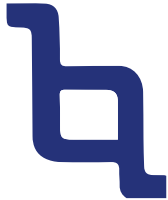


Welcome to the 5th Ataxia Investigators Meeting (AIM 2014)



Sponsored by the National Ataxia Foundation



With generous funding support from:

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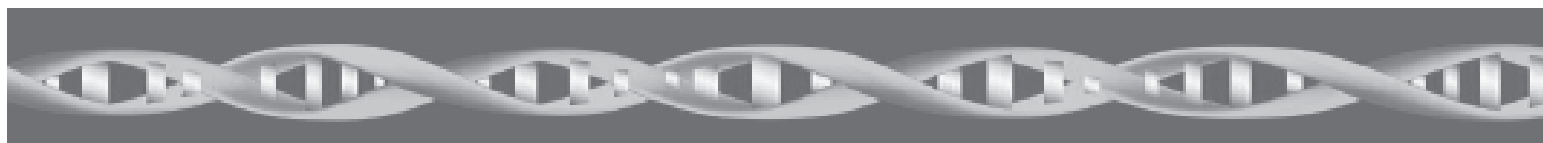
March 18-21, 2014 * Bally's Las Vegas * Las Vegas, NV

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Thank you to
Church Offset Printing, Inc.
for your support of the
2014 Ataxia Investigators Meeting.



THANK YOU

The National Ataxia Foundation thanks this year's AIM co-directors:

Harry T. Orr, Ph.D.

David Lynch, M.D., Ph.D.

and

Steering Committee Members

Henry Paulson, M.D., Ph.D.

Helene Puccio, Ph.D.

Robert Wilson, M.D., Ph.D.

**The National Ataxia Foundation is grateful for the generous support
for the 2014 AIM by our major donors:**

**An Anonymous Donor
The Gordon and Marilyn
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THANK YOU

We are grateful for the generous support of the
5th Ataxia Investigators Meeting from the following Partners:



Research that heals.



Ataxion



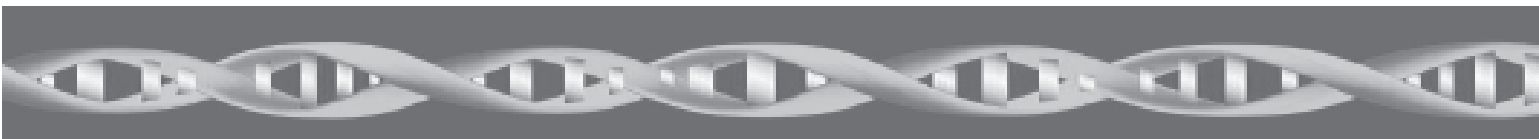
BioMarin



Testing that Makes a Difference.



"Funding for this conference was made possible in part by 1R13 NS086142-01 from the National Institute of Neurological Disorders and Stroke. The views expressed in written conference materials or publications and by speakers and moderators do not necessarily reflect the official policies of the Department of Health and Human Services; nor does mention of trade names, commercial practices, or organizations imply endorsement by the U.S. Government."



WELCOME AND NOTES

March 18, 2014

Dear AIM 2014 Attendee,

Welcome to the 5th Ataxia Investigators Meeting (AIM) sponsored by the National Ataxia Foundation (NAF) in exciting Las Vegas, Nevada.

Ataxia investigators across the country and worldwide typically find the AIM meetings, occurring once every two years, to be extremely helpful both in advancing our understanding of ataxia disease mechanisms and facilitating the push toward therapies. On behalf of the organizing committee, I and David Lynch as co-organizers of AIM 2014 hope you will find this year's meeting exciting and as stimulating as past meetings.

Please note that the format of the AIM 2014 has been modified from previous meetings. The goal is to better facilitate greater discussion and exchange of ideas. 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 quickly towards viable therapeutics. We are looking to all to actively participate.

Built into the meeting are opportunities to interact with patients and family members who will begin arriving this week to attend the NAF Annual Membership Meeting 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.

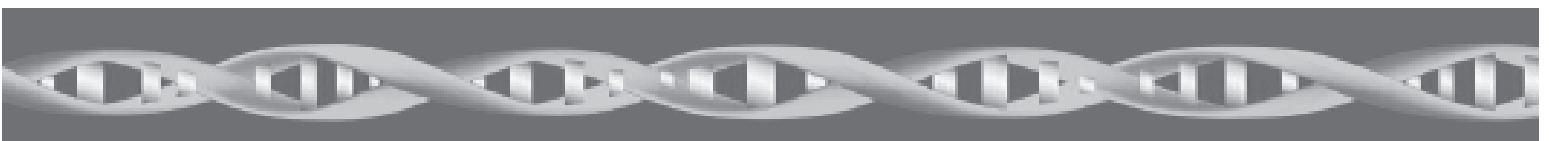
Best regards,

Harry Orr, Ph.D.

AIM 2014 Lead Organizer

Through the generosity of the AIM sponsors and support from NINDS, the 2014 AIM includes more travel-supported Junior Lecturers and poster presenters than any previous meeting. Selected by the Steering Committee members and through review of abstract submissions, these junior investigators are designated in the AIM program with an asterisk * by their names.

We congratulate you and look forward to seeing you at future Ataxia Investigator Meetings.

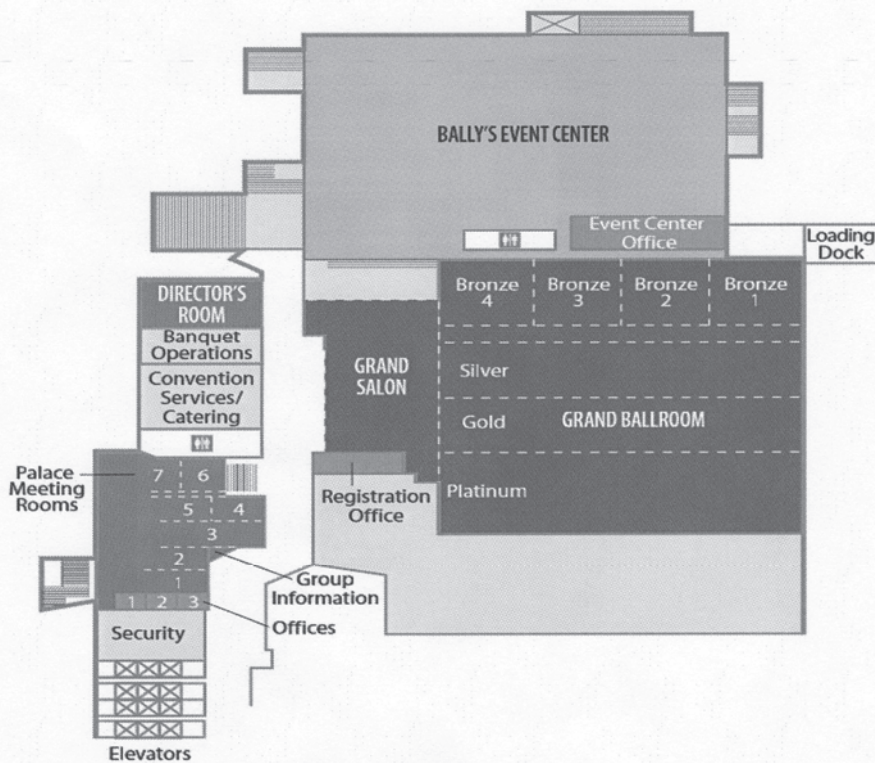


BALLY'S LAS VEGAS FLOOR PLAN

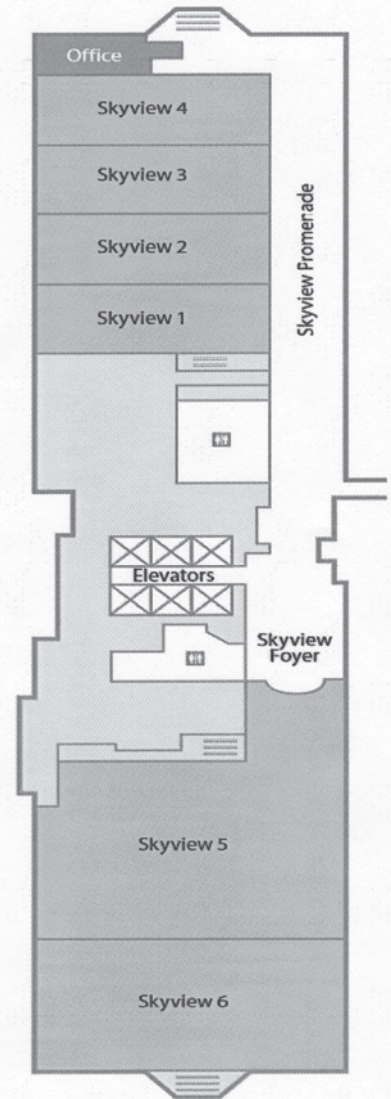
BALLY'S LAS VEGAS
Meeting Facilities Map

All Ataxia Investigators Meeting
functions will take place in meeting
rooms Skyview 5 and 6.

NORTH TOWER



CASINO LEVEL



26TH FLOOR

MEETING SCHEDULE

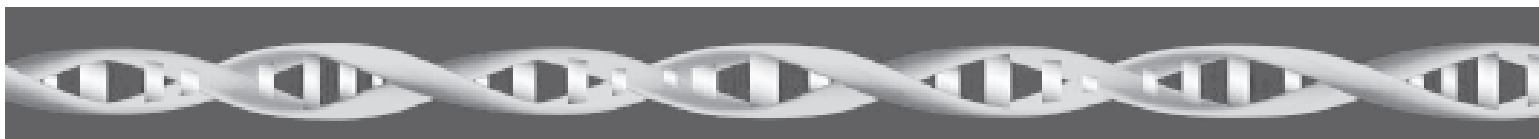
TUESDAY, MARCH 18, 2014

- 4-6:15 PM Check-In and Posters Boards available for hanging posters
- 5:15-6:15 PM** ***Welcome Reception and Check-In continues***
- 6:15 PM** ***Dinner***
- 7:15 PM Opening Remarks – Harry Orr, Ph.D., University of Minnesota
- 7:20 PM **Keynote Address:** Christopher Gomez, M.D., Ph.D. University of Chicago, IL
SCA6: A Bicistronic Gene

WEDNESDAY, MARCH 19, 2014

Posters Up For Viewing from 7:30 AM Wednesday – 6:15 PM Thursday

- 7:30 AM** ***Coffee and Tea Available***
- 8:30 – 12:30** **Theme 1 – Mitochondria in cerebellar function/dysfunction**
- 8:30-8:55 **Key Note:** David C. Chan, M.D., Ph.D., California Institute of Technology
Mitochondrial dynamics and cerebellar function
- Theme 1 Session Chair:** Robert Wilson, M.D., Ph.D., University of Pennsylvania
- 9:00-9:20 Mike Koob, Ph.D., University of Minnesota
Manipulating the mitochondrial genome
- 9:25-9:45 Francesca Maltecca*, Ph.D., San Raffaele Scientific Institute, Milan, Italy
Genetic and pharmacological rescues of Spinocerebellar ataxia in the SCA28 model open to human therapy
- 9:50-10:10 Peter McPherson, Ph.D., McGill University, Montreal, Quebec, Canada
Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay (ARSACS)
- 10:15-10:45** ***Break***
- 10:50-11:10 Pierre-Gilles Henry*, Ph.D., University of Minnesota, Minneapolis, MN
MRS and diffusion MRI of the spinal cord in Friedreich's Ataxia
- 11:15-11:35 Nélio Gonçalves*, Pharm.D., Ph.D., University of Coimbra, Portugal
Caffeine alleviates Progressive Motor Deficits in MJD Transgenic Mice
- 11:40-12:00 Dinesh Deelchand*, Ph.D. University of Minnesota
High field MRS is more sensitive to onset and progression of neurodegeneration than clinical decline in spinocerebellar ataxia type 1



MEETING SCHEDULE

WEDNESDAY, MARCH 19, 2014 *Continued*

12:00-1:00 p.m.

