

4th Ataxia Investigators Meeting (AIM 2012)



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March 13-16, 2012 * Grand Hyatt San Antonio * San Antonio, TX



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Thank You

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

Henry Paulson, M.D., Ph.D.
Christopher M. Gomez, M.D., Ph.D.

and

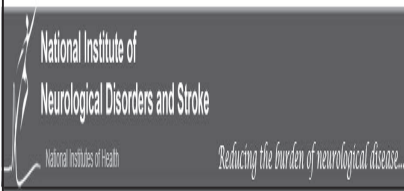
Steering Committee Members:

Joanna Jen, M.D., Ph.D.	Harry Orr, Ph.D.	Helene Puccio, Ph.D.
Stefan Pulst, M.D.	Laura Ranum, Ph.D.	Jeremy Schmahmann, M.D.
George Wilmot, M.D., Ph.D.		Robert Wilson, MD, Ph.D.

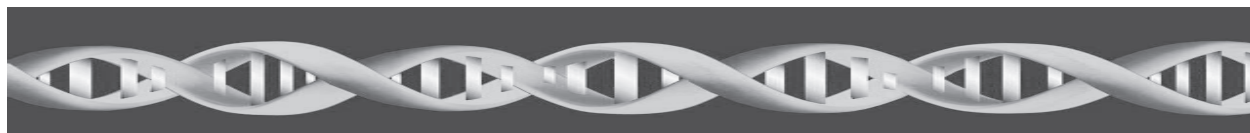
We are grateful for the generous support of the 4th Ataxia Investigators Meeting
from the following partners:

The Gordon and Marilyn Macklin Foundation
An Anonymous Donor

ApoPharma Inc.



Thank you to Church Offset Printing, Inc.
for your support of the
2012 Ataxia Investigators Meeting.



Welcome

Dear AIM 2012 Attendee:

Welcome to the Fourth International Ataxia Investigators' Meeting, "AIM2012: Advancing Toward Therapeutics," hosted by the National Ataxia Foundation with support from the National Institutes of Health, Office of Rare Disease Research, ApoPharma, Inc., Ataxia UK, Ataxia Ireland, Athena Diagnostics, Inc., A-T Children's Project, Bob Allison Ataxia Research Center, Friedreich's Ataxia Research Alliance, the Gordon and Marilyn Macklin Foundation and an anonymous donor. The primary goal of the AIM conferences, which began in 2005, is to bring together a group of international scientific investigators to address the multi-disciplinary nature of ataxia, better define the pathogenic basis of ataxia, explore routes to therapy, and help establish future leaders in ataxia research. Toward that goal, we are very excited with the final AIM2012 program.

AIM2012 will focus on the most recent scientific advances and emerging approaches to therapy, with five objectives: 1) Enhance the open exchange of information related to ataxia and therapeutic strategies; 2) Stimulate collaborative research between investigators worldwide; 3) Improve our understanding of ataxic disorders, including shared disease mechanisms; 4) Establish international protocols for the common investigation and storage of data related to ataxia and its treatment; and 5) Promote junior investigators by giving them an opportunity to present their work, interact with more established scientists, and meet persons with ataxia so they can see firsthand the impact of their work.

The three prior AIM conferences were instrumental in advancing ataxia research. We have every reason to believe that AIM2012 will continue to move ataxia research forward, bringing those affected by ataxia hope for a future in which treatments are available.

As you may know, AIM2012 dovetails with the annual membership meeting of the National Ataxia Foundation, which maximizes the impact of this meeting for scientists and patients alike.

We are pleased to announce a unique opportunity at this year's meeting. You are invited to attend to participate or observe a "Birds of a Feather" break-out sessions on Friday from 2 – 5 pm. The "Birds of a Feather" provides patients and their family members an opportunity to learn more about a particular type of ataxia or a specific topic which relates to their situation. We encourage you to attend one of these "Birds of a Feather" sessions.

Again, we welcome you to San Antonio and look forward to your participation in the conference. We encourage you to take full advantage of the stimulating presentations and networking opportunities that will be present throughout AIM2012.

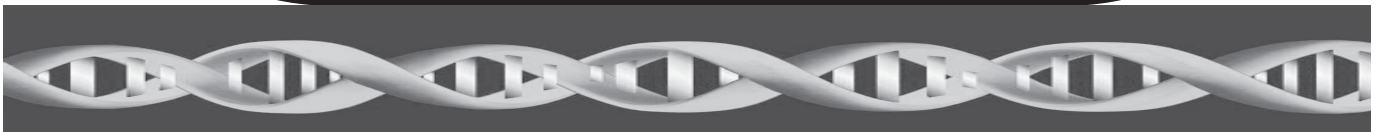
Your AIM 2012 Co-Lead Organizers,

Henry Paulson

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

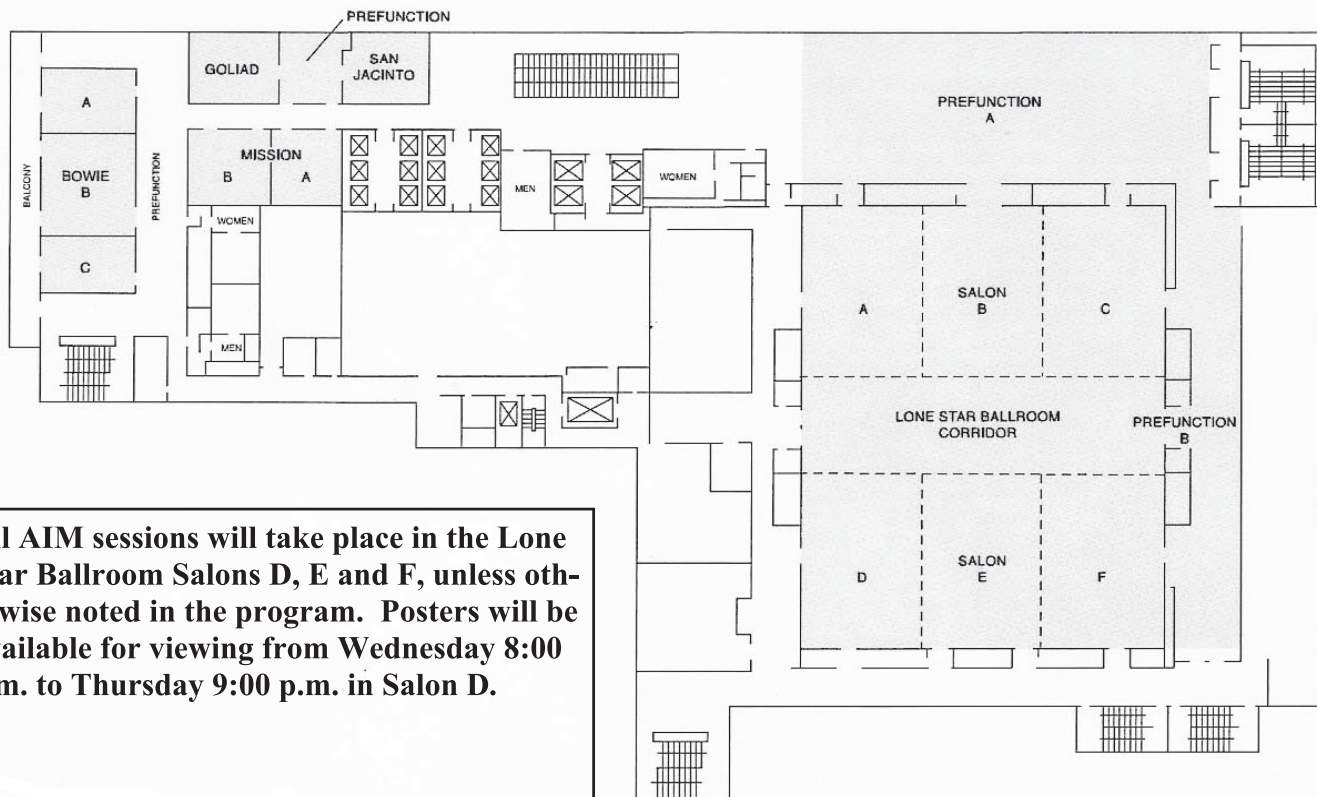
Christopher M. Gomez

Christopher M. Gomez, M.D., Ph.D.

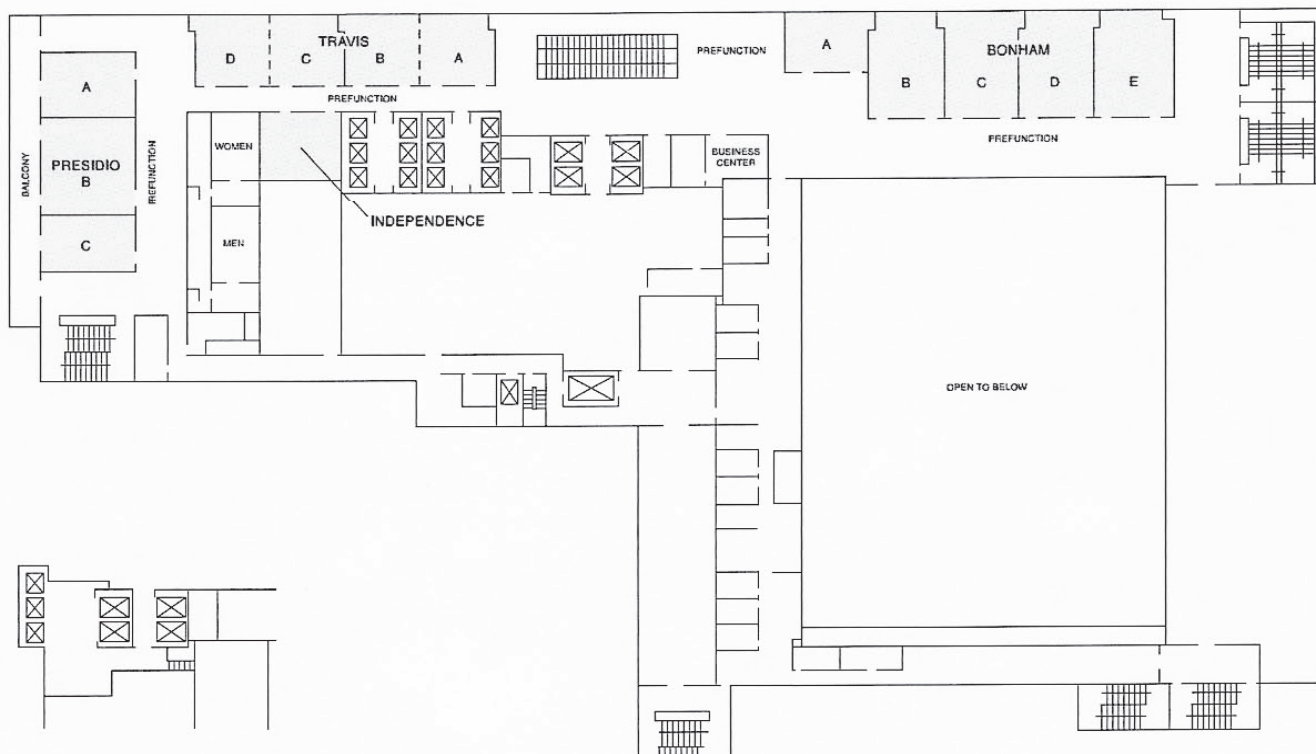


Grand Hyatt San Antonio Floor Plans

SECOND LEVEL



THIRD LEVEL



AIM 2012 Agenda at-a-Glance

TUESDAY MARCH 13

4:00-7:00 p.m.	AIM Registration
4:00-6:00 p.m.	Welcome Reception
6:00 p.m.	Dinner and Opening Remarks: Henry Paulson, M.D., Ph.D.
7:00 p.m.	Keynote Address: Huda Zoghbi, M.D.

WEDNESDAY MARCH 14

7:00-8:00 a.m.	Breakfast
8:00 a.m.	Keynote Address: Nancy Bonini, Ph.D.
8:40 a.m.-12:20 p.m.	Theme 1 Presentations: Cerebellar function and dysfunction in ataxia
12:20-1:00 p.m.	Lunch
1:00	Keynote Address: Christian DeZeeuw, M.D., Ph.D.
1:40-5:00 p.m.	Theme 2 Presentations: Pathogenesis of dominant ataxias
5:00-7:00 p.m.	Dedicated Poster Session with Wine and cheese
Evening	Networking Dinner on your own in San Antonio

Poster Sessions

Wednesday 5:00-7:00 p.m. there will be a dedicated poster session for all AIM attendees. Poster presenters should be available at their posters for the entire evening to present their important ataxia research findings. Wine and cheese will be served.

Thursday 12:20-1:00 p.m. is an opportunity to further review posters during lunch. Feel free to bring your box lunch into the poster session room.

Thursday 5:15-6:15 p.m. is a new offering for NAF's Annual Meeting attendees. All poster presenters should be available at their posters during this time to allow patients and family members to meet you and learn more about ataxia research and the importance of supporting research. This session should be attended by poster presenters only to allow room for wheel chairs and walkers in the poster session room.



