



**SPONSORED BY THE
National Ataxia Foundation**

**WELCOME TO THE
6th Ataxia
Investigators Meeting
(AIM 2016)**



**March 29 – April 1, 2016
Caribe Royale in Orlando, Florida**

Table of Contents

Welcome and AIM Steering Committee	2
Thank You to Sponsors	3
Meeting Schedule	4-8
Patient/Family/Investigator Interaction Opportunities	9
Scientific Poster Sessions	10-15
Become a Professional Member of the NAF	15
NAF's Research Grant Program for FY2017	16
Biographies	17-33
Meeting Attendees	34-40

All AIM conference communications, materials and abstracts are confidential.

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Welcome

March 29, 2016

Dear AIM 2016 Attendee,

Welcome to the 6th Ataxia Investigators Meeting (AIM) sponsored by the National Ataxia Foundation (NAF) in beautiful Orlando, Florida. On behalf of the organizing committee, I and Gulin Oz as co-organizers of AIM 2016 hope you find this year's meeting exciting and as stimulating as past meetings.

Please note that the format of the AIM 2016 again is designed to facilitate discussion and involvement of junior researchers. Each speaker is asked to focus on the cutting edge of their research with their time equally divided between their presentation and questions, from you, the meeting attendees. We believe that robust discussion and brainstorming among both senior and junior investigators, facilitated by the session chairs, will maximize progress toward elucidation of the ataxias and move us quicker towards viable therapeutics. We are looking to all to participate actively.

There also are opportunities to interact with patients and family members who will begin arriving this week to attend the NAF Annual Ataxia Conference later this week. We hope you will take the opportunity to introduce yourself and share your research efforts with them. It can be galvanizing to your career to hear directly from patients and families on how the disease impacts their lives and how much they appreciate your research efforts.

I look forward to an excellent meeting.

Best regards,



David R. Lynch, MD, PhD

The National Ataxia Foundation is grateful to the 2016 AIM Steering Committee members:

David Lynch, MD, PhD – Chair • Gulin Oz, PhD – Co-chair

Alexandra Durr, MD • Albert La Spada, MD • Harry Orr, PhD • Henry Paulson, MD, PhD

The 7th Ataxia Investigators Meeting will be held at the Philadelphia Downtown Marriott Monday, April 2 – Thursday, April 5, 2018. The AIM 2018 Steering Committee members include:

Gulin Oz, PhD – Chair • Albert La Spada, MD – Co-chair

Marija Cvetanovic, PhD • Patricia Maciel, PhD • Jeremy Schmahmann, MD

Vikram Shakkottai, MD, PhD • George (Chip) Wilmot, MD, PhD • Harry Orr, PhD, ex-officio

Thank You to Sponsors

The National Ataxia Foundation is grateful to the following sponsors for their generous support of the 6th Ataxia Investigators Meeting:

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Meeting Schedule

TUESDAY, MARCH 29, 2016

- 4:00-6:15 p.m. AIM Check-in and Poster Boards available to hang posters
5:15-6:15 p.m. Welcome Reception and Check-in continues
6:30 p.m. Dinner
7:15 p.m. Opening Remarks – David Lynch, MD, PhD, Children’s Hospital of Philadelphia
7:25 p.m. Welcome – Mike Parent, The National Ataxia Foundation
7:30 p.m. Keynote: Jennifer Raymond, MD, Stanford University School of Medicine
Neural learning rules in the cerebellum

WEDNESDAY, MARCH 30, 2016

Theme 1 – Cerebellar Circuitry and its role in Ataxia

- 7:30-8:30 a.m. Healthy Start Continental Breakfast
8:30-8:55 a.m. Keynote: Timothy J. Ebner, MD, PhD, University of Minnesota
*Cerebellum Acts as a Forward Internal Model and
Computes Sensory Prediction Errors*

Theme 1 Session Chair: David Lynch, MD, PhD Children’s Hospital of Philadelphia

- 9:00-9:20 a.m. *Hong Lin, PhD Children’s Hospital of Philadelphia
Cerebellar circuit deficits in a frataxin-deficient mouse model of Friedreich’s ataxia
9:25-9:45 a.m. *Abigail Person, PhD, University of Colorado School of Medicine
A feedback pathway from the cerebellar nuclei to cerebellar cortex regulates sensory gain
9:50-10:10 a.m. *Jonathan Schisler, PhD University of North Carolina at Chapel Hill
The characterization of a mouse model of SCAR16 generated with CRISPR gene editing
10:15-10:45 a.m. Morning Break
10:50-11:10 a.m. *Andreia Castro, PhD, University of Minho, Portugal
Serotonergic signaling suppresses SCA3 pathogenesis: tackling the when and how
11:15-11:35 a.m. Liliana S. Mendonça, PharmD, PhD, University of Coimbra, Portugal
*Transplantation of Neural Stem Cells as a therapeutic strategy
in Machado-Joseph disease*
11:40-noon Tycho Hoogland, PhD, The Netherlands
Shining light on aberrant Purkinje cell coding in ataxic mouse mutants
Noon-1:00 p.m. Lunch

Meeting Schedule

WEDNESDAY, MARCH 30, 2016 (continued)

Theme 2 – Biological Basis of Cerebellar Degeneration

1:00-1:25 p.m. Keynote: *Dineke Verbeek, PhD, University Medical Center Groningen
The role of opioid signalling in cerebellar neurodegeneration

Theme 2 Session Chair: Harry T. Orr, PhD, University of Minnesota, Minneapolis

1:30-1:50 p.m. Andrew P. Lieberman, MD, PhD, University of Michigan
Non-cell autonomous mechanisms in SBMA

1:55-2:15 p.m. Olaf Riess, MD, University of Tuebingen, Germany
Mechanisms in SCA3

2:20-2:40 p.m. *Jana Schmidt, PhD, University of Tuebingen, Germany
Assessing in vivo the role of ubiquitination in the pathogenesis of SCA3

2:45-3:15 p.m. Afternoon Break

3:20-3:40 p.m. *Jordi Magrane, PhD, Weill Cornell Medical College, New York
Cellular mechanisms in FRDA

3:45-4:05 p.m. * Sara Duarte-Silva, PhD, University of Minho, Portugal
TUDCA reduces neuroinflammation and improves motor symptoms in a transgenic mouse model of Machado-Joseph disease

4:10-4:30 p.m. Laura Ranum, PhD, University of Florida, Gainesville
Identification of novel spectrin, beta, non-erythrocytic 2 (SPTBN2) mutations in a large patient cohort

4:30- 4:45 p.m. “Hot Chair” Junior Poster Presenters Group A (2 minutes each)
Franziska Hoche, MD Theme 1 – A-T
Massachusetts General
Sharan Paul, PhD Theme 2 – SCA2
University of Utah
Margit Burmeister, PhD Theme 2 – Autophagy
University of Massachusetts
Swati Khare, MS Theme 3 – SCA13
University of Florida
Gautam Rajpal, PhD Theme 4 – SCA3
University of Michigan (presented by Henry Paulson, MD, PhD)
Maureen Daly, PhD Theme 5 – CCAS
Massachusetts General (presented by Jeremy Schmahmann, MD)

5:00-6:30 p.m. Scientific Poster Session A (Wine and Cheese)

6:30 p.m. Networking Dinner on your own

Meeting Schedule

THURSDAY, MARCH 31, 2016

Theme 3 – Genomic and Genetic Approaches to cerebellar function and ataxia and Epigenetics

7:30-8:30 a.m. Healthy Start Continental Breakfast

8:30-8:55 a.m. Keynote: John Hardy, PhD, University College London, Institute of Neurology
Genomic analysis of neurodegeneration

Theme 3 Session Chair: Alexandra Durr, MD, PhD, Hopital de la Salpetriere, Paris, France

9:00-9:20 a.m. *Yogesh K. Chutake, PhD, University of Oklahoma HSC
Epigenetic Silencing of the FXN gene in Friedreich ataxia

9:25-9:45 a.m. *Vikram Khurana, MBBS, PhD, Brigham and Women's Hospital
and Harvard Medical School, Boston, MA
Neurodegenerative Proteinopathies: Genome Scale investigations

9:50-10:10 a.m. * Bing Yao, PhD, Emory University, Atlanta, GA
5-hydroxymethylcytosine-mediated Epigenetic Dysregulation in Cerebellar Degeneration

10:15-10:30 a.m. "Hot Chair" Poster Presenters Group B – 2 minutes each

Matthew Scaglione, PhD Theme 2 – Poly SCAs
Medical College of Wisconsin

Vincenzo Gennarino, PhD Theme 2 – SCA1
Baylor College of Medicine

Wei-Ling Tsou, PhD Theme 3 – SCA6
Wayne State University School of Medicine

Percy Tumbale, PhD Theme 3 – AOA1
Nat'l Inst Environmental Health Sciences

Edgardo Rodriguez, PhD Theme 4 – SCA1
University of Florida

Charles Isaacs, BA Theme 5 – FRDA
Children's Hospital of Philadelphia

10:35 a.m.-noon Scientific Poster Session B (Coffee, Tea, Snack)

Noon Official Group Photo of AIM Attendees will be taken at this time

Noon-1:00 p.m. Lunch



Meeting Schedule

THURSDAY, MARCH 31, 2016 (continued)

Theme 4 – Therapeutic Strategies for ataxia: Translational Research

1:00-1:25 p.m. Keynote: Beverly Davidson, PhD, Children’s Hospital of Philadelphia
Progress in advancing RNAi for the spinocerebellar ataxias to human application

