

WELCOME TO THE
7th Ataxia
Investigators Meeting
(AIM 2018)



SPONSORED BY THE

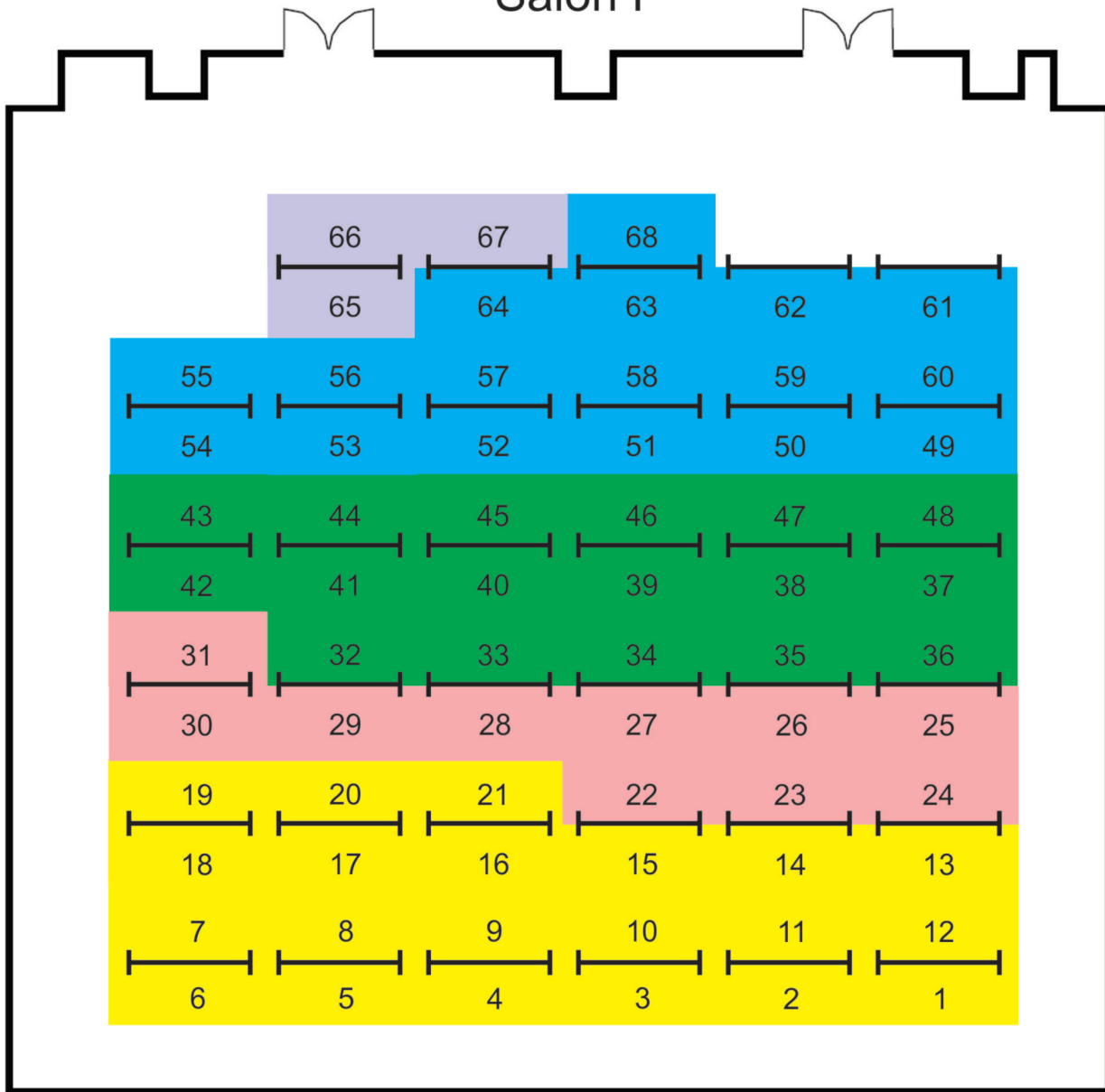
NAF **National Ataxia**
Foundation

April 2 – 5, 2018

Marriott Philadelphia Downtown
in Philadelphia, Pennsylvania

Poster Map:

Salon F



Theme 1

Theme 2

Theme 3

Theme 4

Theme 5

Table of Contents

Welcome	2
Thank You to Sponsors	3
Meeting Schedule	4-8
AIM Exhibitors	8
Patient/Family/Investigator Interaction Opportunities	9
Scientific Poster Sessions	10-15
Become a Professional Member of the NAF	15
2020 AIM Meeting Announcement	16
Biographies	17-33
Meeting Attendees	34-39
AIM Steering Committee	39
Notes	40
History of the AIM & Group Photos	Inside & Outside Back Cover

All AIM conference communications, materials and abstracts are confidential.

Wireless Internet Information

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

Network Name NAF

Password ataxia282942

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.





**NAF National Ataxia
Foundation**

Connecting Ataxia families, researchers,
clinicians and the community

April 2, 2018

Dear AIM 2018 Attendee,

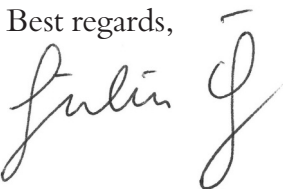
On behalf of the Organizing Committee and my Co-Chair Dr. Al La Spada, I would like to welcome you to the 7th Ataxia Investigators Meeting sponsored by the National Ataxia Foundation. AIM meetings have been essential to bring ataxia investigators from around the world together to exchange ideas and knowledge and move towards effective therapies. This year we continue this tradition, but also make a point of bringing experts from other neurodegenerative disease areas to learn from their experiences.

We have lectures from senior and junior investigators to recognize the brightest minds in ataxia research and follow a format with extended time for questions at the end of each talk. This format facilitated excellent discussion and exchange of ideas at the past two AIMs. This year we also added *Open Discussion* sessions to further facilitate brainstorming as a community after we have heard all presentations in each Theme. If you are a Trainee, we especially want to hear your questions and ideas during Q&A and open discussion. To encourage participation, we will give out raffle tickets for each question/comment from a Trainee for a surprise drawing at the end of AIM 2018! *Hot Chair* sessions that feature two-minute talks by Junior Investigators to advertise their posters were highly enjoyed by the attendees of AIM 2016, so we kept them in the Program. And to add to the fun, AIM 2018 features two *Debate* sessions this year where thought leaders in our field will address pros and cons of issues that the field is tackling; we hope that these debates will demonstrate that even ideas that are widely accepted should be challenged at times. Last but not least, please make the most out of the opportunities to interact with patients and their family members, at the *Poster Session for Patients and Families*, the Wednesday dinner and the *Birds of a Feather* sessions.

We strongly encourage you to complete the meeting survey – we take the feedback to heart and make changes in the planning of the meeting to accommodate the desires of the attendees at the next AIM.

I look forward to stimulating discussions and an engaging meeting as we move towards a new era of ataxia therapeutics.

Best regards,



Gülin Öz, PhD
AIM 2018 Chair

Thank You to Sponsors

The National Ataxia Foundation is grateful for the generous support of the 7th Ataxia Investigators Meeting.

- The Clementz Family Foundation
- National Institutes of Neurological Disorders and Stroke at the National Institutes of Health
- National Center for Advancing Translational Sciences/Office of Rare Diseases

Industry Sponsors

- Biogen Inc.
- Biohaven Pharmaceuticals
- Cadent Therapeutics
- Cydan II
- Ionis Pharmaceuticals, Inc.
- Reata Pharmaceuticals
- Steminent Biotherapeutics Inc
- Takeda Pharmaceuticals International, Inc.
- Ultragenyx

Foundation Sponsors

- A-T Children's Project
- Ataxia UK
- Bob Allison Ataxia Research Center (BAARC)
- Friedreich's Ataxia Research Alliance (FARA)



Ataxia UK



BOB ALLISON
ATAXIA
RESEARCH CENTER



The 7th Ataxia Investigators Meeting is supported by the National Institute Of Neurological Disorders And Stroke of the National Institutes of Health under Award Number R13NS105417. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

Meeting Schedule

MONDAY, APRIL 2, 2018

- 3:30–6:30 p.m. AIM Check-in and Poster Boards available to hang posters
5:30–6:30 p.m. Welcome Reception & Opening Remarks
Gülin Öz, PhD, University of Minnesota
6:30 p.m. Buffet Dinner at Marriott Hotel Salon G

TUESDAY, APRIL 3, 2018

- 7:30–8:30 a.m. Continental Breakfast in Salons I-L

Theme 1 – Basic cerebellar function and dysfunction

Session Chair: Marija Cvetanovic, PhD, University of Minnesota

- 8:30–9:05 a.m. Key Note: Chris de Zeeuw, MD, PhD, Erasmus Medical Center
Common Default Pathways in Ataxia
- 9:05–9:30 a.m. Wade Regehr, PhD, Harvard University
Purkinje cell synapses
- 9:30–9:55 a.m. Baljit Khakh, PhD, University of California, Los Angeles
Cells that tile your brain: astrocyte roles in neural circuits
- 9:55–10:15 a.m. Morning Break
- 10:15–10:40 a.m. Esther Becker, PhD, University of Oxford
Stem cell-derived models to elucidate the disease mechanisms in spinocerebellar ataxia
- 10:40–11:05 a.m. *Adam Avery, PhD, University of Minnesota
Molecular and cellular consequences of SCA5 mutations in β -III-spectrin
- 11:05–11:30 a.m. Ray Truant, PhD, McMaster University
Reactive Oxygen Stress and DNA Damage Define ATM as a Node in Age-Onset Neurodegenerative disease
- 11:30–11:50 p.m. Theme 1 Open Discussion led by Theme Chair
- 11:50–1:00 p.m. Lunch at Marriott in Salons I-L
- ### **Theme 2 – Therapeutic targets to correct neuronal dysfunction**
- Session Chair: Vikram Shakkottai, MD, PhD, University of Michigan*
- 1:00–1:35 p.m. Key Note: Keith W. Caldecott, PhD, University of Sussex
DNA strand breakage and human neurodegenerative disease
- 1:35–2:00 p.m. *Jill Napierala, PhD, University of Alabama at Birmingham
Activating mitochondrial aldehyde dehydrogenases to mitigate oxidative damage in Friedreich's ataxia



*Indicates a Junior Investigator

Meeting Schedule

TUESDAY, APRIL 3, 2018 (continued)

- 2:00–2:25 p.m. * Alanna Watt, PhD, McGill University
Understanding and reversing pathophysiological changes in a mouse model of SCA6
- 2:25–2:45 p.m. Afternoon Break
- 2:45–3:10 p.m. * Matthew Scaglione, PhD, Medical College of Wisconsin
Using a proteostatic outlier to interrogate the polyglutamine diseases
- 3:10–3:35 p.m. * Colleen Stoyas, PhD, Duke University
Sirt1 restores proper calcium homeostasis to achieve neuroprotection in spinocerebellar ataxia type 7
- 3:35–4:00 p.m. * Yalan Zhang, PhD, Yale University
Kv3.3 channels bind the survival protein Hax-1 and activate the TBK1 signaling pathway
- 4:00–4:20 p.m. Theme 2 Open Discussion led by Theme Chair
- 4:20–4:40 p.m. “Hot Chair” Junior Poster Presenters Group 1
(3 minutes each, 2 minute talk + 1 minute questions)
- Terri Driessen, PhD SCA1 Yale University
Lauren Moore, PhD Candidate SCA3 University of Michigan
Eunju Seong, PhD SCAR4 University of Michigan
Austin Ferro, BA SCA1 University of Minnesota
Vincent Francis, PhD ARSACS McGill University
Ha Eun Kong, BA FXTAS Emory University
- 4:40–5:00 p.m. Brief Break
- 5:00–6:30 p.m. Scientific Poster Session (Wine and Cheese)
- 6:30 p.m. Networking Dinner on your own