AIM 2012 Agenda at-a-Glance

THURSDAY MARCH 15

7:00-8:00 a.m.	Breakfast
8:00 a.m.	Keynote Address: Robert (Berch) Griggs, M.D.
8:00 a. m.-12:20 p.m.	Theme 3 Presentations : Pathogenesis of recessive and other ataxias
12:20-1:00 p.m.	Lunch and Posters
1:00-5:15 p.m.	Theme 4 Presentations: Toward therapies through disease mechanisms
5:15-6:15 p.m.	Poster Session for Patients and Family Members
6:30 p.m.	Dinner
7:00 p.m.	Keynote Address: Hélène Puccio, Ph.D.

FRIDAY MARCH 16

7:00-8:00 a.m.	Breakfast
8:40 a.m.-12:40 p.m.	Theme 5 Presentations: Working together to advance ataxia research and therapy
12:40 p.m.	Closing Lunch
2:00-5:00 p.m.	<i>"Birds of a Feather"</i> Small Group Sessions with patients

Each of these small group sessions are facilitated by a researcher, clinician or other appropriate leader. Feel free to attend any session that may be of interest to you. You will be very welcomed by the attendees.

SCA1 in the Bonham C Room
SCA2 in the Bonham D Room
SCA3 in the Presidio B Room
SCA6 in the Bowie B Room
All Other SCAs in the Lone Star E
Sporadic/MSA in the Lone Star B Room
Unknown/Episodic/AOA in Lone Star A Room
Over Age 30 Friedreich's in the Lone Star C
Under age 30 with ataxia in the Lone Star F Room
Parents of Non-Friedreich's in the Bonham E Room
Parents of Friedreich's in the Lone Star D Room

Detailed AIM Schedule

TUESDAY, MARCH 13, 2012

- 3-4 p.m. Junior Lecturers Welcome Reception (By Invitation only)
- 4-6 p.m. Welcome Reception and Registration for all AIM Attendees
- 6-7 p.m. **Dinner**
- 7 p.m. Opening Remarks – Henry Paulson, M.D., Ph.D., University of Michigan
- 7 p.m. **Keynote Address: Huda Zoghbi, M.D.**, Baylor College of Medicine
Toward Therapy in SCA1

WEDNESDAY, MARCH 14, 2012

8:00AM Wednesday – 9PM Thursday Posters Up For Viewing

- 7:00 a.m. **Breakfast**
- Theme 1 – Cerebellar function and dysfunction in ataxia**
Chairs: Drs. Christopher Gomez & Khalaf Bushara
- 8:00 a.m. **Keynote Address: Nancy Bonini, Ph.D.**, University of Pennsylvania
Multiple triggers to neuronal death
- 8:40 a.m. Jeremy Schmahmann, M.D., Massachusetts General Hospital
Cerebellum and Neuropsychiatry: Radical Implications of a New Field
- 9:10 a.m. Vikram Shakkottai, M.D., Ph.D., University of Michigan
Targeting potassium channels for the treatment of cerebellar ataxia
- 9:40 a.m. **Break 30 Minutes**
- 10:10 a.m. Joanna Jen, M.D., Ph.D., UCLA
RNA exosome in cerebellar and spinal motor neuron maldevelopment and degeneration
- 10:40 a.m. Mandy Jackson, Ph.D., University of Edinburgh
Progressive cerebellar dysfunction in a mouse model of SCA5: the beta-III spectrin connection



Detailed AIM Schedule

WEDNESDAY, MARCH 14, 2012, continued

11:10 a.m. Junior Lecturer: Xiaofei Du, M.D., University of Chicago
P/Q calcium channel $\alpha 1A$ C termini regulation of Purkinje cell gene expression, dendritic morphology and synaptic input are abolished with SCA6.

11:30 a.m. Junior Lecturer: Yuanzheng Gao, Johns Hopkins University
(now at University of Florida, Gainesville)
Beta III sepectrin is critical for development of Purkinje neuron dendrites and spines

11:50 a.m. Discussion – All participants

12:20 a.m. **Lunch 40 Minutes**

Theme 2 – Pathogenesis of dominant ataxias

Chairs: Drs. Harry Orr & Joseph Savitt

1:00 p.m. **Keynote Address: Christian De Zeeuw, M.D., Ph.D.**, Erasmus MC, Netherlands
New insights into the role of the cerebellum in health and disease

1:40 p.m. Hirokazu Hirai MD, Ph.D., Gunma University Graduate School of Medicine, Japan
Mutant PKC γ in spinocerebellar ataxia type 14 impairs synapse elimination and synaptic plasticity in Purkinje cells in vivo.

2:10 p.m. Hidehiro Mizusawa, M.D., Tokyo Medical and Dental University, Tokyo
Progress in the research on SCA31

2:40 p.m. Laura Ranum, Ph.D., University of Florida,
Repeat Associated Non-AUG Translation: Lessons from SCA8

3:10 p.m. **Break 15 minutes**

3:25 p.m. Patricia Maciel, Ph.D., University of Braga, Portugal
Modeling MJD/SCA3 in the mouse and in C. elegans: probing pathogenesis towards therapies

3:55 p.m. Junior Lecturer: Keyi Zhu, M.D., Ph.D., Baylor College of Medicine
Identification of Common Pathogenic Mechanisms of Spinocerebellar Ataxias (SCA) Using Drosophila Models

4:15 p.m. Junior Lecturer: Tyisha Hathorn, Ph.D., University of Florida
mGluR1 mislocalization and LTP deficits in a mouse model of spinocerebellar ataxia type 5

4:35 p.m. Discussion – All Participants

5:00-7:00 p.m. **Poster Session** for all AIM Attendees

Evening **Networking Dinner on your own in San Antonio**



Detailed AIM Schedule

THURSDAY, MARCH 15, 2012

7:00 a.m. **Breakfast**

Theme 3 – Pathogenesis of recessive and other ataxias

Chairs: Drs. Rob Wilson and Joanna Jen

8:00 a.m. **Keynote Address: Dr. Robert (Berch) Griggs, M.D.**, University of Rochester
Designing Clinical Trials for Rare Diseases

8:40 a.m. Marek Napierala, Ph.D., University of Texas MD Anderson Cancer Center
Epigenetic silencing in Friedreich's ataxia as target for therapy

9:10 a.m. Yosef Shiloh, Ph.D., Tel Aviv University, Tel Aviv, Israel
Ataxia-telangiectasia: do the known functions of ATM explain the phenotype?

9:40 a.m. Peter Todd, M.D., Ph.D., University of Michigan
Toward understanding Fragile X Tremor Ataxia Syndrome (FXTAS)

10:10 a.m. **Break 30 Minutes**

10:40 a.m. Tetsuo Ashizawa, M.D., University of Florida
Sequence complexity of expanded ATTCT repeats and its phenotypic correlation in SCA10

11:10 a.m. Junior Lecturer: Floriana Licitra, Ph.D Student, IGBMC, Illkirch, France
Pathophysiological Mechanisms Underlying Recessive Ataxia Associated With Coenzyme Q10 Deficit

11:30 a.m. Junior Lecturer: Natalia Gromak, Ph.D., Oxford
Molecular function of human senataxin protein, mutated in AOA2/ALS4 disorders, in the regulation of gene expression

11:50 a.m. Discussion – All Participants

12:20 p.m. **Lunch and Posters 40 Minutes**

Theme 4 – Toward therapies through disease mechanisms

Chairs: Drs. Sarah Ying & Al LaSpada

1:00 p.m. Luis Pereira de Almeida, Ph.D., University of Coimbra, Portugal
Disease-modifying strategies for Machado-Joseph disease

1:30 p.m. Robert Wilson, M.D., Ph.D., University of Pennsylvania
Experimental Therapeutics for Friedreich Ataxia

2:00 p.m. Edgardo Rodriguez-Lebron, Ph.D., University of Iowa
Selective targeting of the polyQ-encoding Cav2.1 splice isoform



Detailed AIM Schedule

THURSDAY, MARCH 15, 2012, continued

- 2:30 p.m. Puneet Opal, M.D., Ph.D., Northwestern University, Chicago, IL
Vascular Endothelial Growth Factor in Spinocerebellar Ataxia
- 3:00 p.m. **Break 30 Minutes**
- 3:30 p.m. Gulin Oz, Ph.D., University of Minnesota
*Surrogate Marker Potential of in vivo
Magnetic Resonance Spectroscopy in Ataxia Trials*
- 4:00 p.m. Junior Lecturer: Peter Breuer, Ph.D., University of Bonn Medical Center, Germany
Calpain-mediated Ataxin 3 Aggregation in Mice and Men
- 4:20 p.m. Junior Lecturer: Sara Lagalwar, Ph.D., University of Minnesota
*Inhibition of Ataxin-1 S776 phosphorylation:
A strong candidate for SCA1 therapeutic*
- 4:40 p.m. Discussion – All Participants
- 5:15-6:15 p.m. **Poster Session for Patients**
- 6:30 p.m. **Dinner**
- 7:00 p.m. **Keynote Address: Helene Puccio, Ph.D.**, Research Director INSERM,
New insights into mechanisms of recessive ataxias

FRIDAY, MARCH 16, 2012

- 7:00 a.m. **Breakfast**
- Theme 5 – Working together to advance ataxia research and therapy**
Chairs: Drs. S.H. Subramony & Wendy Galpern
- 8:00 a.m. Thomas Klockgether, M.D., University Hospital Bonn, Germany
Spinocerebellar ataxias: The prodromal phase
- 8:30 a.m. John Ferguson, M.D. National Institutes of Health
Office of Rare Disease Research
- 9:00 a.m. Ludger Schöls, M.D., Hertie-Institute for Clinical Brain Research, Germany
Characterizing POLG ataxia (and comparison to other mitochondrial ataxias)
- 9:30 a.m. Shoji Tsuji, M.D., Ph.D., University of Tokyo
*Whole genome resequencing to elucidate
the molecular basis of multiple system atrophy*



Detailed AIM Schedule

FRIDAY, MARCH 16, 2012, continued

- 10:00 a.m. **Break 30 Minutes**
- 10:30 a.m. David Lynch, M.D., University of Pennsylvania
Collaborative research efforts in Friedreich ataxia
- 11:00 a.m. Theresa Zesiewicz, M.D., FAAN, University of South Florida
Review of Clinical Trials for Ataxia
- 11:30 a.m. Junior Lecturer: Andreia Castro, Ph.D., University of Minho, Portugal
*Searching for therapeutic compounds for Machado-Joseph disease:
a C. elegans-based screening*
- 11:50 a.m. Junior Lecturer: Anna Duarri, Ph.D., University of Groningen, The Netherlands
*Exome sequencing reveals disease gene underlying
spinocerebellar ataxia 19*
- 12:10 p.m. Discussion – All Participants
- 12:40 p.m. **Closing Lunch**
- 2-5 p.m. Birds of a Feather Sessions

These are small groups of patients and family members who are grouped by type of ataxia .You would be welcome to attend and participate in any of the groups shown below. Patients would be interested in meeting you and hearing about your research initiatives.

SCA1 facilitated by Puneet Opal and Harry Orr in the Bonham C

SCA2 facilitated by Gulin Oz and Joseph Savitt in the Bonham D

SCA3 facilitated by Henry Paulson and Theresa Zesiewicz in the Presidio B

SCA6 facilitated by Christopher Gomez in the Bowie B

All Other SCAs facilitated by Laura Ranum and Al LaSpada in the Lone Star E

Sporadic/MSA facilitated by Jeremy Schmahmann and Tetsuo Ashizawa in the Lone Star B

Unknown/Episodic/AOA Joanna Jen and Larry Schut in Lone Star A

Over Age 30 Friedreich's facilitated by Helene Puccio, Susan Perlman, David Lynch & Mark Payne in the Lone Star C

Under age 30 facilitated with ataxia by Sarah Ying, Matthew Bower and George (Chip) Wilmot in the Lone Star F

Parents of Non-Friedreich's facilitated by S.H. Subramony, Chip Wilmot & Larry Schut in the Bonham E

Parents of Friedreich's Susan Perlman, Mark Payne, David Lynch and parent, David Zilles in the Lone Star D



Poster Presenter Travel Grant Awardees

The following investigators' abstracts were reviewed and selected for a travel grant to attend AIM 2012. These and other invited poster presenters will have their research on display during the poster sessions and can be viewed during meals and breaks.