Lunch

1-5 PM

Theme 2 – Toward therapies through disease mechanisms

1:00-1:25

Key Note: Holly Kordasiewicz*, Ph.D., ISIS Pharmaceuticals, San Diego
ASO-based Therapy in Neurodegeneration

Theme 2 Session Chair: Henry Paulson, M.D., Ph.D. University of Michigan, Ann Arbor, MI

1:30-1:50

Hélène Puccio, Ph.D., INSERM, Illkirch, France
Gene Therapy – Friedreich ataxia

1:55-2:15

Edgardo Rodriguez-Lebron*, Ph.D. University of Iowa
RNAi and SCA3

2:20-2:40

Albert LaSpada, M.D., Ph.D., University of California San Diego
Therapeutic approaches for SCA7

2:45-3:15

Break

3:20-3:40

Maria do Carmo Costa*, Ph.D., University of Michigan
Pharmacological and genetic approaches to reduce levels of mutant protein ATXN3 in MJD/SCA3

3:45-4:05

Megan Keiser*, Ph.D., University of Iowa
Translating RNAi therapy for Spinocerebellar Ataxia 1 (SCA1) to the clinic

4:10-4:30

Chenchen Niu*, M.D., Ph.D., University of California, San Diego
Ataxin-7 knock-down therapy to treat spinocerebellar ataxia type 7: Development of an oligonucleotide-based approach to preclinical trial validation in mice

5-7 PM

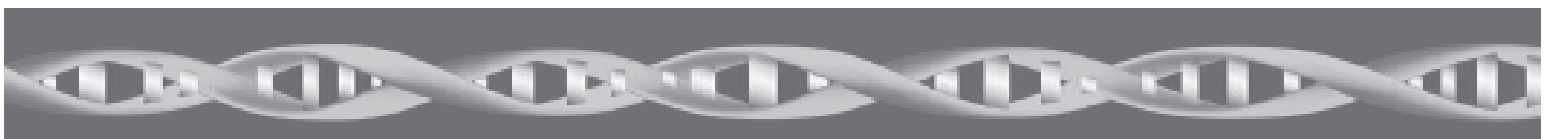
Scientific Poster Session with wine and cheese

5-6 p.m. Theme 1 and 2 presenters will be at their posters

6-7 p.m. Theme 3,4, and 5 presenters will be at their posters

Evening

Networking Dinner on your own in Las Vegas



MEETING SCHEDULE

THURSDAY, MARCH 20, 2014

Posters Up For Viewing from 7:30 AM – 6:15 PM Thursday

7:30 AM

Coffee and Tea Available

8:30 – 12:30

Theme 3 – Genomic Approaches to cerebellar function and ataxia

8:30-8:55

Key Note: Susan Ackerman, Ph.D., Jackson Laboratories, Bar Harbor, ME
Post-transcriptional defects and cerebellar degeneration

Theme 3 Session Chair: Stefan Pulst, M.D. University of Utah

9:00-9:20

Peng Jin, Ph.D., Emory University, Atlanta, GA
TET-mediated Epigenetic Modification(s) in Ataxia

9:25-9:45

Jeehye Park*, Ph.D., Baylor College of Medicine
An unbiased screen for therapeutic targets in SCA1

9:50-10:10

Andrea H. Nemeth, MBBS, DPhil, FRCP, University of Oxford
Genomic Approaches to Diagnosis of Ataxia

10:15-10:45

Break

10:50-11:10

Brent Fogel*, M.D., Ph.D., UCLA David Geffen School of Medicine
Mutation of Senataxin Alters Disease-Specific Transcriptional Networks in Patients with Ataxia with Oculomotor Apraxia Type 2

11:15-11:35

Scott Atwood*, Ph.D., Stanford University School of Medicine
Missing-in-Metastasis is a novel cerebellar ataxia gene that regulates Purkinje cell survival

11:40-12:00

Pavitra S. Ramachandran*, M.S., University of Iowa
RNA interference therapy provides therapeutic benefit in a Spinocerebellar ataxia type 7 mouse model

12:00-1:00 AM

Lunch

1-5 p.m.

Theme 4 – Cerebellar circuitry, function/dysfunction

1:00-1:25

Key Note: Kamran Khodakhah, Ph.D., Albert Einstein College of Medicine
Information coding by Purkinje cells

Theme 4 Session Chair: David Lynch, M.D., Ph.D., University of Pennsylvania

1:30-1:50

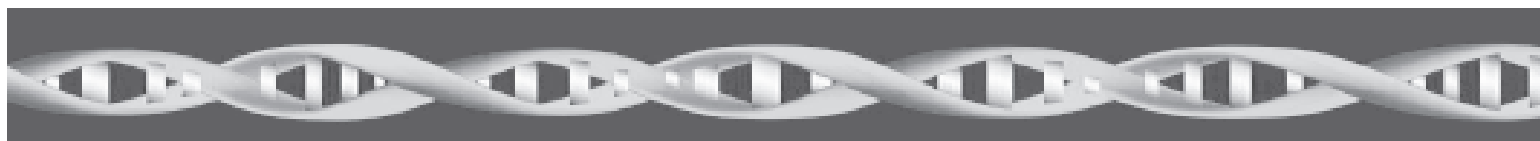
Christophe Lenglet*, Ph.D., University of Minnesota, Minneapolis, MN
Functional connectivity in Friedreich ataxia

1:55-2:15

Javier Medina*, Ph.D., University of Pennsylvania
Acquisition/Consolidation new motor skills

2:20-2:50

Break



MEETING SCHEDULE

THURSDAY, MARCH 20, 2014 *Continued*

- 2:55-3:15 Vikram Shakkottai*, M.D., Ph.D., University of Michigan, Ann Arbor, MI
Ion Channel dysfunction in SCA
- 3:20-3:40 Peter Todd*, M.D., Ph.D., University of Michigan
RAN translation in FXTAS
- 3:45-4:05 Lynn Ulatowski*, Ph.D., Case Western Reserve University
Cellular level mechanism of vitamin E deficiency-induced ataxia
- 5:15-6:15 PM **Poster Session for Patients and Families**
- 6:15 PM *Investigators please remove posters at this time.*
- 6:30 PM Dinner**
- 7:15 PM **Keynote Address** Alexandra Durr, M.D., University Hospital Salpêtrière, Paris, France
An Update from EuroSCA

FRIDAY, MARCH 21, 2014

- 7:30 AM Coffee and Tea Available**
- 8:30 – 12:00 Theme 5 – Robust biology of ataxia**
- 8:30-8:55 Key Note:** Joe Gleeson, M.D., University of California San Diego
Joubert syndrome & disorders of cerebellar development
- Theme 5 Session Chair:** Laura Ranum, Ph.D, University of Florida, Gainesville, FL
- 9:00-9:20 Karl Herrup, Ph.D., Hong Kong University of Science and Technology, Hong Kong
Ataxia Telangiectasia
- 9:25-9:45 Vivian G. Cheung*, M.D., University of Michigan Life Sciences Institute
RNA sequence modifications and neurologic disease
- 9:50-10:10 Ravi Chopra*, B.A., University of Michigan
Compartment-specific changes in excitability are associated with dendritic atrophy in Purkinje neurons in a mouse model of spinocerebellar ataxia type 1
- 10:15-10:45 Break**
- 10:50-11:10 Marija Cvetanovic*, Ph.D., University of Minnesota
Inflammation in SCA
- 11:15-11:35 Cleo Smeets*, MSc, University Medical Center Groningen, Netherlands
Glutamate mimicking mutant Dynorphin A causes spinocerebellar ataxia type 23
- 11:40-12:00 Thorsten Schmidt*, Ph.D., University of Tuebingen Germany
Ataxin-3 isoforms modify the pathogenesis of Spinocerebellar ataxia type 3
- 12:00 Closing Remarks – David Lynch, M.D., Ph.D., University of Pennsylvania

PATIENT/FAMILY/INVESTIGATOR INTERACTION

Linking Ataxia Families and Investigators:

The location of the 2014 Ataxia Investigators Meeting was selected so that it dovetails with the Annual Membership Meeting of the National Ataxia Foundation, the largest ataxia foundation in the country. This will maximize the impact of this meeting for scientists and patients alike to provide opportunities for meaningful interactions between researchers, patients and caregivers, which is considered beneficial for all involved. This has a number of very important effects. First, junior and senior investigators will see that what they work on in the lab does make a difference. Second, it is an opportunity for scientists to communicate with the public 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:

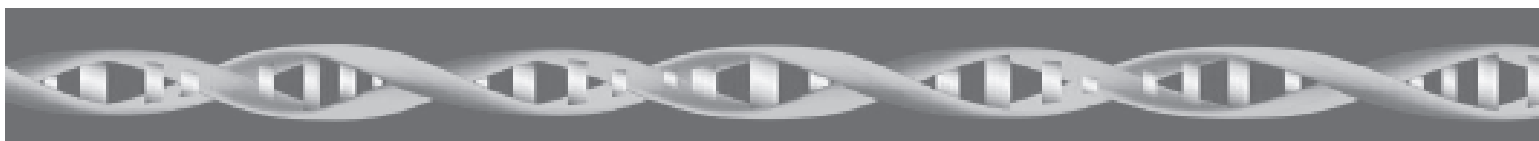
On Thursday, March 20, from 5:15—6:15 p.m. there is a dedicated poster session for patients and families. During this time we ask that all poster presenters be available at their posters so that patients and families can meet you and learn more about your ataxia research efforts. *This session should be attended by poster presenters only to allow room for wheel chairs and walkers in the poster session room.*

Posters must be removed from the poster boards after this session.

“Birds of a Feather” Small Group Session:

On Friday, March 21 from 2:00-5:00 p.m., after the AIM 2014 has adjourned, patients and family members will meet in facilitated small groups based on the type of ataxia that they have or their role, such as parent, spouse, or family member. You are very welcome to attend any session that would be of interest to you.

