

**Welcome to the Third
Ataxia Investigator's Meeting (AIM 2010)
In Conjunction with the University of Chicago
Program in Pathobiology and Translational Neuroscience**



**Sponsored by the
National Ataxia Foundation**



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ApoPharma Inc.
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National Institute of Neurological Disorders and Stroke (NINDS)
Santhera Pharmaceuticals

March 9-11, 2010 * Hyatt Regency O'Hare * Chicago, IL

Dear ATM 2010 Attendee:

Welcome to the Third Ataxia Investigators Meeting (ATM 2010) sponsored by the National Ataxia Foundation in conjunction with the University of Chicago Program in Pathobiology and Translational Neuroscience.

We look forward to one evening and two full days of keynote speakers and presentations from senior ataxia investigators from around the world who will present their latest discoveries in dominant, recessive and sporadic ataxias, as well as 35 poster presenters who were selected by the ATM 2010 steering committee from over 70 excellent abstracts submitted for review. We regret that we had to limit the number of poster presenters who could attend ATM 2010.

We are pleased to announce a unique item of this year's meeting which will be the Junior Lecture speaking slots for four applicants selected from those applying to the abstract session. The intent of the Junior Lectures is for the ATM 2010 Steering committee to recognize and promote new investigators with promising careers in ataxia research.

Since its inception in 2005, when the first ATM was held in Tampa, a new level of interest has been reached with regard to ataxia research. The next ATM is being planned for 2012. We strongly encourage you to complete the survey that is included with your meeting materials. This will provide the Steering Committee with valuable information to make future meetings successful and beneficial as we continue to move ataxia research to the forefront bringing those affected by these disorders hope for a future where viable treatments will be available.

Again, on behalf of the ATM Steering Committee, the National Ataxia Foundation and the generous sponsors of ATM 2010, I welcome you and look forward to the next days of stimulating presentations and networking opportunities.

Yours,

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



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For your support of the
2010 Ataxia Investigators Meeting.

General Overview of AIM 2010 Agenda

March 9-11, 2010

TUESDAY	Event	Location
3:00-6:00 PM	Registration	O'Hare Foyer
4:00-6:00 PM	Reception	O'Hare Ballroom
7:00-8:00 PM	Keynote Speaker: Christian Hansel, PhD	O'Hare Ballroom
7:00-9:30 PM	Dinner	O'Hare Ballroom
WEDNESDAY	Event	Location
Wed & Thu	Posters Displayed	O'Hare Foyer
7:00-8:00 AM	Breakfast	O'Hare Ballroom
8:00-9:30 AM	Theme 1: Cerebellar function (and episodic) dysfunction	O'Hare Ballroom
10:00-12:00 PM	Theme 2: Molecular pathogenesis of autosomal dominant ataxias	O'Hare Ballroom
12:00-1:15 PM	Lunch and Posters	O'Hare Ballroom
1:15-6:00 PM	Theme 2: Continued	O'Hare Ballroom
5:30-6:00 PM	Keynote Speaker: Richard Morimoto, PhD	O'Hare Ballroom
6:30-10:00 PM	River East Art Center Hors d' oeuvre & Dessert Reception	Offsite
THURSDAY	Event	Location
7:00-8:00 AM	Breakfast	O'Hare Ballroom
8:00-12:30 PM	Theme 3: Molecular pathogenesis of recessively inherited ataxias	O'Hare Ballroom
8:00-8:40 AM	Keynote Speaker: Keith Caldecott, PhD	O'Hare Ballroom
12:30-1:45 PM	Lunch and Posters	O'Hare Ballroom
1:45-6:00 PM	Theme 4: Moving towards therapy: Novel strategies and outcomes measures in ataxia	O'Hare Ballroom
6:00-7:00 PM	Dinner and Young Investigator Awards	O'Hare Ballroom
7:00-8:00 PM	Keynote Speaker: Ole Isacson, MD	O'Hare Ballroom

Tuesday, March 9, 2010

All Events take place in the O'Hare Ballroom unless noted.