WEDNESDAY, APRIL 4, 2018

- 7:30–8:30 a.m. Continental Breakfast in Salons I-L
- Theme 3 – Translational research and disease models**
Session Chair: Patricia Maciel, PhD, University of Minho
- 8:30–9:05 a.m. Key Note: Huda Zoghbi, MD, Baylor College of Medicine
Lessons we continue to learn from studying SCA1
- 9:05–9:30 a.m. * Maria do Carmo Costa, PhD, University of Michigan
Drug and target discovery for Spinocerebellar ataxia type 3

Meeting Schedule

WEDNESDAY, APRIL 4, 2018 (continued)

- 9:30–9:55 a.m. Lisa Ellerby, PhD, Buck Institute for Age Research
Disease Modeling and Therapeutic Targets in Huntington's Disease
- 9:55–10:15 a.m. Theme 3 Open Discussion led by Theme Chair
- 10:15–10:35 a.m. “Hot Chair” Junior Poster Presenters Group 2
(3 minutes each, 2 minute talk + 1 minute questions)
- Chandrakanth Edamakanti, PhD SCA1 Northwestern University
- Luis Pereira de Almeida, PhD SCA3 University of Coimbra
- Vincenzo Gennarino, PhD PUM1 Columbia University
- Amy Salovin, MS FRDA Children’s Hospital
of Philadelphia
- Jorge da Silva, PhD Candidate SCA3 University of Minho
- Francesca Tiano, PhD Candidate FRDA University of Rome
“Tor Vergata”
- 10:40 a.m. Official Photo of all AIM Attendees will be taken at this time
- 10:45 a.m. – noon Scientific Poster Session (Coffee, Tea, Snack)
- 12:00–1:30 p.m. Networking Lunch - Offsite
- Theme 4 – Enhancing Clinical Trial Readiness**
- Session Chair: George Wilmut, MD, PhD, Emory University, Atlanta, GA*
- 1:30–2:05 p.m. Key Note: Frank Bennett, CSO, Ionis Pharmaceuticals
Antisense based drugs for the treatment of triplet repeat diseases
- 2:05–2:30 p.m. * Hayley McLoughlin, PhD, University of Michigan (Theme 3)
*Antisense Oligonucleotides provide therapeutic benefit in Spinocerebellar ataxia
type 3 mice*
- 2:30–2:55 p.m. * Ian Harding, PhD, Monash University
*The ENIGMA-Ataxia Global Neuroimaging Research Consortium:
Big Data meets Rare Disorders*
- 2:55–3:20 p.m. Amy Bastian, PhD, PT, Johns Hopkins University
Motor Learning and Rehabilitation for Ataxia
- 3:20–3:30 p.m. Afternoon Break
- 3:30–3:55 p.m. *Pierre-Gilles Henry, PhD, University of Minnesota
*Longitudinal MRS, MRI and DTI in the Spinal Cord in Friedreich’s Ataxia:
24-month follow-up*

Meeting Schedule

WEDNESDAY, APRIL 4, 2018 (continued)

- 3:55-4:20 p.m. Dobrila D. Rudnicki, PhD, NCATS
Catalyzing the Translation of Basic Science Discoveries into the Clinic
- 4:20-4:45 p.m. Debate: "Should treatments be shown to be effective in animal disease models prior to human trials?"
Harry Orr, PhD, University of Minnesota vs. Stefan Pulst, MD, University of Utah
- 4:45-5:05 p.m. Theme 4 Open Discussion led by Theme Chair
- 5:05-5:15 p.m. Albert La Spada, MD, PhD, FACMG, Duke University
Vera Cruz, Mexico – SCA 7 Research Trip
- 5:15-6:45 p.m. Poster Session for Patients and Families
- 6:45 p.m. Posters must be taken down
- 6:45 p.m. Dinner at the Marriott in Salons I-L
- 7:30 p.m. The Patient/Family Perspective
Bill Sweeney, NAF Board President
Michael, Karen and Jennifer Leader
Linda Snider, MD and Kyle Bryant

THURSDAY, APRIL 5, 2018

- 7:30-8:30 a.m. Continental Breakfast in Salons I-L
- Theme 5 – Ataxia Trials: What can we learn from trials in other rare diseases?**
Session Chair: Jeremy Schmahmann, MD, Harvard, Boston, MA
- 8:30-9:05 a.m. Key Note: John Day, MD, PhD, Stanford University
Treating SMA with Gene Modification and Gene Replacement
- 9:05-9:30 a.m. James D. Berry, MD, MPH, Harvard University
Coordinating Ataxia Research: Lessons from the Northeast ALS Consortium
- 9:30-9:55 a.m. Ludy Shih, MD, MMSc, Biogen, Inc.
Focus on biomarkers: preparing for clinical trials in the spinocerebellar ataxias
- 9:55-10:15 a.m. Morning Break
- 10:15-10:40 a.m. *Manuela Corti, PT, PhD, University of Florida
Gene Therapy for Friedreich's Ataxia
- 10:40-11:05 a.m. Robert Berman, MD, Biohaven Pharmaceuticals
Trigriluzole: A Phase 2/3 Randomized Controlled Trial in Patients with Spinocerebellar Ataxia

Meeting Schedule

THURSDAY, APRIL 5, 2018 (continued)

- 11:05-11:30 a.m. Debate: "Catching the horse in the barn – Should treatment trials begin before ataxia symptoms develop?"
*Thomas Klockgether, MD, University of Bonn vs.
George Wilmot, MD, PhD, Emory University*
- 11:30-11:50 a.m. Wrap-up of entire AIM
Summary by chairs Gülin Öz and Al La Spada, followed by open discussion and brainstorming
- 11:50-12:00 p.m. Closing Remarks
Albert La Spada, MD, PhD, FACMG, Duke University

AIM Exhibitors

APDM Wearable Technologies is a digital health company focused on developing and commercializing digital endpoints for neuroscience and balance disorders. APDM has raised \$13MM+ from the NIH, has over 200+ publications, thousands of researchers worldwide using our technology, and are currently involved in five active clinical trials. We are currently focused on creating an instrumented version of the SARA protocol. Matthew Johnson, General Manager is staffing the exhibit table.

MNG Laboratories is an internationally recognized clinical diagnostic leader specializing in neurogenetic and complex biochemical testing. Our portfolio includes movement disorders, epilepsy, muscular dystrophies, intellectual disabilities, metabolic and other inherited disorders.

Our extensive offer of next generation sequencing panels, proprietary Genome MaNaGer™ database and our Neurogenetic Answers™ reporting platform is recognized by genetic experts around the world. We are proud to offer first-in-class reporting and fast turnaround times.

With over 15 years of neurogenetic experience, and powered by a culture of discovery and advancement, MNG Laboratories delivers results that make a difference for patients and their families. MNG Laboratories offers CLIA and CAP certified reports and adheres to current ACMG guidelines and recommendations.



Patient/Family/Investigator Interaction Opportunities

Linking Ataxia Families and Investigators

The location of the 2018 Ataxia Investigators Meeting was selected so that it dovetails with the 61st Annual Ataxia Conference of the National Ataxia Foundation. 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. Important effects are junior and senior investigators will see that their research in the lab and/or clinic makes a significant impact to the ataxia community. In addition, it is an opportunity for scientists to communicate with the ataxia community and explain their ataxia research initiatives, which is invigorating and hopeful for patients and families.

In addition to informal conversations that may take place throughout the meeting, there are three dedicated opportunities to interact with persons affected by ataxia and their family members and caregivers.

Patient and Family Poster Session

On Wednesday, April 4, from 5:15-6:45 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. This session should be attended by poster presenters only to allow room for wheel chairs and walkers in the poster session room. Please bring a chair to your poster so that you may sit down when speaking with a person who uses a wheel chair.

Wednesday AIM Dinner

On Wednesday, April 4, at the AIM dinner, four speakers from the ataxia community will share their stories and challenges of living with ataxia. From the surveys taken after the 2016 AIM, many AIM attendees stated that this was the best part of the meeting. Don't miss this!

“Birds of a Feather” Small Group Sessions

After the AIM 2018 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 interest to you.

Below is the listing of groups meeting on Thursday, April 5 from 2:00-5:00 p.m. and their locations:

- SCA 1 Franklin 4
- SCA 2 Meeting Room 405
- SCA 3 Franklin 2
- SCA 6 Franklin 1
- SCA 7 Meeting Room 406
- All other SCAs (inc. SCA 5 Franklin 3
and SCA 8) & DRPLA
- AOA Meeting Room 404
- Spouses and Partners without ataxia Franklin 13



Scientific Poster Sessions

Poster Numbering

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

Session Times

- Tuesday Scientific Poster Session is from 5:00 to 6:30 p.m.
- Wednesday Scientific Poster Session is from 10:45 a.m. to noon
- Wednesday Patient/Family Poster Session is from 5:15 to 6:45 p.m.
- Posters must be taken down at 6:45 p.m. on Wednesday

Tuesday Scientific Poster Sessions (5:00 – 6:30 p.m.)

- 1_Theme 1_Tue Alaimo, Daniele for Marton MNG Laboratories
Combining repeat expansion testing with NGS phenotype-based panels provides significant diagnostic benefit
- 3_Theme 1_Tue Chen, Dong-Hui, MD, PhD University of Washington
Neurologic manifestations in SAMD9L-related Ataxia-Pancytopenia syndrome
- 5_Theme 1_Tue *Driessen, Terri, PhD Yale University
Transcriptomics Approaches to Understand Tissue Vulnerability in Mouse Models of SCA1
- 7_Theme 1_Tue *Francis, Vincent Gerard, PhD McGill University
Sacsin the protein product of the gene mutated in the recessive ataxia ARSACS regulates organelle positioning
- 9_Theme 1_Tue Issa, Fadi, PhD East Carolina University
Cellular Mechanisms Underlying Pathogenesis of Spinocerebellar Ataxia Type 13
- 11_Theme 1_Tue *Moore, Lauren, PhD Candidate University of Michigan
Autophagic dysfunction may coincide with activated nucleophagy in cellular and mouse models of SCA3
- 13_Theme 1_Tue Mu, Weiyi, ScM, CGC Johns Hopkins University
Diagnostic yield of a comprehensive genetic testing algorithm for ataxia
- 15_Theme 1_Tue Nath, Siddharth, BSc McMaster University
Using a novel spinocerebellar ataxia variant to probe the mechanisms underlying pathology in CAG triplet repeat disorders
- 17_Theme 1_Tue Ranum, Laura, PhD University of Florida
SCA8 RAN polySer protein preferentially accumulates in white matter brain regions and is regulated by eIF3F
- 19_Theme 1_Tue *Seong, Eunju, PhD University of Michigan
Mutations in VPS13D lead to ataxia with spasticity and mitochondrial defects
- 21_Theme 1_Tue Suart, Celest, BHSc McMaster University
Ataxin-1 localizes to DNA damage in an ATM dependent manner



*“Hot Chair” poster presenters

Scientific Poster Sessions

Tuesday Scientific Poster Sessions (continued)