SCA 1 and SCA 2 in the Palace 6 & 7 Room
SCA3 in the Bronze 3 Room
SCA6 in the Palace 2 Room
All other SCAs (including SCA5, SCA7, SCA8, DRPLA) in the Bronze 4 Room
Unknown with Family History - Episodic & AOA in the Skyview 4 room
Unknown without Family History - Sporadic, MSA, and Gluten in the Skyview 2 Room
Over age 30 Friedreich in the Silver Ballroom
Under age 30 with Ataxia in the Skyview 6 Room
Parents (Non-Friedreich) in the Skyview 1 Room
Parents (Friedreich) in the Skyview 3 Room
Spouses & Partners without ataxia in the Gold Ballroom
Family members (over 30) without ataxia in the Palace 1 Room
Family members (under 30) without ataxia in the Conference Room



POSTER PRESENTERS

Theme 1: Mitochondria in cerebellar function/dysfunction

Chuang, Sheng-Fei, M.S. Changhua Christian Hospital Changhua, Taiwan	Far-infrared radiation Phototherapy Improving the Mitochondrial Function in Transgenic Spinocerebellar Ataxia 3 Neuron Cell-line
Cortopassi, Gino, Ph.D. University of California Davis Davis, CA	Small molecule repurposing for Friedreich's ataxia therapeutics. Identification of compounds, mechanism of action and functional recovery
Lagalwar, Sarita, Ph.D. Skidmore College Saratoga Springs, NY	Mitochondrial OXPHOS Dysfunction in Spinocerebellar Ataxia Type 1
Larivière, Roxanne, Ph.D. Montreal Neurological Institute Montreal, Quebec, Canada	<i>Sacs</i> knockout mice present pathophysiological defects underlying autosomal recessive spastic ataxia of Charlevoix-Saguenay
Tsuji, Shoji, M.D., Ph.D. The University of Tokyo, Japan	Mutations of <i>COQ2</i> in Familial and Sporadic Multiple System Atrophy. Implications for development of therapy for MSA

Theme 2: Toward therapies through disease mechanisms

Almeida, Luis Pereira de, Ph.D. University of Coimbra, Coimbra, Portugal	Autophagy activation mitigates motor and neuropathological deficits in mouse models of Machado-Joseph disease
Bayot, Aurélien, Ph.D. INSERM Paris, France	Distal effect of the GAA expansion on <i>PIP5K1B</i> in Friedreich's ataxia: What compound heterozygous patient tells us
Bezprozvanny, Ilya, Ph.D. UT Southwestern Medical Center Dallas, TX	Positive modulators of calcium-activated potassium channels as potential treatment for SCA2 and other SCAs.
Chutake, Yogesh K.*, Ph.D. University of Oklahoma Health Sciences Center Oklahoma City, OK	Altered nucleosome positioning and deficient transcriptional initiation caused by the expanded GAA triplet-repeat in Friedreich ataxia
Delatycki, Martin, Ph.D., FRACP Murdoch Childrens Research Institute Melbourne, Australia	Clinical Management Guidelines for Friedreich ataxia
Delatycki, Martin, Ph.D., FRACP Murdoch Childrens Research Institute Melbourne, Australia	An open label clinical pilot study of resveratrol as a treatment for Friedreich ataxia
Durr, Alexandra, M.D., Ph.D. Hôpital de la Salpêtrière Paris, France	Identification of robust biomarkers of metabolic changes in spinocerebellar ataxia type 1-2-3-7
Esteves, Sofia, Ph.D. Student (Carvalho or Teixeira-Castro will present this poster) University of Minho Braga, Portugal	Lithium chloride therapy fails to improve motor function in a transgenic mouse model of Machado-Joseph disease
Fuselier, Kayla*, M.S. LSU Health Sciences Center New Orleans, LA	MLH3, a minor player in mismatch repair, is a major force in somatic expansion of the Friedreich ataxia GAA·TTC repeat

POSTER PRESENTERS

Gennarino, Vincenzo A., Ph.D.

Baylor College of Medicine
Houston, TX

Gottesfeld, Joel, Ph.D.

(Elisabetta Soragni will present this poster)

Scripps Research Institute
La Jolla, CA

He, Fang*, Ph.D.

University of Michigan Medical School
Ann Arbor, MI

Hermann, Katherine

(Dr. Schmahmann will present this poster)
Massachusetts General Hospital/Harvard
Boston, MA

Hu, Yuan Shih (Jennifer)*, Ph.D.

Northwestern, Chicago
Chicago, IL

Jardim, Laura B., M.D., Ph.D.

Hospital de Clinicas de Porto Alegre, Porto
Alegre, Brazil

Mendonça, Liliana*, Pharm.D, Ph.D.

University of Coimbra
Coimbra, Portugal

Mollema, Nissa*, Ph.D.

University of Minnesota
Minneapolis, MN

Napierala, Marek*, Ph.D.

University of Alabama at Birmingham, AL

Nobrega, Clevio, Ph.D.

University of Coimbra
Coimbra, Portugal

Öz, Gülin, Ph.D.

University of Minnesota
Minneapolis, MN

Puccio, Helene, Ph.D.

Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC)
Illkirch, France

Schmidt, Jana, Ph.D.

Institute of Medical Genetics and Applied Genomics in Tuebingen
Tuebingen, Germany

Simões, Ana T., Ph.D.

Center for Neuroscience and Cell Biology
Coimbra, Portugal

Characterize the molecular mechanisms underlying differential neuronal vulnerability in Spinocerebellar ataxia type 1 (SCA1)

Epigenetic Therapy for Friedreich's Ataxia: Evaluation in Patient iPSC-Derived Neuronal Cells and in a Phase I Clinical Trial

TDP-43 Suppresses CGG Repeat-induced Neurodegeneration through Interactions with HnRNP A2/B1

Key Clinical Features of Multiple System Atrophy of the Cerebellar Type

The histone deacetylase HDAC3 is essential for Purkinje cell function, potentially complicating the use of HDAC inhibitors in SCA1

Lithium trial in Machado-Joseph Disease: subgroup analysis and responsiveness of rating scales

Transplantation of cerebellar neural stem cells alleviates neuropathology and motor impairments in Machado Joseph Disease

ATXN1 expression in SCA1 patient-derived iPS cells

Correction of the GAA repeat expansions in Friedreich's ataxia cells using zinc finger nucleases.

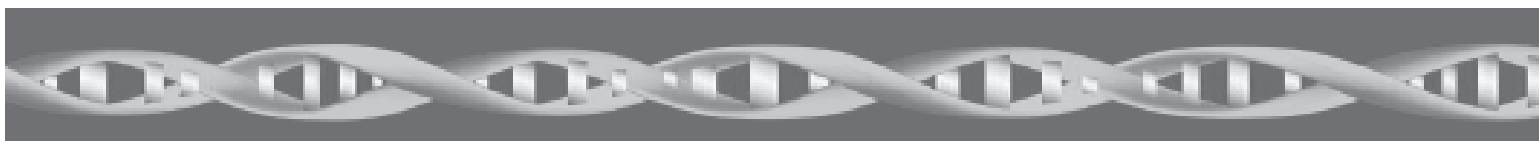
Translational repression in Machado-Joseph disease: ataxin-2 as a new therapeutic target

Assessing Recovery from Neurodegeneration in SCA1: High field MRS vs. Behavioral Testing, quantitative PCR and Histology

Loss of *Adck3* leads to Purkinje cells dysfunction, skeletal muscle defect and Coenzyme Q₉ deficiency, recapitulating the physiopathology of human ARCA2

Assessment of Riluzole as therapeutic option for Spinocerebellar ataxia type 3

Orally-administered calpain inhibitor BDA-410 reduces ataxin-3 cleavage and alleviates neuropathology in a lentiviral mouse model of Machado-Joseph disease



POSTER PRESENTERS

Soong, Bing-Wen, M.D., Ph.D.

National Yang-Ming University
School of Medicine
Taipei, Taiwan

Soragni, Elisabetta*, Ph.D.

Scripps Research Institute
La Jolla, CA

Soriano, Sirena, Graduate student

Universitat de València,
Valencia, Spain

Sowa, Anna*, M.S.

University of Tübingen
Tübingen, Germany

Teixeira-Castro, Andreia*, Ph.D.

University of Minho
Braga, Portugal

Todi, Sokol, Ph.D.

Wayne State University
School of Medicine
Detroit, MI

Truant, Ray, Ph.D.

McMaster University
Ontario, Canada

Yang, Su*, B.S.

Emory University
Atlanta, GA

Safety and efficacy of intravenous administration of allogeneic adipose mesenchymal stem cells in cerebellar ataxia: a phase 1/2a clinical trial

Role of single nucleotide polymorphism (SNP) in the protein deacetylase Sirt6 in Friedreich's ataxia pathogenesis

Proteomic profiling to identify modifier genes in a *Drosophila* model of Friedreich's ataxia

Altered nucleocytoplasmic shuttling of ataxin-3 *via* transport proteins affects the pathogenesis of spinocerebellar ataxia type 3

A whole organism screen identifies serotonergic signaling as modulator of Machado-Joseph disease pathogenesis

New efforts toward therapeutics for polyglutamine-dependent SCAs

Polyglutamine domain flexibility mediates the proximity between flanking sequences in huntingtin and Ataxin-7.

Mesencephalic astrocyte-derived neurotrophic factor (MANF) is protective in spinocerebellar ataxia 17 (SCA17) knock-in mice

Theme 3: Genomic Approaches to cerebellar function and ataxia

Bower, Matthew, M.S., CGC

University of Minnesota
Medical Center
Minneapolis, MN

Burmeister, Margit, Ph.D.

University of Michigan
Ann Arbor, MI

Diaz Garcia, Javier, Graduate student

Baylor College of Medicine
Houston, TX

Figueroa, Pattie*, M.S.

University of Utah
Salt Lake City, UT

Gavrilova, Ralitzta, M.D.

Mayo Clinic,
Rochester, MN

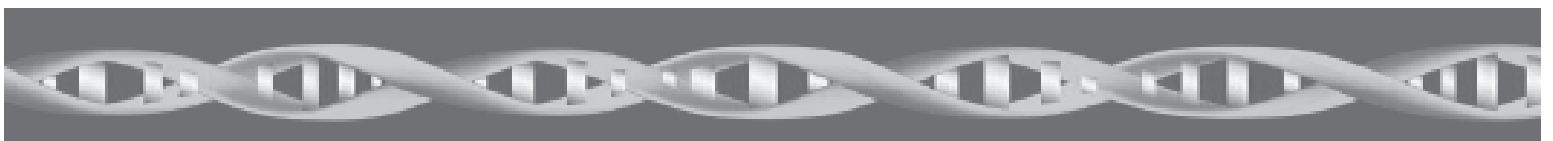
Diagnostic yield of a targeted next generation sequencing approach in a young adult population: high frequency of recessive ataxias and implementation of a next-generation sequencing copy-number variation algorithm.

Gene Identification in Ataxia: Two Years of Experience with Next Generation Exome Sequencing

Screens for genetic modifiers of Spinocerebellar ataxia type 1 in *Drosophila* and cell culture

Spontaneous Rat Mutant – A New Model for X-Linked Ataxia

Comparison of Friedreich Ataxia Patients with Trinucleotide Repeat Expansions and Point Mutations in FXN-Encoded Frataxin

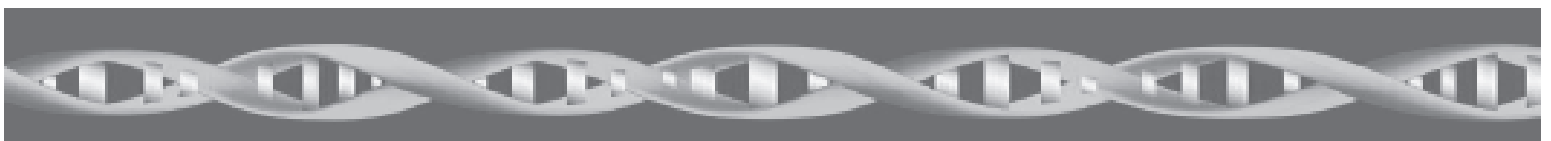


POSTER PRESENTERS

Li, Jiali, Ph.D. Rutgers University Piscataway, NJ	Loss of 5-hmC is a key factor in driving Purkinje cell vulnerability in A-T brain
Moccia, Robert F. Jr., M.D. Massachusetts General/Harvard Boston, MA	A New Cerebellar Syndrome Associated with Novel Mutations in <i>SYNE1</i>
Nibbeling, Esther*, MSc University Medical Center Groningen	Identification of novel spinocerebellar ataxia disease genes using next generation sequencing approaches
Paucar, Martin, M.D. Karolinska University Hospital, Stockholm, Sweden	A family affected by SCA27 caused by an interstitial chromosome 13q33.1 deletion
Scoles, Daniel, Ph.D. University of Utah Salt Lake City, UT	Antisense oligonucleotides for the treatment of spinocerebellar ataxia type 2 (SCA2)
Sun, Miao, Ph.D. University of Chicago Chicago, IL	An exome sequencing based approach for the molecular diagnosis of ataxia
Ward, Jacqueline M., B.S. University of California San Diego San Diego, CA	Generation and characterization of isogenic induced pluripotent stem cell lines as a model for spinocerebellar ataxia type 7
Yao, Bing*, Ph.D. Emory University Atlanta, GA	Genome-wide alteration of 5-hydroxymethylcytosine in a mouse model of fragile X-associated tremor/ataxia syndrome