Time	Event
3:00-6:00 PM	Registration
4:00-6:00 PM	Welcome Reception
6:00-7:00 PM	Dinner
7:00PM	Welcome and Opening Remarks—Christopher M. Gomez, MD, PhD Keynote Speaker: Christian Hansel, PhD, University of Chicago, “Cerebellar Learning and Connectivity”

AIM 2010 Outing

All registered and invited AIM attendees are invited to this off-site social event.

On Wednesday evening a chartered bus will take us to Downtown Chicago to the River East Art Center “... a place of enrichment, expression and inspiration... a place to discover fine art, and the finer things in life... a place of community, ideas, poetry, performances, lavish galas and corporate affairs. Moreover, it is a place of surpassing beauty – breathtaking spaces, dazzling views, daring architecture. In the coming years, River East Art Center will give rise to stunning works of importance – masterpieces to be discovered and cherished by local residents.” Taken *from the River East Art Center website*.

Buses will board from the lobby doors of the hotel between 6:15–6:30 p.m. for an evening of informal networking with other ataxia investigators, and the opportunity to view paintings and sculptures on display that range from the wonderfully avant-garde to the internationally renowned and enjoy hors d'oeuvres, desserts and beverages. Buses will return to the hotel at the end of evening.

We look forward to offering this off-site event to AIM participants.

**If you are unable to attend,
please inform one of the NAF Staff members.
Thank you.**



***The National Ataxia
Foundation has Ataxia
Research Funding Available!***

The National Ataxia Foundation (NAF) began direct funding of ataxia research studies in 1978.

Currently, the National Ataxia Foundation has three ataxia research programs including: 1) NAF Research Program, 2) NAF Research Fellowship Award, and 3) NAF Young Investigator Award.

To find out more about these NAF research programs, guidelines, and application forms, please visit the Foundation's web site at www.ataxia.org or email: naf@ataxia.org.



Wednesday, March 10, 2010

All Events take place in the O'Hare Ballroom unless noted.

Time	Event
7:00-8:00 AM	Breakfast
8:00-9:30 AM	Theme 1: Cerebellar function (and episodic) dysfunction <i>Co-Chairs: Henry Paulson, MD, PhD and Sarah Ying, MD</i>
8:00 AM	Episodic Ataxias: an overview, Joanna C. Jen, MD, PhD University of California-Los Angeles
8:30 AM	Episodic ataxia: physiological mechanisms, Ellen Hess, PhD, Emory University
9:00 AM	The therapeutic mode of action of 4-AP in Episodic ataxia, K. Khodakhah, PhD, Albert Einstein College of Medicine
9:30-10:00 AM	Break
10:00-12:45 PM	Theme 2: Molecular pathogenesis of autosomal dominant ataxias <i>Co-Chairs: Russell Margolis, MD & George Wilmot, MD, PhD</i>
10:00 AM	SCA2 and SCA13, Stefan Pulst, MD, University of Utah
10:30 AM	SCA3, Henry Paulson, MD, PhD, University of Michigan
11:00 AM	SCA6, Christopher M. Gomez, MD, PhD, University of Chicago
11:30 AM	Late Breaking News
12:00-1:15 PM	Lunch and Posters
1:15 - 6:00 PM	Theme 2: Continued
1:15 PM	Invited Junior Lecturer - Paula Ladd, University of California-San Diego
1:40 PM	Invited Junior Lecturer - Mirella Dottori, PhD, University of Melbourne
2:05 PM	SCA 7, Albert LaSpada, MD, PhD, FACMG, University of California-San Diego
2:35 PM	SCA17, Xiao-Jiang Li, MD, PhD, Emory University
3:05-3:30 PM	Break
3:30 PM	SCA5 and SCA8, Laura Ranum, PhD, University of Minnesota
4:00 PM	SCA10, Tetsuo Ashizawa, MD, PhD, University of Florida, Gainesville
4:30 PM	Molecular pathophysiology of SCA 14 caused by gammaPKC mutations, Norio Sakai, MD, PhD Hiroshima University, Hiroshima, Japan
5:00 PM	Perspectives on dominant ataxia: insights from SCA 1 that apply to other spinocerebellar ataxias, Harry Orr, PhD, University of Minnesota
5:30 PM	Keynote Speaker: Richard Morimoto, PhD, Northwestern University, “The Proteostasis Challenge: The Stress of Misfolded Proteins in Aging and Disease”
6:30-10:00 PM	River East Art Center: Hors d' oeuvre & Dessert Reception in Downtown Chicago. Please begin boarding buses at 6:15 PM.