- 23_Theme 2_Tue..... Dong, Yina, PhD Children's Hospital of Philadelphia
Targeting GRP75 as a potential therapy for Friedreich's Ataxia
- 25_Theme 2_Tue*Ferro, Austin, BA..... University of Minnesota
Biphasic function of Bergmann glia in Spinocerebellar ataxia type 1
- 27_Theme 2_Tue..... Lin, Hong, PhD Children's Hospital of Philadelphia
Impaired cerebellar endoplasmic reticulum (ER)-mitochondria contacts and signaling in the KIKO mouse model of Friedreich ataxia
- 29_Theme 2_Tue.....Manek, Rachna H, PhD Student University of Florida
The 5'UTR of ATXN1 is alternatively spliced and post-transcriptionally regulates Ataxin-1 expression
- 31_Theme 2_Tue Zhang, Miao, PhD Chapman University
A Mutant SK2 Channel Hypersensitive to Ca²⁺
- 32_Theme 3_TueNeves-Carvalho, Andreia, PhD for SilvaUniversity of Minho
Neuroprotective effects of creatine in the CMVMJD135 mouse model of spinocerebellar ataxia type 3
- 35_Theme 3_Tue Delatycki, Martin, MD, PhD Murdoch Children's
Keeping the black dog at bay: understanding depression in Friedreich ataxia Research Institute
- 37_Theme 3_TueGennarino, Vincenzo A. PhDColumbia University
A mild PUM1 mutation is associated with adult-onset ataxia, whereas haploinsufficiency causes developmental delay and seizures
- 39_Theme 3_Tue Kuo, Sheng Han, MD Columbia University
Climbing Fiber Synaptic Organization in Tremor and Ataxia: A Novel Mouse Model
- 41_Theme 3_TueMaciel, Patricia, PhD for Campos University of Minho
The neuroprotective effect of Hyptis suaveolens, Hyptis pectinata and Hyptis marrubioides in Caenorhabditis elegans model of SCA3/MJD
- 43_Theme 3_Tue.....*Pereira de Almeida, Luís, PhD..... University of Coimbra
Gene-editing for treatment of Machado-Joseph disease
- 45_Theme 3_Tue*Salovin, Amy, MS Children's Hospital of Philadelphia
Characterization of cardiac phenotype in the KIKO mouse model of FRDA
- 47_Theme 3_TueTodi, Sokol, PhD for Sutton, Joanna PhD.....Wayne State University
Toxicity and aggregation of the polyglutamine disease protein, ataxin-3, is regulated by its binding to VCP/p97 in Drosophila melanogaster

Scientific Poster Sessions

Tuesday Scientific Poster Sessions (continued)

- 49_Theme 4_TueAshizawa, Tetsuo, MDHouston Methodist
Clinical Trial Readiness for SCA1 and SCA3 Research Institute
- 51_Theme 4_Tue..... Cahn, Suzy, BS Emory University
Spinocerebellar Ataxia (SCA) Patients' Perceptions Regarding Reproductive Options
- 53_Theme 4_Tue.....Gupta, Anoopum S., MD, PhDMassachusetts General Hospital
Machine prediction of ataxia severity and class from a simple mouse-based computer task
- 57_Theme 4_Tue Kaas, Bonnie, MD Johns Hopkins University
Antibody Testing and Detection in Ataxia
- 59_Theme 4_Tue O'Keefe, Joanne, PhD..... Rush University
Dual-task cognitive motor interference and fast paced walking exacerbates gait deficits in Fragile X-Associated Tremor/Ataxia Syndrome (FXTAS)
- 61_Theme 4_TueTiano, Francesca, PhD Student University of Rome
*Frataxin deficiency in Friedreich's Ataxia is associated with reduced "Tor Vergata"
levels of HAX-1, a regulator of cardiomyocyte death and survival.*
- 63_Theme 4_Tue Shih, Ludy, MD, MMSc Biogen, Inc.
Focus on biomarkers: preparing for clinical trials in the spinocerebellar ataxias
- 64_Theme 4_TueVogel, Adam P., PhD.....University of Melbourne
Motor speech and swallowing phenotype of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)
- 66_Theme 5_Tue Hall, Deborah A, MD PhD Rush University
Open-label pilot trial of citicoline for fragile X-associated tremor/ataxia syndrome (FXTAS)
- 68_Theme 4_Tue Szmulewicz, David, MD Florey Institute
Cerebellar Ataxia with Neuropathy and Vestibular Areflexia Syndrome (CANVAS), a novel vestibulo-cerebellar ataxia: clinical phenotype, pathology, imaging abnormalities, differential diagnoses and a quantitative bedside test.

Scientific Poster Sessions

Wednesday Scientific Poster Sessions (10:45 a.m. – noon)

- 2_Theme 1_Wed Ashizawa, Tetsuo MD..... Houston Methodist
Internal sequences of large pentanucleotide repeat expansion alleles in SCA10 Research Institute
- 4_Theme 1_Wed Coarelli, Giulia, MD ICM
Motor neuron involvement threatens survival in spinocerebellar ataxia type 1
- 6_Theme 1_Wed Fogel, Brent, MD, PhD for Valera University of California –
Prevalence of spinocerebellar ataxia 36 in a US population Los Angeles
- 8_Theme 1_Wed Guell, Xavier, MD MIT and Harvard
New developments in functional neuroimaging of the cerebellum: macroscale gradients of organization and triple representation of cognitive/affective task processing
- 10_Theme 1_Wed Lorenzo, Damaris N., PhD University of NC at Chapel Hill
Regulation of cerebellar development and connectivity by β -spectrins: implications for spinocerebellar ataxias
- 12_Theme 1_Wed Morgan, Owen, BA Johns Hopkins University
Motor-cognitive Multitasking in Cerebellar Ataxia
- 14_Theme 1_Wed Nakamura, Katsuya, MD, PhD University of Florida
Identification of novel spectrin, beta, non-erythrocytic 2 (SPTBN2) variants in a large cohort of ataxia patients
- 16_Theme 1_Wed Paucar-Arce, Martin, MD, PhD Karolinska Institutet
Ataxia and cerebellar atrophy in Charcot-Marie-Tooth type 4C
- 20_Theme 1_Wed Slapik, Mitchell, BA Johns Hopkins University
A Characterization of Language Impairment in Cerebellar Ataxia
- 22_Theme 2_Wed Bushart, David, Graduate Student University of Michigan
Targeting potassium channels to treat cerebellar ataxia
- 24_Theme 2_Wed *Edamakanti, Chandrakanth, PhD Northwestern University
Mutant Ataxin1 alters cerebellar stem cell behavior and neuronal connectivity in Spinocerebellar ataxia type 1
- 26_Theme 2_Wed *Kong, Ha Eun, BA Emory University
Genetic modifiers of FXTAS CGG toxicity identified through combining metabolic profiling of FXTAS mice with Drosophila genetics
- 28_Theme 2_Wed Maciel, Patricia, PhD for Castro University of Minho
Preclinical evidence supporting early initiation of citalopram treatment in Machado-Joseph disease: findings in two mouse models
- 30_Theme 2_Wed Wilson, Robert, MD, PhD Children’s Hospital
Identification of p38 MAPK as a novel therapeutic target for Friedreich ataxia of Philadelphia

Scientific Poster Sessions

Wednesday Scientific Poster Sessions (continued)

- 33_Theme 3_Wed..... Clark, Elisia, PhD..... University of Pennsylvania
Molecular Mechanisms Underlying the Atypical Mild Phenotype in Friedreich's ataxia Patients with Missense Mutations
- 34_Theme 3_Wed..... Costa, Maria do Carmo, PhD..... University of Michigan
Transgenic mouse models of Machado-Joseph disease show cerebellar neurochemical profiles similar to those of patients
- 36_Theme 3_Wed Delatycki, Martin, MD, PhD Murdoch Children's
Sexual Function, Intimate Relationships and Friedreich Ataxia Research Institute
- 38_Theme 3_Wed..... Keaney, Gregg, PhD Cadent Therapeutics
CAD-1883, a clinical-stage positive allosteric modulator of the small conductance calcium-activated potassium channel (SK channel): effects on cerebellar Purkinje neuron firing, ataxic gait, and tremor in animal models
- 40_Theme 3_Wed Larson, Sarah N., MSc University of Minnesota
Effects of Antisense Oligonucleotide (ASO) Therapy on Neurochemistry in the Atxn1154Q/2Q Mouse Measured with Proton Magnetic Resonance Spectroscopy
- 42_Theme 3_Wed Neves-Carvalho, Andreia, PhDUniversity of Minho
Testing the Therapeutic Potential of Mesenchymal Stem Cells and Their Secretome in an Animal Model of Spinocerebellar Ataxia Type 3
- 44_Theme 3_Wed Pohl, Franziska, PhD Candidate Robert Gordon University
Utilization of Rapeseed Pomace (RSP) Extracts in the Prevention of Neurological Impairment in a C. elegans Model of Machado-Joseph Disease (MJD)
- 46_Theme 3_Wed..... da Silva, Jorge Diogo, PhD Candidate..... University of Minho
Genetic and pharmacological modifiers of ATXN3 proteotoxicity converge to antioxidant response pathways
- 48_Theme 3_Wed Tsou, Wei-Ling, PhD.....Wayne State University
Molecular chaperones and their efficacy in protecting against polyglutamine degeneration
- 50_Theme 4_WedBolzan, Gabriela, MDUniversidade Federal
Causal factors behind early- and late-onset Machado-Joseph Disease Do Rio Grande Do Sul
patients do not interfere with the rate of Neurological Deterioration
- 52_Theme 4_Wed..... El-Gohary, Mahmoud, PhD..... APDM, Inc.
Objective Measures of Ataxic Gait Using Wearable Inertial Sensors
- 54_Theme 4_Wed.....Gomez, Christopher M., MD, PhDUniversity of Chicago
Postural Sway from Inertial Sensors for Quantitative Balance Measures in Ataxia

Scientific Poster Sessions

Wednesday Scientific Poster Sessions (continued)

- 55_Theme 4_Tue..... Hassan, Anhar, MB. BCh, FRACP Mayo Clinic - Rochester
Case series of CANVAS (Cerebellar Ataxia with Neuropathy and bilateral Vestibular Areflexia Syndrome)
- 56_Theme 4_Wed..... Huryn, Laryssa, MD National Eye Institute at NIH
The Natural History of Spinocerebellar Ataxia Type 7
- 58_Theme 4_Wed..... Lenglet, Christophe, PhD University of Minnesota
Longitudinal Brain Diffusion MRI in Friedreich's Ataxia: 24-month Follow-up
- 60_Theme 4_Wed Stephen, Christopher D., MD Massachusetts General Hospital
Oculomotor abnormalities are ubiquitous in the spinocerebellar ataxias
- 62_Theme 4_WedVittal, Padmaja, MD, MS..... Northwestern Medicine
Antisense FMR1 splice variant: a predictor of fragile X-associated tremor/ataxia syndrome
- 65_Theme 5_Wed..... Blair, Ian A., PhD..... University of Pennsylvania
Low apolipoprotein A-I levels in Friedreich's ataxia: effect of statins
- 67_Theme 5_Wed.....Vogel, Adam P., PhD University of Melbourne
Speech Rehabilitation in hereditary ataxias

Wednesday Patient/Family Poster Session is from 5:15 to 6:45 p.m.

Posters must be taken down at 6:45 p.m. on Wednesday.

Become a Professional Member of the National Ataxia Foundation

As a researcher who has an interest in ataxia, we invite you to become a part of an organization that is providing current and reliable information about all forms of ataxia and funding cutting-edge world-wide research to bring new knowledge of the genetics and disease mechanisms of ataxia as well as medical interventions. As a Professional Member you will be included with other scientists and researchers who have made a commitment to researching neurological and genetic diseases and take an active part in furthering a cause that directly affects your career. Professional Membership gives you unique access to the Foundation's services and information.

Become a professional member now for a membership donation of \$65 per year. Please go to our website at www.ataxia.org or scan this QR code to become a member and support the research efforts of the National Ataxia Foundation.



National Ataxia Foundation

8th Ataxia Investigators Meeting

March 3-6, 2020



Sheraton Denver Downtown
Denver, Colorado



NAF
**National Ataxia
Foundation**

Connecting Ataxia families, researchers,
clinicians and the community



The National Ataxia Foundation would like to thank Dr. Albert La Spada who will chair the 8th Ataxia Investigators Meeting. For information on abstract submission, AIM registration and sponsorship opportunities, contact Sue Hagen at susan@ataxia.org

Biographies



Adam W. Avery, PhD – As a postdoctoral fellow, I have developed a strong interest in cytoskeletal mechanisms that underlie neuronal morphogenesis and maintenance, and the molecular mechanisms by which disease mutations disrupt the cytoskeleton to cause neurodegeneration. In my post-doctoral position in Dr. Thomas Hays' lab at the University of Minnesota, in the Department of Genetics, Cell Biology and Development, I have acquired expertise in live imaging and genetic approaches in *Drosophila*. I have effectively combined my training in *Drosophila*, with my undergraduate and graduate training in biochemistry, to demonstrate that the spectrin-actin cytoskeleton, underlying the plasma membrane, is required in dendrites to support branch stability and dendritic arbor outgrowth. My work has led to a model for how a human SCA5 disease mutation in β -III-spectrin disrupts plasticity of the spectrin-actin cytoskeleton and leads to a loss of arborization. I have significant experience in the biotechnology industry and am currently pursuing cell-based assays to identify small molecules that modulate the affinity of spectrin-actin linkages. My long-term goal is to establish my own lab in academia to pursue these interests independently.