Theme 4: Cerebellar circuitry, function/dysfunction

Dell'Orco, James*, B.S. University of Michigan Ann Arbor, MI	Purkinje neuron atrophy in Spinocerebellar Ataxia Type 1 represents a compensatory mechanism for increased membrane excitability
Guell, Xavier, Medical Student Drs. Hoche or Schmahmann will present this poster.	Language impairments in patients with cerebellar degeneration
Hoche, Franziska, M.D. Massachusetts General Hospital Boston, MA	The cerebellar contribution to social cognition
Hoche, Franziska, M.D. Massachusetts General Hospital/Harvard Boston, MA	Prospective evaluation of the sensitivity and specificity of cognitive tasks for diagnosis of the cerebellar cognitive affective syndrome.
Juelich, Richard*, B.A. Massachusetts General Hospital/Harvard Boston, MA	Diffusion MRI detection of anatomic changes in the cerebellar folia in a mouse model of spinocerebellar ataxia type 1
Kuo, Sheng Han, M.D. Columbia University New York, NY	Pathological Findings of Holmes Tremor Associated with Anti-Yo Cerebellar Degeneration
MacMore, Jason*, B.A. Mass General/Harvard	The Brief Ataxia Rating Scale (BARS) - development of an internet accessible video library for education and training



POSTER PRESENTERS

Schmahmann, Jeremy, M.D.

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A comparison of the Brief Ataxia Rating Scale (BARS) and the Scale for the Assessment and Rating of Ataxia (SARA) in the Ataxia Centers of Johns Hopkins University (JH), Massachusetts General Hospital (MGH) and the University of California Los Angeles (UCLA)

Subramony, S. H, M.D.

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Force but not time control is impaired in patients with spinocerebellar ataxia during fast ankle goal-directed contractions and is associated with altered activation of tibialis anterior motor units

Zhang, Yalan*, M.D., Ph.D.

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Regulation of neuronal survival by potassium channel interactions with Hax-1

Theme 5: Robust biology of ataxia

Avery, Adam*, Ph.D.

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Dendritic transport dysfunction in a novel Drosophila model of SCA5

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Molecular Genetics of Spinocerebellar ataxia type 8 (SCA8): RAN proteins and RNA foci

Carvalho, Andreia*, MSc

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Dominant negative effect of polyglutamine expansion perturbs normal function of ataxin-3 in neuronal cells

Chen, Dong-Hui*, M.D., Ph.D.

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Pathogenesis of spinocerebellar ataxia type 14 in mouse models

Matos, Carlos, B.S., M.A.

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Modulation of catalytic activity and polyQ toxicity by phosphorylation in spinocerebellar ataxia type 3

Ramani, Biswa*, B.S.

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Aberrant processing of mutant ataxin-3 transcript accelerates pathogenesis in SCA3

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De novo mutation of an ataxin-7 30 CAG repeat intermediate allele as the cause of juvenile-onset spinocerebellar ataxia type 7

Tsou, Wei-Ling*, Ph.D.

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Understanding the neuro-protective mechanism of the SCA3/MJD protein, ataxin-3

Yoon, Grace, M.D.

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Massive CAG repeat expansion and somatic instability in maternally inherited infantile SCA7

All AIM conference communications, materials and abstracts are confidential.

NAF RESEARCH GRANT PROGRAMS



*The National Ataxia Foundation
office is located in
Minneapolis, Minnesota*



The National Ataxia Foundation offers Five Ataxia Research Grant Programs

The National Ataxia Foundation (NAF) was established in 1957 by Dr. John W. Schut, the brilliant physician and researcher who was determined to find the cause, and cure for the ataxias.

Dr. Schut lost his personal battle with ataxia before his dream was realized but his vision to improve the lives of persons affected by ataxia through support, education, and research continues to be the guiding light of NAF. Due to the generosity of donors and individual and family fundraising efforts, NAF is pleased to provide funding to support research that will bring us closer to finding the answers needed to end ataxia.

The National Ataxia Foundation (NAF) has a commitment to fund the best science with relevance to ataxia, both in basic and translational research, however the emphasis of the Foundation's research program is on Young Investigator and Post-Doc grants: the next generation of scientists.

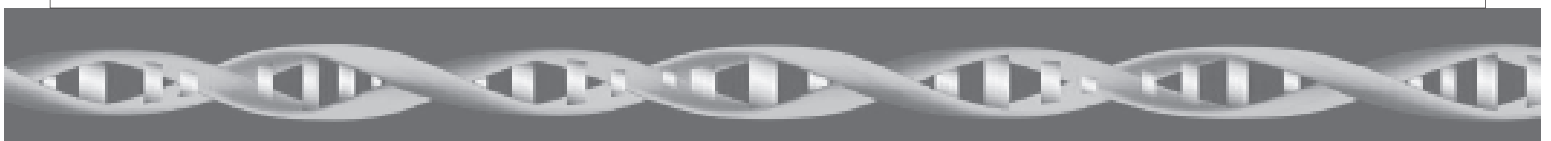
The Foundation supports hereditary ataxia research, but has also placed an emphasis on research into sporadic ataxia and is encouraging submission of research projects that will investigate non-genetic forms of ataxia. More information available at www.ataxia.org. NAF funds research both within the United States and from International investigators.

Application forms for NAF's research programs will be posted on the website in late April 2014.

Research Grant: One year grant of up to \$15,000 but promising proposals up to \$30,000 will be considered. Seed monies in early or pilot phases of studies and ongoing investigations that demonstrate need to attract future funding from other sources.

Letter of Intent with a ½ to one-page abstract with specific aims of your research due: 7/15/14

Full application due: 8/15/14



NAF RESEARCH GRANT PROGRAMS

Young Investigator (YI) Award: One year grants of \$35,000 to encourage young investigators to pursue a career in the field of ataxia research. Candidates must have attained an MD or PhD degree, and have an appointment as a junior faculty member, senior post-doc or clinical fellow. Individuals at the Associate Professor level are not eligible. Clinicians must have finished their residency no more than five (5) years prior to applying. PhDs must have completed their post-doc training within the past five years from the date of application. This award is for new faculty/investigators.

Letter of Intent with a ½ to one-page abstract with specific aims of your research due: 8/1/14

Full application due: 9/2/14

Research Post-Doc Fellowship Award: One year grant up to \$35,000 is intended for an individual to spend a 3rd year in a post-doc position to increase the chance to establish an independent ataxia research program.

Letter of Intent with a ½ to one-page abstract with specific aims of your research due: 8/15/14

Full application due: 9/15/14

Pioneer SCA Translational Research Award: One year grant of \$100,000 focusing on research investigations that will facilitate the development of treatments for the Spinocerebellar Ataxias.

Letter of Intent with a ½ to one-page abstract with specific aims of your research due: 8/15/14

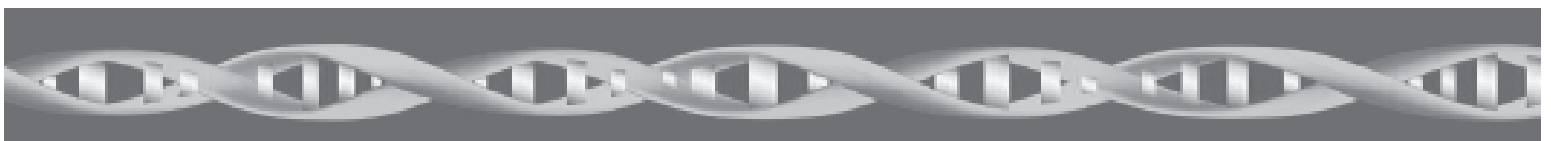
Full application due: 9/15/14

Young Investigator (YI-SCA) Award for SCA Research: One year grant of \$50,000 to encourage young investigators to pursue a career in spinocerebellar ataxia research. Candidates must have attained an MD or PhD degree, and have an appointment as a junior faculty member, senior post-doc or clinical fellow. Individuals at the Associate Professor level are not eligible. Clinicians must have finished their residency no more than five (5) years prior to applying. PhDs must have completed their post-doc training within the past five years from the date of application. This award is for new faculty/investigators.

Letter of Intent with a ½ to one-page abstract with specific aims of your research due: 8/1/14

Full application due: 9/2/14

Research applications and instructions are on NAF's website, www.ataxia.org. If you have specific questions about NAF's research program, you may contact Sue Hagen at susan@ataxia.org or 763-553-0020.



