wed ................................. Ranum, Paul ..................... Children’s Hospital of Philadelphia
Spatiotemporally barcoded single-cell RNA-Seq enables characterization of the murine cerebellum at fine resolution

74_Theme 4_Wed ................................. Pereira de Almeida, Luis ...... University of Coimbra, Portugal
Allele-specific and non-invasive AAV-based silencing of mutant ataxin-3 alleviates neuropathology and motor deficits in Machado-Joseph disease

76_Theme 2_Wed ................................. ‡Rodden, Layne ................ University of Oklahoma
An active-to-inactive state switch of an enhancer in intron 1 contributes to FXN gene silencing in FRDA

78_Theme 2_Wed ................................. Schmidt, Thorsten .......... Eberhard Karls Universität Tübingen, Institute of Medical Genetics and Applied Genomics
Factors modifying the age at onset in Spinocerebellar Ataxia Type 3 / Machado-Joseph disease

80_Theme 2_Wed ................................. Scoles, Daniel .................. University of Utah
ALS-associated genes in SCA2 mouse spinal cord transcriptomes

*“Hot Chair” Poster Presenter  ‡Travel Stipend Recipient
Scientific Poster Sessions

Wednesday Scientific Poster Sessions (continued)

82_Theme 2_Wed .......................... Sheeler, Carrie ............................ University of Minnesota
Ataxin-1 Phosphorylation at Serine 776 in Mouse and Human iPSC-derived Motor Neurons

84_Theme 4_Wed .......................... Simpson, William ........................... McMaster University
Assessment of Ataxia Using Natural Language Processing and Computational Analysis of Speech

86_Theme 1_Wed .......................... Smith, Erin Lea ............................. University of Nebraska Medical Center, Omaha
Evidence-based screening for lower extremity ataxia

88_Theme 4_Wed .......................... Stephen, Christopher ............... Massachusetts General Hospital
Quantitative oculomotor assessment and non-motor biomarkers in late-onset GM2 gangliosidosis

91_Theme 5_Wed .......................... Stuart, Celeste ............................ McMaster University
Evaluating the impact of a knowledge translation platform for spinocerebellar ataxias on its readers and volunteers

93_Theme 4_Wed .......................... *Szmulewicz, David ........................ Monash University
Bioinformatics-Based Identification of Expanded Repeats: A Non-reference Intronic Pentamer Expansion in RFC1 Causes CANVAS

96_Theme 2_Wed .......................... Tsou, Wei-Ling ........................... Wayne State University
The role of the ataxin-7 binding partner, USP22 in glial-directed retinal development and degeneration

97_Theme 1_Wed .......................... Villarin, Colin ............................. Massachusetts General Hospital
Cerebellar Cognitive Affective Syndrome Scale for Children (CCAS-c): development of norms in an exploratory cohort of typically developing children

99_Theme 4_Wed .......................... ‡Vogel, Adam ............................ The University of Melbourne
Speech-ATAXIA: a multinational, multilanguage consortia for speech in hereditary ataxias

101_Theme 2_Wed .......................... Xhako, Eder ............................. Baylor College of Medicine
Studies of mechanisms driving brain region specific neurodegeneration in SCA1

103_Theme 3_Wed .......................... Yrigollen, Carolyn ............... Children’s Hospital of Philadelphia
Assessing CRISPR mediated deletion of CGG repeats in the brains of mice modeling FXTAS

105_Theme 4_Wed .......................... Zhang, Yuan ............................. University of Michigan
Measurement of Repeat Associated non-AUG translated proteins
Scientific Poster Sessions

Thursday Scientific Poster Sessions (11:30 AM-1:00 PM)

1. Theme 2_Thur .......................... ‡Aguado, Julio ................................. University of Queensland
   A novel pathogenic mechanism for premature neurodegeneration in Ataxia-Telangiectasia

3. Theme 2_Thur .......................... ‡Anderson, Collin ............................ University of Utah
   Gene therapy in the Shaker rat model of cerebellar degeneration and ataxia

6. Theme 1_Thur .......................... Banez Coronel, Monica ..................... University of Florida
   Novel RAN proteins accumulate in SCA1,2,3 and 7 brains

9. Theme 5_Thur .......................... Beiner, Melissa ............................... Biohaven
   Results from the Long-Term Open Label Extension Phase Analyses of BHV4157-201: A Phase IIb/III, Randomized, Double-blind, Placebo-controlled Trial of the Safety and Efficacy of Troriluzole in Adult Subjects with Spinocerebellar Ataxia

11. Theme 4_Thur .......................... ‡Chang, Zhuoqing .......................... Duke University
    Assessing oculomotor smooth pursuit function from smartphone video

13. Theme 1_Thur .......................... Chen, Helen ................................. Massachusetts General Hospital
    New pathogenic mutation in the Niemann-Pick C (NPC) Type 1 gene: Confirmation by diagnostic workup of NPC in a 41-year-old woman with idiopathic late onset cerebellar ataxia

15. Theme 2_Thur .......................... Coffin, Stephanie ............................ Baylor College of Medicine
    Brain region specific contribution of the ATXN1/CIC interaction to Spinocerebellar ataxia type 1

16. Theme 1_Thurs .......................... Compton, Andrea .......................... CureDRPLA
    DRPLA: Path to Treatment

17. Theme 2_Thur .......................... ‡Cook, Anna ............................... McGill University
    Exercise acts via BDNF-TrkB signalling to rescue deficits in a mouse model of spinocerebellar ataxia type 6

19. Theme 3_Thur .......................... Corbett, Grant .............................. Exicure, Inc.
    Biodistribution of Spherical Nucleic Acids in the Nonhuman Primate Central Nervous System

21. Theme 1_Thur .......................... ‡Davies, Sarah Perry ...................... Teachers College,
    Characterizing Cough and Swallowing Outcomes in Cerebellar Ataxia  Columbia University

23. Theme 1_Thur .......................... Domingo, Kristine ........................ University of Minnesota
    STUB1 variants are frequent in patients with unexplained adult onset complex ataxia phenotypes

*“Hot Chair” Poster Presenter  ‡Travel Stipend Recipient
Scientific Poster Sessions

Thursday Scientific Poster Sessions (continued)

25_Theme 4_Thur ................................. Durr, Alexandra ........ Sorbonne Universite, Paris, France
Is plasma neurofilament light a longitudinal biomarker for polyglutamine spinocerebellar ataxias?

27_Theme 2_Thur ................................. Elsaey, Mohaned .................... Technische Universität
Zebrafish stable transgenic model for SCA1 Carolo-Wilhelmina zu Braunschweig

29_Theme 4_Thur ................................. *Faber, Jennifer .................... German Center for Neurodegenerative Diseases
Data-driven model of dynamic biomarkers in SCA3 – from early pre-ataxic to late ataxic disease stages

31_Theme 3_Thur ................................. Ferro, Austin ....................... University of Minnesota
Kir4.1 dysregulation, potential common degeneration mechanism between brainstem and cerebellum in SCA1

33_Theme 4_Thur ................................. Gundry, Katie ....................... University of Minnesota
Antisense oligonucleotide gene silencing reverses neurochemical abnormalities in SCA3 mice Medical School

35_Theme 1_Thur ................................. Hamel, Katherine ................... University of Minnesota
Region specific circuitry alterations in SCA1 cerebellum Medical School

37_Theme 3_Thur ................................. Haver, Holly ......................... Duke University
SRCP1 utilizes functional amyloid to suppress polyglutamine aggregation.