Thursday, March 11, 2010

All Events take place in the O'Hare Ballroom unless noted.

Time	Event
7:00-8:00 AM	Breakfast
8:30-10:15 AM	Theme 3: Molecular pathogenesis of recessively inherited ataxias <i>Co-Chairs: SH Subramony, MD and Alexandra Durr, MD, PhD</i>
8:00 AM	Keynote Speaker: Keith Caldecott, PhD, University of Sussex, "Defects in DNA Single-Strand Break Repair and Neurodegenerative Disease—a Matter of Balance"
8:45 AM	The function of ATM in the central nervous system: beyond DNA breaks, Karl Herrup, PhD, Rutgers University
9:15 AM	Senataxin, defective in AOA2, protects against oxidative stress and transcription dysregulation, Martin Lavin, PhD, Queensland Institute of Medical Research, Brisbane
9:45 AM	Novel Recessive ataxias, Michel Koenig, MD, PhD, Institut de Genetique, CNRS, Universite Louis-Pasteur
10:15-10:45 AM	Break
10:45-12:15 PM	Theme 3: Continued
10:45 AM	Overview of Friedreich's ataxia, Robert Wilson, MD, PhD, University of Pennsylvania
11:15 AM	Addressing the Role of Frataxin in Iron-Sulfur Cluster Assembly, Grazia Isaya, MD, PhD, Mayo Clinic of Rochester
11:45 AM	Frataxin gene regulation and pathways affected by frataxin deficiency, Massimo Pandolfo, MD, Service de Neurologie Hôpital Erasme, Brussels
12:15 PM	Late Breaking News
12:30-1:45 PM	Lunch and Posters
1:45-3:35 PM	Theme 4: Moving towards therapy: Novel strategies and outcomes measures in ataxia <i>Co-Chairs: Wendy Galpern, MD, PhD and Jeremy Schmahmann, MD</i>
1:45 PM	Friedreich's ataxia: Mitochondrial therapeutics, Sidney Hecht, PhD, Biodesign Institute, Arizona State University
2:15 PM	Invited Junior Lecturer - Alain Martelli, PhD, Institut de Genetique et de Biologie Moleculaire, Strasbourg, France
2:40 PM	Invited Junior Lecturer - Isabelle Iltis, PhD, University of Minnesota
3:05 PM	Friedreich's ataxia-5 Transcriptional enhancement with HDAC inhibitors, Joel Gottesfeld, PhD, Scripps Institute, San Diego

Thursday, March 11, 2010 continued

All Events take place in the O'Hare Ballroom unless noted.

Time	Event
3:35 - 4:00 PM	Break
4:00 - 6:00 PM	Theme 4: Continued
4:00 PM	Natural history studies and EuroSCA research network: 2010, Thomas Klockgether, MD University Hospital Bonn, Bonn, Germany
4:30 PM	Imaging correlates of SCA 1 pathogenesis: From Biomarkers to Bioexplanations, Ana Solodkin, PhD University of Chicago
5:00 PM	MRS biomarkers for ataxias, Gulin Oz, PhD, University of Minnesota
5:30 PM	Towards therapy for ataxia: how do we get there? Kurt Fischbeck, MD, National Institute of Neurological Disorders and Stroke
6:00 - 8:30 PM	Dinner
6:00 PM	Young Investigator Awards
7:00 PM	Keynote Speaker: Ole Isacson, MD, Harvard University, "Stem cells and iPS cells: Paradigm shifts in cell therapeutics and discovery for neurodegenerative diseases"

Thank you!