Theme 4 Session Chair: Albert LaSpada, M.D., PhD, University of California, San Diego

1:30-1:50 p.m. *Elisabetta Soragni, PhD, The Scripps Research Institute, La Jolla, CA
Identification of biomarkers for clinical trials in Friedreich’s ataxia

1:55-2:15 p.m. *Eric Wang, PhD, University of Florida, Gainesville, FL
Post-transcriptional regulation of gene expression in neuromuscular disease

2:15-2:45 p.m. Afternoon Break

2:50-3:10 p.m. *Hayley McLoughlin, PhD, University of Michigan, Ann Arbor, MI
Antisense Oligonucleotide Therapy for SCA3

3:15-3:35 p.m. *Marek Napierala, PhD, University of Alabama at Birmingham, AL
A next generation perspective on the molecular mechanism of Friedreich’s ataxia

3:40-4:00 p.m. Richard A. Gatti, MD, University of California Los Angeles School of Medicine
Opportunities for treating nonsense mutations in the ATM gene with SMRT drugs

5:15-6:15 p.m. Poster Session for Patients and Families

6:30 p.m. Dinner

7:15 p.m. The Patient/Family Perspective

Meeting Schedule

FRIDAY, APRIL 1, 2016

Theme 5 – Therapeutic strategies for ataxia: Clinical Trials, Outcome Measures and Drug Development

7:30–8:30 a.m. Healthy Start Continental Breakfast

8:30–8:55 a.m. Keynote: Merit Cudkowicz, MD, MSc, Massachusetts General Hospital, Boston
Clinical Trials in ALS and Ataxia – Lessons From Other Fields

Theme 5 Session Chair: Gulin Oz, PhD, University of Minnesota, Minneapolis, MN

9:00–9:20 a.m. Kenneth Fischbeck, MD, NINDS, National Institutes of Health
Clinical trials in neurodegenerative disease

9:25–9:45 a.m. *Fanny Mochel, MD, PhD, University Hospital La Salpêtrière, Paris
Magnetic Resonance Spectroscopy studies in Spinocerebellar Ataxia

9:45–10:15 a.m. Morning Break

10:20–10:40 a.m. *Kathrin Reetz, MD, Aachen University, Germany
Multimodal Imaging in Ataxias

10:45–11:05 a.m. Robert Wilson, MD, PhD University of Pennsylvania Medical Center
Phenotypic drug screening using random shRNA selection

11:10–11:30 a.m. *Christophe Lenglet, PhD, University of Minnesota, Minneapolis, MN
Cross-sectional and longitudinal diffusion MRI and MRS of the spinal cord in Friedreich's Ataxia

11:30–11:45 p.m. Closing Remarks – David Lynch, MD, PhD, University of Pennsylvania

AIM 2016 Adjourns

2–5 p.m. "Birds of a Feather" Sessions at the Annual Ataxia Conference
See page 9 for meeting room assignments.

Junior Investigators are welcome to attend these small groups

Patient/Family/Investigator Interaction Opportunities

Linking Ataxia Families and Investigators

The location of the 2016 Ataxia Investigators Meeting was selected so that it dovetails with the 59th 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 research, which is invigorating and hopeful for patients and families.

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

Poster Session

On Thursday, March 31, from 5:15-6:15 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.

“Birds of a Feather” Small Group Sessions

After the AIM 2016 has adjourned, patients and family members will meet in facilitated small groups to learn of the latest research, meet others and discuss the challenges of living with ataxia. New last year, the Birds of a Feather sessions were divided into groups that met either Friday or Saturday afternoon. You are very welcome to attend any session that would be of interest to you.

Below is the listing of groups meeting on Friday, March 31 from 2:00-5:00 p.m. and their locations:

- SCA 1 Bonaire 4
- SCA 2 Bonaire 5
- SCA 3 Curacao 1 & 2
- SCA 6 Curacao 6
- All other SCAs and DRPLA Curacao 3 & 4
- FRDA over age 30 Antiqua 1 & 2
- FRDA Parents Bonaire 6 & 7
- Unknown without Family History Bonaire 1, 2 & 3
(including gluten ataxia, sporadic ataxia and MSA)

Scientific Poster Sessions

Poster Numbering

The first number of the poster represents the theme; the second number of the poster is the poster number within that theme followed by the letter A or B indicating in what session the poster will be presented.

Session Times

- Poster Session A is 5–6:30 p.m. on Wednesday, March 30
- Poster Session B is 10:35 a.m. – noon on Thursday, March 31

Theme 1 Poster Presenters

- 1-1A* Hoche, Franziska, MD, Massachusetts General, Boston, MA
The Cerebellar Cognitive Affective Syndrome (CCAS) in Children with Ataxia-Telangiectasia (AT)
- 1-2B Cvetanovic, Marija, PhD, University of Minnesota, Minneapolis, MN
Deciphering cognitive deficits in SCA1
- 1-3A Doss, Sarah, MD, University Medicine Berlin, Germany
fMRI-study in a German multicenter SCA14 cohort
- 1-4B Marvel, Cherie, PhD, Johns Hopkins University, Baltimore, MD
Visuospatial Memory Can Be Predicted by Recall Strategies in Individuals with Cerebellar Ataxia
- 1-5A Sathyanesan, Aaron, PhD, Children’s National Medical Center, Washington, D.C.
Motor Learning Deficits in a Mouse Model of Premature Birth Injury
- 1-6B Sonni, Akshata, MS, University of Massachusetts Amherst, MA
Sleep Physiology in Individuals with Cerebellar Disease

Theme 2 Poster Presenters

- 2-1A* Paul, Sharan, PhD, University of Utah, Salt Lake City, UT
*Neurodegeneration and aberrant RNA metabolism:
the role of Stauf1 in spinocerebellar ataxia type 2 (SCA2) pathogenesis*
- 2-2B* Scaglione, Matthew, PhD, Medical College of Wisconsin, Milwaukee, WI
Interrogation of the Dictyostelium discoideum protein quality control network
- 2-3A* Burmeister, Margit, PhD, University of Michigan, Ann Arbor, MI
*Mutation in ATG5 leads to Ataxia with Developmental Delay –
Implications for a role of Autophagy in common Ataxias?*
- 2-4B* Gennarino, Vincenzo, PhD, Baylor College of Medicine, Houston, TX
*Brain Region Resolved Protein-Protein Interaction Network
of Spinocerebellar Ataxia Type 1 (SCA1) disease*

Scientific Poster Sessions

Theme 2 Poster Presenters (continued)

- 2-5A Ayhan, Fatma, BS, University of Florida, Gainesville, FL
Muscleblind modulates RAN protein effects in Spinocerebellar Ataxia Type 8 BAC transgenic mice
- 2-6B Basso, Manuela, PhD, University of Trento, Italy
Spinocerebellar Ataxia 35 and Mutant Transglutaminase 6 Mediated Toxicity
- 2-7A Chopra, Ravi, BS, University of Michigan, Ann Arbor, MI
*Increased dendritic excitability and calcium-dependent PKC activation:
a novel mechanism underlying Purkinje neuron dendritic degeneration in cerebellar ataxias*
- 2-8B Cohen, Rachael, DVM, Johns Hopkins University, Baltimore, MD
Molecular Pathogenesis of Spinocerebellar Ataxia Type 12
- 2-9A Dell'Orco, James, BS, University of Michigan, Ann Arbor, MI
*Altered potassium channel expression in Spinocerebellar ataxia type 2
differentially affects Purkinje neuron spiking and dendritic excitability.*
- 2-10B Diaz Garcia, Javier, PhD, Baylor University, Houston, TX
Analysis of Glucose Metabolism During Pathogenesis of Spinocerebellar ataxia type 1
- 2-11A Esteves, Sofia, PhD, University of Minho, Portugal
(Presented by Carmo Costa, Maria do, PhD)
*Motor function and selective neuronal vulnerability in transgenic mouse models
of Machado-Joseph disease*
- 2-12B Fogel, Brent, MD, PhD, University of California – Los Angeles, CA
Mutation of the Murine Ataxia Gene TRPC3 Causes Cerebellar Ataxia in Humans
- 2-13A Huryn, Laryssa, MD, National Eye Institute, National Institutes of Health
Ophthalmic Findings in Spinocerebellar Ataxia Type 7
- 2-14B Kim, Joo Hyun (Joanne), PhD, University of Minnesota, Minneapolis, MN
The role of astrocytic NF- κ B in SCA1
- 2-15A Kubota, Tomoya, PhD, University of Chicago, Chicago, IL
*A missense mutation in the selective filter of BK channel causes
cognitive impairment and progressive cerebellar ataxia*
- 2-16B Li, Pan, PhD, Johns Hopkins University, Baltimore, MD
ATXN2AS, a gene antisense to ATXN2, is associated with SCA2 and ALS
- 2-17A Nath, Siddharth, MD, McMaster University Hamilton, ON, Canada
*Using a novel spinocerebellar ataxia variant to probe the mechanisms
underlying pathology in CAG triplet repeat disorders*

Scientific Poster Sessions

Theme 2 Poster Presenters (continued)

- 2-18B Nitschke, Larissa, BS, Baylor College of Medicine, Houston, TX
Characterize the molecular and cellular consequences of Atxn1 phosphorylation in Spinocerebellar ataxia type 1 (SCA1)
- 2-19A Perez, Barbara, BS, University of Florida, Gainesville, FL
Patient-derived cell models to understand reduced penetrance of SCA8
- 2-20B Santarriaga, Stephanie, BS, Medical College of Wisconsin, Milwaukee, WI
Investigation of the proteostatic network during Dictyostelium's developmental cycle
- 2-21A Zu, Tao, MD, University of Florida, Gainesville, FL
A novel mouse model of RAN translation