39_Theme 1_Thur ................................. Huang, Haoran ...................... University of Michigan
Examining the basis for age-dependent neuronal dysfunction in inherited cerebellar ataxia

41_Theme 4_Thur ................................. Iannuzzelli, Katherine ............. Johns Hopkins University
The Influences of Socioeconomic Status on SCA 3 Disease Course

43_Theme 2_Thur ................................. Johnson, Sean ....................... Wayne State University
Differential Handling and Toxicity of Ataxin-3 Isoforms in Spinocerebellar Ataxia Type 3

47_Theme 2_Thur ................................. Keiser, Megan S. .......... Children's Hospital of Philadelphia
Translating Gene Therapy for Spinocerebellar Ataxia Type 1: Unforeseen consequences in higher mammals

49_Theme 2_Thurs ................................. *Li, Pan P ......................... Johns Hopkins University School of Medicine
Knock-in mouse model of spinocerebellar ataxia type 12

51_Theme 4_Thur ................................. Lin, Chih-Chun ........ Columbia University Medical Center
A Global Comparison of the Frequencies of Spinocerebellar Ataxias
Scientific Poster Sessions

Thursday Scientific Poster Sessions (continued)

53. Theme 2_Thur ................................ Maciel, Patricia ............................... University of Minho
  Glucocorticoid receptor dysfunction as a SCA3/MJD biomarker and target for bile acid therapy

55. Theme 2_Thur ................................ Maciel, Patricia ............................... University of Minho
  Pre-clinical assessment of Mesenchymal Stem cell-based therapies in Spinocerebellar ataxia type 3

58. Theme 2_Thur ............................... Macopson, Joshua Jones  .... Duke University Medical Center
  Nucleocytoplasmic transport is impaired in Spinocerebellar Ataxia type 7

60. Theme 3_Thur .............................. McLoughlin, Hayley S. .................. University of Michigan
  Assessing Benefits of Antisense Oligonucleotide Therapy in a Presymptomatic Mouse Model of Spinocerebellar
  Ataxia Type 3

63. Theme 2_Thur ............................. Morrison, Logan ............................ University of Michigan
  Hypertrophy and potassium channel dysfunction in the spinocerebellar ataxia type 1 brainstem

65. Theme 1_Thur ............................ Ngo, Kathie ..................................... UCLA
  A Diagnostic Ceiling for Exome Sequencing in Cerebellar Ataxia and Related Neurological Disorders

67. Theme 4_Thur ............................ Oubre, Drandon ............................. University of Massachusetts Amherst
  Estimation of Ataxia Severity using Wrist-Worn Sensors and the Finger-to-Nose Test

69. Theme 4_Thurs ............................ Park, Young Woo .......................... University of Minnesota
  Measurement of cross-sectional differences and longitudinal changes with diffusion tensor imaging in spinocerebellar
  ataxias type 1, 2, 3 and 6

71. Theme 3_Thur ............................. Ranum, Laura ................................ University of Florida
  Targeting PKR pathway reduces RAN proteins and provides a novel therapeutic strategy for multiple repeat
  expansion diseases including the SCAs

73. Theme 4_Thur ............................... Pereira de Almeida, Luis ............. Center for Neuroscience
  and Cell Biology (CNCB)
  Pinpointing cerebellar biomarkers by in vivo Proton-magnetic resonance spectroscopy in a transgenic mouse model of
  Machado-Joseph disease/Spinocerebellar ataxia type-3

75. Theme 3_Thur ............................. Poli, Sonia ................................. Minoryx
  MIN102 (Leriglitazone), a brain penetrant PPAR gamma agonist in clinical development for the treatment of
  Friedreich’s ataxia

*“Hot Chair” Poster Presenter  ‡Travel Stipend Recipient
Scientific Poster Sessions

Thursday Scientific Poster Sessions (continued)

77_Theme 3_Thur ............................. Rosa, Juao-Guilherme .......................... University of Minnesota
Brain Derived Neurotrophic Factor (BDNF) Delays Onset of Pathogenesis in Transgenic and Knock-In (KI) Mouse Models of Spinocerebellar Ataxia Type 1 (SCA1)

79_Theme 2_Thur ............................. Schreiber, Anna ............................... University of Alabama at Birmingham
Friedreich’s ataxia neuronal progenitor cells (NPCs) containing an endogenous FXN-luciferase fusion gene as a novel screening platform

81_Theme 4_Thur ............................. Shah, Vrutangkumar V. ........................ Oregon Health & Science University
Digital Outcomes of Mobility in people with Spinocerebellar Ataxia, and Healthy Control Subjects

83_Theme 2_Thur ............................. Simpson, Bryan ............................. Children’s Hospital of Philadelphia
CRISPR-Cas9 Gene Editing Strategies of ATXN2 for the Treatment of Spinocerebellar Ataxia Type 2

85_Theme 2_Thur ............................. Skariah, Geena ............................... University of Michigan
Endogenous tagging of RAN peptides in Fragile X-associated Tremor/Ataxia Syndrome.

87_Theme 3_Thur ............................. Srinivasan, Sharan Ram ........................ University of Michigan
Augmenting BK Channel Dysfunction for Therapeutic Rescue in Spinocerebellar Ataxia

89_Theme 4_Thur ............................. Stephen, Christopher ........................ Massachusetts General Hospital
Gerstmann-Straussler Scheinker disease presenting as a late onset slowly progressive spinocerebellar ataxia: expanding the phenotypic spectrum of genetic prion disease

90_Theme 2_Thurs ............................. *Suart, Celeste ............................... McMaster University
Ataxin-1 is signalled to DNA damage by ATM kinase

92_Theme 2_Thur ............................. ‡Suh, Jaehong ............................... Massachusetts General Hospital
Distinctive Roles of Ataxin-1 in SCA1 and Alzheimer’s Disease

94_Theme 2_Thurs ............................. *Tejwani, Leon ............................... Yale University
Longitudinal single nucleus transcriptomic profiling of the SCA1 mouse cerebellum

95_Theme 1_Thur ............................. Toscano, Brenda ............................... McGill University
Rescue of motor deficits by a mitochondria-targeted antioxidant in a mouse model of ARSACS

98_Theme 4_Thur ............................. Vogel, Adam ............................... University of Melbourne
Alignment of speech outcomes to traditional approaches towards clinical assessment: using Friedreich ataxia as a model for development
Thursday Scientific Poster Sessions (continued)

100_Theme 4_Thur ........................................ Witek, Natalie ............. Rush University Medical Center
Inpatient versus outpatient evaluation of suspected paraneoplastic cerebellar degeneration: A retrospective cohort analysis

102_Theme 4_Thur ................................. Yang, Chen-Ya ..... Columbia University Medical Center
Dysphagia in Spinocerebellar Ataxias type 1, 2, 3, and 6

104_Theme 2_Thur ................................. ‡Zhang, Yalan ....................................... Yale University
Loss of cell survival protein Hax-1 contributes to the neurodegeneration caused by G592R Kv3.3 mutation

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Or scan this QR code:

*“Hot Chair” Poster Presenter  ‡Travel Stipend Recipient
Biographies

Naiara Akizu, PhD – Naiara Akizu received her PhD by the University of Barcelona for her studies on the role of epigenetic chromatin modifications in the nervous system development, a project that she developed in Dr. Marian Martinez-Balbas's laboratory under the co-mentorship of Dr. Elisa Marti. Motivated by her interest in neural development and neurological diseases, she joined Dr. Joseph Gleeson’s laboratory at the University of California, San Diego, for her postdoctoral studies, where she gained expertise in neurogenetics of pediatric diseases and modeling with pluripotent stem cells. She complemented her training by working with mouse models of neurological diseases under the co-mentorship of Dr. Ulrich Müller at The Scripps Research Institute, La Jolla. She is recipient of the NIH K99/R00 award and NAF young investigator award, among other fellowships that funded her scientific research career.