Amy Bastian, PhD, PT – Dr. Amy Bastian is a neuroscientist and physical therapist who studies the neural control of human movement. She is Chief Science Officer of the Kennedy Krieger Institute and the Director of the Motion Analysis Laboratory. Dr. Bastian holds the rank of Professor of Neuroscience at Johns Hopkins, with a joint appointment in Neurology. Dr. Bastian studies how people with and without neurological damage control movement and learn new patterns. Her laboratory uses computerized movement tracking techniques, non-invasive brain stimulation, novel devices and robotics to control walking and reaching movements. A major focus of her work has been on the role of the cerebellum in moving, sensing and learning. She also focuses on locomotor control and plasticity in adults, children, and people with neurological diseases.



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 interests include the genetic and molecular underpinnings of cerebellar ataxia with a particular focus on the role of the mGluR1-TRPC3 pathway. Her group has recently developed a robust and reproducible protocol to generate cerebellar neurons from human induced pluripotent stem cells.

Biographies (continued)



C. Frank Bennett, PhD – Dr. Bennett is Senior Vice President of Research at Ionis Pharmaceuticals. He is responsible for preclinical antisense drug discovery and antisense technology research. Dr. Bennett is also the franchise leader for the Neurological Programs at Isis. He is one of the founding members of the Company. Dr. Bennett has been involved in the development of antisense oligonucleotides as therapeutic agents, including research on the application of oligonucleotides for inflammatory, neurodegenerative diseases and cancer, oligonucleotide delivery, pharmacokinetics and medicinal chemistry. Dr. Bennett has published more than 175 papers in the field of antisense research and development and has more than 150 issued U.S. patents.

Prior to joining Isis, Dr. Bennett was Associate Senior Investigator in the Department of Molecular Pharmacology at SmithKline and French Laboratories, GlaxoSmithKline (currently, GlaxoSmithKline).



Robert Berman, MD – Robert Berman M.D. is the Chief Medical Officer and co-founder of Biohaven Pharmaceuticals (New Haven, CT), a clinical-stage biopharmaceutical company with a portfolio of innovative, late-stage product candidates targeting neurological diseases. Biohaven is the first pharmaceutical company to advance a compound into late stage registrational trials for the treatment of Spinocerebellar Ataxia. Dr. Berman has over 30 years of experience in neuroscience research, both within academia (Yale School of Medicine) and industry (Pfizer, Bristol-Myers Squibb). He has over 70 peer-reviewed publications and is an Adjunct Professor at the Yale University School of Medicine, Department of Psychiatry.



James Berry, MD, MPH – Dr. Berry is a clinician caring for people with Amyotrophic Lateral Sclerosis (ALS), and clinical researcher with a focus on understanding and developing new therapies for ALS. He is co-director of the Massachusetts General Hospital (MGH) Multidisciplinary Amyotrophic Lateral Sclerosis (ALS) Clinic. He has worked to extend the reach of the MGH clinic team beyond the walls of the physical clinic using novel tools such as video televisits and remote monitoring.

Dr. Berry also works as a research investigator carrying out ALS trials at MGH and as overall investigator of numerous multicenter ALS biomarker studies and trials. He conducts ALS biomarker work focused on the identification of markers of ALS in blood and spinal fluid, with a particular emphasis on markers of abnormal inflammation. He collaborates broadly with researchers around the globe on these biomarker efforts.

Dr. Berry is also the Associate Medical Director of the MGH Neurological Clinical Research Institute (NCRI), and Director of the Partners Neurodegenerative Clinical Research Fellowship. He is a member of the Massachusetts ALS Registry team and CDC/ATSDR National ALS Registry and Biorepository Expert Panels. In addition, he is on the Executive Committee of the NEALS Consortium, a national organization for ALS clinical researchers, where he also leads the NEALS Biorepository and Technology in ALS subcommittee.

Biographies (continued)



Keith Caldecott, PhD – Keith Caldecott obtained his BSc (Hons) at Sheffield University (1987) and developed an interest in how cells repair DNA strand breaks during his PhD in the laboratory of Penny Jeggo, at the Medical Research Council's National Institute for Medical Research (London). Keith's interest in DNA repair continued during postdoctoral fellowships with Larry Thompson (California) and Tomas Lindahl FRS (London), and in 1995 Keith established his own laboratory at the University of Manchester. In 2002 the lab moved to the Genome Damage and Stability Centre (GDSC); a multi-disciplinary centre-of-excellence funded jointly by the MRC and the University of Sussex. Research highlights include the identification of new DNA strand break repair proteins (Whitehouse et al, Cell 2001; Loizou et al, Cell, 2004; Cortes-Ledesma et al, Nature 2009; Rulten et al, Mol Cell, 2011), and that specific hereditary neurodegenerative diseases are associated with cellular defects in DNA strand break repair (El Khamisy et al, Nature, 2005; Shen et al, Nature Genetics 2010; Gomes-Herrerros, Nature Genetics 2014). In 2010 Keith was elected a member of EMBO, in 2012 was elected a member of the Academy of Medical Sciences (FMedSci), and in 2016 was awarded a Rotal Society Wolfson Research Merit Award.



Andreia Castro, PhD – Andreia Castro is a post-doctoral researcher in the laboratory of Professor Patrícia Maciel at School of Medicine, University of Minho in cooperation with Professor Rick Morimoto from Northwestern University. She holds a B.Sc. in Biochemistry from University of Porto, Portugal, received a Msc. in Molecular Genetics and a PhD in Health Sciences from University of Minho, Portugal. Her main research interest is to study the imbalance of protein homeostasis (proteostasis) associated with neurodegenerative diseases and to understand how proteostasis adaptation and/or enhancement may be beneficial to age-related disorders and constitute important therapeutic approaches. During her graduated studies, Andreia has established a *C. elegans* model for the study of MJD/SCA3 pathogenesis that shows protein aggregation and nervous system dysfunction. She identified a number of modifier genes, namely aging-related genes that stalled disease progression, and validated the model as a useful tool for drug screening protocols. Using pharmacogenetics, she identified compounds and cellular targets with therapeutic potential for MJD. Study of the mode-of-action of the promising drugs and how they impact on neuronal cells to sustain a balanced proteome is her current focus.



Manuela Corti, PT, PhD – Dr. Manuela Corti is currently an Assistant Professor at the University of Florida in the Child Health Research Institute and Powell Gene Therapy Center. Dr. Corti is a clinical scientist engaged in translational research focusing on understanding the contribution of neurological impairment in neuromuscular disorders by combining expertise in clinical assessment with novel therapies that rely on correcting the fundamental genetic defect. Her specific research is dedicated at developing AAV gene therapy programs for neuromuscular diseases and immunomodulation strategies to allow for multiple AAV dosing. Her research interests also include outcome measures and clinical trial readiness for neuromuscular diseases.

Biographies (continued)



Maria do Carmo Costa, PhD – Maria do Carmo Costa, Ph.D. is a research assistant professor in the Department of Neurology at Michigan Medicine, University of Michigan.

Carmo received her undergraduate degree in Biochemistry from the University of Porto (2000) and her Ph.D. in Health Sciences from the University of Minho (2008) in Portugal.

Carmo started her research career in 1998 in the laboratory of Patrícia Maciel, Ph.D., and Jorge Sequeiros, M.D., Ph.D., at UnIGENE/ Institute of Molecular and Cellular Biology (IBMC), University of Porto, while conducting her undergraduate thesis on the improvement of molecular diagnosis of Huntington's disease (HD) and of Spinocerebellar ataxia type 3 (SCA3). She continued working as a research assistant at IBMC until 2003, being responsible for the genetic testing of Huntington's disease, participating in several molecular genetic studies of SCA3, HD and related disorders, and in the generation and characterization of *C. elegans* and mouse models of SCA3.

For her Ph.D. thesis, at the University of Minho and under continued guidance of Dr. Patrícia Maciel, Carmo focused on studying the *ATXN3* mouse homologue gene and its protein contributing for a better understanding of its biological function. She also generated and characterized a SCA3 transgenic mouse model that replicates aspects of the human disease.

Given her long interest in SCA3, Carmo joined the laboratory of Henry Paulson, M.D., Ph.D., at the Department of Neurology, University of Michigan, in 2008 as a post-doctoral fellow where she explored pharmacological and RNA interference-mediated strategies to reduce levels of mutant *ATXN3* protein in the brain that can be translated for SCA3 patients.

Carmo joined the faculty of the Department of Neurology in 2013. Using biochemistry, molecular and cellular biology, high-throughput small-molecule and genetic screens, and mouse models, her research currently focuses on drug and target discovery for SCA3 and molecular mechanisms of neurodegeneration.



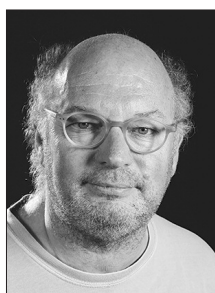
Marija Cvetanovic, PhD – Dr. Marija Cvetanovic received her BSc in Molecular Biology and Physiology from the University of Belgrade in 1998. She completed her PhD at the Department of Immunology, Microbiology and Virology at the University of Illinois in Chicago in 2004, studying recognition of apoptotic 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.

Biographies (continued)



John Day, MD, PhD – John Day is Professor of Neurology and Pediatrics, and Director of the Division of Neuromuscular Medicine at Stanford University. Prior to moving to Stanford in 2011, Dr. Day was Professor of Neurology and Pediatrics, and Director of the Paul and Sheila Wellstone Muscular Dystrophy Center at the University of Minnesota. In Minnesota Dr. Day helped in the identification and characterization of myotonic dystrophy type 2, spinocerebellar ataxia type 5 and spinocerebellar ataxia type 8. As Director of the MDA clinic at both Stanford and the University of Minnesota, Dr. Day has cared for patients and families affected by Friedreich's ataxia as well as other neuromuscular and ataxic disorders for many years. To help develop treatments for neurodegenerative diseases, Dr. Day has spearheaded clinical trials of antisense oligonucleotides for myotonic dystrophy, spinal muscular atrophy and C9Orf72 ALS at Stanford, as well as gene replacement trials for SMA.



Chris I. De Zeeuw, MD PhD – Chair of Department of Neuroscience, Erasmus MC, Rotterdam and vice-director of the Netherlands Institute for Neuroscience of the Royal Dutch Academy of Sciences (KNAW), Amsterdam.

De Zeeuw is focusing on the question how sensorimotor skills can be controlled at high spatiotemporal resolution by the cerebellum, bridging insights obtained at the molecular and circuitry level in mouse mutants up to the psychophysical level in healthy humans and patients. In contrast to the prevailing view that cerebellar learning is controlled by synaptic depression, in his pioneering work he showed that in fact potentiation and suppression can work synergistically, that the dominating mechanisms are module-specific adjusted to the sensorimotor behavior involved, and that deviations in this distributed synergy as well as in synchrony of cerebellar activity can lead to diseases like autism and ataxia. Rather than claiming that the olivocerebellar system controls either timing or learning of movements, he was one of the first to show that it can do both simultaneously. He received personally from Her Majesty, Queen Beatrix, the so-called Beatrix Award, which was given to the Dutch scientist with the best performance in the field of neuroscience and movement control over the 50-year period from 1956 to 2006; in 2012, he received an ERC-advanced grant on neuronal encoding mechanisms; and in 2014 he was elected as an Academy Member (KNAW).