Theme 3 Poster Presenters

- 3-1A* Khare, Swati, MS, University of Florida, Gainesville, FL
A neurodevelopmental, non-progressive form of SCA13 displays neuroplasticity and an association with abnormal Epidermal Growth Factor Receptor trafficking
- 3-2B* Tsou, Wei-Ling, PhD, Wayne State University School of Medicine, Detroit, MI
1ACT-dependent neurodegeneration in Drosophila models of SCA6
- 3-3A Asher, Melissa, BS, University of Minnesota, Minneapolis, MN
A role for ataxin-1 in hippocampal neurogenesis
- 3-4B* Tumbale, Percy, PhD, National Institute of Environmental Health Sciences, Research Triangle Park, NC
Molecular basis of the Aprataxin-linked Cerebellar Degeneration, Ataxia with Oculomotor Apraxia Type 1 (AOA1)
- 3-5A Bower, Matthew, MS, CGC, University of Minnesota, Minneapolis, MN
Next generation sequencing and copy number variation analysis from NGS data in an adult population with unexplained ataxia
- 3-6B Du, Xiaofei, MD, University of Chicago, Chicago, IL
The role of 1ACT in the early stage development of the cerebellum
- 3-7A Maciel, Patricia, PhD, University of Minho, Portugal
Impact of longevity-associated mutations on ATXN3 aggregation and motor impairments in a C. elegans model of Machado-Joseph disease: focus on proteostasis
- 3-8B Manek, Rachna, BS, University of Florida, Gainesville, FL
Unbiased, cell-type specific analysis of transcriptomic activity in SCA1
- 3-9A Stephen, Christopher, MD, MRCP, Harvard Medical School, Boston, MA
X-linked spinocerebellar ataxia (SCA) type 1A: a novel late onset phenotype of ATP2B3 mutation

Scientific Poster Sessions

Theme 4 Poster Presenters

- 4-1A* Rajpal, Gautam, PhD, University of Michigan, Ann Arbor, MI
(Presented by Dr. Henry Paulson)
Compound Screen to Identify Inhibitors of ATXN3 Oligomerization
- 4-2B* Rodriguez, Edgardo, PhD, University of Florida, Gainesville, FL
Cas9 editing of expanded nucleotide repeats in the adult mouse brain.
- 4-3A Pereira de Almeida, Luis, PhD University of Coimbra, Coimbra, Portugal
*Reestablishing Ataxin-2 downregulates translation of mutant ataxin-3
and alleviates Machado-Joseph disease*
- 4-4B Ashizawa, Tetsuo, MD, Methodist Hospital Research Institute, Houston, TX
Intravenously administered novel lipid nanoparticles deliver cargo to cerebellar Purkinje cells
- 4-5A Bushart, David, BS, University of Michigan, Ann Arbor, MI
*Potassium channel activators improve physiological alterations in Spinocerebellar ataxia type 1
by restoring spiking and reducing Purkinje neuron dendritic excitability*
- 4-6B Butler, Jill, PhD, University of Alabama at Birmingham, AL
Defining the molecular signature of Friedreich's ataxia to identify novel biomarkers
- 4-7A Clark, Elisia, BS, University of Pennsylvania, Philadelphia, PA
Distinct Effects of Frataxin Missense Point Mutations on Localization and Protein Processing
- 4-8B Carmo Costa, Maria do, PhD, University of Michigan, Ann Arbor, MI
B screen identifies an atypical antipsychotic as a modulator of abundance of mutant ataxin-3
- 4-9A Friedrich, Jillian, BS, University of Minnesota, Minneapolis, MN
Antisense Oligonucleotides as a Potential Therapeutic for SCA1
- 4-10B Grabczyk, Ed, PhD, LSU Health Sciences Center, New Orleans, LA
*Small molecule directed alternative splicing of mMLH3 to slow repeat expansion
in an FRDA mouse model*
- 4-11A Handler, Hillary, BS, University of Minnesota, Minneapolis, MN
Efficacy of Antisense Oligonucleotides in Reducing Purkinje Cell Ataxin 1 RNA Levels
- 4-12B Hu, Yuan-Shih (Jennifer), PhD, Northwestern University, Chicago, IL
VEGF-based nanotherapy in Spinocerebellar ataxia type 1 (SCA1)
- 4-13A Keiser, Megan, PhD, Children's Hospital of Philadelphia, PA
Translating RNAi therapy for Spinocerebellar ataxia 1 (SCA1) to the clinic
- 4-14B Koeppen, Arnulf, MD, Albany Medical College, Albany, NY
Iron, zinc, copper and calcium in Friedreich Cardiomyopathy

Scientific Poster Sessions

Theme 4 Poster Presenters (continued)

- 4-15A Lee, Won-Seok, MS, Baylor College of Medicine, Houston, TX
Elucidating the reciprocal regulation of ATXN1 and ZBTB7A to identify therapeutic targets for spinocerebellar ataxia type 1
- 4-16B Moore, Lauren, MS, University of Michigan, Ann Arbor, MI
Widespread in vivo suppression of mutant ATXN3 by antisense oligonucleotides in a transgenic mouse model of SCA3
- 4-17A Nobre, Jorge Rui, PhD, University of Coimbra, Portugal
Silencing Machado Joseph-Disease through the Systemic Route
- 4-18B Schmidt, Thorsten, PhD University of Tuebingen, Germany
Influencing the intracellular localization of ataxin-3 as treatment approach for Spinocerebellar Ataxia Type 3 (SCA3)
- 4-19A Scoles, Daniel, PhD, University of Utah, Salt Lake City, UT
Antisense oligonucleotides targeting ATXN2 for treating spinocerebellar ataxia type 2 (SCA2)
- 4-20B Silva-Fernandes, Anabela, PhD, University of Minho, Braga, Portugal
(Presented by Sara Duarte-Silva)
Creatine-supplemented diet delays disease onset and improves the overall phenotype of the CMVMJD135 mouse model of MJD
- 4-21A Sutton, Joanna, BS, Wayne State University School of Medicine, Detroit, MI
Should ubiquitin-binding site 2 of ataxin-3 be targeted for therapy in Spinocerebellar Ataxia Type 3?
- 4-22B Toonen, Lodewijk, MS, Leiden University Medical Center, The Netherlands
Ataxin-3 exon skipping as a treatment strategy for Spinocerebellar Ataxia type 3
- 4-23A Yang, Su, PhD, Emory University, Atlanta, GA
Developing the MANF-based therapeutic approach for Spinocerebellar ataxia 17

Theme 5 Poster Presenters

- 5-1A* Daly, Maureen, PhD, Massachusetts General Hospital, Boston, MA
(Presented by Dr. Jeremy Schmammann)
*The Cerebellar Neuropsychiatric Rating Scale (CNRS):
A New Measure of the Affective Component of the CCAS*
- 5-2B* Isaacs, Charles, BA, Children's Hospital of Philadelphia, PA
Progression of Friedreich ataxia: Quantitative characterization in a large cohort over a 5-year period
- 5-3A Anand, Ravi, MD, EryDel Pharmaceuticals, St. Moritz, Switzerland
Design of a prospective, international study to assess progression of neurological symptoms in patients with Ataxia Telangiectasia using validated neurological and global assessment ratings

Scientific Poster Sessions

Theme 5 Poster Presenters (continued)

- 5-4B Deelchand, Dinesh, PhD, University of Minnesota, Minneapolis, MN
Sensitivity of volumetric MRI and MRS to onset and progression of SCA1
- 5-5A Delatycki, Martin, MBBS, PhD, FRACP, Murdoch Children's Research Institute, Victoria, Australia
A longitudinal study of the SF-36 version 2 in Friedreich ataxia
- 5-6B Ravishankar, Adarsh, BS, University of Minnesota, Minneapolis, MN
Morphological Alterations of Cortical Structures in SCA1
- 5-7A Soong, Bing-Wen, MD, PhD, National Yang-Ming University, Taiwan
Intravenous Administration of Allogeneic Mesenchymal Stem Cells in Patients with Cerebellar Ataxias: A Phase I/IIa Clinical Trial
- 5-8B Vogel, Adam, PhD, University of Melbourne, Australia
Two pilot therapies of intensive home based speech rehabilitation for adults with degenerative ataxia
- 5-9A Kempainen, Jennifer MS, CGC, Mayo Clinic, Rochester, MN
Clinical Features of Cardiomyopathy in Patients with Friedreich Ataxia: Echocardiographic Review and Multivariate Pattern Recognition
- 5-10B Morgan, Jeremy, MS, CoRDS, Sanford Research, Sioux Falls, SD
A Patient Registry for all Rare Diseases

All AIM conference communications, materials and abstracts are confidential.

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 to become a member and support the important work of the Foundation which includes funding ataxia research.

NAF's Research Grant Program for FY 2017

For new and innovative studies that are relevant to the cause, pathogenesis or treatment of ataxia, both hereditary and non-hereditary. More information and grant applications are available on the National Ataxia Foundation website

Research Seed-Money One-Year Grant

Seed monies up to \$15,000 but promising proposals up to \$30,000 will be considered in early or pilot phases of studies and ongoing investigations that demonstrate a need to attract future funding from other sources. Letter of intent due July 15, 2016. Application due August 15, 2016.