Since 2017, Naiara Akizu is a faculty member of the Center for Cellular and Molecular Therapeutics at the Children’s Hospital of Philadelphia and Assistant Professor of Pathology and Laboratory Medicine at the University of Pennsylvania. Her laboratory studies pathogenic mechanisms of neurodevelopmental and neurodegenerative disorders that affect children, some of which involve the cerebellum, with the ultimate goal to contribute to the future development of therapies for currently incurable disorders.

Adam Avery, PhD – I am interested in cytoskeletal mechanisms that support neuronal morphogenesis and maintenance, and understanding how these mechanisms are disrupted in neurodegenerative diseases. Currently my lab is investigating how the spectrin-actin cytoskeleton supports the formation of complex dendritic arbors, and how spectrin function and arborization are disrupted by spinocerebellar ataxia type 5 mutations in β-III-spectrin. Using protein biochemical techniques, together with genetic and live cell imaging approaches in the model organism Drosophila melanogaster, we recently revealed that the spectrin-actin cytoskeleton is required for dendrite stability and arbor outgrowth. Significantly, we also determined that a subset of SCA5 mutations increase the binding affinity of β-III-spectrin for actin. Using a novel FRET assay, we are performing high throughput drug screening to identify small molecules that target mutant spectrin and reduce its affinity for actin. I am an Assistant Professor of Biochemistry at Oakland University.

Monica Banez-Coronel, PhD – Monica Banez-Coronel is an assistant scientist at the Center for NeuroGenetics, University of Florida. Monica received her B.Sc. in Biology from the University of Seville in 1999. She completed her Ph.D. in Biochemistry and Molecular Biology at the Autonomous University of Madrid (2007), studying the inhibition of a lipidic enzyme often overactivated in tumors as a selective strategy for human cancer therapy.

Her interest on understanding the molecular mechanisms of disease took her to join the laboratory of Xavier Estivill, M.D., Ph.D. at the Centre for Genomic Regulation (Barcelona) as a postdoctoral fellow (2007), where she explored small RNA-mediated toxicity in
Huntington’s disease (HD) and the suitability of blocking the RNA toxic species to improve disease symptoms in HD mouse models.

Shortly after the discovery of repeat associated non-ATG (RAN) translation by the Ranum lab, Monica joined the group of Laura Ranum, Ph.D, at the Center for NeuroGenetics, University of Florida (2012) where she focused on studying RAN translation in Huntington’s disease (HD), RAN protein toxicity and the correlation of RAN protein accumulation with disease pathology.

Monica joined the faculty of the Department of Molecular Genetics and Microbiology at the University of Florida in 2017. Her current research focus on exploring RAN translation accumulation across CAG•CTG expansion disorders and understanding the contribution of RAN proteins to disease pathogenesis.

**Esther B.E. Becker, PhD** – Esther Becker is an Associate Professor of Neurobiology in the Department of Physiology, Anatomy and Genetics at the University of Oxford.

Esther received an MSc degree in Medical Biology from the University of Amsterdam in the Netherlands. She obtained her Ph.D. from Harvard University, where she discovered novel signaling pathways regulating neuronal apoptosis. Esther did her post-doctoral training supported by a Human Frontier Science Program Fellowship at the University of Oxford. During this time she characterized a novel mouse model of cerebellar ataxia, the Moonwalker mouse. In 2010, she was awarded a Research Fellowship from the Royal Society to establish her own research program at the University of Oxford. Her research aims to elucidate the genetic and molecular underpinnings of cerebellar disorders and to develop therapeutic interventions. Her work has led to the discovery of novel gene mutations causing spinocerebellar ataxia (SCA), including SCA41 (TRPC3) and most recently SCA44 (GRM1). The Becker group has recently developed a robust and reproducible protocol to generate cerebellar neurons from human induced pluripotent stem cells.

**Andreea C. Bostan, PhD** – Andreea C. Bostan, PhD, is a Research Assistant Professor in the Department of Neurobiology and the Systems Neuroscience Center at the University of Pittsburgh. The overarching goal of her research is to understand the organization and function of neural circuits at the interface between movement, thought, and emotion. A graduate of the University of Toronto, Andreea received her doctorate in Neuroscience from the University of Pittsburgh. Under the mentorship of Peter L. Strick, Ph.D., Andreea used neurotropic viruses as transneuronal tracers to establish that the cerebellum and basal ganglia are interconnected and form an integrated network with the cerebral cortex. These results are published in the Proceedings of the National Academy of Science (2010) and continue to have a profound impact on concepts about contributions of the cerebellum and the basal ganglia to health and disease. As discussed in a recent Nature Reviews Neuroscience (2018) review, Andreea is particularly interested in how changes in the integrated network, which links the cerebellum with the basal ganglia and the cerebral cortex, contribute to the manifestation of neurologic and psychiatric disorders.
Biographies (continued)

David D. Bushart, PhD – David Bushart is a Postdoctoral Research Fellow in the Department of Neurology at the University of Michigan. David currently works in the laboratory of Dr. Vikram Shakkottai, with whom he also completed his Ph.D. training in 2018. For the past six years, David’s research has focused on the relationship between ion channel dysfunction and impaired motor function in degenerative ataxia. David’s work has helped identify a group of ion channels that show impaired expression and function across spinocerebellar ataxias (SCA) of multiple etiologies. In addition, David has helped identify new drug strategies that improve ion-channel activity and neuronal function in SCA. David’s future research will continue to focus on the development of new therapies that target ion-channel dysfunction in SCA and other degenerative disorders.

David was the recipient of the 2018 Davenport Research Award from the Department of Molecular & Integrative Physiology at the University of Michigan. In addition to his research interests, David works part-time as a Clinical Trials Coordinator for ataxia-related studies at Michigan Medicine.

Thomas Cech, PhD, Nobel Laureate – After his Ph.D. in chemistry from the University of California, Berkeley and postdoctoral research at the Massachusetts Institute of Technology, Dr. Cech joined the faculty of the University of Colorado Boulder in 1978. In 1982 Dr. Cech and his research group discovered self-splicing RNA in Tetrahymena, providing the first exception to the long-held belief that biological reactions are always catalyzed by proteins. Because RNA can be both an information-carrying molecule and a catalyst, perhaps a primordial self-reproducing system consisted of RNA alone.

Dr. Cech became a Howard Hughes Medical Institute investigator in 1988 and Distinguished Professor of Chemistry and Biochemistry in 1990. From 2000–2009, he served as president of the Howard Hughes Medical Institute, the largest private biomedical research organization in the U.S.A. He has now returned to full-time research and teaching at the University of Colorado Boulder. Dr. Cech’s work has been recognized by many national and international awards and prizes, including election to the U.S. National Academy of Sciences (1987), the Heineken Prize of the Royal Netherlands Academy of Sciences (1988), the Albert Lasker Basic Medical Research Award (1988), the Nobel Prize in Chemistry (1989), and the National Medal of Science (1995).

Alice Chen-Plotkin, MD – Alice Chen-Plotkin is the Parker Family Associate Professor of Neurology at the University of Pennsylvania. A Phi Beta Kappa graduate and English literature major at Harvard University, she began her scientific training as a Rhodes Scholar at Oxford University. She subsequently joined the faculty at the Perelman School of Medicine at the University of Pennsylvania, where she leads a research group studying the neurodegenerative diseases and maintains a clinical practice caring for Parkinson’s Disease patients. She has won top awards from both the Academy of Neurology (Jon Stolk Award, 2014) and the American Neurological Association (Derek Denny Brown Award, 2018) for her translational research efforts.
Marija Cvetanovic, PhD – Dr. Marija Cvetanovic received her BSc in Molecular Biology and Physiology from the University of Belgrade in 1998. She completed her PhD at the Department of Immunology, Microbiology and Virology at the University of Illinois in Chicago in 2004, studying recognition of apoptotic cells by macrophages. As a postdoctoral fellow she worked in the laboratory of Dr. Puneet Opal, exploring the molecular pathology of Purkinje neurons in the Spinocerebellar Ataxia Type 1 (SCA1).