- | | | |
|----|--------------------------------|--|
| 1 | Alves, Sandro, Ph.D. | Selective molecular autophagic defects in rodent models and in patients of Spinocerebellar ataxia type 7
Université Pierre et Marie Curie-Paris |
| 2 | Collins, Sarah | Disruption of the cerebellar module causes the main clinical phenotype in hereditary ataxia
VA Medical Center, Albany, NY |
| 3 | Costa, Maria do Carmo, Ph.D. | RNAi-mediated suppression of mutant ataxin-3 in a transgenic mouse model of Machado-Joseph disease
University of Michigan |
| 4 | Du, Jintang , Ph.D. | Generation of expanded GAA•TTC triplet repeats in Friedreich's Ataxia iPSCs
The Scripps Research Institute |
| 5 | Dupré, Nicolas, M.D. | A mutation in the RNF170 gene causes autosomal dominant sensory ataxia
Laval University, Quebec City, Canada. |
| 6 | Evans-Galea, Marguerite, Ph.D. | <i>FXN</i> methylation predicts expression and clinical outcome in Friedreich ataxia
Murdoch Children's Research Institute |
| 7 | Hekman, Katherine , B.A. | Loss of Translational Fidelity in SCA26
University of Chicago |
| 8 | Horton, Laura , B.A. | Second abstract - Spinocerebellar Ataxia Type 7: Clinical Course, Phenotype-Genotype Correlations, Neuropathology
Massachusetts General |
| 9 | Ittis, Isabelle, Ph.D. | State-of-the art 1H MR Spectroscopy (1H MRS) on clinical platform for Neuro-chemical characterization of patients with Ataxia with Oculomotor Apraxia Type 2 (AOA2)
University of Minnesota |
| 10 | Kasumu, Adebimpe, B.S. | Suppressing IP3 signaling delays the onset of Spinocerebellar Ataxia 2
University of Texas Southwestern |
| 11 | Ladd, Paula, Ph.D. | Convergent Transcription at the Ataxin-7 Locus Produces dsrna Fragments that are Processed by Dicer
University of California, San Diego |
| 12 | Mason, Amanda, B.S., M.S. | CAG/CTG Repeat Instability Differences In Striatum And Cerebellum Are Paralleled By Marked Expression Variation In DNA Replication And Mismatch Repair Proteins
University Of California, San Diego |
| 13 | Ramani, Biswarathan, B.S. | Characterization Of A Knock-In Mouse Model Of SCA3
University Of Michigan |
| 14 | Scoles, Daniel, Ph.D. | Features of <i>ATXN2</i> expression control
University of Utah |
| 15 | Soragni, Elisabetta, Ph.D. | Understanding The Mechanism Of Action Of 2-Aminobenzamide HDAC Inhibitors In Reversing <i>FXN</i> Gene Silencing
The Scripps Research Institute, La Jolla CA |



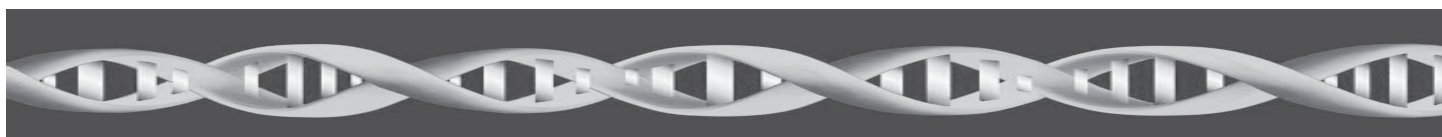
Invited Poster Presenters

- | | |
|---|---|
| 16 Almeida, Bruno, Ph.D. | Ataxin-3:p97/VCP interaction: a route for Ataxin-3 three-dimensional structure determination
Universidade do Porto Portugal |
| 17 Becker, Esther, Ph.D. | TRPC3 signaling in Purkinje cell development and cerebellar ataxia
University of Oxford, UK |
| 18 Bosch AK, Marie, B.S. | FGF14 Regulation of Nav channels in Cerebellar Purkinje Neurons
Washington University in St. Louis, MO |
| 19 Bower, Matthew, MS, CGC | Technical and clinical validation of a next generation sequencing diagnostic assay for rare ataxias.
University of Minnesota |
| 20 Burmeister, Margit, Ph.D. | Identification of a novel recessive ataxia gene using integrated genomic analysis
University of Michigan |
| 21 Busenlehner, Laura, Ph.D. | Over-Expression of Frataxin Results in Severe Growth Inhibition in Fission Yeast
University of Alabama |
| 22 Chen, Dong Hui, M.D., Ph.D. | A spinocerebellar ataxia associated with hematologic cytopenias
University of Washington School of Medicine |
| 23 Collins, Abigail, M.D. | Glut-1 transporter deficiency syndrome, an under-recognized, potentially treatable form of "ataxic cerebral palsy."
University of Colorado |
| 24 Copeland, Brian, M.D. | Abnormal cerebrospinal fluid neurotransmitter profiles in a father and daughter with spinocerebellar ataxia type 1.
University of Texas Health Sciences Center |
| 25 Cvetanovic, Marija, Ph.D. | Glial pathology in SCA1: cellular and molecular mechanisms
Northwestern University |
| 26 Duarte-Silva, Sara
Presented by Patricia Maciel | Lithium Treatment Does Not Ameliorate The Phenotype Of A New Transgenic Mouse Model Of Machado-Joseph Disease
University of Minho, Braga, Portugal |
| 27 Evert, Bernd, Ph.D. | Modulation of ATXN3 gene expression by microRNAs
University Clinics Bonn, Bonn, Germany |
| 28 Fan, Caleb, B.S. | A Graphical User Interface For Automated Evaluation Of Cerebellar Shape In MR Images
Johns Hopkins University |
| 29 Figueroa, Pattie, B.S., M.S. | Prior molecular diagnostic accuracy and age of disease onset variation in the CRC-SCA, a multicenter study of spinocerebellar ataxias
University of Utah |
| 30 Fogel, Brent, M.D., Ph.D. | Mutations In Rare Ataxia Genes Are Uncommon Causes of Sporadic Cerebellar Ataxia
UCLA |



Invited Poster Presenters

- | | | |
|----|---|--|
| 31 | Giunti, Paola, M.D., Ph.D. | Detecting retinal changes in autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) and other genetic ataxias using optical coherence tomography (OCT)
UCL Institute of Neurology, London, UK |
| 32 | Hansen, Stephen, Ph.D. | SCA6 Epistatic modulation of the SCA2 phenotype
University of Utah |
| 33 | Hearst, Scoty, Ph.D. | The Design and Delivery of Thermally Responsive PKA and 14-3-3 Inhibitory Peptides to Treat SCA1
University of Mississippi Medical Center |
| 34 | Hunegs, Lisa
Presented by Jennifer Farmer, MS, CGC | The National Institute of Neurological Disorders and Stroke (NINDS) Common Data Elements (CDEs) for Use in Friedreich's Ataxia Clinical Studies
KAI Research, Inc |
| 35 | Koeppen, Arnulf, M.D. | Tissue Donations provide research opportunities in hereditary ataxia
Albany Research Institute , Albany, NY |
| 36 | Mahishi, Lata, Ph.D. | MicroRNAs in Friedreich Ataxia
Cornell University New York, NY |
| 37 | Nobrega, Clevio, Ph.D.
Presented by de Almeida | RNA interference rescues cerebellar abnormalities and ataxia in a lentiviral-based model of Machado-Joseph disease
University of Coimbra Portugal |
| 38 | Seki, Takahiro, Ph.D. | Effects Of The SCA14 Disease Protein Gpkc On Cellular Protein Quality Control Systems
University Of Michigan |
| 39 | Smith, Danielle, Ms.CC. | The polyglutamine SCAs in a developing African country: Past Perspectives and Future Prospects
University of Cape Town, Cape Town, South Africa |
| 40 | Subramony, S.H., M.D. | National Ataxia Registry
University of Florida Gainesville, FL |
| 41 | Takahashi, Emi, Ph.D. | Identification of Cerebellar Cortex and White Matter Pathways in Adult Humans Revealed by High Angular Resolution Diffusion Tractography
Massachusetts General |
| 42 | Watson, Lauren, Ph.D. Student | Therapeutic Gene Silencing For SCA7
University Of Oxford, UK |
| 43 | Xia, Guangbin, M.D., Ph.D. | Generation of disease-specific human induced pluripotent stem(iPS) cell as in vitro model of spinocerebellar ataxia type 2
University of Florida, Gainesville |
| 44 | Ying, Sarah, M.D. | Principal Component Analysis Of Brain MR Images May Be Used To Classify Cerebellar Shape Changes
Johns Hopkins University |



Ataxia Research Grant Information



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

The National Ataxia Foundation has 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 please 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 science.

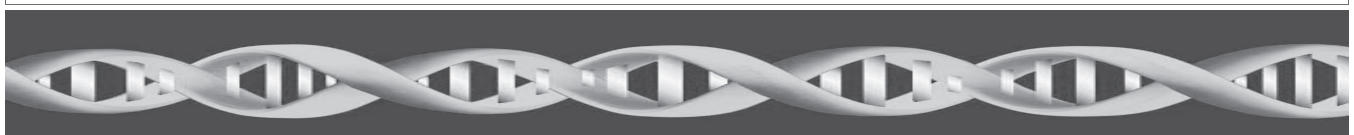
The Foundation supports hereditary ataxia research, but has also placed an emphasis on research into sporadic ataxia and is encouraging submission of research projects that will investigate non-genetic forms of ataxia. More information available at www.ataxia.org.

Applications forms for NAF's research programs will be posted on the website in April 2012.

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: 7/15/12

Full application due: 8/15/12



Ataxia Research Grant Information

Young Investigator (YI) Award: One year grant between \$35,000- \$50,000 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 and PhDs must be no more than five (5) years from end of the completion of their post doc training.

Letter of Intent with a ½ to one-page abstract with specific aims of your research due: 8/1/12

Full application due: 9/1/12

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: 8/15/12

Full application due: 9/15/12

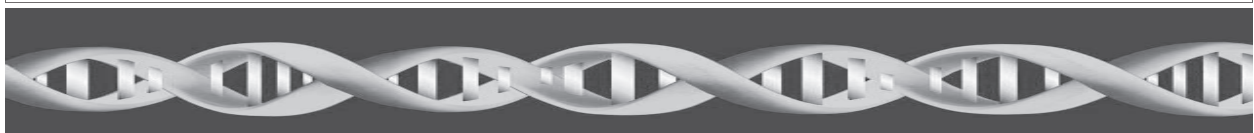
Pioneer SCA Translational Research Award: One year grant of \$100,000 focusing on research investigations that will facilitate the development of treatments for the Spinocerebellar Ataxias.

Instructions and due dates for letter of intent and full application will be announced on the website in April 2012.

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 and PhDs must be no more than five (5) years from end of the completion of their post doc training.

Instructions and due dates for letter of intent and full application will be announced on the website in April 2012.

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.



AIM 2012 Biographies



Luis Pereira de Almeida, Ph.D.

Luís Pereira de Almeida received his Ph.D. in Pharmaceutical Sciences from the University of Coimbra in 2003. During his Ph.D. studies, he has spent three years in the laboratory of Patrick Aebischer and Nicole Déglon at the Gene Therapy Center, CHUV in Lausanne, Switzerland. He integrated the Center for Neurosciences and Cell Biology of the University of Coimbra (CNC), Portugal in 1996 where he is principal investigator and member of the board. He spent short sabbatical leaves at Commissariat à l'Énergie Atomique, Saclay in France (2005) and at the Massachusetts Institute of Technology (2010).

Luis is assistant professor at the Faculty of Pharmacy, University of Coimbra and for some years has been involved in the coordination of the Doctoral Program in Experimental Biology and Biomedicine of CNC and in the organization of advanced courses on “Neurodegenerative diseases”, “Principles and Practice in Drug Development” (MIT Portugal -PhD Program)”, “Nuclear biology and gene therapy” and “Gene and cell therapy for the CNS”.

Luis has been responsible for research projects funded by the Portuguese Foundation for Science and Technology, the Association Française de Myopathies, and the National Ataxia Foundation (USA) and leads one of the twelve European research groups that constitute the Initial Training Network “TreatPolyQ” within the 7th Framework Program of the European Union. Currently supervises 4 Pos doctoral fellows and 5 PhD students.

His research has been focused on molecular therapy approaches for neurodegenerative disorders with a focus on Machado-Joseph disease/spinocerebellar ataxia type 3, including disease modifying and gene silencing approaches, autophagy activation and proteolysis inhibition, works published in thirty papers cited over 600 times, in journals such as Brain, Human Molecular Genetics, PLoS One, Human Gene Therapy and The Journal of Neurosciences, awarded with prizes by the Portuguese Society for Neurosciences (2009 and 2011) and the Portuguese Society of Human Genetics (2009).

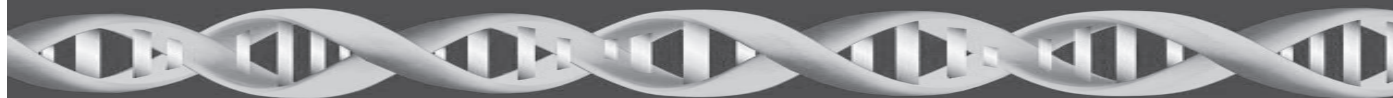


Tetsuo Ashizawa, M.D., Ph.D.