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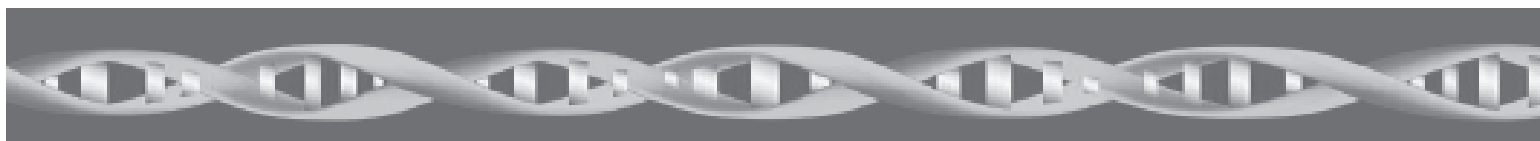
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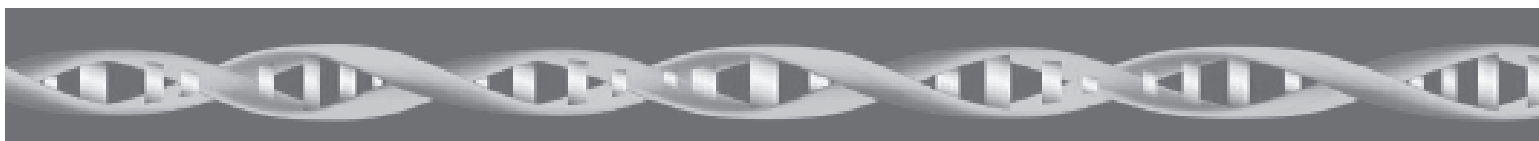
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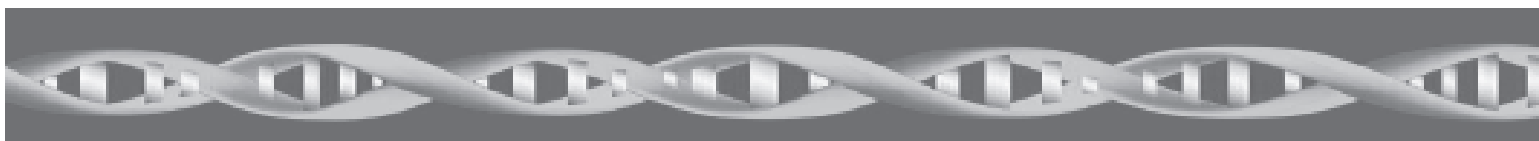
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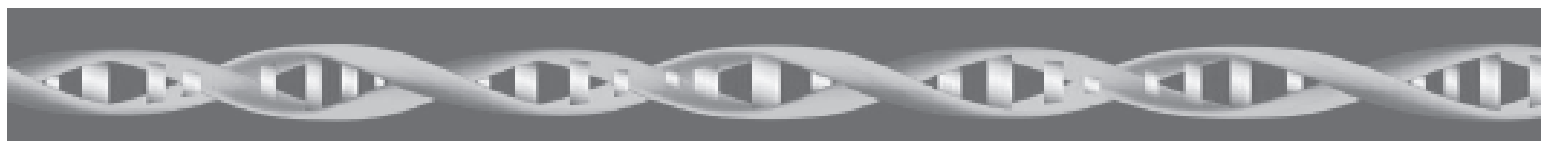
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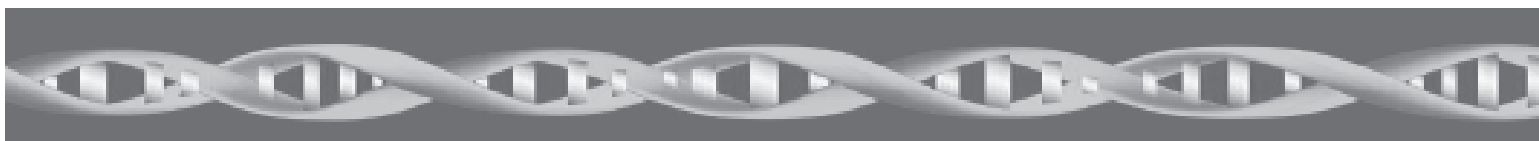
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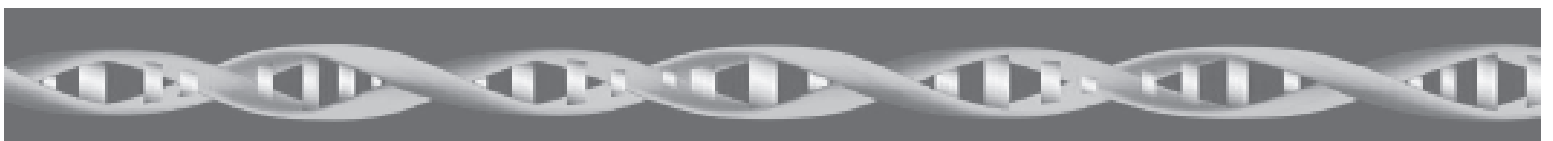
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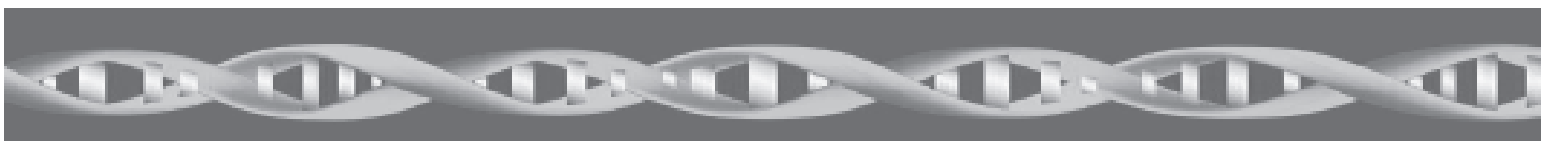
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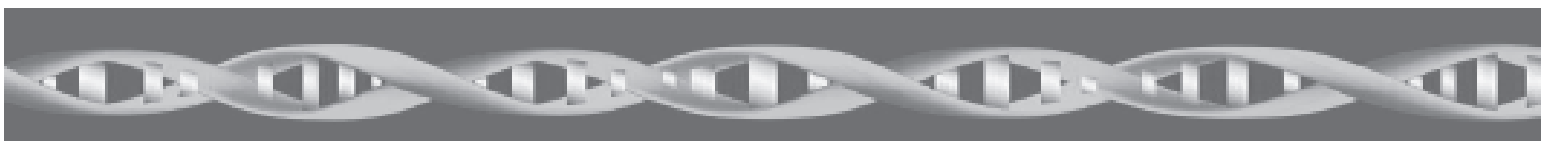
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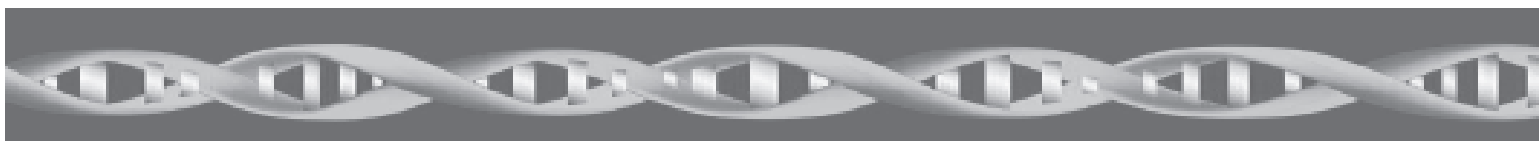
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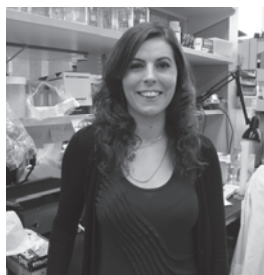
The following biographies of Jr. and Sr. Speakers were submitted for publication in the AIM Program.



Dr. Susan Ackerman is a professor at The Jackson Laboratory and a Howard Hughes Medical Institute investigator. Research in Dr. Ackerman's laboratory is focused on the identification of the genes, pathways and networks involved in both the development of the cerebellum and the degeneration of cerebellar neurons. Dr. Ackerman received her Ph.D. in biology from the University of California in Los Angeles, and did her undergraduate work in biology and chemistry at the California State University, Chico. She is also an adjunct professor at University of Maine, Orono and Tufts University School of Medicine, and an associate geneticist at Massachusetts General Hospital, Boston.



Dr. Scott Atwood is a postdoctoral research fellow at Stanford University School of Medicine. He received a B.S. degree in biology from the University of Utah and earned his Ph.D. in molecular biology at the University of Oregon where he studied how kinase activity influences cell-fate specification of neural stem cells. He is currently training in the laboratory of Dr. Anthony Oro in the Program in Epithelial Biology at Stanford University's dermatology department where he investigates kinase regulation of cell fate and cancer. Dr. Atwood has discovered how polarity proteins necessary for neural stem cell fate are critical amplifiers of Hedgehog signaling during basal cell carcinoma progression. He is currently investigating the pathogenesis of novel cerebellar ataxia gene *Mtss1*, a Hedgehog component that contributes to Purkinje cell homeostasis. He is a recent recipient of the National Institutes of Health Pathway to Independence Award.



Dr. Costa started her research career in 1998/1999 by working with Dr. Maciel in the establishment and improvement of the molecular diagnosis of MJD/SCA3 and HD for her undergraduate thesis in Biochemistry at the University of Porto. Post-graduation, she continued working with Dr. Maciel and Prof. Sequeiros in molecular genetic studies of MJD, HD and HD-like disorders, and in animal models of MJD at the University of Porto. In 2003, she moved with Dr. Maciel's lab to the University of Minho where she completed her PhD in Health Sciences. As a graduate student, she studied the *ATXN3* mouse homologue gene and protein and participated in the generation and characterization of a novel transgenic mouse model for MJD. In 2008, she moved to Ann Arbor, Michigan to do her post-doctoral training with Dr. Paulson in the development of therapeutics for MJD/SCA3 using RNA interference and small-molecules to reduce levels of mutant protein ATXN3. Currently, as a research investigator in Dr. Paulson's lab she is pursuing her interest in therapeutic approaches for MJD/SCA3 and its molecular mechanisms. She is planning to expand her work in translation research to other neurological diseases.

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David Chan is Professor of Biology at the California Institute of Technology and an Investigator of the Howard Hughes Medical Institute. Since 2000, his lab's main interest is in understanding the role of mitochondrial dynamics in cellular function and human physiology. Mitochondria are dynamic organelles that have many important functions in cells, including energy generation, metabolism, and regulation of cell death. A key feature of mitochondria is that they undergo cycles of fusion and fission, and his lab studies the role of these processes in controlling their function. In addition, several human diseases arise from a perturbation of these processes, and his lab is trying to understand the cellular mechanisms involved in disease pathogenesis.

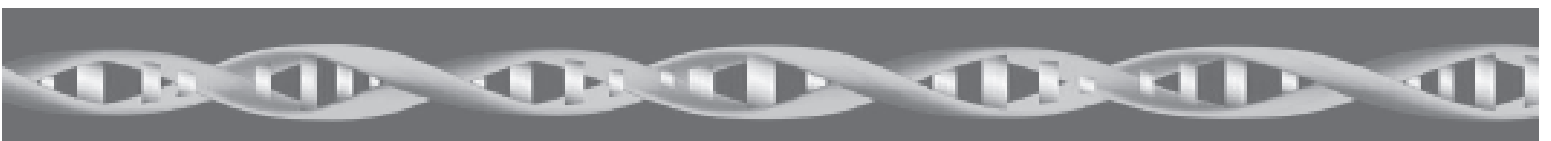


Dr. Cheung is an Investigator of the Howard Hughes Medical Institute and the Frederick G.L. Huetwell Professor of Pediatrics (Division of Neurology) and Professor of Genetics at the University of Michigan. Her laboratory studies genetic variation in gene regulation. They combine genetic mapping with genomic and molecular methods to study regulation of gene expression. They are particularly interested in understanding how RNA processing plays a role in maintaining normal cellular functions and in response to cellular stress, particularly those that underlie neurodegenerative diseases. They study genetic variation in chromatin modification, signal transduction and transcriptional processing; and how these affect disease susceptibility.

More recently, her group uncovered a surprising finding of differences between RNA and its corresponding DNA sequences beyond the known RNA editing mechanisms. They found all 12 types of RNA-DNA (RDDs) sequence differences including transversions. There is individual variation in RDDs, thus this discovery extends genetic variation beyond DNA sequences. In addition, it adds RNA sequence modification to 5' capping and splicing as key steps in RNA processing. The Cheung lab is working to determine the mechanisms that mediate RDDs and their roles in gene expression and regulation of cellular functions.



Ravi Chopra is an MD/PhD student at the University of Michigan, currently in his third year of the program. Ravi received his B.A. degree in Biological Sciences from Northwestern University in 2011, where he studied neuronal regulation of the cellular heavy metal stress response in *Caenorhabditis elegans*. He subsequently matriculated in the University of Michigan MSTP program and completed two years of medical school coursework before moving on to the graduate phase of his training. He is now a member of Dr. Vikram Shakkottai's research group. Ravi's primary research interest is in the structure and function of Purkinje neuron dendrites in spinocerebellar ataxia type 1. In particular, he is interested in how altered dendritic function may cause pathologic changes in Purkinje neuron dendrites, contributing to the progressive deterioration in motor performance that is seen in this disease.