On behalf of the ataxia families we serve, the National Ataxia Foundation thanks each AIM participant for taking time from their busy schedules to attend the

Third Ataxia Investigators Meeting.

Thank You to the AIM 2010 Steering Committee:

Alexandra Dürr, MD, PhD

Harry Orr, PhD

Henry Paulson, MD, PhD

Laura Ranum, PhD

Robert Wilson, MD, PhD

Stefan Pulst, MD

A Special Thank You to Christopher M. Gomez, MD, PhD, the Lead Organizer for AIM 2010

From the National Ataxia Foundation's Board of Directors

You are invited to attend the NAF Annual Membership Meeting for those affected by ataxia and their families. Registration is just \$55 to attend all events which includes a Friday Chicago Style Pizza buffet reception and Saturday evening banquet with a plated meal and general sessions on ataxia treatments and research. Register in Grand Ballroom D where you will pick up your Annual Membership Meeting materials and name badge which will serve as your entry to all events.

Registration Hours:

Thursday: 8:00 AM - 9:00 PM

Friday: 7:30 AM - 5:30 PM

Saturday: 7:30 AM - 5:30 PM



Chicago

53rd Annual Membership Meeting
March 12-14, 2010
Hosted by the Chicago Area Ataxia Support Groups

AIM 2010 Biographies

Key Note Speakers



Keith Caldecott, Ph.D.

Keith Caldecott is a Professor of Biochemistry at the Genome Damage and Stability Centre in Sussex. Professor Caldecott conducted his PhD studies at the National Institute for Medical Research in London, before spending postdoctoral periods in California at the Lawrence Livermore National Laboratory and in London at the Cancer Research UK Clare Hall Laboratories.

Professor Caldecott's research is focused on characterising novel human DNA strand break repair pathways, and investigating the link between defects in these pathways and human genetic disease. This work has identified a number of novel human DNA single-strand break repair and DNA double-strand break repair proteins, and has highlighted the relationship between defects in chromosomal single-strand break repair and hereditary neurodegenerative disease. Future work in the Caldecott laboratory will address the molecular mechanisms by which un-repaired single-strand breaks impact on neural function and human health.



Christian Hansel, Ph.D.

Dr. Christian Hansel is Professor of Neurobiology at the University of Chicago and Chair of the University of Chicago Graduate Program in Neurobiology. Dr. Hansel performed his graduate work at the Max-Planck-Institute for Brain Research in Frankfurt (Germany), and obtained his Ph.D. from the University of Zurich (Switzerland), where he also studied as an undergraduate student. He worked for 3 years as a postdoctoral fellow at Johns Hopkins University (Baltimore), after which he accepted his first faculty position at the Erasmus University Medical Center in Rotterdam (The Netherlands). After 8

years in Rotterdam, Dr. Hansel joined the Department of Neurobiology at the University of Chicago. Dr. Hansel's research focuses on molecular and cellular mechanisms of motor adaptation and learning in the cerebellum. His laboratory studies various forms of synaptic and non-synaptic plasticity in the cerebellum of rats and genetically modified mice using whole-cell patch-clamp recordings and fluorometric calcium imaging techniques. Clinically relevant aspects of his research include studies on alterations in Purkinje cell physiology in cerebellar ataxias, the use of memory enhancing drugs in the cerebellum as well as the effects of alcohol consumption on cerebellar computation.



Ole Isacson, M.D.

Dr. Ole Isacson is Professor of Neurology (Neuroscience) at Harvard Medical School and Director of the Center for Neuroregeneration Research and the Neuroregeneration Laboratory at McLean Hospital, an NIH Udall Parkinson's Disease Research Center of Excellence. He is also a member of the Scientific Advisory Board of the Harvard NeuroDiscovery Center and Principal Faculty of the Harvard Stem Cell Institute. Dr. Isacson received his Medical Bachelor (1984) and Doctor of Medicine (a research doctoral degree in Medical Neurobiology, 1987) from the University of Lund, Sweden. In 1989, after a 2-year postdoctoral neuroscience fellowship position at Cambridge University, England,