Young Investigator (YI) \$35,000 One-Year Award

Young Investigator Award for SCA Research (YI-SCA) \$50,000 One-Year Award

To encourage young investigators to pursue a career in ataxia research or spinocerebellar ataxia research. Must have MD or PhD and have appointment as a junior faculty member. Letter of intent due August 1, 2016. Application due September 1, 2016.

Post-Doc Fellowship Award \$35,000 One-Year Award

Applicants should have completed at least one year of post-doctoral training, but not more than two at the time of application, and have a commitment to ataxia research. Letter of intent due August 15, 2016. Application due September 15, 2016.

Pioneer SCA Translational Research Award \$100,000 One-Year Award

This grant is intended for research investigations that will facilitate the development of treatments for the spinocerebellar ataxias (SCAs). Letter of intent due August 15, 2016. Application due September 15, 2016.

Research Training Fellowship in Ataxia Award

A two-year award funded in partnership with NAF and the AAN and American Brain Foundation. The award provides salary and tuition to support formal education in research methodology in ataxia. Alternatives to clinical and translational proposals (more basic proposals) are encouraged if the primary goal of those proposals is to support the development of tools to facilitate the development of treatments for the ataxias. The two-year award will consist of an annual salary of \$55,000 plus \$10,000 per year for tuition. A letter of intent is not required. Application due October 1, 2016.

Biographies



Margit Burmeister, Ph.D., is a Research Professor at the Molecular & Behavioral Neuroscience Institute and Vice Chair and Professor of Computational Medicine & Bioinformatics as well as a Professor of Psychiatry and Human Genetics at the University of Michigan. She received her Ph.D. in Biology from the European Molecular Biology Laboratory and the University of Heidelberg and did postdoc training at the University of California San Francisco, where she co-invented Radiation Hybrid mapping with David R. Cox and Richard M. Myers, a technique that was important during the early Human Genome Project.

Using an integrative genomic approach, she has identified several ataxia genes, as well as other rare neurological disease genes. She also investigates gene x environment interaction in addictions and depression. Currently, she is interested in further investigating the involvement of autophagy in ataxia.



David Bushart – I am interested in studying how altered neuronal membrane excitability contributes to neurological diseases at both the single-cell and network levels. There is growing evidence that altered cerebellar Purkinje neuron membrane excitability may contribute to early symptoms in degenerative ataxias, and I am therefore particularly interested in whether restoring normal Purkinje neuron membrane excitability may be an effective strategy for the symptomatic treatment of cerebellar ataxias. In my current project, I am investigating how altered ion channel expression influences membrane excitability in a mouse model of Spinocerebellar ataxia type 1, the ATXN1[82Q] model. I have determined that modulators of calcium-activated potassium channels and subthreshold-activated potassium channels, when applied simultaneously, can restore normal firing to Purkinje neurons in acute cerebellar sections from ATXN1[82Q] mice. Ongoing studies will investigate the interaction between dendritic and somatic Purkinje neuron excitability, dendritic integrity, and motor function in ATXN1[82Q] mice. My long-term research interests include the identification of novel ion channel modulators which improve neuronal function in neurodegenerative diseases. I am currently a graduate student in the Department of Molecular and Integrative Physiology at the University of Michigan.



Andreia Castro is a post-doctoral fellow researcher in the laboratory of Professor Patrícia Maciel at School of Health Sciences, 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 (2007) and a PhD in Health Sciences from University of Minho, Portugal (2011). 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

Biographies

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.



Yogesh Chutake received his Bachelor's in Pharmacy degree from the University of Pune (India) and a Master of Technology degree from the University of Mumbai (India). After receiving his PhD in Biochemistry & Molecular Biology from the University of Oklahoma Health Sciences Center he is currently doing his postdoctoral training in the Bidichandani lab in the Department of Pediatrics, Section of Genetics at the University of Oklahoma Health Sciences Center. His research is centered on investigating the mechanism of the epigenetic defect in Friedreich ataxia, with the goal of testing and developing therapeutic strategies that reverse this epigenetic defect. His ongoing research is supported by the Translational Neuroscience Research program of the Oklahoma Center for Neuroscience, and a grant from the Million Dollar Bike Ride (MDBR) program of the Orphan Disease Center at the University of Pennsylvania.



Dr. Merit Cudkowicz is the Chief of the Massachusetts General Hospital Neurology Service and the Julieanne Dorn Professor of Neurology at Harvard Medical School in Boston.

Dr. Cudkowicz's research and clinical activities are dedicated to the study and treatment of patients with amyotrophic lateral sclerosis (ALS). She serves as director of the Massachusetts General Hospital ALS Clinic and the Massachusetts General Hospital Neurological Clinical Research Institute. She is one of the founders and co-directors of the Northeast ALS Consortium, a group of more than 100 clinical sites in the United States and Canada dedicated to performing collaborative, academic-led clinical ALS trials.

She is Principal Investigator of the Clinical Coordination Center for the National Institute of Neurological Disorders and Stroke's Neurology Network of Excellence in Clinical Trials (NeuroNEXT). She is currently leading efforts at 25 different centers that conduct high-impact Phase 2 trials in neurology, hoping to speed trials by seven months to a year. The NeuroNEXT network established an academic central internal review board led by Massachusetts General and serves as a system for efficient study start-ups.

A dedicated educator, Dr. Cudkowicz mentors many young neurologists in clinical investigation of ALS and related neurodegenerative disorders. She also serves on the medical advisory board for the Muscular Dystrophy Association and the Massachusetts Amyotrophic Lateral Sclerosis Association.

Dr. Cudkowicz completed her medical degree in the Health Science and Technology program of Harvard Medical School. She served her internship at Beth Israel Hospital in New York and her neurology residency and fellowship at Massachusetts General. She also obtained a master's degree in Clinical Epidemiology from the Harvard School of Public Health. She is fluent in Italian.

Biographies



Javier Díaz-García – Baylor College of Medicine and Jan and Dan Duncan Neurological Research Institute (Texas Children’s Hospital). I was born in the south-west of Spain, in a region called Extremadura. I moved to Madrid when I was 18 years old and went to the university. I obtained my bachelor’s degree in Biology in the Autonomous University of Madrid. One year before finishing my bachelor, I firstly started to work with neurological diseases. After I graduated, I came to U.S. to work with Polyglutamine Diseases at Baylor College of Medicine (BCM) in Dr. Botas Lab. One year later, I went back to Madrid to study a Master Degree in Cell Biology and Genetics. In 2011, I came back to Baylor (US) to follow my research in SCA1, SCA2 and SCA7. I joined to an International PhD Students Program at the Autonomous University of Madrid, doing all my training at BCM and Jan and Dan Duncan Neurological Research Institute (NRI) (Texas Children’s Hospital). I became a Doctor in February 2016. Currently, I am continuing my studies with Ataxias as a postdoc at BCM in Dr. Botas Lab at BCM-NRI.



Beverly L. Davidson is the Director of The Raymond G. Perelman Center for Cellular and Molecular Therapeutics and holds the Arthur V. Meigs Chair in Pediatrics at The Children’s Hospital of Philadelphia. She is also Professor in the Department of Pathology and Laboratory Medicine, Perelman School of Medicine, University of Pennsylvania, Philadelphia, Pennsylvania.

Dr. Davidson received her Ph.D. in Biological Chemistry from the University of Michigan and in 1994 was recruited to the University of Iowa where she was promoted to Associate Professor in 1999 and Professor in 2001. From 1999–2014 she held the Roy J. Carver Chair in Biomedical Research, and was named Vice Chair for Research, Internal Medicine from 2004–2014. Professor Davidson’s research is focused on inherited brain disorders and the development of novel therapies to treat these fatal diseases. In 2007, she was named a Fellow by the American Association for the Advancement of Science, received a University of Iowa Regents Award for Faculty Excellence, and was named a University of Iowa Carver Research Program of Excellence (through 2014). In 2008 she was an Iowa Women of Innovation Nominee for Research Innovation and Leadership. In 2009, Dr. Davidson received the Mathilde Solowey Award, NIH, and was named a Member, Electorate Nominating Committee, Medical Sciences, AAAS. In 2011, Dr. Davidson was the S.J. Armond Lecturer for the AANP, and the University of Iowa Presidential Lecturer. In 2012 she received the Carver College of Medicine Faculty Service Award, along with the University of Iowa Innovator Award. In 2012 she was elected to the Advisory Council for the ASGCT, and in 2014 she was voted Chair, Medical Sciences Committee for the AAAS. She has served as Co-Chair of the TAG study section (NIH) and co-chair of the Editors Panel, Transformative Award Review Committee from the Office of the Director (NIH), and in 2014 was asked to serve on the Blue Ribbon Panel Review. In 2014 she was appointed to the National Advisory Council of the NINDS. In 2015 Dr. Davidson was appointed a member of the Scientific Advisory Board of the Huntington Study Group and the Medical Research Advisory Board of the National Ataxia Foundation, and was awarded the Leslie Gehry Brenner Prize for Innovation in Science.

Biographies

Dr. Davidson is a co-founder of Spark Therapeutics, Inc., and serves on the advisory board of Serepta Therapeutics, Wave Life Sciences and Marina Biotech.