Dr. Tetsuo Ashizawa is Professor and Chairman of the Department of Neurology at the University of Florida (UF), Gainesville, Florida. Dr. Ashizawa also holds the Melvin Greer Professorship of Neurology. Dr. Ashizawa received his medical degree from the Keio University School of Medicine in Tokyo in 1973. He completed his neurology residency training and subsequent clinical and basic science fellowships at Baylor College of Medicine. In 1981 he joined the faculty at Baylor, where he climbed to the academic rank of tenured Professor in 1997. In 2002 Dr. Ashizawa was recruited to the University of Texas Medical Branch (UTMB) in Galveston, Texas to chair the Neurology Department, and then moved to Gainesville, Florida in April 2009 as Chair of the Department of Neurology at UF. He has

published over 180 papers in leading scientific and clinical journals and books.

Dr. Ashizawa's basic science research projects have primarily been focusing on neurogenetic disorders caused by expanded short tandem repeats, including myotonic dystrophy, Friedreich's ataxia and autosomal dominant spinocerebellar ataxias. His current research is to investigate the pathogenic mechanism of spinocerebellar ataxia type 10 (SCA10). Dr. Ashizawa is also the principal investigator of a nation-wide consortium for clinical research on SCA1, SCA2, SCA3 and SCA6. This consortium is one of the Rare Disease Clinical Research Consortia (RDCRC) organized and funded by NIH. This consortium will establish the infrastructure and database to prepare for future clinical trials of new therapies for SCAs.



AIM 2012 Biographies



Khalaf Bushara, M.D., FRCP (London)

Dr. Bushara is a movement disorders specialist with special interest in ataxia and diseases of the cerebellum. Dr. Bushara is a graduate of the University of Khartoum, Sudan. He was trained in internal medicine and neurology in the United Kingdom, where he was elected as a fellow of the Royal College of Physicians (London). He completed a neurology residency at the University of Wisconsin-Madison and a fellowship in Movement Disorders at the human motor control section at the National Institutes of Health where he gained experience with ataxia patients and functional imaging research. Dr. Bushara's current research focuses on the use of functional MRI to investigate the functions of the cerebellum, including movement control and cognition.



Christian DeZeeuw, M.D., Ph.D.

Prof. Dr. C.I. (Chris) De Zeeuw is professor and chair at the Dept. of Neuroscience of Erasmus Medical Centre in Rotterdam, and scientific co-director of the Netherlands Institute for Neuroscience of the Royal Dutch Academy of Arts & Sciences (KNAW) in Amsterdam. Chris De Zeeuw started his career in Rotterdam (lab Jan Voogd) where he received in 1990 his PhD degree in medicine *Cum Laude*. Following his medical studies at both the University of Amsterdam and University of Rotterdam he got his medical degree (MD) one year later, again *Cum Laude*. As a first-year PhD-student at his first international lecture in Turin, Italy, he surprised the international neuroscience community right from the start by showing a new technology, by which one could demonstrate for the first time simultaneously the connectivity of nerve fibers and the identity of their neurotransmitters. This new technology led to numerous discoveries of new functional connections in the brain. Together with two other major technical developments in the field of neuro-anatomy that he introduced in his thesis, he provided a basis for deciphering neural networks at the ultrastructural level. After obtaining his PhD, he received the prestigious Huygens Science Award – KNAW fellowship from the Royal Dutch Academy of Arts & Sciences, which allowed him to move to New York (Rodolfo Llinas) and enter the field of neurophysiology. Here he unraveled the circuitry of the vestibulocerebellum underlying compensatory eye movement control and he showed that this part of the brain can create predictions that are required for motor learning. In the meantime, he discovered a new cell organelle, the dendritic lamellar body, that is widely distributed in the brain and implicated in the functional control of electrical synapses, which form next to chemical synapses the main substrate of communication between neurons in the central nervous system. This discovery plus his desire to use transgenics for elucidating brain function inspired him to move, once again, to another field, the field of molecular biology (Frank Grosveld). In this field he discovered new genes, one of which was CYLN2, which turned out to be one of the main genes responsible for the neurological and cognitive symptoms of patients with Williams Syndrome; today, diagnostic screens for translocations of this gene are used worldwide. In 1998 he was invited to merge the Dept. of Anatomy and Dept. of Physiology at Erasmus MC into the Dept. of Neuroscience that he has been chairing since. Over the past decade the Dept. of Neuroscience has flourished in that it gained about ten times more scientists and staff, and that many of the postdocs and faculty received important awards and fellowships. For example, in the prestigious VENI, VIDI and VICI program of NWO and ZonMw, the main Dutch grant agencies, this Dept. alone received more than 20 fellowships, of which more than 30% were ranked number 1 in the country including the entire medical field and the field of life sciences. His PhD, C.C.H., was the first of Erasmus MC and Erasmus University to receive a EU-RYI Award. More than 40 PhDs successfully defended their thesis and more than five young scientists al-



AIM 2012 Biographies

ready left the Dept. to become chair and/or full professor elsewhere. By participating actively in the lab, De Zeeuw has not only inspired many of his students to become successful, but he himself also received several important personal awards. In 2001, he received the PIONIER Award of NWO and ZonMW, being ranked number 1 of all medical scientists in the Netherlands. In 2006, he received personally from Her Majesty, Queen Beatrix, the so-called Beatrix Award, which was given to the Dutch scientist with the best performance in the field of neuroscience and movement disorders over the 50-year period from 1956 to 2006. In addition, he obtained many prestigious grants from many national and international science agencies, such as NWO, ZonMw, FES, HFSP, EU, and KNAW, and he has been presiding over national and international consortia governing multimillion-Euro programs such as the NeuroBSIK Mouse Phenomics and NeuroBasic Pharma Phenomics programs for valorization of animal models for brain disorders and pharmaceutical products in the Netherlands, and the European SEN-SOPAC program for Bio-Inspired networks supporting Robotics control. Many of the technologies he developed for academic neuroscience research are nowadays commercially available via Neurasmus BV. In 2007 he became, next to being chairman at Erasmus MC, co-director of the Netherlands Institute for Neuroscience to determine its new scientific focus and start up a new group on cerebellar cognition. So along with the managerial tasks, he kept focusing on his greatest ambition, multidisciplinary neuroscience research. Over the past decade, he has been able to combine all fields mentioned above including neuro-anatomy, neurophysiology, molecular biology, psychology, neurology, computational modeling and robotics. Due to this unique set of approaches, his research has led to unprecedented insights of how the brain, in particular the cerebellum, may control learning behavior. These insights have led to the revolutionizing hypothesis that temporal coding must play an imminent role in this control, which forms the topic of this proposal. His papers have now been cited more than 5000 times. Many of them have been published in high-ranked Journals such as Nature Genetics, Cell, Nature Neuroscience, Neuron, Science, TINS, Nature Reviews, etc. He published three books on the cerebellum and he has been co-organiser of virtually all international cerebellar meetings over the past 15 years. He has been invited for over 250 international lectures, and visited virtually all prestigious neuroscience institutes in the world.



Wendy Galpern, M.D., Ph.D.

Dr. Galpern is a Program Director in the Office of Clinical Research at the National Institute of Neurological Disorders and Stroke at the National Institutes of Health in Bethesda, MD. She is involved with clinical research in movement disorders including the ataxias.

Dr. Galpern earned her medical and doctoral degrees from the University of Massachusetts Medical School and conducted her doctoral studies in the laboratory of Dr. Ole Isacson. She completed her internship in medicine at the Massachusetts General Hospital followed by neurology residency and a clinical and basic science fellowship in movement disorders at the Massachusetts General Hospital and Brigham and Women's Hospital in Boston, MA. Prior to joining NINDS, Dr. Galpern was a clinical fellow in movement disorders at the Toronto Western Hospital in Toronto, ON.



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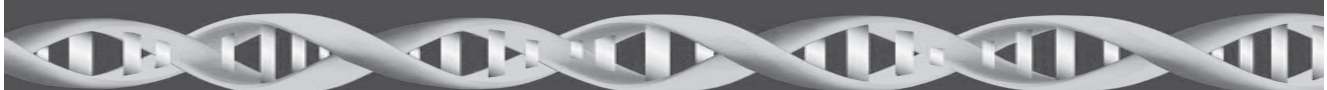
Christopher Gomez, M.D., Ph.D.

Dr. Gomez received his medical degree from the Pritzker School of Medicine in Chicago, Illinois, and his Ph.D. in Immunology from the University of Chicago. He also served his residency in Neurology at the University of Chicago. Until December 2005, Dr. Gomez served as Professor of Neurology and Associate Head for Research in the Department of Neurology, where he established and directed the University of Minnesota Ataxia Clinic until December 2005. Dr. Gomez is presently Professor and Chair of the University of Chicago, Department of Neurology and directs the University of Chicago Ataxia Center. His research interests are in SCA6, and the newly identified SCA26 and SCA32, as well as in developing disease biomarkers for ataxia. Dr. Gomez also serves on the Medical Research Advisory Board for the National Ataxia Foundation.



Robert C. Griggs, M.D.

Dr. Griggs is Professor of Neurology, Medicine, Pathology and Laboratory Medicine and Pediatrics at the University of Rochester School of Medicine and Dentistry. He was Chair of the Department of Neurology and Neurologist-in-Chief at Strong Memorial Hospital (1986-2008). He received his BA from the University of Delaware and his M.D. from the University of Pennsylvania. He received training in Internal Medicine at Case Western Reserve University and the University of Rochester where he was Chief Resident in Medicine and Fellow in Immunology. He trained in Neurology at the National Institute of Neurological Disorders and Stroke (NINDS) and the University of Rochester where he was Chief Resident in Neurology. Dr. Griggs is an internist/neurologist specializing in neuromuscular diseases with a focus on experimental therapeutics. He has directed an NIH-funded training program in the Experimental Therapeutics of Neurological Disease since 1989. This program has trained over 50 clinical neuroscientists who are in positions around the world. He has published over 350 scientific papers and 24 texts which span the fields of medicine and neurology. He served as Editor-in-Chief of *Neurology* (1997-2007). He is Neurology Editor of *Cecil Textbook of Medicine* and an Editor of *Cecil Essentials of Medicine*. He was elected to the Institute of Medicine of the National Academy of Sciences in 1998. He was President (2009-2011) of the American Academy of Neurology. Since 1998, Dr. Griggs has chaired the Executive Committee of the Muscle Study Group (MSG), an international consortium of investigators focused on developing new treatments for neuromuscular disease. He is the Principal Investigator of the Consortium for the Investigation of Neurological Channelopathies (CINCH) in the Rare Disease Network. CINCH is focused on developing new and better treatments for the channelopathies: Andersen-Tawil syndrome (a form of periodic paralysis), other periodic paralyses, the episodic ataxias and the non-dystrophic myotonias. CINCH has also trained 22 fellows at Rochester and in other channelopathy centers around the world: London(UK), Harvard, NINDS, UCLA, UCSF, UT (Dallas), and the U of Kansas.



AIM 2012 Biographies



John H. Ferguson M.D.

Dr. John H. Ferguson received a BS degree in Physics from Case Institute of Technology, an MD degree from Case Western Reserve University (CWRU) School of Medicine, and completed residency training in neurology at University Hospitals of Cleveland, Ohio. After two years as an Air Force neurologist in Japan, and a Fellowship in Neurophysiology at the Université de Montréal, he joined the neurology faculty of CWRU School of Medicine conducting research in epilepsy. Ten years later, he moved to Waco, Texas, to begin a private practice in neurology and was appointed Associate Clinical Professor of Neurology and Family Practice by the Baylor College of Medicine. After ten years in private practice, Dr. Ferguson was appointed Director of the Office of Medical Applications of Research at the National Institutes of Health where he directed the NIH Consensus Development Program for a little over a decade. He has served as Chairman of the Laboratory and Diagnostics Panel and was a member of the Executive Committee of the Medical Coverage Advisory Committee (MCAC) for the Centers for Medicare and Medicaid Services (CMS) and Chairman of the Therapeutics and Technology Assessment Committee of the American Academy of Neurology. He is presently a medical and health research consultant to the NIH Office of Rare Diseases Research.



Katrina Gwinn, M.D.

Dr. Katrina Gwinn is a Program Director and Senior Scientific Officer at the National Institute of Neurological Disorders and Stroke (NINDS), at the National Institutes of Health (NIH). At NIH she manages a number of extramural research programs and projects including the NIH Ataxia Portfolio. She worked at NIH from 2001 through 2007, when she moved to become the Vice Chair of Research in the Genetics Department at Baylor College of Medicine in Houston, Texas; she returned to NIH in 2010. She earned her BA in psychobiology from Wellesley College (1982) and subsequently worked in the field of molecular biology in Nobel Laureate Phil Sharps laboratory at MIT and Bob Kingston's at MGH. She earned her MD degree from Vanderbilt School of Medicine followed by a neurology residency at the University of Michigan and a Movement Disorders Fellowship at Mayo Clinic Scottsdale. She continued an academic career in gene discovery in Movement Disorders and Neurodegenerative disease, at the Mayo Clinic (Scottsdale, Jacksonville) and subsequently NIH (intramural). Recent honors and awards have included NINDS Merit awards, the NIMH Directors award, and twice, the NIH Directors Award for her work in genetics. She continues to pursue her interests in helping researchers navigate the NIH grants and contracts system to allow the translation of laboratory discoveries into the clinic, genetics of neurological disorders, and perhaps most importantly, mentoring junior faculty and students.