In 2012, Marija joined the Department of Neuroscience and Institute for Translational Neuroscience at the University of Minnesota as the Assistant professor. Research in her laboratory aims to increase our understanding of how glial cells modulate pathogenesis of SCA1 as well as of the etiology of cognitive and mood deficits in SCA1.

Sascha Du Lac, PhD – Sascha du Lac, PhD, is a Professor of Otolaryngology, Neurology, and Neuroscience at the Johns Hopkins University School of Medicine. She received her BA in Biology from University of Chicago and PhD in Neuroscience from Stanford University. After a postdoctoral fellowship in Physiology at UCSF, she joined the faculty of the Salk Institute and was appointed as an Investigator of the Howard Hughes Medical Institute prior to moving to Johns Hopkins University. Her laboratory studies the structure, function, and plasticity of brainwide cerebellar circuits.

Jennifer Faber, MD – Dr. Jennifer Faber is resident physician at the Department of Neurology, University Hospital Bonn, Germany, and clinical investigator and research assistant at the German Center of Neurodegenerative Diseases (DZNE) in the group of Prof. Thomas Klockgether. She received a prediploma in Mathematics from Bonn University, and graduated from Bonn University Medical School with an M.D. and a doctor of medicine degree. After a research stay at the Department of Psychology at Stanford University, she continued her postdoctoral training at the DZNE.

Dr. Faber’s research focuses on MR imaging of ataxias. In particular, she uses diffusion tensor imaging to assess structural alterations of the white matter beyond atrophy patterns. She received the second prize of the Mähler-Linke-Foundation for an imaging study in sporadic adult-onset ataxias. Within the ongoing European Spinocerebellar Type 3/Machado-Joseph Disease Initiative (ESMI) she co-leads the longitudinal collection of standardized multimodal MRI. Here, the particular focus lies on mapping structural alterations over the whole course of the disease including the pre-ataxic stage. Together with Dr. Heike Jacobi she initiated the SCA Global Young Investigator Initiative, a platform for scientific exchange amongst young researchers and for education and training purposes. Since 2020 is Dr. Faber fellow of the Hertie Network of Excellence in Clinical Neuroscience.
Phyllis Faust, MD, PhD – Dr. Phyllis Faust is Professor of Pathology and Cell Biology at Columbia University Medical Center, NY, NY, where she is a board-certified neuropathologist and conducts research focused on the neuropathology of human cerebellar neurodegenerative diseases. Dr. Faust received her MD-PhD in Cell and Molecular Biology at Washington University School of Medicine, St. Louis, MO. She completed pathology residency training at Barnes-Jewish Hospital in St. Louis (anatomic pathology) and Columbia University Medical Center (neuropathology), followed by a Howard Hughes postdoctoral fellowship in the developmental neurobiology laboratory of Dr. Mary E. Hatten at Rockefeller University.

Dr. Faust has a longstanding interest in cerebellar biology from both a developmental and neurodegenerative perspective. Her laboratory developed the first mouse knockout model for a peroxisomal biogenesis disorder defect, which models a human neuronal migration disorder, Zellweger syndrome. Her analyses in this knockout model further fostered her interest in cerebellar pathology and neuronal degeneration. Since 2006, a series of seminal studies in her laboratory has identified a cluster of quantifiable morphologic and molecular changes in the human postmortem essential tremor cerebellum that distinguish them from controls, and are centered mainly in/around the Purkinje cell. We have now formally studied the degree to which several of these pathologies occur in primary cerebellar neurodegenerative diseases such as spinocerebellar ataxias (e.g., SCA 1, 2, 3 and 6) and multiple system atrophy, and in other tremor disorders where cerebellar dysfunction is postulated (e.g., Parkinson’s disease, dystonia). Using a unique quantitative morphologic approach, we have defined that a somewhat distinctive morphologic signature of cerebellar degenerative changes marks these disorders of ataxia and tremor.

Mandi Gandelman, PhD – Mandi Gandelman, PhD is a postdoctoral fellow at the Stefan Pulst lab, Department of Neurology, University of Utah. Mandi received her Master’s and PhD degree from the Universidad de la Republica, in Montevideo, Uruguay. During her PhD she worked on motor neuron and astrocyte signaling that contributes to death of motor neurons in Amyotrophic Lateral Sclerosis. In 2017 she joined Dr. Pulst lab, which focuses on Spinocerebellar Ataxia type 2, with research ranging from the basic mechanisms of disease to therapy development. Mandi currently studies neuronal stress pathways involved in Purkinje cell death in Spinocerebellar Ataxia type 2, with a special focus on neurodegeneration caused by mutated ATAXIN-2.

Joel M. Gottesfeld, PhD – Joel Gottesfeld is a Professor of Molecular Medicine at The Scripps Research Institute in La Jolla, California. The major effort in our laboratory for the past decade has been to develop a novel therapeutic approach for the neurodegenerative disease Friedreich’s ataxia (FRDA). At present, there is no approved treatment for this disease. While the genetic basis for FRDA is well established (loss of the essential mitochondrial protein frataxin due to a GAA triplet repeat expansion in the FXN gene), our laboratory was the first to demonstrate that gene silencing is due to heterochromatin formation on pathogenic FXN alleles. Importantly, we showed that members of the 2-aminobenzamide class of histone deacetylase inhibitors reverse gene silencing in cells,
Biographies (continued)

and in collaborative studies, in mouse models for FRDA. Our laboratory developed a human neuronal cell model for FRDA, based on differentiation of patient induced pluripotent stem cells, and this model offers the first opportunity to screen compounds in a cell type relevant to the human disease. A Phase Ib clinical trial of one of our compounds has shown biochemical efficacy in reversing FXN gene silencing in patients but pointed to pharmacological limitations of this compound class. Current efforts are aimed at identification of compounds with improved efficacy and improved pharmacological properties. We hope to take these newer compounds into human clinical studies in the near future.

Viviana Gradinaru, PhD – Dr. Viviana Gradinaru completed her B.S. at Caltech and her Ph.D. research at Stanford University and is now a Professor of Neuroscience and Biological Engineering at Caltech. Dr. Gradinaru’s research interests focus on developing tools and methods for neuroscience (optogenetic actuators and sensors; tissue clearing and imaging; gene delivery vehicles) and using them to characterize circuits underlying locomotion, reward, and sleep, with the goal to inform deep brain stimulation (DBS) and better understand the underlying mechanisms of action.

Prof. Gradinaru has received the NIH Director’s New Innovator and Pioneer Awards and a Presidential Early Career Award for Scientists and Engineers, and has been honored as a World Economic Forum Young Scientist and as one of Cell’s 40 under 40. Gradinaru is also a Sloan Fellow, Pew Scholar, Moore Inventor, Valleé Scholar, and Allen Brain Institute NGL Council Member, and received the inaugural Peter Gruss Young Investigator Award by the Max Planck Florida Institute for Neuroscience. In 2017 she was the Early-Career Scientist Winner in the Innovators in Science Award in Neuroscience (Takeda and the New York Academy of Sciences) and in 2018 she received a Gill Transformative award.

Viviana Gradinaru has also been very active in teaching and service, participating with lab members in regular technology training workshops at Caltech and for summer courses at Cold Spring Harbor Laboratory as well as running the CLOVER Center (Beckman Institute for CLARITY, Optogenetics and Vector Engineering), which provides training and access to the group’s reagents and methods for the broader research community.