Sara Duarte-Silva is a post-doctoral researcher in the laboratory of Prof. Patrícia Maciel at the Life and Health Sciences Research Institute (ICVS), University of Minho, Braga, Portugal. She holds a B.Sc. in Applied Biology (2006), and an MSc. in Molecular Genetics (2011) from the University of Minho, and a PhD in Health Sciences from the Inter-University Doctoral Programme in Ageing and Chronic Disease (2015). Her main research interest is the search of new therapeutic strategies for Machado-Joseph disease (MJD). During her graduated studies, Sara contributed to the development and characterization of two MJD transgenic mouse models one of which was shown to closely mimic the human condition, both at phenotypic and pathologic levels. Sara's main interest in the search for new therapeutic strategies for MJD, for which she has been performing preclinical trials using the CMVMJD135 mouse model. Using pharmacological approaches targeting proteostasis, she identified some compounds that might be helpful for the treatment of MJD and she also validated the CMVMJD135 mouse model as a valuable tool for preclinical trials. These studies allow, concomitantly to the drug testing, the study of possible pathogenic mechanisms that might be involved in the disease, including autophagy, mitochondrial abnormalities, molecular chaperones and ER stress. One of her recent challenges is to study the role of glia in the pathogenesis of MJD.



Alexandra Durr is a full professor for medical genetics, specialized in neurogenetics in in the Genetic Department at the University Hospital Salpêtrière in Paris-France. After her medical degree obtained 1992 in Germany, University of Ulm, she joined the INSERM (Institut national de la santé et de la recherche médicale) research laboratory on experimental therapy in neuro-degenerative disorders at the Salpêtrière University Hospital in Paris in the Brain and Spine Institute (ICM icm-institute.org). Following her neurological degree, she obtained her PhD in medical genetics in 1998 at the University Denis Diderot Paris VII, in 2009 the Habilitation to supervise research at the University Pierre et Marie Curie Paris VI. Her research interests are genotype-phenotype correlation and understanding the variable clinical expression and therapeutic approaches of genetic disorders focused on cerebellar ataxias, spastic paraplegias and inherited conditions of other movement disorders. She opened the first predictive testing facility for late, adult-onset neurogenetic disorders in France. She is a member of Scientific Advisory Board of INSERM (National Institute of Health and Medical research in France), Scientific Board of the Fondation pour la Recherche Médicale, and coordinator of the International Network SPATAX (SPAstic paraplegias and cerebellar ATAXias (<http://spatax.wordpress.com>)). She is member of the executive committee of the Ataxia Study Group (www.ataxia-study-group.net), and president of the Medical and Paramedical Council for the lay organizations CSC (Connaître les syndromes cérébelleux), AFAF (Association Française de l'Ataxie de Friedreich) and ASL (Spastic paraplegias).

Biographies



Timothy J. Ebner earned his M.D./Ph.D. at the University of Minnesota, completing his degrees in 1979. He began his career in the Department of Neurosurgery in 1979 and became Director of the Neurosurgery Laboratory in 1984. Dr. Ebner served as the Director of the Graduate Program in Neuroscience from 1995–1998 and has served as the Head of the Department of Neuroscience since its inception in 1998. He presently holds the Max E. and Mary LaDue Pickworth Endowed Chair in Neuroscience. He also chairs the MnDRIVE Brain Conditions Steering Committee that oversees a major state investment in neuromodulation for brain disorders.

Dr. Ebner has published extensively on how information is represented spatially and temporally in populations of neurons in the cerebellum and motor cortex. Using optical imaging and electrophysiology techniques, he has investigated properties of the cerebellar circuitry. He also uses these techniques to examine abnormalities in the cerebellar cortex in animal models of the spinocerebellar and episodic ataxias. He has a long history of federally funded research grants and training programs.



Dr. Kenneth Fischbeck received A.B. and A.M. degrees from Harvard University and an M.D. degree from Johns Hopkins. After a medical internship at Case Western Reserve University and a neurology residency at the University of California in San Francisco, he did postdoctoral research on muscular dystrophy at the University of Pennsylvania. In 1982 he joined the faculty in the Neurology Department at the University of Pennsylvania Medical School. In 1998 he came to the NINDS as Chief of the Neurogenetics Branch. He received the Cotzias Award from the American Academy of Neurology and the Jacoby Award from the American Neurological

Association, and he was elected to the Institute of Medicine. His research group is identifying the causes and studying the mechanisms of hereditary neurological and neuromuscular diseases with the goal of developing effective treatment for these disorders.



Vincenzo A. Gennarino studied molecular biology at the University of Palermo and graduated with Laude in 2005. In 2009, he received a PhD in Medical Genetics from TIGEM. During his PhD studies he became interested in how microRNAs regulate gene targets in humans and he introduced a new paradigmatic tools to infer the biological pathways for each human miRNA based on the co-expression of its targets. Those tools, HOCTAR and CoMeTa (*Genome Research*, 2009 and 2012), were later used to identify miR-128 in controlling TFEB (*Science* 2009), and miR-483-5p in regulating MECP2 levels during human fetal development (*Genes & Development*, 2013).

In the lab of Dr. Huda Zoghbi he sought to understand how post-transcriptional modification of the gene, ATXN1, causing SCA1 could impact the disease onset. He discovered that PUM1 negatively regulates ATXN1 and that a modest increase of wild-type Atxn1 levels is enough to cause cerebellar degeneration similar to SCA1 disease (*Cell*, 2015). Recently, he identified NUDT21 as a new neuropsychiatric disease gene by regulating the levels of MeCP2 through alternative

Biographies

polyadenylation (*eLife*, 2015). Now he is working on identifying a brain region resolved protein-protein interaction network of SCA1 disease.



Richard A. Gatti, MD – UCLA David Geffen School of Medicine, Department of Pathology & Laboratory Medicine and Human Genetics, Distinguished Professor Emeritus, Los Angeles 90095-1732. For the past 40 years, my laboratory has studied the pathogenesis of primary immunodeficiency diseases, cancer susceptibility, and radiosensitivity. Twenty years ago, we hypothesized that by isolating and characterizing the gene responsible for ataxia-telangiectasia (ATM), we would at once gain understanding about these relationships and possibly uncover new therapeutic approaches for cancer and primary immunodeficiencies. These efforts were fruitful, opening up new mechanistic connections between cell signaling and DNA repair of double strand breaks, the role of ATM in tumorigenesis, and cancer therapy that employs “synthetic lethality.” a clinical strategy of blocking both homologous DNA repair and non-homologous end joining pathways to make a cancer more sensitive to chemotherapeutic agents. We have also come to appreciate that A-T is not only a prototype for genomic instability, but also represents an overarching syndrome of XCIND (X-ray sensitivity, cancer susceptibility, immunodeficiency, neuropathology, double strand break deficiency). This has revealed that many forms of severe immunodeficiency are also radiosensitive. To address these issues, we have developed several large panels of transformed LCLs that have helped to define: 1) the full spectrum of mutations in the ATM gene, 2) those ATM mutations associated with breast cancer, 3) radiosensitive LCLs of other diseases that are reagents for defining the cellular biomarkers and genes underlying ionizing radiosensitivity, and 4) LCLs that reveal a new class of small molecule read through (SMRT) drug-like compounds that restore ATM protein to cells carrying primary nonsense mutations in the ATM gene, the topic of today’s presentation. Many aspects of the latter project have far-reaching translational potential for treating rare genetic diseases.



Franziska Hoche, M.D. is a postdoctoral fellow in the Ataxia Unit and Jeremy D. Schmahmann’s Laboratory for Neuroanatomy and Cerebellar Neurobiology at the Massachusetts General Hospital (MGH) Department of Neurology. Dr. Hoche received her MD at the Goethe – University in Frankfurt, Germany, where she completed her doctoral thesis on neurodegeneration of auditory brainstem nuclei and fiber tracts in spinocerebellar ataxia under Professor Udo Rüb.

Dr. Hoche’s research focuses on understanding the influences and contributors to motor and cognitive dysfunction in cerebellar disorders in children and adults. She has a particular interest and expertise in ataxia-telangiectasia (AT), a neurodegenerative, hereditary cerebellar disease of childhood. Current studies are focused on the cerebellar cognitive affective syndrome (CCAS) in adults and children including the development of a CCAS rating scale, the adaptation of the Brief Ataxia Rating Scale for children, and the development of cognitive training tools for children with the CCAS.

Biographies

Dr. Hoche has received a number of awards for her work on the cerebellar role in cognition, including the young investigator award of the Neuroscientific Society in Frankfurt, Germany (2011), clinical and scientific fellowship awards from the German Neuropediatric Society (2012, 2013), a travel award from the American Neurological Association (2014), and the Young Investigator Award of the German Neuropediatric Society (2015). She has been funded by the National Ataxia Foundation and the Ataxia Telangiectasia Children's Project, as well as MGH's MINDlink Foundation.



Tycho Hoogland is Assistant Professor in the Department of Neuroscience at Erasmus Medical Center in Rotterdam. He has worked with Chris de Zeeuw at the Netherlands Institute for Neuroscience and Erasmus MC leading projects that looked into the role of complex spike coherence in locomotion and reflexive movements. One present research focus is the functional connectivity between cerebellum and cerebral cortex and how these regions interact to execute properly timed and coordinated movements. Another how aberrant coding in the olivo-cerebellar circuit of ataxic and dystonic mouse mutants affects motor behavior. To this end complex spike dynamics is investigated in cerebellar microzones using two-photon microscopy in head-fixed mice during concurrent behavioral phenotyping on a transparent disk treadmill. Miniaturized head-mounted microscopes are currently in development to perform high-throughput screening of olivo-cerebellar circuit dysfunction in freely moving ataxic mouse mutants on the ErasmusLadder, a device that allows quantification of motor performance and motor learning. Using this approach, our ultimate goal is to test interventions that could rescue olivo-cerebellar circuit and motor function.