Hirokazu Hirai M.D., Ph.D. is Professor of Department of Neurophysiology, Chairman of Division of Neuroscience, and Director of Biological Genome Resource Center at Gunma University Graduate School of Medicine in Japan.

I graduated from Kobe University School of Medicine, Japan, in 1989, and experienced a residency in the university hospital for one year. Then, I proceeded to the Ph.D. course to become a radiologist. I always had six to seven patients suffering from end stage cancers including brain tumors in the hospital, but could do nothing effective for them, and thought that basic research, not clinical medicine, could save patients of incurable diseases and their families. This belief pushed me to decide shifting my carrier to a neu-



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roscientist. After obtaining a Ph.D. in 1994, I did post-doctoral training in Max-Planck Institute for Brain Research in Frankfurt/M, Germany for two years, and RIKEN Brain Science Institute in Japan for five years. In the latter institute, I started to study the cerebellar physiology in a laboratory of Prof. Masao Ito who is famous for Albus-Marr-Ito theory and as a discoverer of cerebellar long-term depression. He is also famous for a discovery of Purkinje neuron being a GABAergic inhibitory neuron. After a five year-stay in Prof. Ito's laboratory, I moved to St. Jude Children's Research Hospital, Memphis, TN in 2001 to focus my research on cerebellar diseases as my initial motivation was to save patients of incurable diseases. Since then, I have been studying pathology and treatment for spinocerebellar ataxia.



Mandy Jackson, Ph.D.

Dr. Jackson obtained her BSc in Molecular Biology from the University of Edinburgh in 1994 and her DPhil from the University of Oxford in 1997. She then moved to carry out postdoctoral research in the Neurology Department, Johns Hopkins University (1998-2001). In 2002 she was awarded a Caledonian Research Fellowship and returned to Scotland to join the neuroscience community at The University of Edinburgh. Dr Jackson subsequently became a RCUK Research Fellow in 2005 and a lecturer at The University of Edinburgh in 2010. Over the years her research has centered on the molecular and physiological mechanisms underlying motor neuron disease and spinocerebellar ataxia.



Joanna C. Jen, M.D., Ph.D.

Joanna C. Jen is Professor of Neurology at UCLA School of Medicine. She obtained her medical and graduate degrees from Yale University, completed neurology residency at UCLA, and pursued fellowship training in neurotology (neurology of balance), physiology, and human genetics, also at UCLA. Her clinical interest in neurotology is complemented by research performed in her laboratory, which focuses on the genetic and physiological bases of disorders affecting balance and eye movement control in neurodevelopment and neurodegeneration. She collaborates with Dr. Robert Griggs at the University of Rochester on an FDA-sponsored phase II trial on 4-aminopyridine in patients with episodic ataxia type 2.



Thomas Klockgether, M.D.

Dr. Klockgether is Director of the Department of Neurology at the University of Bonn and Director of German Center for Neurodegenerative Diseases (DZNE). He received his clinical training in Oldenburg (1981 – 1983) and Tübingen (1987 – 1991). From 1983 to 1987 he worked as a postdoc at the Max-Planck-Institute of Experimental Medicine in Göttingen. Dr. Klockgether has been the coordinator of several national and international collaborative research projects in the field of ataxia. Currently, he is principal investigator of the RISCA study and head of the Ataxia Study Group (ASG). His Research fields include: Molecular Genetics and Molecular Pathogenesis of Neurodegenerative Disorder, Clinical Neurology of Hereditary Ataxias, Neuropharmacology of Parkinson's disease and Structural

brain imaging.



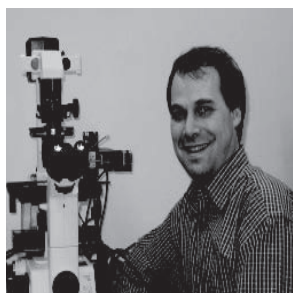
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Albert LaSpada

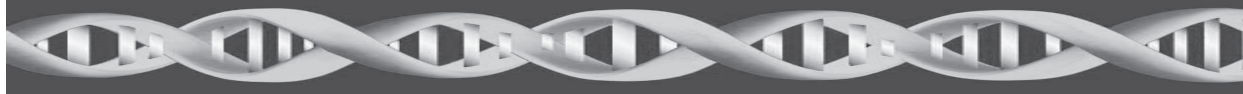
Albert La Spada graduated *Summa Cum Laude* from the University of Pennsylvania with a degree in Biology in 1986. As a recipient of a Medical Scientist Training program award, he pursued combined M.D. - Ph.D. training at the University of Pennsylvania School of Medicine. His 'Molecular Biology' doctoral 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 tract, this discovery of a novel type of genetic mutation has led to the emergence of new field of study in neurodegenerative disease. 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 in the Division of Medical Genetics. He pursued postdoctoral fellowship training as a Howard Hughes Medical Institute Physician Fellow, continuing to focus upon neurodegenerative disease. He joined the faculty in the Department of Laboratory Medicine at the University of Washington Medical Center in 1998, and was 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 the molecular basis of 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 also begun to use this mechanistic knowledge to develop therapies to treat this disorder. Dr. La Spada has been the recipient of numerous grants and awards from the National Institutes of Health, Howard Hughes Medical Institute, Muscular Dystrophy Association, Hereditary Disease Foundation, CHDI, Coulter Foundation, and American Federation for Aging Research. Among his funding awards is the prestigious Paul Beeson Physician Faculty Scholar Aging Research Award. In 2006, Dr. La Spada was inducted into the American Society for Clinical Investigation. In 2007, he was bestowed with the Lieberman Award by the Hereditary Disease Foundation for excellence in Huntington's Disease research. Dr. La Spada sits on a variety of editorial boards and grant review committees.



David Lynch, M.D.

Dr. Lynch received his undergraduate degree from Yale University, where he was a classmate of AIM meeting organizer Henry Paulson, and received his MD and PhD degree at Johns Hopkins University School of Medicine. He finished neurology residency and Fellowship at the University of Pennsylvania. He remained as a faculty member and was promoted to Professor in 2011. His major research is in Friedreich ataxia, as well as on NMDA receptors and antiNMDA receptor encephalitis. He has an international reputation for speaking very, very fast.



AIM 2012 Biographies



Patricia Maciel, Ph.D.

Patricia Maciel has a B.Sc. in Biochemistry and a Ph.D. in Genetics from the University of Porto, Portugal. During her Ph.D. she spent six months in the lab of Prof. Arnold Munich, in Paris, France, and four years in the lab of Prof. Guy Rouleau, at McGill University, Montreal, Canada. She was involved in the effort to map and clone the Machado-Joseph disease causative gene, and then studied genotype-phenotype correlations and genetic instability in this disease, under the supervision of Profs. Guy Rouleau and Jorge Sequeiros.

She then returned to Portugal to help start a new neurogenetics laboratory and continue research in MJD and other neurological disorders, including Huntington disease and related disorders, multiple sclerosis, migraine and Rett syndrome, among others. Her work has led to the publication of over 75 scientific articles in peer-reviewed journals.

In addition to research, Patricia has since 2007 been involved in teaching Genetics and Biochemistry, and is currently Associate Professor at the School of Health Sciences of the University of Minho, located in Braga, north of Portugal. There, she is also coordinator of the International Program of Post-graduation Courses, coordinator of the Neurodevelopment Research Line (within the Neurosciences Research Domain of the Life and Health Sciences Research Institute) and coordinator of the Molecular Diagnosis Services. Her main contributions to the study of ataxias have been in the characterization of the function of the ataxin-3 protein, identification of its molecular interactors, and the generation of animal models for the study of this disease in the mouse and in a small worm named *C. elegans*. These models are useful to understand the mechanisms of disease and to test therapeutic strategies.



Hidehiro Mizusawa, M.D., Ph.D.

Dr. Hidehiro Mizusawa is Professor and Chair of the Department of Neurology and Neurological Sciences at the Graduate School of Medical and Dental Sciences of Tokyo Medical and Dental University, where he is also Director of the Center for Brain Integration Research, Director of the School of Medicine, Vice Director of the Medical Hospital, and Associate Managing Trustee for Research. Dr. Mizusawa graduated with an MD in 1976 from the Faculty of Medicine of the University of Tokyo, where he received his PhD in 1983. After training in internal medicine and neurology at the Tokyo University Hospital and related institutes, he became a

Junior Assistant Professor of the Department of Neurology of Tokyo University in 1982. He then joined the Department of Neurology of Tsukuba University as a Senior Assistant Professor in 1984, becoming an Associate Professor there in 1990. Dr. Mizusawa then joined Tokyo Medical and Dental University as Professor and Chair of the Department of Neurology in 1996. Regarding ataxia, he has contributed particularly to research on molecular genetics and pathogenesis of AVED, SCA6, SCA31 and other ataxias including MSA. Dr. Mizusawa was Chair of the Research Committee on Prion Disease and Slow Virus Infection, Ministry of Health, Welfare and Labor, Japan from 2002 until 2010, and has been Chair of the Research Committee on Prion Disease Surveillance and Infection Control since 2010. He is now also serving as Leader of the Strategic Research Program for Brain Science, Field E, Ministry of Education, Culture, Sports, Science and Technology, Japan (since 2010), President of the Japanese Society for Neuroinfection (since 2009) and President of the Japanese Society of Neurology (since 2010).



Biographies



Marek Napierala, Ph.D.

Dr. Marek Napierala is currently an Assistant Professor at the Department of Molecular Carcinogenesis, University of Texas M.D. Anderson Cancer Center, Science Park, Texas. Dr. Napierala received his Ph.D. degree from the Institute of Bioorganic Chemistry Polish Academy of Sciences, Poznan, Poland. Since the earliest stages of his career Dr. Napierala research has been devoted to mechanisms of diseases caused by expansion of short repeating sequences. He completed his postdoctoral training at the Institute of Biosciences and Technology Texas A&M University in Houston, Texas where his research was focused on mechanisms of expansion of DNA repeats. During this time Dr. Napierala became involved in studies on molecular mechanisms of Friedreich's ataxia. In 2007 he received Young Investigator Award from the National Ataxia Foundation for his studies on mechanism of FRDA pathogenesis. In 2008 Dr. Napierala moved to the University of Texas M.D. Anderson Cancer Center as an Assistant Professor at the Department of Biochemistry and Molecular Biology.

Currently, his research is primarily focused on the mechanism of epigenetic silencing leading to Friedreich's ataxia. He recently completed a high throughput screening campaign aimed to uncover new therapeutic compounds capable of reactivating expression of the Friedreich's ataxia gene. He is also developing new neuronal and cardiac models of FRDA using induced pluripotent stem cell technology as well as cell therapy approaches for this disease. Dr. Napierala studies are supported by NIH, Friedreich's Ataxia Research Alliance and National Ataxia Foundation.

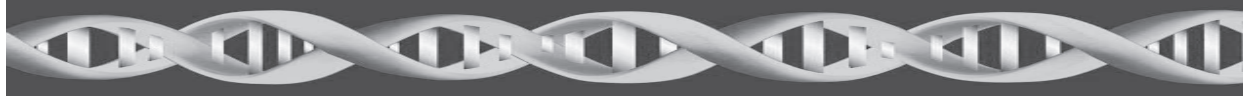


Puneet Opal, M.D, Ph.D is an Associate Professor of Neurology at Northwestern University Feinberg School of Medicine. He holds additional faculty appointments in the department of Cell and Molecular Biology and Interdepartmental Program in Neuroscience. At Northwestern Memorial Hospital, Dr. Opal runs a neurological practice focusing on movement disorders and disorders of the cerebellum. In addition, Dr. Opal heads a research laboratory dedicated to understanding neurodegenerative diseases, including ataxias. His current focus is on elucidating the molecular mechanisms underlying genetic movement disorders.



Harry Orr, Ph.D.

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

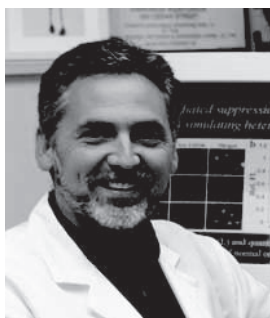


Biographies



Gülin Öz, Ph.D.