Gilles J. Guillemin, PhD, OM – Professor Gilles Guillemin has been working in the fields of Neuroimmunology and tryptophan metabolism for more than 22 years. Professor Guillemin’s team is one of the world’s leading research groups working on the involvement of the tryptophan catabolism (via the kynurenine pathway) in human neurodegenerative diseases.

He demonstrated the roles of this pathway in multiple sclerosis, Alzheimer’s disease, amyotrophic lateral sclerosis, cancers and used the kynurenine pathway metabolites as biomarkers for disease sub-types, severity, progression and response to treatments.

He is the President of the International Society for Tryptophan Research and Editor-in-Chief of the International Journal for Tryptophan Research.

Guillemin has published more than 200 peer reviewed scientific articles. His current h-index is currently 60 and he has more than 14,000 citations.
Biographies (continued)

**Karl Herrup, PhD** – Karl Herrup received his Bachelor’s degree from Brandeis University and his Ph.D. in Neuroscience from Stanford. After two postdoctoral fellowships – in Neurogenetics at Harvard Medical School, and in Neuropharmacology at the Biozentrum in Basel, Switzerland – he joined the faculty of the Human Genetics Department of Yale Medical School. He became Director of the Division of Developmental Neurobiology at the E. K. Shriver Center in 1988. In 1992 he moved to the Department of Neurosciences at Case Western Reserve University Medical School. While there, he directed the University Alzheimer’s Center from 1999 through 2005. In 2006 he moved to Rutgers University to become Chair of the Department of Cell Biology and Neuroscience. In July 2012, he moved to Hong Kong to become the Head of Life Science at The Hong Kong University of Science and Technology. In March 2019, with his contract in Hong Kong coming to an end, he returned to the US and is now a Professor of Neurobiology at the University of Pittsburgh.

**Henry Houlden, MD, PhD** – Our laboratory works on neurogenetics with a particular interest in spinocerebellar ataxia and movement disorders, particularly in diverse populations. We integrate new gene discovery such as CANVAS, SCA11, SCA15, genetics modifiers in the SCAs, using the latest genomic techniques with functional experimental validation in human tissue and other model systems. Our goal is to develop new therapeutics based on an improved understanding of disease mechanisms in children and adults. We recently established a collaborative project to genotype and carry out a GWAS in SCA patients with repeat expansions from around the world to identify genetic modifiers of age at onset and severity. As a group we are very keen to collaborate and keen to analyze further families, possible CANVAS and SCA patients in the age at onset GWAS.

**Heike Jacobi, MD** – Dr. Heike Jacobi works as consultant and chief resident at the Department of Neurology, University Hospital Heidelberg, Germany. After she graduated from Cologne University Medical School with an M.D. and a doctor of medicine degree she successfully completed her specialist training in Neurology at the Department of Neurology of the University Hospital in Bonn. In parallel she worked as a research fellow at the German Center of Neurodegenerative Diseases (DZNE). Since 2016 she continues her work at the Department of Neurology, University Hospital in Heidelberg, Germany.

Dr. Jacobi’s research focuses on the clinical characteristics and natural history of the most common spinocerebellar ataxias with a special interest in the pre-ataxic stage. During her time at the Bonn University Hospital and the German Center of Neurodegenerative Diseases (DZNE) Dr. Jacobi coordinated two international multicenter cohort studies (EUROSCA & RISCA) lead-managed by Prof. Thomas Klockgether. For her work she was awarded the German Heredo-Ataxia Price in 2015 and the Hans-Jörg Weitbrecht Price in 2017. Together with Dr. Jennifer Faber she initiated the SCA Global Young Investigator Initiative, a platform for scientific exchange amongst young researchers and for education and training purposes.
Biographies (continued)

**Holly Kordasiewicz, PhD** – Dr. Holly Kordasiewicz is Vice President and Head of Neurology Research at Ionis Pharmaceuticals, a company that specializes in RNA therapeutics. Holly heads a team focused on identifying antisense oligonucleotide therapeutics for currently untreatable neurodegenerative diseases, including drugs now in clinical trials for ALS, Alzheimer’s disease, Parkinson’s disease and Huntington’s disease. Holly joined Ionis eight years ago after working on the preclinical validation for Ionis’ Huntington’s disease program as a post-doctoral fellow in the laboratory of Dr. Don Cleveland at UCSD. Holly began her work on understanding and developing therapies for neurodegenerative diseases at the University of Minnesota, where she received her Ph.D. in Neuroscience.

**Walter J. Koroshetz, MD** – Dr. Koroshetz became Director of the National Institute of Neurological Disorders and Stroke in 2015. He joined NINDS in 2007 as Deputy Director and has held leadership roles in a number of NIH and NINDS programs including the NIH’s Brain Research through Advancing Innovative Neurotechnologies® (BRAIN) Initiative, the NIH Blueprint for Neuroscience, and the Helping to End Addiction Long Term (HEAL) Initiative.

Before joining NINDS, Dr. Koroshetz served as Vice Chair of the neurology service and Director of stroke and neurointensive care services at Massachusetts General Hospital (MGH). He was a professor of Neurology at Harvard Medical School (HMS) and led neurology resident training at MGH between 1990 and 2007. A major focus of his clinical research career was to develop measures in patients that reflect the underlying biology of their disorders. This led to clinical research using brain imaging techniques including MR spectroscopy in Huntington’s disease, diffusion/perfusion MR and CT imaging, CT angiography, and acute clot removal for large artery stroke.

**Sheng-Han Kuo, MD** – Sheng-Han Kuo is a physician-scientist focusing on the translational research for ataxia and tremor. His group identified key pathological features of human cerebellum of ataxia and tremor and using mouse modeling and physiological tools to probe the cerebellar circuitry of ataxia and tremor. In addition, he organized the Initiative for Columbia Ataxia and Tremor, to engage clinicians, scientists, and engineers at Columbia University to collaborate to find therapy for cerebellar disorders. He is also one of the tri-leaders for Clinical Research Consortium for Spinocerebellar Ataxias and has been analyzing the natural history data to advance our understanding of clinical characteristics and genetic modifiers for spinocerebellar ataxias.
Biographies (continued)

Albert La Spada, MD, PhD, FACMG – Albert La Spada graduated Summa Cum Laude from the University of Pennsylvania with a degree in Biology in 1986. While a M.D. - Ph.D. student at the University of Pennsylvania School of Medicine, La Spada identified the cause of X-linked spinal & bulbar muscular atrophy (SBMA) as an expansion of a trinucleotide repeat in the androgen receptor gene. As the first disorder shown to be caused by an expanded repeat tract, this discovery of a novel type of genetic mutation led to the emergence of a new field of study. After completing training as a Clinical Genetics fellow and a Howard Hughes Medical Institute Physician Post-doctoral Fellow, he joined the faculty at the University of Washington Medical Center in 1998, and became a Professor of Laboratory Medicine, Medicine (Medical Genetics), Pathology, and Neurology (Neurogenetics). In 2009, Dr. La Spada accepted the position of Professor and Division Head of Genetics in Pediatrics, Cellular & Molecular Medicine, and Neurosciences at the University of California, San Diego, and was a founding faculty member of the UCSD Institute for Genomic Medicine and Sanford Consortium for Regenerative Medicine. In 2017, Dr. La Spada was selected as the founding Director of the Duke Center for Neurodegeneration & Neurotherapeutics, and is a Distinguished Professor of Neurology, Neurobiology, and Cell Biology, and the Lincoln Financial Endowed Chair at the Duke University School of Medicine, where he serves as the Vice Chair for Research in the Department of Neurology.