Charles Isaacs is a research analyst for Dr. David Lynch and a certified phlebotomy technician at the Center of Excellence at the Children's Hospital of Philadelphia. He holds a Bachelor's Degree in Philosophy from the University of Pennsylvania. Research focus includes predictors of progression in Friedreich ataxia, features that characterize vision and listening impairments in Friedreich ataxia, and the role of single nucleotide polymorphisms and gene expression levels.



Megan S. Keiser currently holds the title of Project Manager, Translational Neuroscience for the Raymond G. Perelman Center for Cellular and Molecular Therapeutics at The Children's Hospital of Philadelphia. She obtained his doctoral degree in neuroscience from the University of Iowa in the laboratory of Dr. Beverly L. Davidson, where she performed her early work in viral-based RNA interference as a therapy for Spinocerebellar Ataxia type 1 in mouse models. Following the completion of her graduate work in 2013, Megan began her post-doctoral appointment in the Davidson lab. She focused her research on translating the RNAi-based therapy

Biographies

from mice to non-human primates. Her current research focuses on the clinical translation of RNAi-based therapies in Spinocerebellar Ataxias and Huntington's Disease.



Swati Khare is a first year graduate student under the mentorship of Dr. Michael F. Waters at the University of Florida, Gainesville. She completed her Bachelor's degree in Biotechnology from India, and holds a Master's degree in Biomedical Engineering from the University of Florida. During her Master's degree she worked on a *Drosophila* model for Alzheimer's disease after which she transitioned into Dr. Waters' lab to characterize a *Drosophila* model for Spinocerebellar Ataxia 13. She has since then been exploring cell and mammalian models of SCA13.



Vikram Khurana is on the faculty at Harvard Medical School and Massachusetts General Hospital, and a co-founder of Yumanity Therapeutics. His clinical and research interests relate to neurodegenerative diseases, including Parkinson's disease, multiple system atrophy (MSA) and the cerebellar ataxias. He is a First Class Honors medical graduate of the University of Sydney, Australia, and came to Boston as a Fulbright Scholar and American Australian Association Merck Company Foundation Fellow in 2001, obtaining his Ph.D. in neurobiology from Harvard University in 2006. He completed his residency and Fellowship in Neurology at Brigham and Women's and Massachusetts General Hospitals in Boston. His current research uses stem cells to investigate and develop treatments for neurodegenerative diseases. In 2013, he led a study that succeeded in identifying and reversing pathologies in stem cell models derived from Parkinson's disease patients (Chung^{*}, Khurana^{*} et al. *Science* 2013). In 2014, the MSA Coalition awarded him an inaugural grant to apply similar methods to MSA.



Dr. Pan Li is currently a postdoctoral fellow in Department of Psychiatry at Johns Hopkins University School of Medicine. Her research interest is in RNA neurotoxicity and bidirectional transcription in neurodegenerative and neuropsychiatric diseases.

Dr. Andrew Lieberman is the Gerald D. Abrams Professor in the Department of Pathology and Director of Neuropathology at the University of Michigan. Dr. Lieberman received his BS from Duke University and his MD, PhD from the University of Maryland Medical School. He completed residency training in Anatomic Pathology and fellowship training in Neuropathology at the University of

Biographies



Pennsylvania. He trained as a research fellow with Dr. Kenneth Fischbeck at the Neurogenetics Branch NINDS, NIH, and then joined the University of Michigan Medical School faculty in 2001. Dr. Lieberman's research has focused on the mechanism of neurodegeneration in inherited neurological disorders. His laboratory uses cell culture and mouse models to explore the pathogenesis of Kennedy's disease, a polyglutamine expansion disorder, and Niemann-Pick C, a lysosomal storage disease resulting from impaired lipid trafficking.



Dr. Hong Lin received her Ph.D. in Physiology from Chinese Academy of Medical Sciences and obtained postdoctoral training in Molecular Neurobiology and Neuropathology with Dr. William Schlaepfer at the University of Pennsylvania Perelman School of Medicine. Since 2001, Dr. Lin has been studying the role of protein aggregation in the pathogenic mechanisms of neurodegenerative diseases in order to identify new therapeutic targets. In 2008, she joined Dr. David Lynch's lab at the Children's Hospital of Philadelphia. Over the past 18 years, the Lynch lab has developed an emerging research portfolio for understanding, treating and eventually curing the rare neurodegenerative disease Friedreich Ataxia (FRDA). Beginning with clinical research studies, they have moved into translational studies biomarker studies and now basic science. Using the FRDA mouse models, Dr. Lin focuses on understanding cerebellar synaptic deficits in FRDA and development of therapies for this disease.



David R. Lynch, M.D. Ph.D., received his undergraduate training at Yale College, followed by medical school and Graduate School in Neuroscience at Johns Hopkins University School of Medicine. His PhD these was from the laboratory of Dr. Solomon H. Snyder on mechanisms of endogenous opioid processing. He completed residency in Neurology at the Hospital of the University of Pennsylvania, and joined the faculty at the University of Pennsylvania in 1995. His research interests include Friedreich ataxia, and glutamatergic neurotransmission. He was promoted to Associate Professor in 2004 and Full Professor in 2011.



Jordi Magrané, Ph.D. – Weill Cornell Medicine, Feil Family Brain and Mind Research Institute. I am a cell biologist with an interest in mechanisms of neurodegeneration. I am intrigued by the involvement of mitochondria in neurodegenerative diseases and, more specifically, in how impaired mitochondrial function and abnormal mitochondrial axonal transport cause loss of proprioception in Friedreich's Ataxia. Over the last few years, I have dedicated my efforts toward to developing novel microscopy-based, live imaging assays to assess the role of mitochondrial dynamics in neurodegenerative disease pathogenesis, both in vitro (isolated neurons) and in vivo

Biographies

(whole mouse). My research lab is using pharmacological and genetic approaches in isolated sensory neurons and in vivo models of Friedreich's Ataxia to dissect the molecular mechanisms of sensory loss and to evaluate therapeutic strategies to prevent or reverse the identified disease phenotypes.



Liliana Simões Mendonça is a senior Post-Doc in the Vectors and Gene Therapy Group of the Center for Neuroscience and Cell Biology (CNC) of the University of Coimbra, Portugal. She has a Ph.D. in Pharmaceutical Technology and a Pharm. D both from University of Coimbra. She has been working in gene therapy approaches using viral and non-viral vectors and in the neuroprotective and neuroregenerative potential of neural stem cells (adult and induced pluripotent stem cells-derived) transplantation in Machado Joseph Disease (MJD). Moreover, she has been assessing the potential neuroprotective role of anti-inflammatory therapy in MJD. She has several publications including a patent and papers in peer-reviewed journals and two Research Seed Money Awards from National Ataxia Foundation.



Fanny Mochel, MD, PhD is an associate professor of genetics at the University Pierre and Marie Curie (UPMC). She received her MD in Genetics in 2005 at the University Paris Descartes, her PhD in Neuroscience in 2010 at UPMC and is board certified in inborn errors of metabolism. Dr Mochel runs a neurometabolic clinic at La Pitié-Salpêtrière university hospital and is the head of the Neurometabolic research group. She is co-chair of the French society for inborn of errors of metabolism in adults and a scientific board member of the Fondation Lejeune. Her research is focused on the characterization and treatment of brain energy deficiencies in neurodegenerative diseases, especially polyglutamine disease. Her major areas of expertise are the identification of neurometabolic biomarkers in vitro (metabolomics) and in vivo (nuclear magnetic resonance spectroscopy) as well as therapeutic approaches targeting the Krebs cycle.



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.

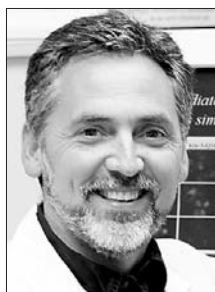
Biographies



Dr. Gulin 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.



Dr. Sharan Paul received his PhD in Biochemistry and Molecular Biology in 2001 from the University of Gunma, Japan. From 2001-2009, he worked with Dr. Sita Reddy at University of Southern California in Los Angeles, where he used mouse genetics and biochemical approaches to understand the molecular basis of myotonic dystrophy, DM1, a multi-symptom disorder characterized by a wide range of muscle and neurological defects. Currently, in Dr. Stefan Pulst's laboratory at the University of Utah, he is studying Spinocerebellar Ataxia Type 2 (SCA2), which is an autosomal dominant disorder characterized by progressive degeneration of cerebellar Purkinje cells and other neurons caused by expansion of the glutamine tract in the ATXN2 protein. Using biochemical and stem cell approaches, he focuses on understanding the molecular basis of SCA2 and on developing therapies for this disease. Dr. Paul is a member of the American Academy of Neurology.



Henry L. Paulson, MD, PhD is the Lucile Groff Professor of Neurology at the University of Michigan (UM), where he co-directs the UM Protein Folding Diseases Initiative and directs the Michigan Alzheimer's Disease Center. He received his MD and PhD from Yale University in 1990 and completed neurology residency and neurogenetics/movement disorders fellowships at the University of Pennsylvania in 1997. After a decade on the faculty at University of Iowa, he joined the Neurology faculty at Michigan in 2007, where he has spearheaded rapid growth in the study of the ataxias and other degenerative brain diseases. Dr. Paulson's research and clinical interests concern the causes and treatment of age-related neurodegenerative diseases, with a focus on hereditary ataxias and Alzheimer's disease. Using test tube, cell-based and animal models his lab has contributed to advances in the understanding of various neurodegenerative diseases, with a particular focus on Spinocerebellar Ataxia type 3 (SCA3). Efforts in his lab increasingly are seeking to find treatment for SCA3 and related ataxias. Nationally, Dr. Paulson has directed ataxia courses at the annual American Academy of Neurology meeting, has served on the scientific advisory boards of numerous disease-related organizations including the National Ataxia Foundation, and is the past Chairperson of the Board of Scientific Counselors at the National Institute for Neurological Disorders and Stroke at

Biographies

the National Institutes of Health. Among his awards, Dr. Paulson is a past Ellison Medical Foundation New Scholar in Aging, semifinalist for the W.M. Keck Foundation Young Scholars in Medical Research, and recipient of the Paul Beeson Physician Faculty Scholar in Aging Award from the American Federation for Aging Research.