Dr. Öz is a brain imaging scientist who specializes in magnetic resonance spectroscopy (MRS) in degenerative brain diseases with special interest in spinocerebellar ataxias. She graduated from Bosphorus University in Istanbul, Turkey with B.S. degrees in Physics and Chemistry and obtained her Ph.D. 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 as assistant professor in 2006. Dr. Öz's research focuses on the application of MRS techniques using MRI scanners with higher magnetic fields than the routine clinical scanners to delineate the chemical alterations in the cerebellum in ataxias. MRS techniques non-invasively quantify many neurochemicals including neurotransmitters and antioxidants in affected brain regions. Such information is expected to facilitate early detection of neurodegeneration and to provide an objective means to monitor disease progression and response to therapies.



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

Dr. Paulson received his M.D. and Ph.D. from Yale University in 1990, and then completed a neurology residency and neurogenetics/movement disorders fellowships at the University of Pennsylvania. In 2007, after a decade at the University of Iowa, he joined the Neurology faculty at the University of Michigan. 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. His lab also has helped pioneer the use of RNA interference as potential therapy for hereditary neurological disorders caused by "toxic" mutant genes. Nationally, Dr. Paulson directs an ataxia course at the annual American Academy of Neurology meeting, serves on the scientific advisory boards of numerous disease-related organizations including the National Ataxia Foundation, and belongs to the Board of Scientific Counselors at the National Institute for Neurological Disorders and Stroke at 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.

Helene Puccio, Ph.D.

Dr. Puccio's laboratory focuses on progressive recessive ataxias, where disease onset, usually during childhood or adolescence, and progression result from neurodegenerative pathways that affect primarily the cerebellum and/or the spinocerebellar tracts. In the past, her main focus was Friedreich ataxia (FRDA), the most common progressive recessive ataxia. She is currently expanding to other recessive ataxia linked to mitochondrial dysfunction. Through conditional knockout approaches, her lab developed the first mouse models for FRDA, which reproduce important progressive features of the human disease, which have shown that the primary deficit in the disease is the deficiency in ISC proteins followed by a secondary mitochondrial iron accumulation and that oxidative stress is not a main feature of the disease. She has provided the first direct evidence that frataxin is necessary for the proper assembly of extra-mitochondrial ISC proteins in mammalian tissues, notably both cytosolic and nuclear proteins,



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including the ISC protein NTH1 involved in DNA repair. More recently, her group has shown biochemically that frataxin is involved in the early ISC assembly protein complex. After obtaining her PhD degree in Genetics from Harvard University in 1998, Dr. Hélène Puccio joined the group of Professor Michel Koenig the Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC, Strasbourg, France) to work on the molecular pathogenesis of Friedreich ataxia. In 2000, she obtained a permanent scientifique position at INSERM (Institut National de la Santé et de la Recherche Médicale), and is now research director of the Pathophysiology of recessive ataxia group at the IGBMC. In 2005, she was the recipient of “Prix Senior de Pathologie Pédiatrique 2004” the first prize from the Association pour l’étude de la Pathologie Pédiatrique. This prize is destined to honor scientific achievement in the comprehension of causes or mechanisms in a pediatric disorder. In 2005, her leadership and group was designated “Equipe FRM” by the french Medical Research Foundation (FRM), 1 of 25 groups that received this label in 2005. This label, first established in 2005, has been awarded to approximately 20 groups/year to recognize and encourage innovative and outstanding research projects that have a potential important medical impact in the long term. In december 2007, she was awarded the prestigious European Research Council award designed to boost the careers of researchers, who may be working in any area of science or scholarship, at the time they are establishing themselves as independent research leaders.



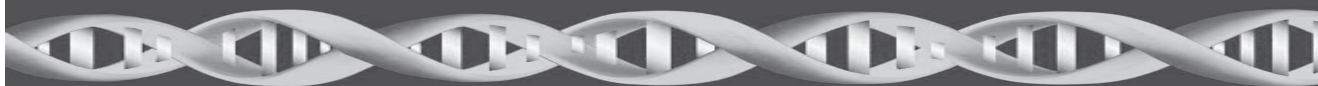
Laura Ranum, Ph.D

Dr. Ranum received her Ph.D. 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 and Research Advisory Board and serves as a reviewer for numerous scientific journals and funding agencies including the Muscular Dystrophy Association and the National Institutes of Health.



Edgardo Rodriguez, Ph.D.

Edgardo Rodriguez-Lebron is a research assistant professor at the University of Iowa. Edgar was born in Arroyo, Puerto Rico. In 1994 he moved to the United States to attend undergraduate school at the University of Central Florida. In 1999 he was admitted to the University of Florida graduate school and during this time 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, Edgar joined the laboratory of Dr. Henry L. Paulson at the University of Iowa in 2005 where he furthered the development of RNA interference as a therapeutic approach to neurological disease. He continued his training under the mentorship of Dr. Paulson at the University of Michigan where 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. His research focuses on the pathogenesis of Spinocerebellar Ataxia type-6 with the goal of developing novel therapies for this currently untreatable disease.



Biographies



Joseph Savitt, M.D., Ph.D. is an Assistant Professor of Neurology at the Johns Hopkins School of Medicine who works at the Parkinson and Movement Disorder Center of Maryland. He has studied at Cornell University and obtained an MD/PhD degree from the University of Maryland. He did his subsequent training at Johns Hopkins completing a neurology residency and a fellowship in neurodegenerative disease. At Johns Hopkins he was the director of the ataxia center, and he now maintains a large neurology practice at a clinic in the Baltimore and Washington suburbs. His research interests include the production of animal models to study neurodegenerative disease and trials of new medications on Humans to better treat conditions like dystonia and Parkinson disease. Currently he cares for more than 300 patients with ataxia including many with the genetic forms of the disease. He has received funding from the National Institutes of Health, the Michael J Fox Foundation, the National Parkinson Foundation and the National Ataxia Foundation among others.



Jeremy Schmahmann

Jeremy D. Schmahmann received his medical degree in 1980 at the University of Cape Town, completed residency in the Neurological Unit of the Boston City Hospital, and trained as a postdoctoral fellow in the Department of Anatomy and Neurobiology at Boston University School of Medicine. He joined the faculty of the Massachusetts General Hospital in 1989, where he is presently Director of the Ataxia Unit, a member of the Cognitive and Behavioral Neurology Unit, and Director of the Laboratory for Neuroanatomy and Cerebellar Neurobiology. Dr. Schmahmann is Professor of Neurology at Harvard Medical School. He has maintained an active teaching role throughout his career, is a Scholar in the Academy at Harvard, and Neurology Clerkship Director and Chair of the Neurology Education Committee at the Massachusetts General Hospital. He was awarded the Norman Geschwind Prize in 2000 for research in behavioral neurology from the American Academy of Neurology and the Behavioral Neurology Society, and the Distinguished Neurology Teacher Award from the American Neurological Association in 2008. He is an elected Fellow of the American Academy of Neurology, and of the American Neuropsychiatric Association, is on the scientific advisory board of the National Ataxia Foundation, medical advisor to the New England Ataxia Support Group, and has been cited in *The Best Doctors in America* since 1998. His research and clinical efforts are focused on the anatomical substrates of cognition, and in particular, on the role of the cerebellum in intellect and emotion. He has over 150 publications in peer-reviewed journals and academic texts, and his books include *The Cerebellum and Cognition* (Academic Press), *MRI Atlas of the Human Cerebellum* (Academic Press), and *Fiber Pathways of the Brain* (Oxford University Press), and he has most recently co-authored and edited *Cerebellar Disorders in Children* (Mac Keith Press) and *Handbook of the Cerebellum and Cerebellar Disorders* (Springer).



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Ludger Schöls, M.D.

I was trained as a medical doctor and did my doctoral thesis in biochemistry. Back to clinical medicine I specialized in Neurology and focussed in my Habilitation on genetics of cerebellar ataxias. My main research interest is still the genetic basis of neurological diseases, especially hereditary movement disorders. I contributed to establishing alpha-synuclein as the first gene causing monogenetic Parkinson disease, to LINGO as the first gene involved in the pathogenesis of essential tremor and – in cooperation with Michel Koenig - to the cloning of senataxin as the cause of ataxia with oculomotor apraxia type 2. We evaluated the phenotypic spectrum of the disease and discovered gene dosage alterations of senataxin as a mutational mechanism causing AOA2 (Arning et al., 2008). A longstanding interest addresses Friedreich's ataxia by characterizing its mitochondrial pathophysiology, demonstrating respiratory defects *in vivo* and initiating respective therapeutical trials (Schulz et al., 2009). More recently my group concentrated on mutations in the mitochondrial polymerase gamma (POLG). Here, we elaborated the broad phenotypical spectrum caused by POLG mutations and studied mitochondrial morphology and function in human fibroblasts and myoblasts (Schulte et al., 2009). Other major efforts concern the clinical characterisation, natural history and genetics of spinocerebellar ataxias (Schols et al., 2004). Here we contributed to the highly successful European Integrated Project EUROSCA (www.euroscala.org/), e.g. (Globas et al., 2008). We evaluated an active coordinative training as an effective treatment in degenerative ataxias which leads to long-term improvements in motor performance and achievements in activities of daily life which persisted over one year when performed on regular basis (Ilg et al., 2009).

Since 2004 I am full professor at the Department of Neurology, University of Tübingen. Here I am heading the Clinical Neurogenetics Unit including specialised outpatient clinics, a ward for neurodegenerative diseases and a molecular genetic lab at the Hertie-Institute for Clinical Brain Research (www.hih-tuebingen.de/klinische-neurogenetik/). In 2010 we funded the first German Center for Rare Diseases at the University Hospital in Tübingen (www.medizin.uni-tuebingen.de/ZSE/).



Vikram Shakkottai, M.D., Ph.D

Dr. Shakkottai 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 St. 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



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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 for cerebellar ataxia.



Yosef Shiloh, Ph.D.

Dr. Yosef Shiloh is Myers Professor of Cancer Genetics in the Department of Human Molecular Genetics and Biochemistry at the Sackler School of Medicine, Tel Aviv University. He obtained his B.Sc. degree at the Technion - Israel Institute of Technology in his native Haifa, and continued his graduate studies in Human Genetics at The Hebrew University of Jerusalem for both his M.Sc. and Ph.D. He trained further at Harvard Medical School and the University of Michigan and was a Fogarty Fellow at the U.S. National Institutes of Health. He is a member of The Israel National Academy of Sciences and Humanities and won the 2005 EMET Prize in Life Sciences, the 2011 American Association of Cancer Research G.H.A. Clowes Award for outstanding accomplishments in basic cancer research, and the 2011 Israel Prize in Life Sciences. He has dedicated most of his scientific career to understanding the severe human genetic disorder, ataxia-telangiectasia (A-T), caused by a defect in a central mechanism that maintains the stability and integrity of the DNA molecule. He began his quest to understand this disease while working on his Ph.D. thesis. This quest culminated in 1995 in the identification of the gene mutated in A-T patients, the *ATM* gene, in his lab. The Shiloh lab has since been studying the function and mode of action of the ATM protein, the product of the A-T gene, and the complex signaling network that ATM activates in response to DNA damage. In addition to his research, Prof. Shiloh devotes considerable time to giving popular scientific lectures to the general public and high school students on the medical, social and ethical implications of the genome revolution and its effect on cancer research and therapy.



S.H. Subramony, M.D.

S H Subramony, is currently Professor of Neurology at the University of Florida College of Medicine and the McKnight Brain Institute, Gainesville, FL where he co-directs the ataxia clinic with Dr Ashizawa. Prior to his current appointment, he was Billy Guyton Professor of Neurology at University of Mississippi Medical Center and Charlotte Warmoth Professor of Neurology at the University of Texas Medical Branch in Galveston, TX. Dr. Subramony received his medical degree from the Maulana Azad Medical College, Delhi University, New Delhi, India, in 1974. After his postgraduate work and clinical training in his home country, Dr. Subramony did further training at MacNeal Memorial Hospital, Berwyn, Illinois and completed his residency in Neurology and a Fellowship in Electromyography at the Cleveland Clinic Foundation, Cleveland, Ohio. He is an internationally recog-



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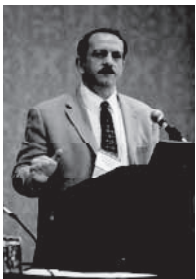
nized expert in spinocerebellar ataxia and neuromuscular disease and is actively involved in many clinical research projects in ataxia. Dr. Subramony is a member of the NAF Medical and Research Advisory Board.

Peter Todd, M.D., Ph.D.