Terri M. Driessen, PhD – Terri M. Driessen is a postdoctoral researcher in the laboratory of Dr. Janghoo Lim at the Department of Genetics, Yale University. She holds a B.S. in Biology and Zoology, and a Ph.D. in Integrative Biology from the University of Wisconsin-Madison. Her broad research interests are in identifying altered molecular mechanisms that are common across affected brain regions and neurodegenerative diseases. She is currently focused on identifying the similarities and differences in the transcriptional profiles of two SCA1 affected brain regions, the cerebellum and inferior olive. This study found distinct genetic pathways that are differentially expressed in each tissue, which offers preliminary insights into tissue-specific pathogenesis in SCA1. Her other ongoing research is focused on elucidating the roles of specific signaling pathways involved in cerebellar degeneration, including the further characterization of Nemo-like kinase in a cell-type specific context of SCA1.

Biographies (continued)



Chandrakanth Edamakanti, PhD – I am Chandrakanth Edamakanti. I received my PhD from the University of Würzburg in neurology. In 2014, I joined the lab of Dr. Puneet Opal at Northwestern University as a Post-Doc. My research focuses on understanding the early pathological derailments that drive the disease pathogenesis in adult-onset, neurodegenerative disorders, specifically a polyglutamine disorder called Spinocerebellar Ataxia Type 1 (SCA1). My primary focus is to explore the existence of plausible, non-cell autonomous toxicity (non-neuronal or neuronal) of Purkinje cells before the disease onset, which is what sets the stage for lateral vulnerability in the disease. I am also interested in unraveling the existence of compensatory mechanisms/secondary cascades that determine the disease onset. Targeting these novel cascades could lead to potential therapeutic targets in the SCA1 field and other adult-onset neurodegenerative disorders.



Lisa M. Ellerby, PhD – Lisa M. Ellerby, Ph.D., is founding faculty member at the Buck Institute for Research on Aging, and has been working on Huntington's disease (HD) for over 15 years. She received both undergraduate and graduate degrees in Chemistry from University of California, Santa Cruz. Her postdoctoral training at UCLA was with National Academy member, Dr. Joan Valentine, studying superoxide dismutase function and its role in ALS. Dr. Ellerby identified several mechanisms by which expanded polyglutamine proteins confer toxicity such as cleavage by caspases and calpain. Her laboratory is using a number of approaches, including siRNA screens in cell culture models of HD and production of new mouse models to evaluate therapeutics in HD. She has considerable expertise in analyses of genetically modified cell culture and mouse models to explore mechanisms involved in neurodegenerative disease including the role of stem cells in HD. Recently, the laboratory has developed as isogenic, allelic induced pluripotent stem cell model of HD using HD patient fibroblasts and demonstrated that the damage imposed by mutant huntingtin (HTT) can be restored after the genetic mutation is corrected by homologous recombination.



Austin Ferro – I completed my undergraduate degree with a major in Neuroscience at Skidmore College where I had the great opportunity to work in Dr. Sarita Lagalwar's lab. While in Dr. Lagalwar's lab, I studied mitochondrial dysfunction in spinocerebellar ataxia type 1 (SCA1), which developed into my undergraduate thesis topic, and two manuscripts (Ferro et al., 2017 *Jove*, Ferro et al., 2017 *PLoS One*). To continue my research in SCA1, I joined the Graduate Program of Neuroscience (GPN) at the University of Minnesota to be a part of the fantastic ataxia group at the UMN, as well as be able to work closely with researchers such as Dr. Harry Orr, Dr. Gülin Öz, and Dr. Marija Cvetanovic. After developing a love for electrophysiology, I joined the labs of Dr. Marija Cvetanovic and Dr. Alfonso Araque to study the role of glia in SCA1, and focus on combining molecular neuroscience with electrophysiology to comprehensively understand how glial reactivity may contribute to disease states. I am currently a third year PhD candidate in the GPN, where I am continuing my research on astroglia contributions to SCA1, and focusing on glial control of homeostasis.

Biographies (continued)



Vincent Gerard Francis – Vincent Gerard Francis, Ph.D. is postdoctoral fellow at Montreal Neurological Institute, McGill University at Montreal. Francis received his Bachelor's and Master's degree from Osmania University and University of Hyderabad at India in Biotechnology. He completed his Ph.D. with a prestigious fellowship from Council of scientific and Industrial Research at the Indian Institute of Technology Madras in Biochemistry working on human phospholipid scramblases which regulate membrane asymmetry. He then joined Dr. Peter McPherson's group in 2015 which focusses on molecular mechanisms regulating membrane trafficking with emphasis on neurodegenerative diseases. He currently works on elucidating the etiology of Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS) disorder and is a recipient of Fonds de recherche du Québec postdoctoral fellowship.



Vincenzo A. Gennarino, PhD – Vincenzo A. Gennarino studied molecular biology at the University of Palermo and graduated *cum laude* in 2005. In 2009, he received a PhD in Medical Genetics from TIGEM. During his doctoral work he became interested in how microRNAs regulate gene targets in humans. He devised new bioinformatic tools to infer the biological pathways for each human miRNA based on the co-expression of its targets. These tools, HOCTAR and CoMeTa (*Genome Research*, 2009 and 2012), were later used to identify miR-128 controlling TFEB (*Science* 2009). For his postdoctoral work, Dr. Gennarino joined the lab of Huda Y. Zoghbi. There he studied the regulation of the epigenetic factor *MECP2* during neurodevelopment (*Genes & Development*, 2013), the post-translational regulation of a neurodegenerative disease protein, Ataxin1 (*Cell*, 2015 and *Cell*, 2018), and alternative polyadenylation (*eLife*, 2015). This work forms the basis of his lab's current projects at the Department of Genetics & Development at Columbia University Medical Center, where he joined the faculty in January 2018 as Assistant Professor.



Ian Harding, PhD – Dr. Harding is a neuroscientist based at the Institute of Cognitive and Clinical Neurosciences at Monash University (Melbourne, Australia). His expertise lies in human neuroimaging (Magnetic Resonance Imaging and Positron Emission Tomography) and computational neuroscience approaches to discovering markers and investigating mechanisms of psychiatric and neurological disorders. He completed in PhD in cognitive neuroscience from the University of Melbourne in 2013, before commencing a post-doc investigating the neural expression and progression of Friedreich Ataxia (FRDA) at Monash University. In 2016, Dr. Harding was awarded a research fellowship from the Australian National Health and Medical Research Council (NIH-equivalent) to commence an independent research career and continue his neuroimaging work in FRDA and other cerebellar and subcortical disorders. He is the founding principal investigator of the ENIGMA-Ataxia neuroimaging research consortium, which aims to pool MRI data and expertise in FRDA and dominant Spinocerebellar Ataxias from more than 20 sites globally. He also currently serves as the convener of the Monash University Dementia & Neurodegeneration Research Network.

Biographies (continued)



Pierre-Gilles Henry – Dr. Henry holds dual degrees in Electrical Engineering (MS) and in Neuroscience (MS, PhD). He uses Magnetic Resonance Spectroscopy (MRS) to study brain metabolism in health and disease. He is currently an Assistant Professor at the Center for Magnetic Resonance Research at the University of Minnesota, one of the world's leading laboratories in MR research

The goal of Dr. Henry's current research is to identify biomarkers of disease progression in neurodegenerative diseases, particularly Huntington's Disease and Friedreich's Ataxia. Identification of such biomarkers, particularly during the presymptomatic phase or at an early stage of the disease, would make it possible to assess the efficacy of prospective treatments in clinical trials.

In addition, he is pursuing integration of MRS with other modalities, such as MR anatomical imaging, MR diffusion imaging, and metabolic modeling. Combination of these multiple modalities using mathematical models is expected to lead to more sensitive assessment of disease progression than each marker considered separately.



Baljit Khakh, PhD – Baljit S. Khakh received a Ph.D. degree from the University of Cambridge in 1995. During his graduate studies, he also spent some time at the Geneva Biomedical Research Institute. Dr. Khakh completed a postdoctoral fellowship in the laboratory of Dr. Graeme Henderson at the University of Bristol, followed by a fellowship at the California Institute of Technology, working in the laboratories of Drs. Henry A. Lester and Norman Davidson as a Wellcome Trust International Prize Traveling Research Fellow, and Senior Research Fellow in the Division of Biology. In 2001, Dr. Khakh returned to Cambridge in the Division of Neurobiology

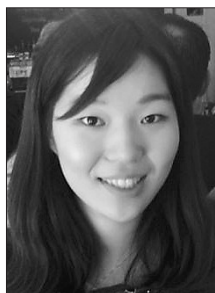
at the MRC Laboratory of Molecular Biology as a Group Leader. Dr. Khakh relocated to UCLA in 2006 and is now Professor of Physiology and Neurobiology. The Khakh lab explores fundamental astrocyte biology.



Professor Thomas Klockgether, MD, PhD – Prof. Klockgether studied medicine at the University of Göttingen and during this time also carried out research at the Max Planck Institute for Experimental Medicine. After graduating, he went to Oldenburg for clinical training and then returned to the Max Planck Institute to work in basic research on Parkinson's disease. He completed his neurology training in Tübingen, where he also began to focus on degenerative ataxias, in addition to pursuing research on Parkinson's disease. These research lines evolved very successfully during his appointment in Bonn as Chairman of Department of Neurology. Prof.

Klockgether has been the Dean of the Medical Faculty of the University of Bonn from 2008 to 2011. Since February 2010 he has been Speaker of the Center for Rare Diseases Bonn (ZSEB) and since May 2011 Director of Clinical Research at the DZNE.

Biographies (continued)



Ha Eun Kong, MD, PhD Student – Ha Eun received her Bachelor's degree in Chemistry from Princeton University, where she did her undergraduate thesis work in the Chemical Biology laboratory of Dr. Tom Muir. After working as a research assistant in the laboratory of chromatin biology and epigenetics under the supervision of Dr. David Allis at Rockefeller University, she moved to Emory, where she is currently pursuing an MD/PhD degree at Emory University School of Medicine. For her PhD studies in Genetics and Molecular Biology, she is investigating the molecular basis for the pathogenesis of Fragile X Tremor and Ataxia Syndrome (FXTAS) under the supervision of her advisor, Dr. Peng Jin, at Emory University.



Albert La Spada, MD, PhD – 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 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 Postdoctoral 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 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 Director of the newly created Duke Center for Neurodegeneration & Neurotherapeutics, and is a Professor of Neurology, Neurobiology, and Cell Biology at the Duke University School of Medicine, where he also 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, 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 to be a Gund-Harrington Scholar for his translational research accomplishments.

Biographies (continued)



Patrícia Maciel, PhD – Patrícia Maciel obtained a B.Sc. in Biochemistry (1993) and a Ph.D. in Biomedical Sciences - Genetics (1998) at the University of Porto, Portugal. Her doctoral studies included an initial training period at the Hôpital Necker-Enfants Malades, Paris, France, and four years in the Rouleau lab at the Centre for Research in Neuroscience, McGill University, Montreal, Canada.

Dr. Maciel is currently an Associate Professor of Biochemistry and Genetics at the School of Medicine and a Senior Researcher at the Health and Life Sciences Research Institute of the University of Minho – Braga, Portugal, where she develops works in the field of Neurogenetics, addressing molecular mechanisms of neuronal function and dysfunction, in connection to human neurodegenerative and neurodevelopmental diseases.

Her major scientific contributions have been towards the mapping and cloning of the spinocerebellar ataxia type 3/Machado-Joseph disease (SCA3/MJD) causative gene, the study of genotype-phenotype correlations in this and other inherited neurological diseases, the identification of the normal cellular function of the protein ataxin-3 and its potential links to pathogenesis, as well as the development of transgenic mouse and *C. elegans* models of SCA3/MJD, useful for therapeutic drug discovery and development. This has led to an interest in studying specific cellular processes such as protein regulation by the ubiquitin-proteasome system in the nervous system. Recently, the Maciel team has contributed actively to the identification of drugs with important therapeutic effects in animal models of SCA3/MJD, through candidate testing and unbiased screening approaches.