Dr. La Spada’s research is focused upon neurodegenerative disease, and he is seeking the molecular events that underlie neurodegeneration and neuron cell death in spinocerebellar ataxia type 7 (SCA7), SBMA, Huntington’s Disease, ALS, and Parkinson’s disease. He and his team have uncovered evidence for transcription dysregulation, perturbed bioenergetics, and altered protein quality control as contributing factors to neuron dysfunction. By reproducing molecular pathology in mice and in neurons derived from human patient stem cells, Dr. La Spada has begun to develop therapies to treat these disorders. Dr. La Spada has been the recipient of grants and awards from the National Institutes of Health, Howard Hughes Medical Institute, Muscular Dystrophy Association, Hereditary Disease Foundation, CHDI, Coulter Foundation, American Federation for Aging Research, Packard Center for ALS Research, Michael J. Fox Foundation, and Harrington Discovery Institute. Among his awards is the Paul Beeson Physician Faculty Scholar Aging Research Award. In 2006, Dr. La Spada was inducted into the American Society for Clinical Investigation. In 2007, he was bestowed with the Lieberman Award by the Hereditary Disease Foundation for excellence in Huntington’s Disease research, and in 2011, he received the Molecular Mechanisms of Neurodegeneration Distinguished Research Award in Milan, Italy. In 2013, Dr. La Spada was inducted into the Association of American Physicians, and in 2015, Dr. La Spada was selected as a Gund-Harrington Scholar for his translational research accomplishments. In 2018, Dr. La Spada became a funded member on one of eleven teams that comprise the Chan Zuckerberg Initiative’s Neurodegeneration Challenge Network, after an international competition.
Biographies (continued)

Pan Li, PhD – Dr. Pan Li is currently an Assistant Professor from Division of Neurobiology, Department of Psychiatry and Behavioral Sciences at Johns Hopkins University School of Medicine. She obtained her Ph.D. in Biology (with a concentration on molecular medicine) from the Hong Kong University of Science and Technology, and then she moved to Johns Hopkins for postdoctoral training with a focus on RNA toxicity and RNA-binding proteins in Huntington’s disease (HD) and spinocerebellar ataxia type 2 (SCA2).

Her current research interest is in the molecular pathogeneses of spinocerebellar ataxia types 2 and 12 (SCA2 and SCA12). She received the Post-Doc fellowship award from National Ataxia Foundation in 2016, and then the YI-SCA research award in 2018, for which she feels very grateful. Using CRISPR/Cas9 approach, she has recently generated the first humanized knock-in mouse model of SCA12.

Owen Morgan, BA – Owen Morgan is a Research Assistant in Dr. Cherie Marvel’s Cognitive Neuropsychiatric Research Laboratory at the Johns Hopkins School of Medicine, Department of Neurology. Owen’s research uses experimental cognitive testing and neuroimaging methods to understand the mechanisms underlying the non-motor effects of cerebellar ataxia. He graduated from St. John’s College in 2017 with a BA in Philosophy and the History of Science and Mathematics.

Larissa Nitschke, BSc – Larissa Nitschke is a Ph.D. candidate in the Integrative Molecular and Biomedical Sciences Program at Baylor College of Medicine in Houston, Texas. She obtained her Bachelor of Science degree in Biology from the Heinrich-Heine University in Düsseldorf, Germany. After completing her thesis research in Germany, Larissa was awarded a prestigious scholarship to perform additional research in genetics and molecular biology at Michigan State University.

For her graduate studies, she joined the laboratory of Dr. Huda Zoghbi to investigate the post-transcriptional and post-translational regulation of Ataxin-1, the causative gene of the neurodegenerative disease Spinocerebellar Ataxia Type 1. Her work has identified a miRNA that regulates ATXN1 expression via interaction with its 5’UTR and demonstrated that disruption of a key phosphorylation site that affects the stability of the ATXN1 protein could mitigate SCA1 pathogenesis in a mouse model of SCA1.
**Biographies (continued)**

**Harry Orr, PhD** – Harry Orr, PhD directs the Institute for Translational Neuroscience and is the Tulloch Professor of Genetics in the Department of Laboratory Medicine and Pathology at the University of Minnesota Medical School. Dr. Orr received a BA degree from Oakland University in Rochester, Michigan. He earned his PhD in neurobiology at Washington University, St. Louis, Missouri and completed a Research Fellowship at Harvard University. Dr. Orr is known as the researcher who, along with Dr. Huda Zoghbi, found the first gene for ataxia, now known as SCA1. Dr. Orr’s research program is focused on the molecular genetics of mammalian development and neurodegenerative diseases. He is a published author of more than 120 articles, many on the genetics of ataxia. Dr. Orr is a member of the National Ataxia Foundation’s Board of Directors and Research Director on NAF’s Medical and Research Advisory Board.

**Young Woo Park, PhD** – Young Woo Park received his Bachelor’s degree from the University of Michigan – Ann Arbor in Electrical Engineering, and his Master’s degree in Biomedical Engineering from the University of Southern California. After working as a software engineer at Bionet Co. Ltd. as a substitute for military service in Seoul, South Korea, he went on to doctoral program at Korea Advanced Institute of Science and Technology (KAIST) in Daejeon, South Korea. Since earning his PhD in Electrical Engineering in 2018, he joined the Center for Magnetic Resonance Research at the University of Minnesota, and works as a post-doctoral research associate.

**Leonard Petrucelli, PhD** – Leonard Petrucelli, Ph.D., is a Ralph B. and Ruth K. Abrams Professor and enterprise chair of the Department of Neuroscience at Mayo. Dr. Petrucelli earned his Bachelor of Science degree at Barry University, Miami, and his Ph.D. degree in molecular and cellular biochemistry at Loyola University and Stritch School of Medicine, Chicago. He came to Mayo Clinic’s Florida campus as a research fellow in 2000 and joined the neurosciences research staff two years later.

Dr. Petrucelli and his research team are at the forefront of their field, researching the cellular mechanisms that cause neurodegeneration in Alzheimer’s disease, amyotrophic lateral sclerosis (ALS), or Lou Gehrig’s disease, and frontotemporal dementia (FTD) and more recent movement disorders and stroke. By combining expertise in drug discovery, cell biology and induced pluripotent stem cell (iPSC) modeling, his lab aims to develop therapies and biomarkers for the treatment of diseases characterized by abnormal protein aggregation. Dr. Petrucelli’s team recently discovered a new therapeutic target and biomarker with the aim of improving the diagnosis and prognosis for patients suffering from FTD and ALS. His team’s research has been published in top tier journals including Science, Nature Medicine, Nature Neuroscience, Neuron, Journal of Clinical Investigation and Annals of Neurology.

Dr. Petrucelli is principal investigator for several grants funded by the National Institutes of Health (NIH) including R35 and is director of two funded NIH programs focused on c9orf72 and Tau Center without Walls. He serves on the Scientific Advisory Board of Science Translational Medicine. He is also the Chief Scientific Advisor to Target.
Stefan Pulst, MD – Stefan M. Pulst is professor and chair of the Department of Neurology at the University of Utah in Salt Lake City. Research in his group focuses on inherited diseases of the nervous system with an emphasis on spinocerebellar ataxias, ALS, and Parkinson disease. The overall goal of his research is to proceed from gene identification in human pedigrees to modeling of the disease in vitro and in rodents. At the mechanistic level, his group is interested in the interplay between UPR, autophagy and RNA granules in neurodegeneration and translating these insights into using antisense oligonucleotide-based therapies for these disorders. Pulst has received numerous awards, most recently the George C. Cotzias award from the American Academy of Neurology, a Senator Jacob Javits Award for Neuroscience from NINDS, and the Singhal Movement Disorders Award from the World Federation of Neurology. Pulst is the founding editor of Neurology® Genetics.