Laura Ranum, PhD received her PhD from the University of Minnesota in 1989 and did her postdoctoral work with Dr. Harry Orr on the identification and characterization of the SCA1 gene (1989-1994). Dr. Ranum is currently the Director of the Center for NeuroGenetics and a Professor of Molecular Genetics and Microbiology in the College of Medicine at the University of Florida. Dr. Ranum's group has focused on the identification and characterization of genes that cause ataxia and muscular dystrophy and has mapped and identified the genes for SCA5, SCA8 and myotonic dystrophy type 2. Current efforts are focused on characterizing mouse models to better understand these diseases and to improve mapping and genetic screening strategies to allow the identification of disease genes from small families. Dr. Ranum is a member of NAF's Board of Directors and Medical Research Advisory Board and serves as a reviewer for numerous scientific journals and funding agencies including the National Institutes of Health.



Dr. Jennifer Raymond is an Associate Professor of Neurobiology at Stanford University. She earned a BA in Mathematics from Williams College and a PhD in Neuroscience from the University of Texas Health Science Center at Houston. She completed postdoctoral training with Dr. Steve Lisberger at the University of California, San Francisco. Her laboratory is working to understand the algorithm used by the cerebellum to implement motor skill learning. Her research approach integrates the use of tightly controlled behavioral paradigms, in vivo and in vitro physiology, computational methods, and molecular-genetic techniques in mice. She is an award-winning teacher, and has worked on best practices in scientific training at the national level. She also has served as an Associate Dean for Faculty Development and Diversity at the Stanford School of Medicine, and her efforts to promote diversity in the scientific workforce have been reported extensively in both scholarly journals and international news outlets.



Kathrin Reetz, M.D. is a clinical scientist at the RWTH Aachen University. She completed her medical and doctoral studies at the University in Cologne in Germany in 2004 (summa cum laude). She started her residency at the Department of Neurology at the University Hospital in Lübeck in Germany. Dr. Reetz was awarded with Postdoctoral Fellowships at the Institute for Systemic Neurosciences at the Medical University Hamburg-Eppendorf, Hamburg, Germany (2006) and at the Center for Neurosciences, Feinstein Institute for Medical Research, North Shore University Hospital, New York, USA (2007-2009). In 2009 she became a junior-JARA-BRAIN

Biographies

professorship for a clinical scientist position at the Department of Neurology at the RWTH Aachen University and Research Centre Jülich. Dr. Reetz became an Assistant Professor at the Department of Neurology, RWTH Aachen University (2014). Her clinical and research interest focus is on (rare) neurodegenerative diseases and the identification of disease-specific neuroimaging biomarkers, through innovative neuroimaging methods and their correlations with clinical and genetic parameters to better understand the pathophysiology of neurodegenerative diseases and enable patients a better risk and progression prognosis.



Olaf Riess, MD, is full professor for Medical Genetics, director of the Institute of Medical Genetics and Applied Genomics, and founder and acting director of the Rare Disease Center Tübingen. He has more than 20 years of experience in clinical genetics and research of genetically caused disorders. Main focus on neurodegenerative and syndromal diseases; both from the clinical as well as from the basic research perspective. Special focus on genetically inherited movement disorders such as ataxias, Huntington's disease, Parkinson's disease, and dystonia, and on the application of genomic HTP technology in the clinical practise. To faster transfer genomic medicine into university settings he is co-founder of the Center for personalized medicine of the University of Tübingen. His group is also well known for genetically modified rat models of neurodegenerative diseases and for preclinical studies. Current research approaches also include rare cancer and the development of biomarkers for rare diseases. Olaf Riess is founder and owner of the SME "Genes and Therapy GmbH" with the focus to accelerate personalized targeted treatment in cancer. He currently is and has been coordinator of numerous international, European and national funded consortia such as EUROSCA, MEFOPA, TECHGENE, RATstream and Neuromics. OR serves in numerous advisory boards such as the German initiative for Rare Diseases (NAMSE), the EFSN task force on spinocerebellar ataxias, the executive member of the Ataxia study group (ASG), as a board member of the study section Neuroscience (Fachgutachter) of the German Research Foundation (DFG), as associated Member of the Commission on genetic diagnostics (Gendiagnostik-Kommission) of the Ministry of Health (BfG), or as a board member of the International Rare Disease Research Consortium IRDiRC (Diagnostics Scientific Committee). He was recently elected as President-elect of the European Society of Human Genetics (ESHG). Published over 350 papers.



Matthew Scaglione – My laboratories research interests focus on understanding how protein quality control pathways recognize and handle proteotoxic species. My laboratory has two distinct, but related projects. The first project investigates how protein quality control pathways determine to refold or degrade misfolded proteins in neurodegenerative diseases. These studies focus on understanding how the ubiquitin ligase CHIP (C-terminus of Hsc70 Interacting Protein) coordinates with chaperones to target misfolded proteins for degradation. Additionally, mutations in CHIP cause autosomal recessive spinocerebellar ataxia16 (SCAR16). We are currently investigating

Biographies

how these mutations disrupt CHIP function. The second project investigates protein quality control pathways in *Dictyostelium discoideum*, an organism that normally expresses ~10,000 homopolymeric amino acid tracts, including ~2,500 polyglutamine tracts. We have recently identified as being highly resistant to polyglutamine aggregation, and are currently investigating mechanisms utilized by *Dictyostelium* to suppress protein aggregation.



Jonathan Cummings Schisler – I am an assistant professor in the Department of Pharmacology and McAllister Heart Institute. The focus of my lab is on reverse translational approaches for diseases involving the dysfunction in protein quality control pathways. I was the co-first author of the research paper that identified the causal gene of spinocerebellar autosomal recessive 16 and my lab is keenly interested in how perturbations in protein homeostasis play a role in cerebellar ataxias and related phenotypes.



Jeremy D. Schmahmann, MD is the Founding Director (1994) of the Ataxia Unit at the Massachusetts General Hospital, Director of the Laboratory for Neuroanatomy and Cerebellar Neurobiology, and Professor of Neurology at Harvard Medical School. Together with collaborators across departments, institutions and countries, he uses anatomical tract tracing tract studies in animals, morphometric, tractography and functional MRI in animal models and humans, and the tools of clinical neurology and cognitive neuroscience to explore the anatomical substrates of intellect and emotion. A central focus of the lab is the clinical neurology and basic science of the ataxias and other cerebellar disorders. Dr. Schmahmann pioneered the role of the cerebellum in cognition and emotion, described the Cerebellar Cognitive Affective Syndrome (CCAS), introduced the dysmetria of thought theory, developed the Brief Ataxia Rating Scale, and has now introduced the CCAS-Rating Scale and Cerebellar Neuropsychiatric Rating Scale.

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 the American Neuropsychiatric Association (of which he is also President). He is a member of the Medical and Scientific Research Advisory Board of NAF, medical advisor to NAF's New England Chapter, and member of the Cerebellar Research Consortium. He has authored over 200 original papers, chapters and clinical contributions, and written or co-edited six books – *The Cerebellum and Cognition*, *MRI Atlas of the Human Cerebellum*, *Fiber Pathways of the Brain*, *Cerebellar Disorders in Children*, *Handbook of the Cerebellum and Cerebellar Disorders*, and *Essentials 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. He has been funded by the NIH, NAF, AT Children's Project,

Biographies

NORD, Sidney R. Baer Jr. Foundation, and MINDlink Foundation. Dr. Schmahmann is actively engaged in mentoring students, residents and fellows in the clinic and lab, and deeply committed to patients with ataxia and their families.



Jana Schmidt, Postdoctoral fellow. Jana Schmidt (née Jana Boy) was born 1977 in Kuehlungsborn, Germany. She studied biology at the University of Rostock, Germany and finished with the Diploma. She then moved to the University of Tuebingen to do her Ph.D. at the Institute of Medical Genetics and Applied Genomics and finished with her thesis entitled “Analysis of transgenic mouse models of spinocerebellar ataxia type 3” in 2009. After characterizing several mouse models of SCA3 her work now focuses mainly on testing potential therapeutic agents using these models, but also on learning more about pathological mechanisms in SCA3.



Elisabetta Soragni is a staff scientist at the Scripps Research Institute in La Jolla in the laboratory of Professor Joel Gottesfeld. Dr. Soragni completed her Ph.D. and undergraduate studies in Italy at the University of Parma. She moved to the U.S. for post-doctoral training with Professor Peter Geiduschek at the University of California San Diego, on the mechanism of RNA polymerase III transcription. At Scripps she has been studying the mechanism of FXN gene silencing in Friedreich’s ataxia and is contributing to the development of HDAC inhibitors as treatment for this disease. Dr. Soragni was awarded several fellowships from NAF, FARA, Ataxia UK, Ataxia Ireland and GoFAR to study the mechanism of action of HDAC inhibitors and has been collaborating with several pharmaceutical companies.