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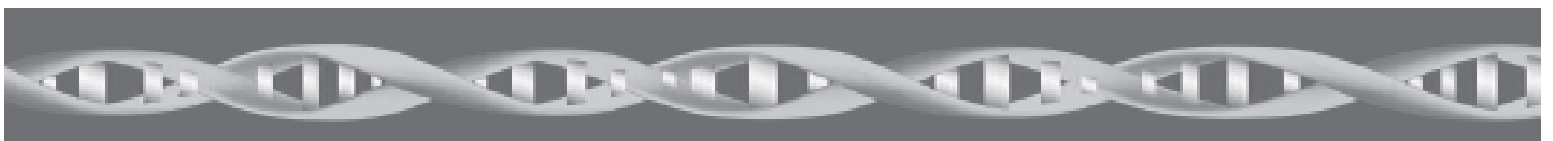
Dr. Marija Cvetanovic received her BSc in Molecular Biology and Physiology from the University of Belgrade in 1998. She completed her PhD at the Department of Immunology, Microbiology and Virology at the University of Illinois in Chicago in 2004. For her PhD research in the laboratory of Dr. David Ucker, Marija examined the interaction of dead cells with the macrophages, cells that are responsible for the clearance of apoptotic cells in the body. She discovered that anti-inflammatory response triggered upon the recognition of apoptotic cells leads to abrogation of inflammatory gene transcription in macrophages. As a postdoctoral fellow in the laboratory of Dr. Puneet Opal, Dr. Cvetanovic has undertaken an exploration of the molecular causes of the Spinocerebellar Ataxia Type1 (SCA1). Marija has discovered that the one of the genes that is transcriptionally misregulated in SCA1 is the angiogenic and trophic vascular endothelial growth factor (VEGF). She has performed detailed mechanistic experiments that demonstrated that mutant ataxin-1 directly represses VEGF promoter via its effects on histone acetylation. More importantly, she has discovered that delivering VEGF either genetically or pharmacologically can improve the SCA1 phenotype *in vivo*. During these studies Dr. Cvetanovic started wondering what happens to the non-neuronal cerebellar cells, such as astrocytes and microglia, during SCA1 pathology. In 2012 Marija joined the Department of Neuroscience at the University of Minnesota as the Assistant professor. The goal of her research is to combine her doctoral and post-doctoral expertise to investigate the involvement of non-neuronal cells in SCA1. Specifically, her lab examines the changes in the function of astrocytes and microglia in SCA1 and what role neuroinflammation plays in SCA1.



Dr. Dinesh Deelchand is a Research Associate/Assistant Professor at the Center of Magnetic Resonance Research (CMRR) at the University of Minnesota and specializes in developing and validating new approaches to study brain metabolism and neurotransmission non-invasively in normal and diseased conditions using NMR spectroscopy (MRS). He obtained a BSc in Physics from the University of Mauritius (Mauritius) in 2000 and a PhD in Physics for his work on carbon-13 MRS studies of carbohydrate and lipid metabolism in type 2 diabetes patients from the University of Nottingham (UK) in 2004. He joined CMRR as a postdoctoral associate to continue his research in brain metabolism using both carbon-13 and proton MRS techniques. Dr. Deelchand current research focuses on using proton MRS to investigate changes in many chemicals in spinocerebellar and Friedreich ataxias. This information would help to assess progression of disease and treatment responses in these patients.



Alexandra Durr works as a consultant in Neurogenetics in the Genetic Department at the University Hospital Salpêtrière in Paris-France. She trained in Germany and France in Neurology and Genetics. 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. *continued . . .*



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Alexandra Durr, *continued*, Her research interest are phenotypical expression and therapeutical approaches of genetic disorders focused on cerebellar ataxias, spastic paraplegias and inherited conditions of other movement disorders. She opened the first presymptomatic testing facility for late, adult-onset neurogenetic disorders in France and is a member of Scientific Advisory Board of INSERM, Scientific Board of the Fondation pour la Recherche Médicale, Scientific Planning Committee of the European Huntington Disease Network 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), AFAP (Association Française de l'Ataxie de Friedreich) and ASL (Spastic paraplegias).



Brent L. Fogel, M.D., Ph.D., Dr. Fogel obtained both his M.D. degree and a Ph.D. in Microbiology and Molecular Genetics from the Medical College of Wisconsin. He completed residency training in Neurology at UCLA as well as a fellowship in Neurogenetics. He is currently an Assistant Professor in the Neurogenetics Program of the Department of Neurology at the David Geffen School of Medicine at UCLA. He is also the current Vice-Chair of the American Academy of Neurology Section on Neurogenetics. Dr. Fogel's research studies basic molecular mechanisms of neuronal function to better understand how impairment leads to neurodegeneration, particularly of the cerebellum. Clinically, Dr. Fogel treats patients with various disorders of balance and coordination, including spinocerebellar ataxia, at the UCLA Ataxia Center. He is also Director of the UCLA Ataxia and Neurogenetics Biobank Program and uses genome-wide methods, including exome sequencing, to identify rare and novel causes of cerebellar ataxia. Dr. Fogel's work has been funded by the American Academy of Neurology / American Brain Foundation, the National Institutes of Health, and the National Ataxia Foundation. The accomplishments of Dr. Fogel's laboratory can be followed on Twitter (@FogelLab).



Christopher M. Gomez, M.D., Ph.D. I have a longstanding interest in the field of genetic disorders of the cerebellum (ataxias) and neuromuscular junction (congenital myasthenic syndromes) for more than 20 years. My long term goals are to help characterize the pathogenic mechanisms and develop treatments for disorders of synaptic and Purkinje cell degeneration. We employ a wide array of research tools including genetic analysis, gene and protein expression studies, novel cell and transgenic mouse models, and in vitro and vivo electrophysiological studies. My group has published on at least 7 different transgenic mouse lines and mouse genetic models for different neuromuscular and cerebellar disorders, and that we developed and or characterized in over 12 papers. My group identified and functionally confirmed some of the first mutations in the acetylcholine receptor that cause the slow-channel congenital myasthenic syndrome (SCS), and developed and extensively analyzed an animal model of the SCS. My group helped characterize the pathological mechanisms in spinocerebellar ataxia type 6 (SCA6) SCA26 and episodic ataxia type 2. We recently discovered that the gene product responsible for SCA6 is translated from newly recognized second cistron within the CACNA1A gene, a novel gene expression mechanism for mammalian systems. I have been involved since 1993 in genetic, phenotypic, and genotype-phenotype and studies of patients with a wide range of ataxia types. In 2006 I moved my lab to the University of Chicago to assume the Chair of the Department of Neurology.

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Nélio Gonçalves, Pharm.D., Ph.D. has received his Ph.D. in Pharmaceutical Sciences from the University of Coimbra, Portugal (2013). During his Ph.D. training, he successfully completed several advanced courses of the Doctoral Program in Experimental Biology and Biomedicine organized by the Center for Neuroscience and Cell Biology of Coimbra (CNC), Portugal, and he is currently conducting a postdoctoral research in CNC.

His research has been focused on the manipulation of a neuromodulation system operated by adenosine A2A receptors (A2AR) in different mouse models of Machado-Joseph Disease (MJD)/spinocerebellar ataxia type 3, namely lentiviral-based and transgenic, and on further investigation of whether A2AR antagonism is able to reduce the associated morphological (Gonçalves *et al.* (2013) *Ann Neurol*) and behavioral modifications, respectively. The candidate strategy was the chronic administration of caffeine (*per os*), based on its ability to antagonize the adenosine receptors, especially of the A2AR, which have been demonstrated to be effective controlling and alleviating neurodegeneration in diverse brain diseases. This study, headed by Prof. Luís Pereira de Almeida (Vectors and Gene Therapy lab) in tight collaboration with Prof. Rodrigo A. Cunha (Purines in Brain Diseases lab, from CNC), may provide the first realistic and safe promising life style prophylactic strategy to delay the onset and interfere with the inexorable evolution of MJD.



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 and 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 metabolomics. Combination of these multiple modalities using mathematical models is expected to lead to more sensitive assessment of disease progression than each marker considered separately.



Karl Herrup received his Bachelor's degree from Brandeis University in Waltham, MA and his Ph.D. in Neuroscience from Stanford University in 1974. After two postdoctoral fellowships – in Neurogenetics at Children's Hospital/Harvard Medical School, and in Neuropharmacology at the Biozentrum in Basel Switzerland – he joined the faculty of the Human Genetics Department of Yale Medical School in 1978 as an Assistant, then Associate, Professor. He became Director of the Division of Developmental Neurobiology at the E. K. Shriver Center in Waltham, MA in 1988. In 1992 he moved to the Departments of Neurosciences and Neurology at Case Western Reserve University Medical School and University Hospitals of Cleveland. While there, he directed the University Alzheimer's Center from 1999 through 2005. In 2006 he moved to the Piscataway/New Brunswick campus of Rutgers University to become Professor and Chair of the Department of Cell Biology and

Neuroscience. *Continued. . .*

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Karl Herrup *continued*: In addition to this leadership role in a large public university, he helped to found the Brain Health Institute, a unique public/private partnership devoted to basic research with relevance to clinical neuroscience. In July 2012, he moved to Hong Kong to become the Head of Life Sciences at Hong Kong University of Science and Technology. His laboratory research is focused on the biology of nerve cell death and the paradoxical role that failed cell cycle regulation plays in the process. His work includes a strong translational interest that directs his studies towards a few select human neurodegenerative diseases including Alzheimer's, a very common late-life dementia, and ataxia-telangiectasia, a very rare multisystem disorder of childhood. Dr. Herrup has authored a large number of highly cited papers and until 2010 served as the founding Senior Editor of the Neurobiology of Disease section of the *Journal of Neuroscience*.



Peng Jin, Ph.D., is Professor in the Department of Human Genetics at Emory University School of Medicine. Dr. Jin received his doctorate degree in Molecular and Developmental Biology from Cincinnati Children's Hospital/University of Cincinnati, and postdoctoral training at Emory University. At Emory, Dr. Jin is interested in the roles of epigenetic and noncoding RNAs in neurodevelopment/aging and brain disorders. Dr. Jin is the recipient of Beckman Young Investigator Award, Basil O'Connor Scholar Research Award from March of Dimes, Alfred P. Sloan Research Fellow in Neuroscience and NARSAD Independent Investigator Award.



Megan Keiser is a first year post-doctoral scholar that was born and bred in Iowa. She received her Bachelor's of Science degree in Integrative Physiology at the University of Iowa. In May, 2013 she received her doctorate in Neuroscience for her work in the Davidson lab at the University of Iowa where she focused on RNAi based therapies for Spinocerebellar Ataxia Type 1 (SCA1) in two different mouse models of SCA1. Currently, she is transitioning her therapies into non-human primates with the hopes of moving toward clinical applications.



Dr. Kamran Khodakhah obtained his bachelors (1989, Pharmacology, Kings College, University of London) and Ph.D. (1992, Pharmacology, University College, London) in the United Kingdom. His Ph.D. thesis research focused on examining the properties of InsP_3 -mediated calcium release in cerebellar Purkinje cells. He then performed a post-doctoral fellowship with Dr Clay Armstrong at the University of Pennsylvania, and subsequently assumed his first independent academic position in the department of Physiology and Biophysics at the University of Colorado Health Sciences Center in 1998. Dr Khodakhah moved to Albert Einstein College of Medicine in 2001 where he currently holds the rank of Professor as the Harold and Muriel Block Chair in Neuroscience. His interests revolve around understanding the role of the cerebellum and basal ganglia in motor coordination and movement disorders. His laboratory combines a multitude of techniques from electrophysiology and optophysiology in vitro and in vivo, to behavioral examination of transgenic animal models.

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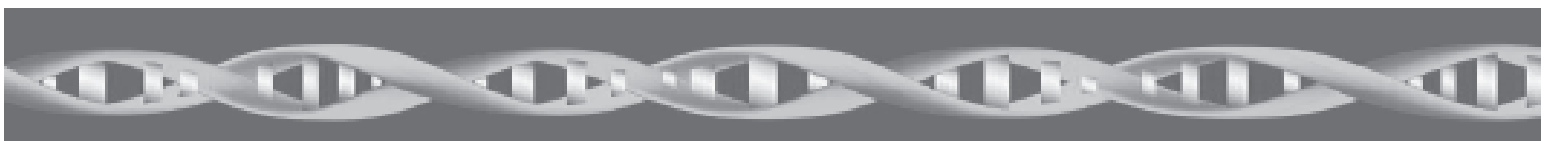


Dr. Holly Kordasiewicz is an Associate Director in the Neuroscience Drug Discovery Department at Isis Pharmaceuticals, a company that specializes in RNA therapies. She heads a team focused on identifying antisense oligonucleotide therapeutics for neurodegenerative diseases, including Huntington's disease, Alzheimer's disease, Parkinson's disease and inherited ataxias. Holly joined Isis 2 years ago after working on the pre-clinical validation for Isis' Huntington's disease program as a post-doctoral fellow in the laboratory of Dr. Don Cleveland at UCSD. Holly began her work on neurodegenerative diseases at the University of Minnesota, where she received her PhD in Neuroscience and studied the underlying disease mechanisms of SCA6 under the direction of Dr. Christopher Gomez.