Paul Ranum, PhD – Paul Ranum, PhD completed his doctorate in Molecular and Cellular Biology at the University of Iowa with renowned geneticist Dr. Richard Smith, where he co-developed the first RNAi based gene therapy approach capable of preventing hearing loss in mice. To better characterize the rare cell types targeted by this therapy, he spearheaded efforts to isolate single cells from the inner ear for single-cell RNA-Sequencing. This project revealed new pathways, novel exons and isoform configurations that provide new targets for therapeutic development.

As a postdoc in Dr. Beverly Davidson’s lab at the Children’s Hospital of Philadelphia and University of Pennsylvania he is implementing a barcode based single-cell RNA-Seq protocol capable of mapping the mouse and human brain at fine spatial and temporal resolution. In the context of spinocerebellar ataxia, this approach will be used to characterize patterns of transcriptional changes that precede degeneration in the cerebellum. He is also an integral team member on projects directed at in vivo AAV evolution in non-human primates, and innovations in drug inducible switches for the control of gene therapy products.

Paul is a trainee in the University of Pennsylvania T32 program in Genomic Medicine and his research is supported by a postdoctoral fellowship from the Hereditary Disease Foundation.

Liana Rosenthal, MD, PhD – Liana Rosenthal, MD, PhD is an Assistant Professor at Johns Hopkins focusing on improving the care and treatment of individuals with ataxia, Parkinson’s disease and other movement disorders. She serves as the Director of the Johns Hopkins Ataxia Clinic as well as the Clinical Core of the Morris K. Udall Parkinson’s Disease Research Center of Excellence amongst other leadership positions. Her career focuses on the development of a multidisciplinary care model for individuals with ataxia and the identification of biomarkers for ataxia and Parkinson’s disease, with the goal of using those biomarkers to identify disease-modifying therapies.
**Biographies (continued)**

**Matthew Scaglione, PhD** – I am currently an Assistant Professor in the Department of Molecular Genetics and Microbiology at Duke University. My lab focuses on understanding the molecular mechanisms that counteract protein aggregation in neurodegenerative diseases. One half of my lab focuses on the social amoeba Dictyostelium discoideum, an organism that we have identified as a proteostatic outlier that is naturally resistant to polyglutamine aggregation. We have gone on to identify a novel type of molecular chaperone that selectively recognizes aggregation-prone polyglutamine-expanded proteins and targets them to the proteasome for degradation. In addition to providing resistance to polyglutamine aggregation to this amoeba we have also found that this chaperone is sufficient to impart resistance to polyglutamine aggregation in human neurons. We are currently working on developing small molecules that mimic this protein and may be useful for treating the polyglutamine aggregates. The second half of my lab focuses on investigating the neuroprotective E3 ligase CHIP that is mutated in Spinocerebellar ataxia, autosomal recessive type 16 (SCAR16). Our work has identified how mutations in CHIP cause SCAR16 and we are currently using this information to develop small molecule regulators of CHIP that may be useful for treating a wide variety of neurodegenerative diseases.

**Vikram Shakkottai, MD, PhD** – Dr. Shakkottai received his medical degree from the Christian Medical College, Vellore, India. He then completed a Ph.D. in biological sciences at the University of California, Irvine and a residency in neurology at Washington University in Saint Louis. He subsequently did a fellowship in movement disorders at the University of Michigan. He is currently Associate Professor of Neurology, Molecular and Integrative Physiology at the University of Michigan. Dr. Shakkottai sees patients with cerebellar ataxia and other balance disorders in the Ataxia Clinic at the University of Michigan. He is the current Director of the Ataxia Program, and he also directs the Michigan Brain Bank.

Dr. Shakkottai’s research involves understanding alterations in neuronal function in cerebellar ataxia. Using mouse models of cerebellar ataxia, his work has established that alterations in the electrical firing patterns of neurons in the cerebellum precedes the loss of neurons and contributes to impaired coordination. His research also suggests that this aberrant neuronal activity contributes to neurodegeneration in ataxia. A major goal of his laboratory is to develop agents targeting ion-channels, molecules that are important for maintaining normal neuronal activity, in order to correct aberrant patterns of electrical activity in cerebellar neurons, and treat symptoms of ataxia.

Dr. Shakkottai has received numerous awards in medical school and was ranked #1 in his medical class. He was awarded the Dorothy Penrose Stout Award for the Best Predoctoral Fellowship application from the American Heart Association Western States Affiliate, the Leonard Berg award for resident research at Washington University, and a Young Investigator award from the National Ataxia Foundation.
Biographies (continued)

**Divya Singhal MD, FAAN** – Dr. Singhal is currently Associate Professor of Neurology at University of Oklahoma, TBI Program Director and Residency Director VA in Oklahoma City. She is ABPN Board certified in Neurology, Epilepsy and Clinical Neurophysiology. As Vice Chair of Women’s issues in Neurology section of American Academy of Neurology (AAN) – she is collaborating on national projects for mentoring neurologists and gender equity educational webinar development. She has done numerous invited national and regional presentations – through National VA Women's Health, AAN, American College of Osteopathic Obstetricians and Gynaecologists (ACOOG); Regional presentations through Epilepsy foundation, VA VISN 19 Women’s Health and community outreach programs.

Her other professional interests include Physician wellbeing, healthcare leadership and clinical education. She is a member of AAN’s joint coordinating council for Wellbeing. She received the prestigious A.B. Baker Teacher Recognition Award from AAN in 2019 for achievement and dedication to education within the field of Neurology. She has also completed Harvard Kennedy School’s Senior Executive Fellows Program – a four-week executive education program for managers seeking to advance to executive leadership positions, through Department of Veteran Affairs’ nationwide corporate leadership development initiative.

**Celeste Suart, PhD Student** – Celeste Suart is a Biochemistry and Biomedical Sciences PhD student in the laboratory of Dr. Ray Truant at McMaster University in Hamilton, Canada. Celeste received her BHSc in Biomedical Discovery & Commercialization from McMaster University in 2017. Her thesis in the Truant Lab focuses on oxidative DNA damage in the context of polyglutamine diseases, primarily SCA1. Her current research focus is on the DNA damage response of the ataxin-1 protein. In 2018, Celeste co-founded SCAsource.net, a knowledge translation website where scientific articles on ataxia are transformed into lay summaries to make research findings accessible to all. In 2019, she was elected to co-chair the 2021 Triplet Repeat Disorders Gordon Research Seminar.

**Pawel M. Switonski, PhD** – Pawel M. Switonski, Ph.D., is a postdoc in Dr. Albert La Spada lab at Duke Center for Neurodegeneration & Neurotherapeutics at Duke University. Dr. Switonski received his Ph.D. in Biochemistry from the Institute of Bioorganic Chemistry, Polish Academy of Sciences, where he completed his thesis on modeling spinocerebellar ataxia type 3 in mice, under the mentorship of Dr. Wlodzimierz Krzyzosiak. In 2016 he was awarded a Postdoctoral Fellowship from Polish Ministry of Science and Higher Education. Dr. Switonski’s research is focused on understanding the molecular mechanisms contributing to the pathogenesis of polyglutamine expansion disorders. He is particularly interested in spinocerebellar ataxia type 7 (SCA7) – an inherited neurodegenerative disease characterized by progressive cerebellar and retinal degeneration. His current research is focused on the role of DNA damage and DNA repair pathways, as well as epigenetic modulation in SCA7.
Biographies (continued)

David Szmulewicz, MD, PhD – David Szmulewicz is an Australian Neurologist, Neuro-otologist and medical researcher. He holds a PhD from the University of Melbourne and his clinical and research interests include balance disorders that affect the cerebellum, vestibular system and the combination of the two. David is the head of the Balance Disorders & Ataxia Service at the Royal Victorian Eye & Ear Hospital, founder of the Alfred Hospital Cerebellar Ataxia Clinic, honorary consultant Neurologist at St Vincent’s Hospital and Lecturer at Melbourne University. David is lead investigator on research defining a novel ataxia – Cerebellar Ataxia with Neuropathy and Vestibular Areflexia Syndrome (CANVAS), a project to develop instrumented objective ataxia metrics, as well as the development of an objective oculomotor test of imbalance – the video VVOR.