Dr. Wei-Ling Tsou is a Senior Post-Doctoral Fellow/Research Associate at Wayne State University School of Medicine in Detroit. She received her B.S. degree in Nutrition and Health Sciences and M.S. degree in Medical Science from Taipei Medical University, Taiwan. She earned her Ph.D. in Neuroscience from National Yang-Ming University in Taiwan. During her Ph.D training, she received grants from the Graduate Students Study Abroad Program and joined the laboratory of Dr. Henry L. Paulson at the University of Michigan in Ann Arbor to further the development of RNAi as a therapeutic approach to Spinocerebellar Ataxias, with a special focus on SCA6. In 2011, Dr. Tsou pursued her postdoctoral fellowship training in Dr. Sokol Todi’s laboratory at Wayne State University. Dr. Tsou has a long-standing interest in Spinocerebellar Ataxias, having conducted her Ph.D. work and postdoctoral training in two of these diseases, SCA3 and SCA6. Her work with SCA3 led to new and important clues into its biology of disease and into potential routes of therapy for this type of ataxia. In SCA6, she developed a novel human miR124-based RNAi delivery platform to successfully and selectively target the polyQ-related disease isoforms. Recently, she generated the first *Drosophila* model of SCA6 to use it as screening platform for potential therapies. She is currently using genetic and biochemical techniques to define molecular mechanisms of neuroprotection for SCA6.

Biographies



Dr. Percy Tumbale received her M.Sc. in Biomedical Science and Ph.D. in Integrative Biology from Florida Atlantic University. She then joined the National Institute of Environmental Health Sciences, and is currently a research fellow at NIEHS. Her research has focused on structure–function studies of Aprataxin, a DNA repair factor implicated in the neurodegenerative disorder, Ataxia with Oculomotor Apraxia 1 (AOA1). She has published 11 journal articles including two high-impact papers on Aprataxin structure and function in *Nature* and *Nature Structure and Molecular Biology*. Dr. Tumbale’s research has established a platform for understanding novel Aprataxin functions and Aprataxin-linked neurodegeneration. She is particularly interested in Ataxia research and hopes that the findings from her studies will lead to a better understanding of the disease and development of strategies for improved diagnosis and treatment.



Dineke Verbeek, PhD graduated from the University of Utrecht (NL) with a MSc in Medical Biology in 2000 (with satisfaction). Then she joined the research group of Prof. Richard Sinke at the Department of Genetics, from the University of Utrecht (NL) to work on her thesis “The localization and identification of novel spinocerebellar ataxia genes in the Dutch autosomal dominant cerebellar ataxia population.” During her PhD studies, she contributed significantly to the field of human genetics on spinocerebellar ataxia by the identification of 2 novel SCA types (SCA19 and SCA23) and increased insights in the pathogenesis of SCA14 in cell model systems. For this last study, she was awarded a pre-doctoral fellowship from the National Institute of Health (NIH) to work in the laboratory of Prof. Kenneth Fischbeck (NIH, Bethesda, USA).

After completing her PhD in 2005, Dineke joined the laboratory of Dr. Eric Reits, Department of Cell biology and Histology at the Academic Medical Center in Amsterdam (NL), where she successfully followed up on her PhD studies work on SCA14. In 2008, Dineke was awarded a prestigious Rosalind Franklin Fellowship by the University of Groningen (NL) to establish her independent research group focusing on unraveling the genetics and biological mechanisms underlying movement disorders with special attention to spinocerebellar ataxias (SCAs).



Dr. Robert B. Wilson, MD, PhD, received his B.A. in Music and his B.S. in Biochemistry from Brown University, and his M.D. and his Ph.D. in Genetics from the University of Pennsylvania. He completed his residency training in Clinical Pathology, and his fellowship training in Transfusion Medicine, at the Hospital of the University of Pennsylvania, and he was then a post-doctoral researcher in the Howard Hughes Medical Institute. He joined the Department of Pathology and Laboratory Medicine at the University of Pennsylvania as an Assistant Professor in 1992 and is now a Full Professor. His primary research interests are in the neurodegenerative disorder Friedreich’s ataxia; in the development of therapeutics for genetic disorders using random shRNA screening approaches; and in the statistics of genetic risk assessment. He is the Co-Director of the University of

Biographies

Pennsylvania and Children's Hospital of Philadelphia Center of Excellence for Friedreich's Ataxia Research and serves on the Scientific Advisory Board of the Friedreich's Ataxia Research Alliance and on the Medical Research Advisory Board of the National Ataxia Foundation. In addition to research, he is a practicing molecular genetic pathologist, signing out cases in the Molecular Pathology Laboratory of the Hospital of the University of Pennsylvania. He also teaches general pathology to medical and graduate students, and molecular pathology to residents and fellows.



Bing Yao, PhD received his bachelor and master in biology from Ocean University of China. As a graduate student at the University of Florida, I made important contributions to understanding the molecular mechanisms of microRNA-mediated gene regulation by characterizing functional domains of the key translational repressor GW182. As a National Ataxia Foundation postdoctoral fellow in Dr. Peng Jin's laboratory at Emory University, my work suggests cytosine modification derivatives, especially 5-hydroxymethylcytosine (5hmC), play central epigenetic roles in neurodevelopment and neuronal functions. The dysregulation of cytosine modification dynamics could contribute to neurodegenerative disorders such as Fragile X-associated tremor/ataxia syndrome (FXTAS) and Spinocerebellar ataxia (SCA). RNA-binding proteins, such as hnRNP A2/B1, bridge RNA gain-of toxicity to ectopic transcription control by coordinate with epigenetic modifiers in neural cells. My long-term research interests are to understand the pivotal roles of epigenetic regulation in mammalian neurogenesis, and how dysregulation of these processes may contribute to neural pathology.

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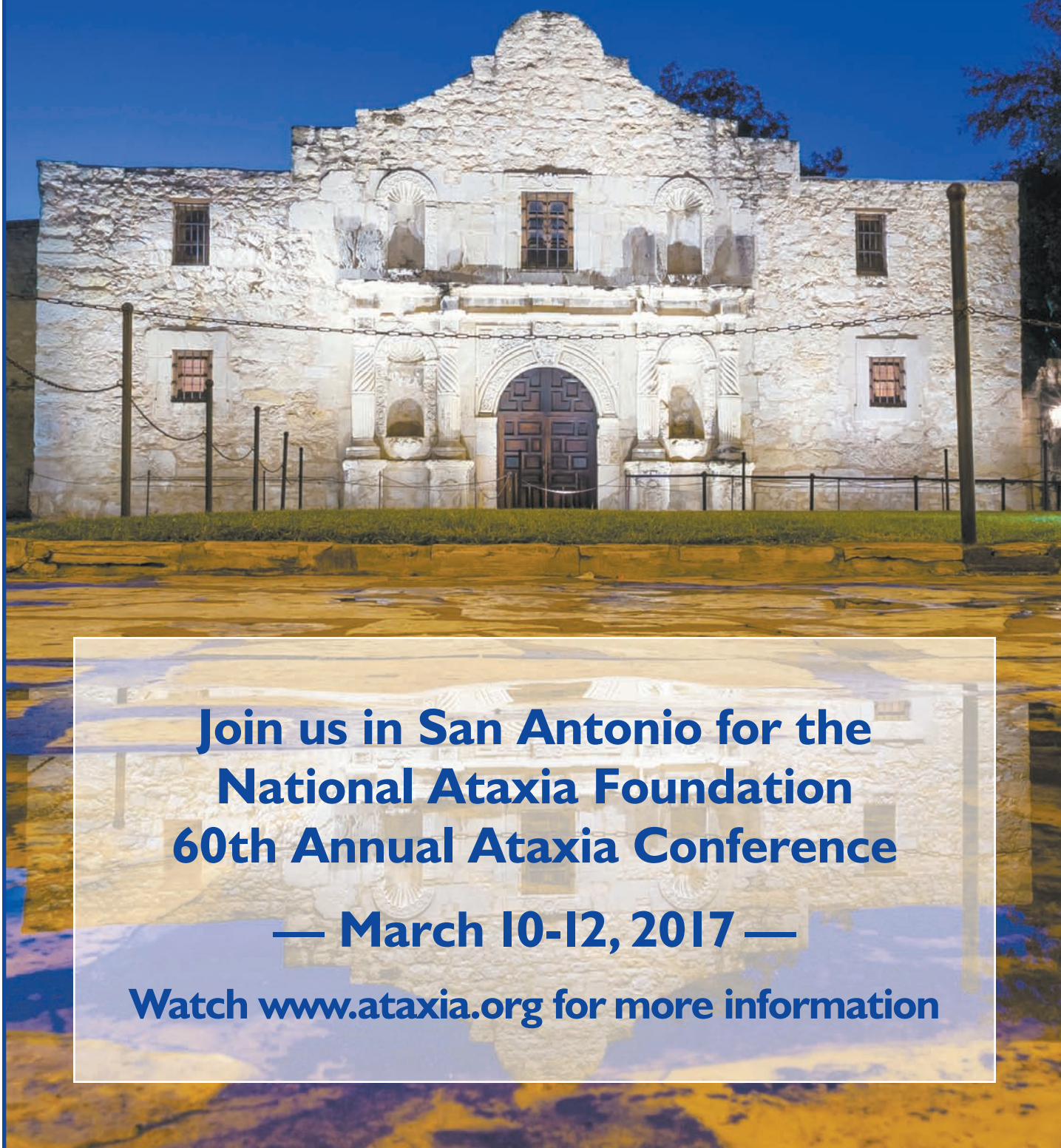
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