Albert La Spada graduated *Summa Cum Laude* from the University of Pennsylvania with a degree in Biology in 1986. As a recipient of a Medical Scientist Training Program award, he pursued combined M.D. - Ph.D. training at the University of Pennsylvania School of Medicine. His 'Molecular Biology' doctoral thesis research focused upon a neuromuscular disorder known as X-linked spinal & bulbar muscular atrophy (SBMA) or Kennedy's disease. While a graduate student, La Spada identified the cause of SBMA as an expansion of a trinucleotide repeat in the androgen receptor gene. As the first disorder shown to be caused by an expanded polyglutamine repeat tract, this discovery of a novel type of genetic mutation led to the emergence of new field of study. After completing his M.D. - Ph.D. training in 1993, Dr. La Spada became a Laboratory Medicine resident at the University of Washington Medical Center and then a Clinical Genetics fellow. He pursued postdoctoral fellowship training as a Howard Hughes Medical Institute Physician Fellow, joined the faculty at the University of Washington Medical Center in 1998, and became a Professor of Laboratory Medicine, Medicine (Medical Genetics), Pathology, and Neurology (Neurogenetics). From 2004-2009, he was Director of the Center for Neurogenetics and Neurotherapeutics at the University of Washington. In 2009, Dr. La Spada accepted the position of Professor and Division Head of Genetics in the Departments of Pediatrics and Cellular & Molecular Medicine at the University of California, San Diego, and is a founding faculty member of the UCSD Institute for Genomic Medicine.

Dr. La Spada's research laboratory remains focused upon neurodegenerative disease. Dr. La Spada's laboratory is attempting to understand the molecular events that underlie the processes of neurodegeneration and neuron cell death in spinocerebellar ataxia type 7 (SCA7), and has found a number of connections between pathways involved in transcription and neuron dysfunction. By reproducing molecular pathology in model organisms such as mice, he has begun to use this knowledge to develop therapies to treat SCA7 and related disorders. Dr. La Spada has been the recipient of the Paul Beeson Aging Research Award, as well as grants and awards from the National Institutes of Health, Muscular Dystrophy Association, Hereditary Disease Foundation, CHDI, and the National Ataxia Foundation, including a Pioneer 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. Dr. La Spada sits on a variety of editorial boards and grant review committees, and he was inducted into the Association of American Physicians in 2013.

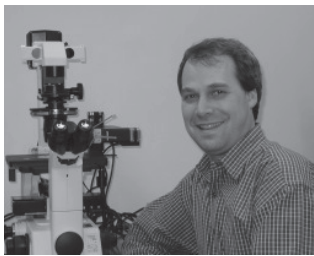


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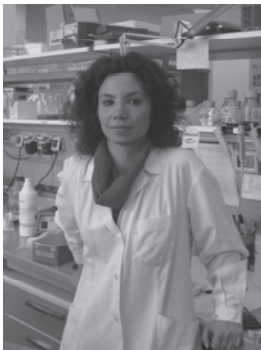


Dr. Lenglet is an Assistant Professor at the Center for Magnetic Resonance Research (Department of Radiology) and a Scholar of the Institute for Translational Neuroscience at the University of Minnesota. After receiving a M.Sc. in Computer Science & Engineering from the Compiègne University of Technology (Compiègne, France, 2003), and a M.Sc. in Applied Mathematics from École Normale Supérieure de Cachan (Cachan, France, 2003), he earned a Ph.D. in Biomedical Imaging and Neuroscience from INRIA Sophia Antipolis - Méditerranée (Sophia Antipolis, France, 2006). He then joined the Imaging and Visualization Department at Siemens Corporate Research in Princeton, New Jersey as a Research Scientist. In 2008, he moved to the University of Minnesota as a Research Associate in the Department of Electrical and Computer Engineering. In 2010, he became a faculty member of the Center for Magnetic Resonance Research.

Dr. Lenglet's group develops computational tools to harness the power of high-field Magnetic Resonance Imaging (MRI) for neuroscience and clinical applications. His research aims at better understanding the structural and functional alterations of brain connections in neurodegenerative disorders such as spinocerebellar and Friedreich's ataxias.

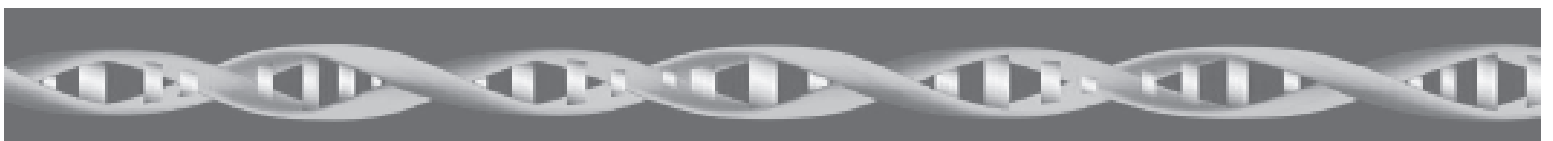


David Lynch, M.D., received his Bachelors degree in Molecular Biophysics and Biochemistry from Yale College in 1981, his PhD and MD in Neuroscience at Johns Hopkins University in 1988. His post-graduate training includes an internship in Internal Medicine, Hospital of the University of Pennsylvania, Philadelphia, PA, 1988-1989. His residency was in Neurology at the Hospital of the University of Pennsylvania, Philadelphia, PA, 1989-1992. In addition, Dr. Lynch was awarded a Postdoctoral Fellowship at the Departments of Pharmacology and Neurology, Clinical Specialties: Movement Disorders, Neurogenetics, University of Pennsylvania School of Medicine, Philadelphia, PA, 1992-1995.



Francesca Maltecca graduated *cum laude* in 2003 in Biotechnologies at the University of Milan under the supervision of dr. Servadio and then obtained a PhD in Molecular Medicine at the University Vita-Salute San Raffaele in Milan in 2008 under the supervision of prof. Casari. From 2007 to 2008 she was a visiting scientist in the Research Unit for Molecular Medicine headed by prof. Bross at the University Hospital Skejby in Aarhus, Denmark. After a post-doc in prof. Casari's lab (2009-2013), she recently got a tenure as university researcher at the University Vita-Salute San Raffaele in Milan. The main focus of her research is the study of the mitochondrial defects underlying Purkinje cell degeneration in spinocerebellar ataxia type 28 (SCA28), through *in vitro* and *in vivo* approaches. She is at present the recipient of the second year renewal of a NAF Young Investigator Award for the SCA Research. She also recently obtained the grant "Young Researchers" launched by the Italian

Minister of Health, a program promoting the startup of an independent career.



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Dr. Peter S. McPherson received his Ph.D. in Neuroscience from the University of Iowa in 1992 working with Howard Hughes Medical Institute Investigator Dr. Kevin P. Campbell. He performed post-doctoral training at Yale University in the laboratory of Dr. Pietro De Camilli, also an Investigator of the Howard Hughes Medical Institute. Dr. McPherson is currently the James McGill Professor in the Departments of Neurology and Neurosurgery and Anatomy and Cell Biology, McGill University.

Dr. McPherson's laboratory uses biochemical, molecular, structural, cellular, and proteomic approaches to identify and functionally characterize proteins that function in membrane trafficking with a focus on clathrin-mediated endocytosis. The study of proteins identified in Dr. McPherson's laboratory has led to conceptual breakthroughs in the mechanisms of endocytic vesicle formation and how endocytosis is linked to the actin cytoskeleton and signaling mechanisms. Moreover, several proteins under study in Dr. McPherson's laboratory are mutated in neurological diseases, specifically Parkinson disease, amyotrophic lateral sclerosis, and autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS). Indeed, dysregulation of membrane trafficking has emerged as a central theme in neurodegenerative diseases and Dr. McPherson is at forefront of this field.



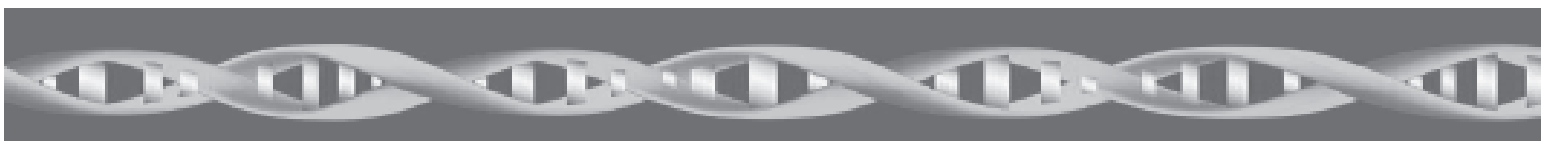
Dr. Medina was born in Madrid, but grew up in sunny Malaga on the Mediterranean coast of Spain. After high school, he was awarded a full-tuition scholarship to study in the United States. He received dual BS degrees in Physics and in Computer Science from Drexel University (Philadelphia) in 1993, and a PhD in Neuroscience from The University of Texas (Houston) in 2000. His thesis work and subsequent post-doctoral studies at The University of California (San Francisco) have a common goal that continues to drive his research program today: to achieve a full mechanistic understanding of how the brain learns to control our movements with precision. Since 2008 Dr. Medina has been an assistant professor in the Psychology Department at The University of Pennsylvania (Philadelphia). His laboratory is currently using computational

approaches and tools from pharmacology, *in vivo* neurophysiology and optogenetics to examine how olivo-cerebellar circuits contribute to simple forms of motor learning in behaving animals.

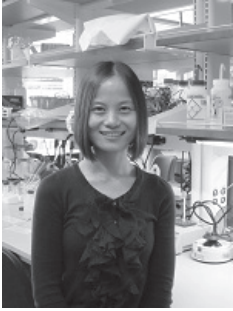


Dr. Andrea H Németh is a Clinician Scientist at the University of Oxford. She completed her medical training at the Royal Free Hospital in London and moved to Professor Kay Davies lab, completing her DPhil at the University of Oxford in 1995. Following this she was awarded a prestigious MRC Clinician Scientist Fellowship and during this time developed her interest in ataxias, going on to characterise several novel genetic ataxia syndromes. Dr Németh became a Consultant in Neurogenetics at the Oxford University Hospitals and recently joined the Nuffield Department of Clinical Neurosciences at the University of Oxford.

Her research interests focus on understanding genetic mechanisms underlying normal function and disease within the central nervous system. Recent work has used next generation sequencing (NGS) to characterize novel ataxia syndromes and this research has also led to the development of genetic testing for ataxias which is available as a clinical service.



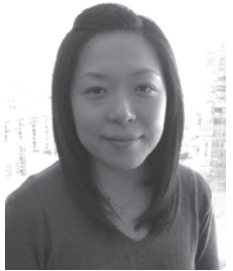
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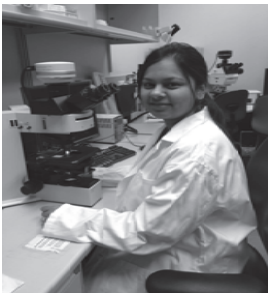
Dr. Chenchen Niu received her M.D. from the school of medicine, Soochow University, China. Then she obtained her Ph.D. in neuroscience from University of Hong Kong in 2010. Her thesis work focused on the neuroprotective signaling mechanisms of telomerase via the induction by Brain-derived Neurotrophic Factor (BDNF) in nervous system injury. In 2012 she joined Dr. Albert La Spada's lab as a postdoctoral research fellow and is currently working on developing gene silencing therapies to treat spinocerebellar ataxia type 7 (SCA7) and related neurodegenerative disorders.