Hayley McLoughlin, PhD – Dr. Hayley McLoughlin is a Research Investigator in the department of Neurology at the University of Michigan. She received her PhD in Neuroscience from the University of Iowa in 2013 under the mentorship of Dr. Beverly Davidson. She completed postdoctoral work with Dr. Henry Paulson (2014-2016) at the University of Michigan, focusing on gene therapy approaches for Spinocerebellar Ataxia type 3/Machado-Joseph disease (SCA3/MDJ). In the fall of 2016, she joined the faculty of the Neurology Department at the University of Michigan, where she is continuing her research focused on therapeutic strategies for neurodegenerative diseases. In January of 2018, she was awarded a National Ataxia Foundation SCA Young Investigator grant to seek SCA3 biomarkers through proteomic and transcriptional profiling. In 2018, she was awarded NINDS CREATE Bio U01 funding to lead a collaboration with Ionis Pharmaceuticals in developing antisense oligonucleotide therapy for SCA3.



Lauren Moore – Lauren Moore is a neuroscience Ph.D. candidate in the laboratory of Dr. Henry Paulson at the University of Michigan. Lauren earned her B.S. in biomedical physics at Northeastern University in Boston in 2013. Her dissertation research in the Paulson lab has focused on investigating mechanisms of neurodegeneration in spinocerebellar ataxia type 3 (SCA3) using human embryonic stem cell and mouse models of SCA3, with particular interests in the role of autophagy and nuclear dysfunction in disease. In collaboration with Ionis Pharmaceuticals and Dr. Hayley McLoughlin, her research has also focused on preclinical assessment of antisense

Biographies (continued)

oligonucleotide therapy in cellular and mouse models of SCA3. Lauren has received several awards for her work on SCA3, including best poster presentations at the EMBO Mechanisms of Neurodegeneration Meeting (2017) and the CAG Triplet Repeat Disorders Gordon Research Seminar (2015), and has been awarded several fellowships including the MiBrain Initiative Neuroscience Predoctoral Fellowship. She intends to complete her dissertation research in fall 2018.



Jill Napierala, PhD – Jill Sergesketter Napierala, Ph.D. is an assistant professor in the Department of Biochemistry and Molecular Genetics at the University of Alabama at Birmingham. She received her B.S. in Biology and Chemistry from the University of Indianapolis and her Ph.D. in Biochemistry and Molecular Biology from Indiana University in the laboratory of Dr. David Skalnik. She went on to complete a post-doctoral fellowship at the University of Texas MD Anderson Cancer Center with Dr. Sharon Dent where her work centered on characterizing chromatin modifying enzyme complexes in leukemia models. She then joined the laboratory of Dr. Marek Napierala and transitioned her research focus to defining molecular mechanisms underlying the neurodegenerative disorder, Friedreich's ataxia (FRDA). Currently, Jill's research interests cover several aspects of FRDA molecular pathogenesis, including identification of gene expression biomarkers, defining the pathogenic impact of Frataxin point mutant proteins in cell and animal models, and determining the role of mitochondrial aldehyde dehydrogenases in mitigating oxidative stress in FRDA neuronal cell models.

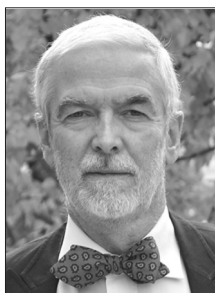


Harry Orr, PhD – Harry Orr, PhD directs the Institute for Translational Neuroscience and is the Tulloch Professor of Genetics in the Department of Laboratory Medicine and Pathology at the University of Minnesota Medical School. Dr. Orr received a BA degree from Oakland University in Rochester, Michigan. 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.



Gülin Öz, PhD – Dr. Öz is a brain imaging scientist who specializes in magnetic resonance spectroscopy (MRS). She graduated from Bosphorus University in Istanbul, Turkey with BS degrees in Physics and Chemistry and obtained her PhD in Biochemistry at the University of Minnesota. She continued with postdoctoral training at the Center for Magnetic Resonance Research at the University of Minnesota where she joined the faculty in 2006. Dr. Öz's research focuses on the application of MRS techniques to detect chemical changes in the cerebellum in ataxias. MRS non-invasively measures levels of many brain chemicals including neurotransmitters and antioxidants. Such information can facilitate early detection of neurodegeneration and provide an objective means to monitor disease progression and response to therapies.

Biographies (continued)



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 the Pulst laboratory 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. His group is pioneering 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 and a Senator Jacob Javits Award for Neuroscience from NINDS. Pulst was chair of the AAN Science Committee from 2006 to 2011 and now chairs the AAN Meeting Management Committee. He is also the founding editor of *Neurology*® Genetics.

Wade Regehr, PhD – Wade Regehr has long studied synapses in the cerebellum. These studies initially focused on calcium regulation of neurotransmitter release and short-term synaptic plasticity. His lab went on to describe endocannabinoid release by Purkinje cells and interneurons and retrograde regulation of synapses. His lab subsequently focused on Purkinje cell collaterals that allow the output of the cerebellar cortex to provide feedback to other Purkinje cells, granule cells and inhibitory interneurons. His lab has recently described molecular specializations that allow Purkinje cells to provide frequency invariant synaptic transmission to deep cerebellar nuclei.



Dobriła Rudnicki, PhD – Dr. Doda Rudnicki is a program director on the Special Initiatives team, Office of the Director, at NCATS. Dr. Rudnicki's primary scientific and programmatic interest is in the creation of novel tools and technologies relevant to cost-effective and accelerated drug discovery and development across many disease phenotypes. Prior to joining NCATS, Dr. Rudnicki was a principal investigator in the Department of Psychiatry, Johns Hopkins School of Medicine. Her laboratory has performed seminal work on the role of sense and antisense RNA transcripts in the pathogenesis of Huntington disease-like 2 (HDL2), Huntington disease (HD) and spinocerebellar ataxia type 2 (SCA2). She has received multiple NIH grants, as well as support from national and international private foundations, including the Hereditary Disease Foundation, CHDI Foundation, and Advocacy for Neuroacanthocytosis Patients. Dr. Rudnicki has obtained her Ph.D. degree in neurobiology from the University of Aachen (Germany), and her B. Sc. degree in biochemistry and microbiology from King's College, University of London (UK).

Biographies (continued)



Amy Salovin, MS – Amy holds a Master’s of Science in Cellular and Molecular Biology from the University of New Haven, and works as a research assistant in the laboratory of Dr. David Lynch at the Children’s Hospital of Philadelphia. The Lynch lab is driven to help develop a cure for Friedreich Ataxia, and focuses on research ranging from basic mechanisms of disease pathogenesis to preclinical drug assessment and clinical trials. While initially focusing on neurological deficits in Friedreich Ataxia, the Lynch lab has begun expanding their work in other systems to garner a deeper understanding of the disease as a whole. Amy’s current research projects focus on characterizing the cardiac phenotype of Friedreich Ataxia mouse models with the goal of treating these models with preclinical agents to reverse, or at least halt, cardiac disease progression. We hope these pre-clinical drug assessments will translate into more drugs entering the treatment pipeline, and ultimately translate into therapy.



Matthew Scaglione – I am currently an Assistant Professor in the Department of Biochemistry and the Neuroscience Research Center at the Medical College of Wisconsin. 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.



Jeremy D. Schmahmann, MD – Dr. Schmahmann is Professor of Neurology at Harvard Medical School, and a Neurologist at the Massachusetts General Hospital where he is the Founding Director (1994) of the Ataxia Unit, Director of the Laboratory for Neuroanatomy and Cerebellar Neurobiology, and a member of the Cognitive Behavioral Neurology Unit. His research and clinical practice focuses on the neurology and basic science of the ataxias and other cerebellar disorders, and he has pioneered the role of the cerebellum in cognition and emotion.

Dr. Schmahmann graduated with distinction from the University of Cape Town, South Africa, completed residency in the Neurological Unit of Boston City Hospital, and postdoctoral fellowship in the Department of Anatomy and Neurobiology at Boston University School of Medicine. He is a Fellow of the American Academy of Neurology, the American Neurological Association, and

Biographies (continued)

the American Neuropsychiatric Association of which he is Immediate Past President. He is a member of the Medical and Scientific Research Advisory Board of the National Ataxia Foundation, Medical Advisor to the New England Chapter of NAF, the Cerebellar Research Consortium for the Study of Cerebellar Ataxias, and on the Executive of the Society for Research on the Cerebellum and Ataxias. He has authored over 200 papers, chapters and clinical contributions, and written or co-edited six books – including most recently *Essentials of the Handbook of the Cerebellum*. His awards include the Norman Geschwind Prize for research in behavioral neurology, Distinguished Neurology Teacher Award (American Neurological Association), Special Prize for Sustained Excellent in Teaching (Harvard Medical School), and he has been cited in *The Best Doctors in America* since 1996.



Eunju Seong, PhD – Eunju Seong, Ph.D., received her doctoral degree in Neuroscience at the University of Michigan Ann Arbor. While taking courses in human genetics, statistics and neuroscience, she was mentored for her Ph.D. by Margit Burmeister, Ph.D. to study mouse mutants with seizures and motor defects. Upon completion of her study she took a break in her career to tend her growing family. She resumed her research in biomedical science as a postdoc, first at the University of Nebraska Medical Center studying neuronal differentiation, then she joined Dr. Burmeister’s team again as a postdoc to become a key member of her ataxia gene discovery team. Dr. Seong has focused on applying novel genetic, cell and stem cell technologies on ataxia-related research, often in collaboration with experts around the world. Throughout her research career, Dr. Seong has focused molecular and cellular mechanism of movement disorders, most recently experimental validation of novel ataxia candidate genes.



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 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.

Biographies (continued)

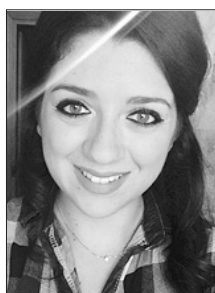
He was awarded the Dorothy Penrose Stout Award for the Best Predoctoral Fellowship application from the American Heart Association Western States Affiliate and the Leonard Berg award for research done as a resident at Washington University. He also holds a patent related to his work on an ion channel gene used to generate a mouse model of cerebellar ataxia.



Jorge Silva – Jorge is an MD/PhD student at the School of Medicine, University of Minho in Braga, Portugal. He has completed five out of six years of medical school and would like to follow a specialization in Medical Genetics. He is currently pursuing his PhD studies in the field of Neurogenetics, under the supervision of Professor Patrícia Maciel, at the Life and Health Sciences Research Institute (ICVS), in Braga, Portugal. He has previously focused his research interests in the modelling of neurologic and genetic disorders using *Caenorhabditis elegans* models, such as Alzheimer's disease and spinal muscular atrophy. Currently, his studies focus on Machado-Joseph's disease (MJD). More specifically, he is addressing how we can use hormones that bind to nuclear receptors as therapeutic strategies, what are the underlying mechanisms of these compounds, and what are the protective responses elicited by drugs that have positive effects in the phenotype of the nematode model of the disease. Since nuclear hormone receptors (NHR) have been associated with various neurodegenerative conditions (both as causes and risk factors), his goal is to take advantage of the nematode model to perform pharmacological and genetic modulations tackling NHRs to improve MJD.



Colleen A. Stoyas, PhD – Colleen A. Stoyas completed her PhD at the University of California, San Diego in 2017 in the laboratory of Dr. Albert La Spada. Her work focuses on the transcriptional and metabolic changes underlying SCA7 disease pathology. Additionally she has orchestrated collaboration with researchers in Mexico working with one of the world's largest SCA7 patient populations, and continues to work with Dr. La Spada to enhance communications between these research groups. In San Diego, Colleen previously implemented a novel NAF and ataxia awareness program and hopes to soon revive the local Walk n' Roll. After some time at Duke University with Dr. La Spada, Colleen began her formal post doctoral training in Musculoskeletal Biology and Neuromuscular Disease at the Genomics Institute of the Novartis Research Foundation.