J. Paul Taylor, MD, PhD – Dr. Taylor is an Investigator of the Howard Hughes Medical Institute, Director of Translational Neuroscience and Edward F. Barry Endowed Chair in Cell and Molecular Biology St. Jude Children’s Research Hospital. Dr. Taylor received his M.D. and Ph.D. degrees from Jefferson Medical College, trained as a resident in Neurology at the University of Pennsylvania and as a Fellow in Neurogenetics at NINDS/NIH where he trained with Kurt Fischbeck. Dr. Taylor’s research focuses on the molecular basis of inherited neurological disorders. Among Dr. Taylor’s contributions are the discovery of disease-causing mutations in RNA-binding proteins; the co-discovery that low complexity sequences in proteins – frequently found in RNA-binding proteins – mediate phase transitions that assemble membrane-less organelles such as RNA granules; and revealing mechanisms as to how disturbance of RNA granule dynamics contributes to diseases, such as ALS-FTD.

Leon Tejwani, MS – Leon Tejwani is a graduate student in Interdepartmental Neuroscience Program at Yale University. He received his BS and MS degrees from UC San Diego, where he worked in Dr. Alysson Muotri’s lab to develop new human stem cell models for various neurodevelopmental disorders. At Yale, Leon has worked with Dr. Janghoo Lim on understanding both the pathological mechanisms that underlie the spinocerebellar ataxias, as well as novel therapeutic approaches to several other neurodegenerative disorders. More specifically, Leon has identified a novel approach to increasing functional lysosome biogenesis, which can enhance clearance of toxic TDP-43 species and modulate pathological phenotypes in multiple mouse models of ALS/FTD. In his efforts to better understand spinocerebellar ataxia type 1 (SCA1), Leon has performed longitudinal single nucleus transcriptomic profile of the SCA1 knock-in mouse cerebellum, generating a comprehensive roadmap of the transcriptional dynamics of each cell type throughout the disease.
Biographies (continued)

**Sophie Tezenas du Montcel, MD, PhD** – Sophie Tezenas du Montcel is a public health doctor (Assistance Publique-Hôpitaux de Paris, Sorbonne University) with a PhD in genetic Epidemiology. She has an expertise in biostatistics and epidemiology. For the past 15 years, she has analyzed data concerning SCA patients and related disorders (Friedreich ataxia, Huntington disease). Since then she has developed collaborations locally and internationally. As part of these collaborations, she has developed a quantitative tool to evaluate the severity of the cerebellar syndrome, the CCFS (Cerebellar Composite Functional Score) and a semi-quantitative clinical scale for cerebellar assessments, SARA (Scale for the Assessment and Rating of Ataxia). She analyzes the EUROSCA natural history, a large multinational European cohort of SCA patients, and the genetic modifier study of the EUROSCA study. She also analyzes the RISCA cohort, a European cohort of SCA for at-risk subjects. As part of the READISCA study, granted by the NIH, she is in charge to design the best trial for SCA patients and to analyze the clinical data.

**Sokol V. Todi, PhD** – Sokol Todi and his laboratory have a long-standing interest in age-related neurodegeneration, with special emphasis on polyglutamine-dependent Spinocerebellar Ataxias. As a postdoctoral fellow, and later in his independent laboratory, Dr. Todi has investigated the biochemical and physiological properties of ataxia-causing proteins and has utilized the model organism, *Drosophila melanogaster*, as a sentinel of processes that underlie the biology of SCAs and to find protective pathways against them. Through various collaborations, Dr. Todi has also researched therapeutic solutions for non-ataxia disorders, including Parkinson’s and Alzheimer’s Diseases. Presently, Dr. Todi and his lab are expanding their work on SCAs and on the role that the wild-type versions of ataxia-related proteins have during development and in adults.

**Adam Vogel, PhD** – Adam Vogel, PhD is a behavioral scientist with training in speech and neuroscience. He is Professor at The University of Melbourne, Australia and a National Health & Medical Research Council (Australia) Fellow. Adam is Director of the Centre for Neuroscience of Speech where his team work on improving how we recognize, describe and treat communication and swallowing deficits in people with progressive neurological disorders including ataxias.

Adam is also Founder and Chief Science Officer of Redenlab Inc, a neuroscience technology company that uses speech and language biometrics to enhance decision making in clinical trials.
Biographies (continued)

R.J. Wessells, PhD – Wessells received his PhD from the Ohio State University and did postdoctoral fellowships at University of Michigan and the Sanford Burnham Prebys Medical Discovery Institute, where we worked to develop the Drosophila system as a model to study adult cardiac function. He returned to University of Michigan in 2005 to start his own lab, where he developed the first endurance exercise program in an invertebrate genetic model. Since moving to Wayne State in 2014, his lab has focused on identification of therapeutic mimetics that provide the benefit of exercise to sedentary animals. The lab has successfully identified two single-gene mimetics that mimic the adaptive changes induced by exercise when overexpressed in muscle. In addition, they have discovered a subset of neurons that are activated during exercise, and if stimulated genetically, can provide benefits of exercise to flies that are completely restrained from moving. These results have been expanded into humans, where the lab is engaged in an ongoing study to investigate the effects of virtual reality-simulated exercise on human cardiac function.

The Wessells lab has recently opened a second focus to examine the effects of exercise mediating genes as a treatment for mitochondrial diseases and ataxias using the Drosophila system. Specifically, PolyQ disease models in the fly are being used to test the efficacy of both chronic exercise and genetic and pharmacological exercise mimetics for slowing disease progression and preserving mobility and survival in affected animal models.
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# Meeting Attendees

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Biohaven is a proud sponsor of the AAC. Please visit the Biohaven booth during the conference to learn more about Biohaven’s ongoing ataxia research. Biohaven would like to express our gratitude to all clinical trial volunteers. Advances in the ataxia field would not be possible without your dedication. Our ataxia team continues to be inspired by the compassion and commitment of the entire ataxia community.
Connecting families, clinicians and researchers to further DRPLA research and work towards a treatment for DRPLA.

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For DRPLA patients and caregivers:
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Exicure, Inc. is a clinical stage biotechnology company developing a new class of spherical nucleic acids (SNA™).

Our proprietary SNA architecture is designed to unlock the potential of therapeutic oligonucleotides in a wide range of cells and tissues. Exicure is excited to utilize this platform in various ataxias and recently announced the launch of its first neurological program in Friedreich's Ataxia.

Founded in 2011, Exicure, Inc. is Nasdaq-listed and headquartered in Chicago, Illinois and Cambridge, Massachusetts.

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History of the Ataxia Investigators Meeting

In 2004, under the direction of Dr. John Day, the National Ataxia Foundation planned to host the first in a series of scientific meetings which would be called: Ataxia Investigators Meeting (AIM). The first of these meetings was held in conjunction with the 2005 Annual Membership Meeting in Tampa, FL. Following the 2005 AIM, meetings have been held biennially in 2008 in Las Vegas, 2010 in Chicago, 2012 in San Antonio, 2014 in Las Vegas, 2016 in Orlando, and 2018 in Philadelphia. These three-day conferences have brought leading ataxia investigators, both junior and senior, from around the world together to share both clinical and scientific research.

Planning is underway to partner with the Friedreich’s Ataxia Research Alliance (FARA) and Ataxia UK to facilitate one global ataxia meeting that will take place in Orlando, Florida on March 15-18, 2022.