Dr. Todd is an assistant professor of neurology in the University of Michigan Medical School. Dr. Todd obtained his medical and doctoral degrees through the Medical Scientist Training Program at the University of Wisconsin. His Ph.D. research focused on the molecular pathogenesis of Fragile X Syndrome. After completing his medical internship and neurology residency training at the University of Pennsylvania in Philadelphia, he came to the University of Michigan in 2008 to as fellow in movement disorders and neurogenetics, where he trained with Dr. Henry Paulson. In 2010, he joined the U-M faculty as an assistant professor of neurology. Dr Todd's research is focused primarily on neurological disorders where RNA and RNA processing are thought to play a primary role in disease pathogenesis, including fragile X-associated tremor ataxia syndrome (FXTAS) and myotonic dystrophy. Clinically, Dr. Todd is co-founder of the Fragile X Foundation research consortium clinic at the University of Michigan and also sees patients with movement and neurodegenerative disorders at the Ann Arbor VAMC. Dr. Todd has won numerous awards and honors. He was an AAN Foundation clinical research fellow and received the 2010 S. Weir Mitchell Alliance award from the American Academy of Neurology. He is funded by a Clinician Scientist (K-08) Career Development Award from the National Institute for Neurological Diseases and Stroke and was recently appointed as the inaugural Bucky and Patti Harris Collegiate Professor of Neurology.

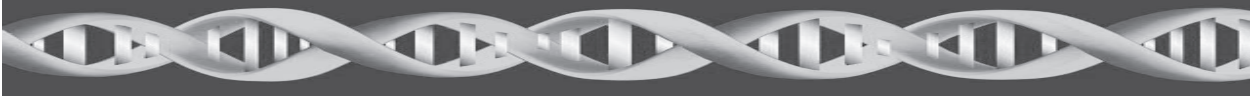
Shoji Tsuji, M.D.

Dr. Tsuji received his medical degree from the University of Tokyo, Tokyo, Japan, and his PhD from the University of Tokyo. He served his residency in Neurology at Jichi Medical School, Tochigi, Japan. Until July 2002, Dr. Tsuji served as Professor of Neurology and Director at Brain Research Institute, Niigata University, Niigata, Japan. Dr. Tsuji is presently Professor and Chair of Neurology, the University of Tokyo. He has identified a number of causative genes for hereditary ataxias and other neurodegenerative diseases. He has created excellent animal models for hereditary ataxias and is trying to develop new therapeutic strategies. The University of Tokyo Hospital recently established Medical Genome Center with installation of 5 next generation sequencers and server systems for genome informatics. As the Director of this center, he is applying these new technologies to elucidate molecular basis of sporadic neurodegenerative diseases including multiple system atrophy as well as hereditary diseases.



George (Chip) Wilmot, M.D., Ph.D.

Dr. George Wilmot is Assistant Professor in the Department of Neurology at Emory University in Atlanta, GA. He directs the Emory University Ataxia Center and has a very active clinic specializing in ataxia. Dr. Wilmot received his MD and PhD from the University of Michigan and then went on to neurology residency at Emory University. He joined the faculty at Emory directly after residency and is currently a member of the Movement Disorders section. He has been actively involved in the formation and early leadership of the Cooperative Ataxia Group and has participated in the Collaborative Clinical Research Network in Friedreich Ataxia since its inception. His research focus is on the natural history of the ataxias and development of clinical trials.



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

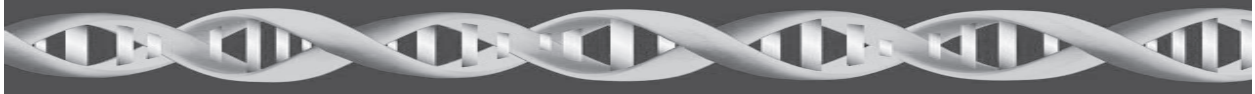
Dr. Wilson received his B.A. in Music and his B.S. in Biochemistry from Brown University, and his M.D. and his Ph.D. in Genetics from the University of Pennsylvania. He completed his residency training in Clinical Pathology, and his fellowship training in Transfusion Medicine, at the Hospital of the 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 Friedreich's ataxia, and in the development of small RNA therapeutics and biological tools. In addition to research, he is practicing clinical 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, molecular pathology and molecular genetic pathology to residents and fellows, and topics related to neurodegenerative disease to neuroscience graduate students.



Sarah H. Ying, M.D.

Dr. Sarah Ying is an Assistant Professor of Radiology in the Johns Hopkins University School of Medicine, with a secondary appointment in Neurology and Ophthalmology. She is the medical director of the Laboratory for Medical Image Computing in the Division of Neuroradiology. She received her B.A. from Harvard-Radcliffe College in Cambridge, Massachusetts, and her M.D. from The Johns Hopkins University School of Medicine in Baltimore, Maryland. Following internship training at The Johns Hopkins Hospital, she completed residency training in Neurology at Barnes Hospital in St. Louis, Missouri. She pursued additional training in neuro-otology and neuroimaging with Dr. Robert Baloh and Dr. Arthur Toga at the University of California, Los Angeles, then returned to Johns Hopkins for a clinical and research fellowship in neuro-otology and neurophthalmology with Dr. David Zee.

Now, Dr. Ying's research focuses on neurodegenerative syndromes, particularly hereditary ataxia syndromes. The Ying laboratory is developing neurophysiology and neuroimaging biomarkers to elucidate systems-level control of movement, balance, and cognition. In particular, they are studying how longitudinal, morphological changes in connected elements of cerebellar and extracerebellar circuits can reflect patterns of disease-specific degeneration, and how these changes can help guide therapeutic and rehabilitative approaches, ranging from EEG-based brain-computer interfaces to dance movement therapy. As a member of the Image Analysis and Communications Lab, she is also involved in the development of image processing techniques for automated identification of cerebellar and brainstem structures using multi-modality magnetic resonance imaging, including diffusion tensor imaging.



AIM 2012 Biographies



Huda Zoghbi, M.D.

Huda Zoghbi is Professor of Pediatrics, Neurology, Neuroscience, and Molecular and Human Genetics at Baylor College of Medicine and serves as an Investigator with the Howard Hughes Medical Institute. She is also the Director of the Jan and Dan Duncan Neurological Research Institute at Texas Children's Hospital.

Zoghbi's interest is in using the tools of modern genetics to understand the proper development of the brain as well as what goes awry in specific neurodevelopmental and neurodegenerative conditions. She has published seminal work regarding the molecular basis of Rett syndrome and of late-onset neurodegenerative diseases. Together with Dr. Harry Orr, Zoghbi has identified the cause and has been studying the pathogenesis of spinocerebellar ataxia type 1. The Orr-Zoghbi team has provided insight into factors critical for neurodegeneration in SCA1, and is now focused on finding ways to uncover suppressors of disease. Dr. Zoghbi is a member of several professional organizations including the NIH Scientific Management Review Board, the McKnight Foundation Neuroscience Board, and the Lasker Jury. She received several honors for her contributions and has been elected to the Institute of Medicine, and the National Academy of Sciences.



Junior Lecturer Biographies

Peter Breuer, Ph.D.

Peter Breuer is a postdoctoral fellow with Prof. Ullrich Wüllner in the Department of Neurology headed by Prof. Thomas Klockgether at the University of Bonn, Germany.

Peter Breuer studied biology with a focus on yeast genetics and graduated in 1994. He joined the lab of Prof. Kurt von Figura in Göttingen as a PhD student to study intracellular trafficking of proteins and vesicles. He obtained his doctoral degree in biochemistry in 1998. He pursued molecular cell biology studies and was awarded a scholarship at the Max Planck Institute of Biochemistry in Martinsried near Munich to study the influence of molecular chaperones on polyglutamine aggregation in the department of Prof. F.U. Hartl. Here, Dr. Breuer identified a potential key mechanism for the cellular malfunction of expanded ataxin 3 (ATXN3), the diseased protein in Spinocerebellar Ataxia Type 3 (SCA3). He identified ATXN3 as sensitive calpain substrate and proved the involvement of calpain proteolysis in SCA3 pathogenesis in subsequent experiments. In collaboration with Dr. Philipp Koch at the Institute of Reconstructive Neurobiology in Bonn, headed by Prof. OliveBrüstle, Dr. Breuer used induced pluripotent stem cell derived neurons to further dissect the pathogenic mechanism of ATXN3 in patient-specific neuronal cells. With this elegant approach, Dr. Breuer and his colleagues found, that calpain-dependent aggregation of ATXN3 could be induced by channel-mediated raise in intraneuronal calcium levels supporting the idea of calpain being a key protease in neuronal dysfunction observed in SCA 3. r Brüstle, Dr. Breuer used induced pluripotent stem cell derived neurons to further dissect the pathogenic mechanism of ATXN3 in patient-specific neuronal cells. With this elegant approach, Dr. Breuer and his colleagues found, that calpain-dependent aggregation of ATXN3 could be induced by channel-mediated raise in intraneuronal calcium levels supporting the idea of calpain being a key protease in neuronal dysfunction observed in SCA 3.



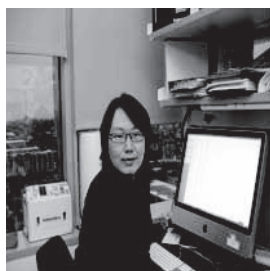
Andreia Castro, Ph.D.

Andreia Castro is a researcher in the field of neurodegenerative diseases, focusing on the molecular mechanisms that regulate neuronal dysfunction and aggregation in SCA3. She holds a B.Sc. in Biochemistry from the University of Porto, Portugal, received a Msc. in Molecular Genetics (2007) and a Ph.D. in Health Sciences from the University of Minho, Portugal (2011). She is currently conducting a postdoctoral research in the laboratory of Professor Patrícia Maciel at School of Health Sciences, University of Minho in cooperation with Professor Rick Morimoto from the Northwestern University, Department of Molecular Biosciences.

During her graduated studies, Andreia has established a *C. elegans* model for the study of MJD/SCA3 pathogenesis and identified a number of modifier genes, namely aging-related genes that stalled disease progression. More recently, she has been dedicated to the identification of small molecules that ameliorate mutant ataxin-3-mediated toxicity in *C. elegans*, and therefore may be useful for therapy development.



Junior Lecturer Biographies



Xiaofei Du, M.D., M.S.

Dr. Du received an M.D. at Shanxi Medical University in 2001 and an M.S. in Pediatrics at Shanghai Second Medical University with an M.S. In 2005 she received an Excellent Woman scientist awarded by the Shanghai Municipal Government. I have a strong background in research on control of gene expression by androgen receptor (AR), which functions as a testosterone-activated transcription factor. The genomic research and target gene expression studies I have done to study AR transcription factor activity were extremely relevant as background to the study of spinocerebellar ataxia type 6 (SCA6). The goal of my present research is to investigate the role of a calcium channel subunit, CACNA1A in the pathogenesis of SCA6. Specifically, I want to elucidate a new mechanism underlying the production of C terminus of CACNA1A gene (a1ACT) and its related gene expression regulation in neuronal cells. As a young investigator working in the area of ataxia, with the continued guidance and mentorship of Dr. Gomez, my research will focus on the following key areas: (1) the function of a1ACT in regulating gene expression as a transcriptional factor; (2) the role of a1ACT in Purkinje cell differentiation, viability and degeneration; and (3) developing the potential therapeutic strategies for SCA6 targeting a1ACT bearing the pathological polyQ. My current position is Research Associate (Assistant Professor) at the University of Chicago, Chicago, IL

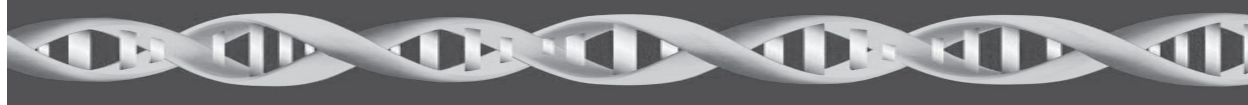


Anna Duarri, Ph.D.

Dr. Duarri started her research career in 2005 at the Biochemistry Department of UAB (Barcelona, Spain), studying the implications of protein phosphorylation in metabolic regulation, under the supervision of Dr. E. Itarte. After gaining her M.Sc. in 2006, she obtained an ISCIII project fellowship and a CIBER-rare diseases position to work on the pathophysiological pathway of Megalencephalic Leukoencephalopathy with subcortical cysts. This work was performed under Dr. R. Estévez and Dr. V. Nunes at the Departments of Medical Genetics and Physiological Sciences, IDIBELL (Barcelona). It led to her Ph.D. thesis, completed in 2010. In July 2011, she became a postdoctoral researcher in the Ataxia research group led by Dr. D. Verbeek, Department of Genetics, UMCG (Groningen, the Netherlands). This group recently discovered the gene causing Spinocerebellar ataxia type 19, and Dr. Duarri is now working on elucidating the molecular pathological mechanism leading to development of the disease.

Yuanzheng, Gao, M.D., Ph.D.