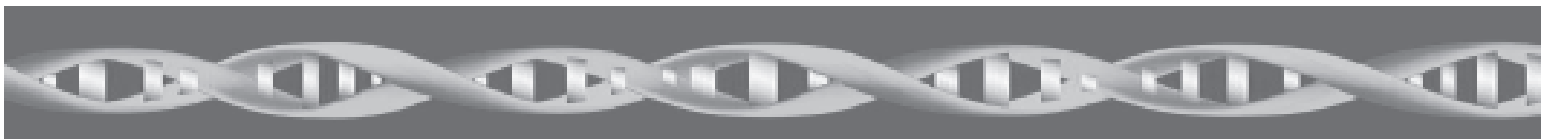
Harry Orr, Ph.D., directs the Institute for Translational Neuroscience and is the Tulloch Professor of Genetics in the Department of Laboratory Medicine and Pathology at the University of Minnesota Medical School. Dr. Orr received a BA degree from Oakland University in Rochester, Michigan. He earned 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, the Research Director of NAF's Medical Research Advisory Board and served as the lead organizer for the AIM 2014.



Dr. Park graduated with a Ph.D. in the Department of Biology from Korea Advanced Institute of Science and Technology (Daejeon, South Korea). In Dr. Jongkyeong Chung laboratory, her thesis focused on investigating the underlying pathogenic mechanism of Parkinson's disease by studying the function of the causative genes *PINK1* and *Parkin* in *Drosophila*. She continued her postdoctoral studies in Dr. Huda Zoghbi laboratory at Baylor College of Medicine and devised an unbiased screen strategy to identify the therapeutic targets for Spinocerebellar Ataxia type 1 (SCA1) and found that components in the MAPK/MSK1 pathway could potentially be therapeutic targets for SCA1. She is also investigating the function of ATXN1 interactors to further understand SCA1 pathogenesis.



Pavitra Ramachandran is a fifth year graduate student at the University of Iowa. She grew up in Chennai, India, where she received a BS in Zoology and an MS in Human Genetics. At the University of Iowa, she is pursuing a Ph.D. under the mentorship of Dr. Beverly Davidson. Her research work focuses on identifying therapies for Spinocerebellar ataxia types 2 and 7. She will be graduating in May 2014.



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Edgardo Rodriguez-Lebron was born in Arroyo, Puerto Rico and currently holds the title of research assistant professor at the University of Iowa Carver College of Medicine. He obtained his doctoral degree in neuroscience from the University of Florida where he performed some of the early work in support of viral-based RNA interference as a therapy for Huntington's disease. Following the completion of his graduate work in 2005, Edgar joined the laboratory of Dr. Henry L. Paulson at the University of Iowa to further the development of RNA interference as a therapeutic approach to neurological disease. Under the mentorship of Dr. Paulson, he began to focus his research on the Spinocerebellar Ataxias, specifically Spinocerebellar Ataxia types 3 and 6. He joined the faculty at the University of Iowa Department of Internal Medicine in the summer of 2010 to work closely with Dr. Beverly L. Davidson in the development of novel therapeutic approaches to Spinocerebellar Ataxias. His current research focuses on the development of genome editing as a molecular therapeutic approach to nucleotide repeat diseases.



Thorsten Schmidt, Ph.D., born in 1973, studied Biochemistry at the Ruhr-University Bochum in Bochum/Germany and graduated in 1997 (Dipl. Biochem.). He received his Ph.D. (Dr. rer. nat.) in 2003 from the University of Rostock/Germany for his thesis entitled „The Pathogenesis of Spinocerebellar Ataxia Type 3“ which was awarded to him with distinction „*summa cum laude*“. Since 2006 he is heading the SCA3 research group at the Institute of Medical Genetics and Applied Genomics of the Eberhard Karls University in Tuebingen/Germany. Moreover, he is responsible for the institute's teaching program for students of natural sciences. His research focusses on pathogenic mechanisms of Spinocerebellar ataxia type 3. In order to dissect these mechanisms, Dr. Schmidt generated one of the first antibodies against ataxin-3 as well as several different transgenic mouse models of this disease. He studies modifying factors of the disease and develops treatment strategies for SCA3 using molecular biological, protein biochemical and cell biological methods.



Vikram Shakkottai, MBBS, Ph.D., received his medical degree from the Christian Medical College, Vellore, India in 2000. He then completed a Ph.D. in biological sciences at the University of California, Irvine in 2004 and a residency in neurology at Washington University in Saint Louis in 2008. He subsequently did a fellowship in movement disorders at the University of Michigan. He was appointed Assistant Professor of Neurology at the University of Michigan in July 2010.

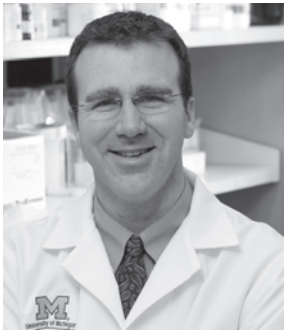
Dr. Shakkottai's research and clinical interests concern understanding the physiologic changes in the cerebellum that accompany cerebellar ataxia. Using mouse models of cerebellar ataxia he has contributed to understanding changes in firing patterns of neurons in the cerebellum that underlie aberrant coordination. His work suggests that reestablishing normal patterns of firing in cerebellar neurons, even in the presence of neuronal loss, might have therapeutic potential.

Dr. Shakkottai has received numerous awards in medical school and was ranked #1 in his medical class. He was awarded the Dorothy Penrose Stout Award for the Best Predoctoral Fellowship application from the American Heart Association Western States Affiliate 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.

AIM 2014 SPEAKER BIOGRAPHIES



Cleo Smeets is completing her PhD work in the field of spinocerebellar ataxia with Dr D Verbeek at the Department of Genetics of the University Medical Center Groningen and Rijksuniversiteit Groningen in the Netherlands. Before starting her PhD, she obtained a Bachelor's degree in Molecular Life Sciences and a Master's degree in Clinical Molecular Sciences at Maastricht University in the Netherlands. During this time, she developed an interest in neuroscience in general and the neurodegenerative field in particular. She explored these topics by carefully choosing internships in major depressive disorder with Dr J Prickaerts at the Department of Mental Health and Neuroscience of Maastricht University, and in Huntington's disease with Prof Dr G Bates at the Department of Medical & Molecular Genetics of King's College London School of Medicine, for which she received a scholarship from the Prinses Beatrix Fund. Currently, Cleo is focusing on elucidating the molecular mechanism of spinocerebellar ataxia type 23, using cell and mouse models. She recently received a grant from the De Cock Foundation to study the behavioral and intrinsic neurotoxic effects of SCA23-mutant Dynorphin A peptide in mice.



Peter Todd, M.D., Ph.D. is the Bucky and Patti Harris professor of neurology in the University of Michigan Medical School. Dr. Todd graduated from the University of California, San Diego and obtained his medical and doctoral degrees at the University of Wisconsin. He completed a medical internship and residency in neurology at the Hospital of the University of Pennsylvania, and a research intensive fellowship in movement disorders and neurogenetics at the University of Michigan. Dr. Todd's research targets the mechanisms by which RNA and RNA processing contribute to neurodegenerative disorders, with a specific interest in Fragile X-associated tremor ataxia syndrome (FXTAS), myotonic dystrophy, and C9orf72-associated ALS and FTD. In addition, Dr. Todd is a co-director of the multidisciplinary Ataxia Clinic at the University of Michigan and sees adult patients with all Fragile X Spectrum Disorders.



Lynn Ulatowski, Ph.D. For her undergraduate studies Lynn attended Westminster College in New Wilmington PA, where she obtained a B.S. in Molecular biology and played on the basketball team. After Westminster Lynn worked as a research assistant at Case Western Reserve University in Cleveland Ohio. During this time she earned her Master's degree in Nutrition while studying the regulation of the cystic fibrosis (CFTR) gene. Lynn's Ph.D. thesis initially focused on vitamin E status in the lysosomal storage disorder Niemann Pick type C (NPC) disease, and the mechanisms by which the tocopherol transfer protein (TTP) regulates vitamin E status. In recent years, Lynn focused her research on understanding the molecular mechanisms that underlie a debilitating neurological disorder, ataxia with vitamin E deficiency (AVED). Specifically, Lynn deciphered the molecular routes of vitamin E transport in the central nervous system, and how these are perturbed in humans afflicted with this familial disease. Lynn is currently a postdoctoral fellow at CWRU, and aspires for an independent research career that centers on the role of oxidative stress in the initiation and progression of ataxias. She also looks forward to teaching and mentoring students. Lynn has a teenager daughter to keep her grounded. For balance, Lynn enjoys being outdoors, hiking, biking and running, including completing three marathons.

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You are invited to become a Professional Member of the National Ataxia Foundation!

It is the mission of the Foundation to improve lives through support, education and research. As a professional who has an interest in ataxia, we invite you to become a part of this 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 mechanism of ataxia as well as medical interventions. Through membership, NAF is able to provide the following:

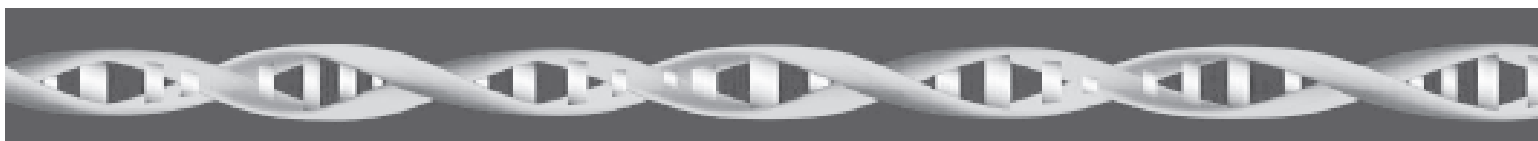
- Education and support services for patients who are affected by ataxia
- Ataxia Investigator Meetings, such as the one you are attending, which bring researchers together to accelerate ataxia research
- Current and accurate medical and genetic information on its website, www.ataxia.org
- Local and on-line support groups to help patients cope with emotional issues related to ataxia
- Annual Membership Meetings with world-leading ataxia researchers and neurologists
- Representation at various Ability Expos and medical conferences to raise ataxia awareness
- Publication and distribution of the Foundation's quarterly newsletter, *Generations*
- Neurologist, movement disorder and ataxia clinic referral information

...and much, much more.

As a Professional Member you will be included with other scientists and researchers who have made a commitment to researching neurological and genetic diseases. Your Professional Membership provides you with uninterrupted issues of *Generations*, registration discounts for NAF's annual membership meeting, and information on research funding opportunities. Joining NAF gives you unique access to our services and information.

Professional Membership enables neuroscientists to take an active part in furthering a cause that directly affects their careers. You can become a professional member now for a membership donation of as little as \$55.00 per year. Please go to our secure website at www.ataxia.org to become a member and support the important work of the Foundation which includes funding ataxia research.

Join us in supporting ataxia research by aligning yourself with the only national organization focusing 100% of its efforts on all types of ataxia.



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ANNOUNCING

58th NAF Annual Membership Meeting

March 13-15, 2015

Sheraton Denver Downtown

Denver, CO