Francesca Tiano, PhD Student – I am a Ph.D. Student in Molecular Medicine, Immunology and Applied Biotechnology in the Laboratory of Signal Transduction, directed by Professor Roberto Testi, at the University of Rome Tor Vergata. I was born in the south of Italy in Calabria. I obtained my bachelor's degree in Biological Sciences at the University of Calabria in 2011. Subsequently, I decided to keep specialized in Cell and Molecular Biology at the University of Rome Tor Vergata and graduated *cum laude* in 2015. Currently, our research group is focusing on a monogenic disease – Friedreich's Ataxia (FRDA) – in which the deficiency of the mitochondrial protein frataxin causes enhanced

Biographies (continued)

susceptibility to stress-induced cell death. In FRDA the heart is frequently affected with typical manifestation of hypertrophic cardiomyopathy, which can progress to heart failure and cause premature death.

Specifically, my research is centered on identifying new genes involved in FRDA pathogenesis and finding new prognostic or diagnostic biomarkers for early diagnosis and clinical monitoring of cardiomyopathy in Friedreich's Ataxia.



Ray Truant – Ray Truant is a native of Toronto, Canada where he received his undergraduate and PhD degrees at the University of Toronto on the biochemistry of the P53 protein. Following his PhD work, he was a Research Associate at the Howard Hughes Medical Institute at Duke University. He started his own lab at McMaster University in 2000, focused on the cell biology of polyglutamine disease proteins. He is currently a Professor at McMaster and Director of the McMaster Biophotonics Facility, dedicated to advanced light microscopy and nanoscopy, as well as High Content Drug screening.

He has been Chair of the Scientific Board of the Huntington Society of Canada since 2007, and on the HSC Board of Directors. He has won the 2012 Queen Elizabeth II Diamond Jubilee Medal, as well as the 2014 Michael Wright Award for community service in HD. He was the founding scientific Editor of HDbuzz.net. His lab now has projects encompassing HD, SCA1, SCA3, SCA7, SCA17 and spontaneous ataxia derived from the Neuromuscular Disease clinic at McMaster. He is on the Scientific Advisory Board on Mitokinin LLC, a company with projects in Parkinson and Huntington diseases, and has current and past collaborations with Novartis AG and PTC Therapeutics, NJ. His current focus is on an inter-disease approach to CAG expansion diseases, focusing on the roles of these proteins in DNA repair and RNA processing.

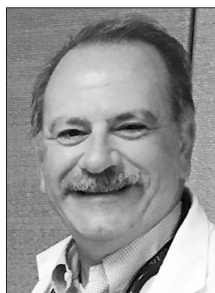


Alanna Watt, PhD – Alanna Watt is an Associate Professor at McGill University in Montreal, Canada. Using state-of-the-art electrophysiological and optical techniques combined with behavioral assays the Watt lab aims to understand pathophysiological alterations in mouse models of two forms of ataxia (SCA6 and ARSACS), and to develop therapeutic interventions.

Prof. Watt received her PhD in Neuroscience at Brandeis University, USA, and pursued postdoctoral research at University College London in London, UK. She joined McGill University in 2011 and was voted “Professor of the Year” in 2016 by the Biology Undergraduate Student Association.

Prof. Watt has published nearly twenty journal articles and book chapters, of which six publications have been cited more than 100 times. She has organized symposia and given lectures at national and international meetings and universities. Dr. Watt has received funding from the British Royal Society, EMBO, Canadian Institutes of Health Research, the Scottish Rite, and the ARSACS Foundation. She is a member of the college of reviewers for national and international funding agencies, serves on the editorial board of Scientific Reports, Open Biology, and Frontiers in Synaptic Neuroscience, and is a board member of the Canadian Association for Neuroscience (CAN).

Biographies (continued)



George Wilmot, MD, PhD – George (Chip) Wilmot, MD, PhD is Associate Professor in the Movement Disorders division of the Department of Neurology at Emory University. He graduated with an MD, PhD from the Medical Scientist Training Program at the University of Michigan, did his neurology residency at Emory, and then joined the faculty at Emory where he is currently Associate Professor. Dr. Wilmot had early career experience in laboratory research working on disease mechanisms of the ataxia and on factors affecting axonal stability, but for the past 15 years has focused primarily on clinical research in ataxia and on the clinical care of ataxia patients.



Yalan Zhang – I received my Ph.D. from the Chinese Academy Medical Sciences and Peking Union Medical College. I then came to the United States to work as a postdoc in the laboratory of Dr. Leonard Kaczmarek to study how ion channels control neuron excitability. Initially I used *Aplysia* as a model neuronal system. In this lab, I received very strong training in molecular biology and aspect of electrophysiology. I cloned *Aplysia* Slo and Slack genes and their isoforms and submitted these to NIH gene bank. In 2012, I began to investigate the mechanism of Spinocerebellar Ataxia type 13 (SCA13), which caused by mutation in the Kv3.3 channel, I discovered that the Kv3.3 channel differ from other potassium channels in that it binds Hax-1, an anti-apoptotic protein that is required for the survival of cerebellar neurons. Our work has led to the growing importance of SCA13 treatment. The paper was published in the journal Cell in 2016. Moreover, I also collaborated with the lab of Dr. Arthur Horwich and found that ALS-linked mutant SOD1 may downregulate a key sodium-gated potassium channel, known as SLACK, through an apoptosis signal-regulating kinase 1 (ASK1) based mechanism.



Huda Zoghbi, MD – Huda Zoghbi grew up in Beirut, Lebanon where she obtained a Bachelor of Science and started medical school at the American University of Beirut before transferring to Meharry Medical College during the Lebanese civil war. She trained in Pediatrics, Neurology, and Molecular Genetics at Baylor College of Medicine where she is now the Ralph D. Feigin Professor of Pediatrics, Neuroscience, and Molecular and Human Genetics and an Investigator with the Howard Hughes Medical Institute. She is the founding Director of the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital.

Her patient-inspired research led to the discovery of the spinocerebellar ataxia type 1 gene and mechanisms mediating neurodegeneration (with Harry Orr), and the discovery of the Rett syndrome gene and its effects on the brain. Her cross-species studies with Juan Botas are leading to potential therapeutic entry points for Alzheimer and Parkinson. Her curiosity-driven research led to the discovery that *Atoh1* governs the development of several components of the balance, hearing, and breathing circuits. She is a member of the National Academy of Medicine and National Academy of Sciences. Among Dr. Zoghbi's honors are the 2017 Canada Gairdner International Award and the 2017 Breakthrough Prize in Neurodegeneration.

Meeting Attendees

Akshay Alaghatta, BS
Duke University
akshay.alaghatta@duke.edu

Daniele Alaimo, MSc
Medical Science Liaison
MNG Laboratories
dalaimo@mnglabs.com

Moore Arnold, PhD
Research Director
Biogen, Inc.
moore.arnold@biogen.com

Tee Ashizawa, MD
Houston Methodist Research
Institute
tashizawa@houstonmethodist.org

Adam Avery, PhD
University of Minnesota
aavery@umn.edu

Naiara Akizu, PhD
Children's Hospital of
Philadelphia
aquizun@email.chop.edu

Kristin Baranano, MD, PhD
Johns Hopkins
kwb@jhmi.edu

Amy J. Bastian, PhD, PT
Kennedy Krieger Institute
bastian@kennedykrieger.org

Esther B.E. Becker, PhD
University of Oxford
esther.becker@dpag.ox.ac.uk

Carole Ben-Maimon, MD
Chief Executive Officer
Chondrial Therapeutics
cbenmaimon@chondrialtherapeutics.com

C. Frank Bennett, PhD
Ionis Pharmaceuticals, Inc.
fbennett@ionisph.com

Robert Berman, MD
Biohaven Pharmaceuticals
robert.berman@snet.net

James Berry, MD
Massachusetts General Hospital
jdberry@mgh.harvard.edu

Ian A. Blair, PhD
University of Pennsylvania
ianblair@upenn.edu

Gabriela Bolzan, MD
Universidade Federal Do Rio
Grande Do Sul
gbolzan94@gmail.com

Jessica Bowman, BS
University of Florida
jbowman1@ufl.edu

Daniel P. Bradley, PhD
Biogen, Inc
daniel.bradley@biogen.com

Margit Burmeister, PhD
University of Michigan
margit@umich.edu

Khalaf Bushara, MD
University of Minnesota
busha001@umn.edu

David Bushart, BS
University of Michigan
dbushart@umich.edu

Barry Byrne, MD, PhD
University of Florida
aatzinger@ufl.edu

Suzy Cahn, BS
Emory University School of
Medicine
suzy.cahn@gmail.com

Keith Caldecott, PhD
University of Sussex
k.w.caldecott@sussex.ac.uk

Ryan Chang, MBA
General Manager
Steminent Biotherapeutics Inc.
ryan.chang@steminent.com

Dong-Hui Chen MD, PhD
University of Washington
cdhui@uw.edu

Elisia Clark, PhD
University of Pennsylvania
eliclark@pennmedicine.upenn.edu

Giulia Coarelli, MD
ICM/Hospitals of Paris
giulia_coarelli@hotmail.it

Manuela Corti, PhD
University of Florida
m.corti@peds.ufl.edu

Maria Do Carmo Costa, PhD
University of Michigan
mariadoc@umich.edu

Meeting Attendees

Marija Cvetanovic, PhD
University of Minnesota
mcvetano@umn.edu

Jorge Diogo Da Silva,
PhD Candidate
University of Minho
id6522@alunos.uminho.pt

Ibrahim Dagher, MD
Cadent Therapeutics
bdagher@cadenttx.com

John W. Day, MD, PhD
Stanford University
juwday@stanford.edu

Chris de Zeeuw, MD, PhD
Erasmus Medical Center
c.dezeeuw@erasmusmc.nl

Marissa Dean, MD
University of Alabama at
Birmingham
mn dean@uabmc.edu

Martin Delatycki, MD, PhD
Murdoch Children's Research
Institute
martin.delatycki@vcgs.org.au

Yi Na Dong, PhD
The Children's Hospital of
Philadelphia
yinayuan@gmail.com

Terri Driessen, PhD
Yale University
terri.driessen@yale.edu

Alexandra Durr, MD, PhD
ICM
alexandra.durr@upmc.fr

Chandrankanth Edamakanti,
PhD
Northwestern University
chandu@northwestern.edu

Kara Eichelkraut
Manager, Medical Field
Operations
Reata Pharmaceuticals
*Kara.Eichelkraut@
reatapharma.com*

Mahmoud El-Gohary, PhD
APDM, Inc.
mahmoud@apdm.com

Lisa M. Ellerby, PhD
Buck Institute for Research
on Aging
lellerby@buckinstitute.org

Jennifer Farmer, MS
Executive Director
Friedreich's Ataxia Research
Alliance
jen.farmer@curefa.org

Austin Ferro, BA
University of Minnesota
ferro029@umn.edu

Kurt Fischbeck, MD
NIH/NINDS
fischbeck@ninds.nih.gov

Brent Fogel, MD, PhD
UCLA
bfogel@ucla.edu

Vincent Francis, PhD
McGill University
vincent.francis@mail.mcgill.ca

Ralitza Gavrilova, MD
Mayo Clinic-Rochester
gavrilova.ralitza@mayo.edu

Vincenzo A. Gennarino, PhD
Columbia University Medical
Center
vag2138@cumc.columbia.edu