Dr. Yuanzheng Gao is a postdoctoral associate in Dr. Laura Ranum's lab at Department of Molecular Genetics and Microbiology, University of Florida College of Medicine in Gainesville Florida. Dr. Gao received his medical degree from Bengbu Medical College Anhui China in 1991. He completed his medical residency and fellowship in Anhui and Shanghai China from 1991-1999. Dr. Gao came to the United States as a visiting scholar sponsored by University of Connecticut in 1999. He was awarded Ph.D in Biomedical Science from University of Connecticut in 2007. During his Ph.D study, Dr. Gao did his thesis research with Dr. John Carson. Dr. Gao's thesis work was about memory related mRNA trafficking in hippocampal neurons. In 2007, Dr. Gao joined Dr. Jeffrey Rothstein's lab as a postdoctoral fellow in Department of Neurology at Johns Hopkins University School of Medicine. His research was focused on Spinocerebellar Ataxia 5 (SCA5). SCA5 is caused by mutations of beta III spectrin. His research showed that beta III spectrin is critical for development of Purkinje neuron dendrites and spines. Dr. Gao is working on mechanisms underlying SCA5, especially how mutations of beta III spectrin cause degeneration of Purkinje neurons.



Junior Lecturer Biographies



Natalia Gromak, Ph.D.

Dr. Gromak received her BSc (Hons) in Molecular Biology from the University of Edinburgh in 1997. She completed her Ph.D. in the lab of Prof. Chris Smith in the Department of Biochemistry, University of Cambridge (UK), in 2001, working on regulation of alternative splicing of muscle-specific genes. In 2002, she joined the University of Oxford as a Post-Doctoral fellow in the lab of Prof. N.J. Proudfoot. Here she investigated the fundamental mechanisms of gene expression and interconnections between transcription and RNA processing in human cells. In 2011 Natalia Gromak was awarded a Royal Society University Research Fellowship allowing her to establish an independent lab in the University of Oxford (UK). In her current research programme, Dr. Gromak is investigating the defects in RNA transcription and processing, associated with neurodegenerative diseases. In particular she is interested in understanding the function of senataxin protein, mutated in AOA2 and ALS4 disorders, in the regulation of human gene expression.



Sara Lagalwar, Ph.D.

I graduated from the University of St. Thomas in St. Paul, Minnesota with degrees in political science and biology. After college, I began a research internship at the Alzheimer's Research Center in St. Paul and the Mayo Clinic, which sparked my interest in neurodegeneration research. I pursued this interest at Northwestern University, where I received my PhD in Neuroscience studying tau phosphorylation and pathology in Alzheimer's disease and frontotemporal dementia. I later joined the lab of Dr. Harry Orr at the University of Minnesota as a postdoctoral associate. In Harry's lab, I have been focusing on the phosphorylation pathways involved in SCA1 and collaborating on a high-throughput drug screen to develop a potential therapeutic. In addition to neurodegeneration research, my other major professional interest is in undergraduate teaching. I will be pursuing both this fall when I join the faculty at Skidmore College. I will be teaching courses in the Neuroscience program and conducting research in SCA1 and Alzheimer's disease.



Floriana Licitra, Ph.D. student

Floriana Licitra studied Biology at the University of Naples (Italy) "Federico II". There she obtained her master's degree in Molecular Biology in Valeria Ursini's laboratory at the Institute of Genetics and Biophysics (IGB-CNR) of Naples. Her main interest was to characterize a poly-Alanine expansion in the homeobox gene, ARX, previously shown to be responsible for mental retardation. In 2009, Floriana was admitted to the International PhD program at the IGBMC in Strasbourg (France), and decided to join H       Puccio's laboratory. Since then, she has been studying a recent form of recessive ataxia, ARCA2, caused by mutations in the ADCK3 gene. In particular, she uses both *in vivo* and *in vitro* approaches to characterize the pathophysiological mechanisms of ARCA2. Notably, she works with a knock out mouse for *Adck3* as well as with neuronal and muscle cell models.

Junior Lecturer bios continued on next page



Junior Lecturer Biographies



Keyi Zhu, M.D., Ph.D.

Dr. Keyi Zhu is a postdoctoral associate in the Department of Molecular and Human Genetics at Baylor College of Medicine in Houston. She received her M.D. degree in 1999 and M.S. degree in 2002 in China. She earned her Ph.D. in 2007 from the University of Texas Health Science Center at Houston, Graduate School of Biomedical Sciences. During all these years of training, she has received numerous awards to recognize her effort and talent. She has published several papers in peer-reviewed journals with her thesis work on proteasome inhibition and cancer therapy. Currently Dr. Zhu's research focuses on the identification of therapeutic targets for spinocerebellar ataxias (SCAs) by understanding the pathogenic mechanisms of the diseases. Her work has been recognized by colleagues in the field, and she was awarded a research fellowship by the National Ataxia Foundation in 2010. She is also a winner of the Young Investigator Award in Neurobiology of Disease at the Children Symposium: Childhood Ataxia in 2011.



NAF 2012 Annual Membership Meeting

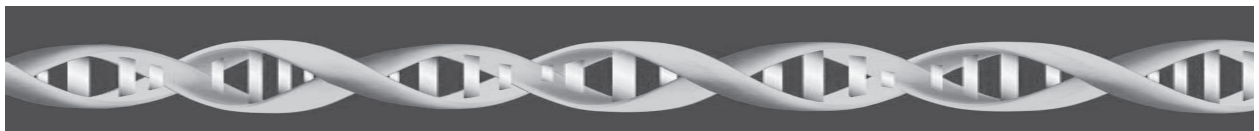
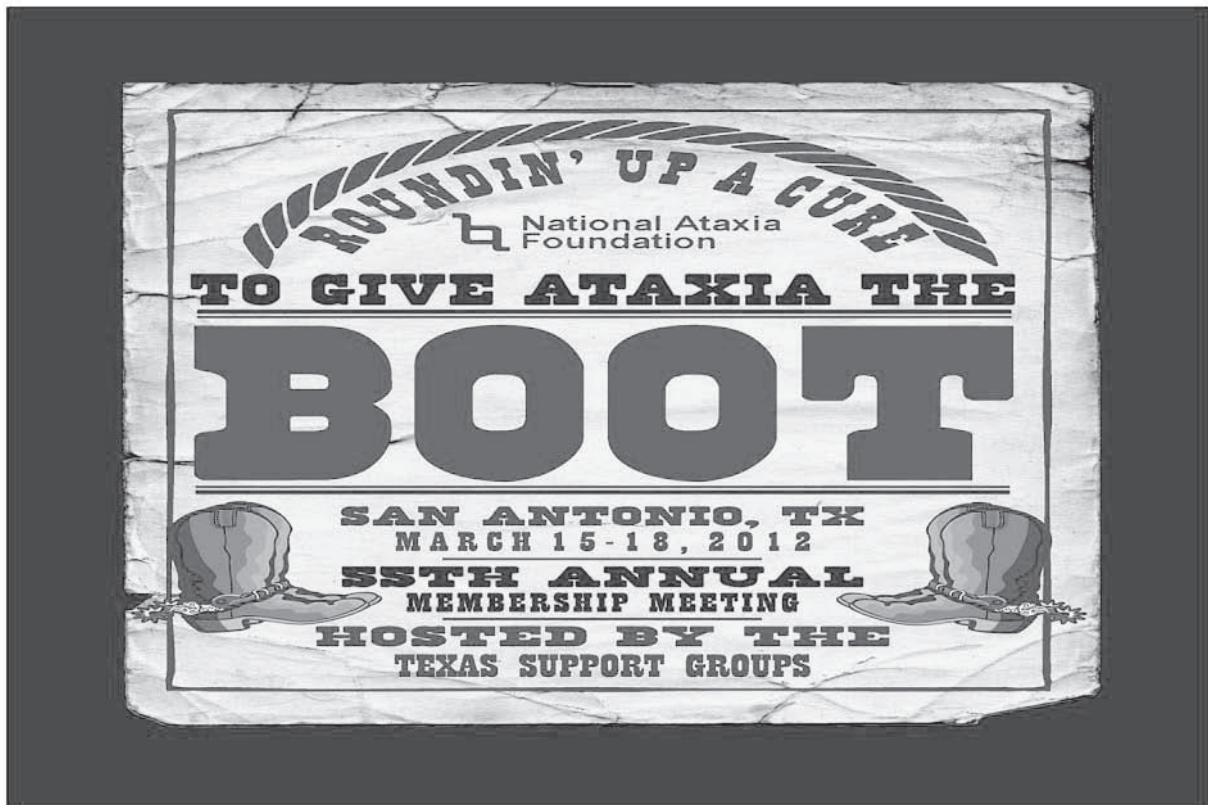
The National Ataxia Foundation's Annual Membership Meeting begins on Friday morning and ends Sunday at 12:30 p.m. This meeting provides patients and their families the opportunity to hear presentations about on-going research initiatives and clinical and practical information for managing ataxia.

Social events include the Welcome Reception on Friday night and Saturday Evening Banquet. NAF is offering a reduced registration fee of \$95 for AIM attendees to attend this meeting which includes admission to all General Sessions and Social events.

To register, please go to the Goliad Room during registration hours which are:

Thursday	9:00 a.m. to 8:00 p.m.
Friday	8:00 a.m. to 5:30 p.m.
Saturday	8:00 a.m. to 5:00 p.m.

Please mention that you are an AIM attendee to receive the reduced registration fee.



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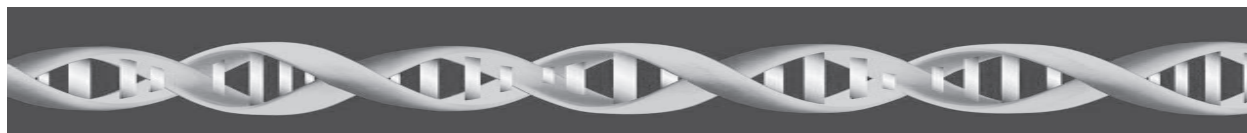
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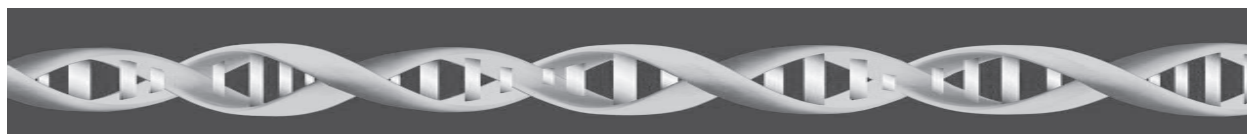
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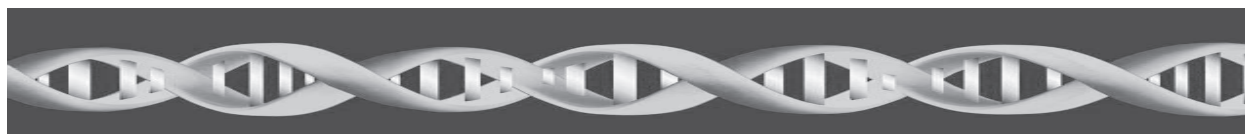
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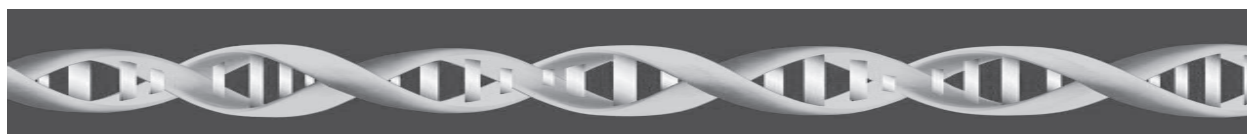
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Join the National Ataxia Foundation

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:

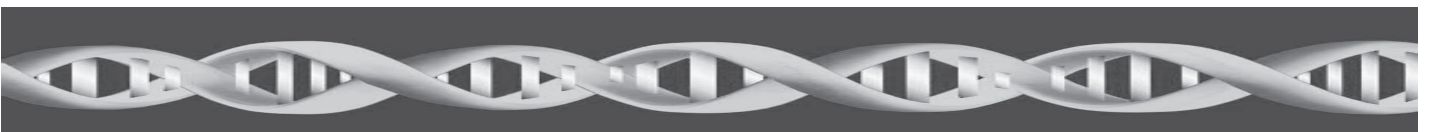
- Education and support services for patients who are affected by ataxia
 - Ataxia Investigator Meetings, such as the one you are attending, which bring researchers together to accelerate ataxia research
 - Current and accurate medical and genetic information on its website, www.ataxia.org
 - Local and 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 researcher by aligning yourself with the only national organization focusing 100% of its efforts on all types of ataxia.

The National Ataxia Foundation is dedicated to improving the lives of persons affected by ataxia through support, education, and research.



Announcement of the 2013 Patient Meeting

56th NAF Annual Membership Meeting

March 15 - 17, 2013

Detroit Marriott at the Renaissance Center Detroit, MI

The Detroit Marriott is situated on the River Walk in downtown Detroit, overlooking the Detroit River and Canada with easy access to shopping, restaurants, entertainment, and the People Mover.

Reservation information will be announced soon

About Detroit

- only a 90-minute flight from 60 percent of the nation's population.
- Detroit is an international destination - located only one mile from Windsor, Ontario, Canada.
- Detroit has world-class cultural attractions, entertainment and dining. In fact, the city was recently named by *AmericanStyle* readers as one of the top 25 arts destinations in the nation.