Geetanjali Gera, PhD
University of Kentucky
geetanjali@uky.edu

Geraerts, Martine, PhD
Lab Manager
UCB Biopharma SPRL
martine.geraerts@ucb.com

Christopher M. Gomez,
MD, PhD
The University of Chicago
gomez001@uchicago.edu

Xavier Guell, MD
MIT and Harvard
xaviergp@mit.edu

Sarah Gunther, MA
UCLA
SGunther@mednet.ucla.edu

Anoopum Gupta, MD, PhD
Massachusetts General Hospital
and Harvard Medical School
agupta20@partners.org

Deborah Hall, MD, PhD
Rush University
deborah_a_hall@rush.edu

Ian Harding, PhD
Monash University
ian.harding@monash.edu

Meeting Attendees

Anhar Hassan, MB, BCh,
FRACP
Mayo Clinic
Hassan.Anhar@mayo.edu

Trevor Hawkins, MD
University of Colorado
trevor.hawkins@ucdenver.edu

Pierre-Gilles Henry, PhD
University of Minnesota
henry044@umn.edu

Laryssa Huryyn, MD
National Eye Institute, NIH
laryssa.huryyn@nih.gov

Diane Hutter, RN
University of Minnesota
hutte019@umn.edu

Fadi Issa, PhD
East Carolina University
issaf14@ecu.edu

Matthew Johnson, BSc
APDM Wearable Technologies
matt@apdm.com

Bonnie Kaas, MD
Johns Hopkins University
bkaas1@jhmi.edu

Gregg Keaney, PhD
Cadent Therapeutics
gkeaney@cadenttx.com

Megan S. Keiser, PhD
Children's Hospital of
Philadelphia
keisermk@email.chop.edu

Baljit Khakh, PhD
UCLA
bkhakh@mednet.ucla.edu

Thomas Klockgether, MD,
PhD
DZNE Bonn
thomas.klockgether@ukbonn.de

Dione Kobayashi, PhD
President
Akeola
dione.kobayashi@gmail.com

Ha Eun Kong, BA
Emory University
haeun.kong@emory.edu

Sheng-Han Kuo, MD
Columbia University
sk3295@columbia.edu

Al La Spada, MD, PhD
Duke University
al.laspada@duke.edu

Sarah N. Larson, MSc
University of Minnesota
lars4070@umn.edu

Jong Seo Lee, BS
Yale University
jongseo.lee@yale.edu

Christophe Lenglet, PhD
University of Minnesota
clenglet@umn.edu

Pan Li, PhD
Johns Hopkins University
Ple5@jhmi.edu

Janghoo Lim, PhD
Yale University
janghoo.lim@yale.edu

Hong Lin, PhD
Children's Hospital of
Philadelphia
linh@email.chop.edu

Damaris Lorenzo, PhD
University of North Carolina
at Chapel Hill
damaris_lorenzo@med.unc.edu

Sarah Luchansky, PhD
Vertex Pharmaceuticals
sarah_luchansky@vrtx.com

Patrícia Maciel, PhD
University of Minho
pmaciel@med.uminho.pt

Florence Malisan, PhD
University of Rome
Tor Vergata
malisan@med.uniroma2.it

Rachna Manek, MS
University of Florida
mrach8@ufl.edu

Caterina Mariotti, MD
Foundation-IRCCS Neurolog-
ical Institute Carlo Besta
caterina.mariotti@istituto-besta.it

Angelical Martin, PhD
Friedreich's Ataxia Research
Alliance
angel.martin@curefa.org

Char Martins, RN
University of Minnesota
charlotte_martins@hotmail.com

Meeting Attendees

Cherie Marvel, PhD
Johns Hopkins
cheriemarvel@gmail.com

James McArthur, PhD
Cydan II, Inc
vcourtney@cydanco.com

Hayley McLoughlin, PhD
University of Michigan
hayleymc@umich.edu

Clementina Mesaros, PhD
University of Pennsylvania
mesaros@upenn.edu

Uwe Meya, MD, PhD
Minoryx Therapeutics
Medical Officer
umeya@minoryx.com

Dan Miller, PhD
NINDS
daniel.miller@nih.gov

Bill Milligan, BSc
Vice President
Steminent Biotherapeutics Inc.
bill@steminent.com

Lauren R. Moore, PhD
Candidate
University of Michigan
lrmo@umich.edu

Owen Morgan, BA
Johns Hopkins University
School of Medicine
omorgan2@jhmi.edu

Weiyi Mu, ScM, CGC
Johns Hopkins University
wmu2@jhmi.edu

Katsuya Nakamura, MD, PhD
University of Florida
katsuyanakamura@ufl.edu

Jill S. Napierala, PhD
University of Alabama
at Birmingham
jsbutler@uab.edu

Marek Napierala, PhD
University of Alabama
at Birmingham
mnapiera@uab.edu

Siddharth Nath, MD, PhD
Candidate
McMaster University
naths3@mcmaster.ca

Andreia Neves-Carvalho, PhD
University of Minho
andriacarvalho@med.uminho.pt

Joan Ann O`Keefe, PhD
Rush University
joan_a_okeefe@rush.edu

Chiadi Onyike, MBBS
Johns Hopkins University
conyike1@jhmi.edu

Puneet Opal, MD, PhD
Northwestern University
p-opal@northwestern.edu

Harry T. Orr, PhD
University of Minnesota
orrxx002@umn.edu

Gülin Öz, PhD
University of Minnesota
gulin@cmrr.umn.edu

Martin Paucar, MD, PhD
Karolinska Institutet
martin.paucar-arce@sll.se

Henry Paulson, MD, PhD
University of Michigan
henryp@umich.edu

Luis Pereira de Almeida, PhD
University of Coimbra
luispa@cnc.uc.pt

Susan L. Perlman, MD
UCLA Medical Center
sperlman@mednet.ucla.edu

Tim Piser, PhD
Chief Scientific Officer
Cadent Therapeutics
tpiser@cadenttx.com

Franzi Pohl, PhD Candidate
Robert Gordon University
f.pohl@rgu.ac.uk

Stefan M Pulst, MD
University of Utah
stefan.pulst@hsc.utah.edu

Laura Ranum, PhD
University of Florida
ranum@ufl.edu

Wade Regehr, PhD
Harvard Medical School
wade_regehr@hms.harvard.edu

Liana S. Rosenthal, MD
Johns Hopkins School
of Medicine
liana.rosenthal@jhmi.edu

Meeting Attendees

Cynthia Rothblum-Oviatt, PhD
Science Coordinator
A-T Children's Project
cynthia@atcp.org

Jonathan Rubin, MD, MBA
Chondrial Therapeutics
jrubin18@verizon.net

Dobрила Rudnicki, PhD
National Center For Advancing
Translational Sciences
dobрила.rudnicki@nih.gov

Amy Salovin, MS
Children's Hospital of
Philadelphia
arattelle@gmail.com

Jack Sandoe, PhD
Vertex Pharmaceuticals
jlsandoe@gmail.com

Matt Scaglione, PhD
Medical College of Wisconsin
mscaglione@mcw.edu

Noreen Scherer, BA
VP of Clinical Operations
Chondrial Therapeutics
nscherer@chondrialtherapeutics.com

Jeremy Schmahmann, MD
Massachusetts General and
Harvard Medical School
jschmahmann@partners.org

Larry Schut, MD
National Ataxia Foundation
lschut@lakedalelink.net

Lauren Seeberger, MD
University of Colorado
lauren.seeberger@ucdenver.edu

Eunju Seong, PhD
University of Michigan
Ann Arbor
ejseong@umich.edu

Vikram Shakkottai, MD, PhD
University of Michigan
vikramsh@med.umich.edu

Ludy Shih, MD
Biogen, Inc.
ludy.shih@biogen.com

Hannah Shorrock, PhD
University of Florida
hannah.sherrock@ufl.edu

Bryan Simpson, BS
Children's Hospital of
Philadelphia
bryansim@upenn.edu

Mitchell Slapik, BA
Johns Hopkins School of
Medicine
msslapik1@jhu.edu

Anne Smith, PhD
Ionis Pharmaceuticals
asmith@ionisph.com

Christopher D. Stephen,
MD, MRCP
Massachusetts General Hospital
cstephen@mgh.harvard.edu

Colleen Stoyas, PhD
Duke University
cstoyas2@gmail.com

Celeste Suart, BSc
McMaster University
suartce@mcmaster.ca

Sub Subramony, MD
University of Florida
s.subramony@neurology.ufl.edu

Niels Svenstrup, PhD
Vice President
Cydan II, Inc
vcourtney@cydanco.com

Pawel Switonski, PhD
Duke University
pms53@duke.edu

David Szmulewicz, MD
Florey Institute
dsz@me.com

Leon Tejwani, MS
Yale University
leon.tejwani@yale.edu

Francesca Tiano, PhD
Candidate
University of Rome
Tor Vergata
fratiano66@gmail.com

John Tilton
Chief Business Officer
Biohaven Pharmaceuticals
john.tilton@biohavenpharma.com

Sokol Todi, PhD
Wayne State University
stodi@wayne.edu

Brenda Toscano, PhD
McGill University
brenda.toscanom Marquez@mail.mcgill.ca

Meeting Attendees

Ray Truant, PhD
McMaster University
truant@mcmaster.ca

Wei-Ling Tsou, PhD
Wayne State University
School of Medicine
wtou@med.wayne.edu

Padmaja Vittal, MD, Ms
MD
Northwestern Medicine
Regional Medical Group
padmaja.vittal@nm.org

Adam Vogel, PhD
The University of Melbourne
vogela@unimelb.edu.au

Elizabeth Wagner, PhD
Wave Life Sciences
hoconnor@wavelifesci.com

Hao Wang, PhD
Takeda Pharmaceuticals
hao.wang2@takeda.com

Nathan Warren, BS
Children's Hospital of
Philadelphia
warren@email.chop.edu

Alanna Watt, PhD
McGill University
alanna.watt@mcgill.ca

George "Chip" Wilmot,
MD, PhD
Emory University
gwilmot@emory.edu

Robert Wilson, MD, PhD
Children's Hospital of
Philadelphia
wilsonr@mail.med.upenn.edu

Talene Yacoubian, MD, PhD
University of Alabama at
Birmingham
tyacoubian@usbmc.edu

Miao Zhang, PhD
Chapman University
zhang@chapman.edu

Yalan Zhang, PhD
Yale University
yalan.zhang@yale.edu

Huda Zoghbi, MD
Baylor College of Medicine
hzoghbi@bcm.edu

The National Ataxia Foundation is grateful to the 2018 AIM Steering Committee members:

Gülin Öz, PhD – Chair
Albert La Spada, MD – Co-chair
Marija Cvetanovic, PhD
Patricia Maciel, PhD
Harry Orr, PhD
Jeremy Schmahmann, MD
Vikram Shakkottai, MD, PhD
George (Chip) Wilmot, MD, PhD

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 medical 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. The three-day conference brought leading ataxia investigators from around the world together to share both clinical and scientific research. Travel grants were provided to 10 young investigators, so they could have the opportunity to partake in these discussions. These young investigators are tomorrow's leaders.

In 2008, immediately preceding the NAF Annual Membership Meeting in Las Vegas, more than 120 ataxia investigators from around the world assembled for the 2nd Ataxia Investigators Meeting. An exciting aspect of the AIM 2008 was the involvement of clinical and basic science investigators from Australia, Belgium, Brazil, Canada, China, England, Finland, France, Germany, Italy, Japan, and the United States. An additional important aspect of the 2008 AIM is that it included more than 40 young investigators.



AIM 2010 – Chicago, Illinois – Christopher Gomez, MD, PhD, AIM 2010 Chair



AIM 2012 – San Antonio, Texas – Henry Paulson, MD, PhD, AIM 2012 Chair



AIM 2014 – Las Vegas, Nevada – Harry Orr, PhD, AIM 2014 Chair



AIM 2016 – Orlando, Florida – David Lynch, MD, PhD, AIM 2016 Chair

8th Ataxia Investigators Meeting – March 3-6, 2020

AIM 2020 – Denver, Colorado – Albert La Spada, MD, PhD – AIM 2020 Chair