Dr. Isacson joined Harvard as an Assistant Professor of Neuroscience. Over the last two decades, his laboratory has grown to an internationally recognized academic research center for Parkinson's disease and related disorders, funded by the NIH, DOD and private foundations. Dr. Isacson's scientific models and studies of conceptually new therapies for neurodegenerative diseases have resulted in many new findings and clinical trials for Parkinson's and Huntington's disease. He is a founding member and past President of the American Society for Neural Therapy & Repair, and the past President of the international Cell Transplant Society, CTS (branch of The Transplantation Society, TTS). He serves as a scientific reviewer and advisor to medical and scientific journals, to the NIH, ESF, and many Parkinson community groups. Dr. Isacson has received several international prizes, research awards and lectureships. He is author or co-author of over 250 scientific research articles and 3 books in his field, and Editor-in-Chief of Molecular and Cellular Neuroscience.

Biographies



Richard Morimoto, Ph.D.

Richard Morimoto is the Bill and Gayle Cook Professor of Biology, Professor of Biochemistry, Molecular Biology and Cell Biology, and Director of the Rice Institute for Biomedical Research at Northwestern University. He holds a B.S. from the University of Illinois at Chicago, received his Ph.D. in Biology from the University of Chicago, and did postdoctoral research at Harvard University in Cambridge, MA. Widely recognized for his research on the regulation of the heat shock stress response and the function of molecular chaperones, the focus of his current research is to understand how organisms sense and respond to physiologic and environmental stress through the activation of genetic pathways that integrate stress responses with molecular and cellular responses that determine protein quality control, the health of the cell, and the lifespan of the organism. Consequently, these studies provide a molecular basis to understand the molecular mechanisms that maintain the stability of the proteome in health and the basis of diseases of protein conformation including neurodegenerative, metabolic, and oncological diseases. Morimoto has published over 200 papers and edited three monographs and two books. He has received many academic honors and awards including a MERIT award from the National Institutes of Health, membership in the American Association for the Advancement of Science (AAAS) and is currently funded by the National Institutes for General Medical Science, National Institutes of Aging, National Institute of Neurological Disorders and Stroke, Huntington Disease Society of America, and the ALS Association. At Northwestern University, Morimoto has served as the Dean of the Graduate School and Associate Provost of Graduate Education and as the Chair of the Department of Biochemistry, Molecular Biology, and Cell Biology. He has a long-standing interest in education and training includes directing the undergraduate biology program, the formation of the Interdepartmental Biological Sciences Graduate Program, and the Medical Scientist Training Program. On a national level, Morimoto has served on the NIH Molecular Biology Study Section, the NIGMS Molecular and Cellular Basis of Disease Panel, the AAAS Scientific Program Committee, the Beckman Scholars Advisory Panel, and the National Institute for General Medical Sciences Advisory Board where he was actively involved with the assessment of minority training programs. He currently serves on the Advisory Boards at the University of Heidelberg, RIKEN Brain Science Institute, Roswell Park Cancer Institute, BioCity Turku, and the Institute for Frontier Medical Science at Kyoto University. He is a co-founder of Proteostasis Therapeutics, Inc. a biotech recently formed in Cambridge, MA to discover small molecule therapeutics for diseases of protein conformation.

Session Presenters



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

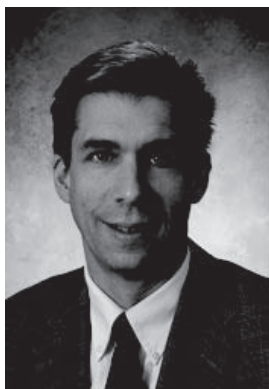
Dr. Tetsuo Ashizawa is Professor and Chairman of the Department of Neurology at the University of Florida (UF), Gainesville, Florida. Dr. Ashizawa also holds the Melvin Greer Professorship of Neurology. Dr. Ashizawa received his medical degree from the Keio University School of Medicine in Tokyo in 1973. 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 nationwide 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.

Biographies



Kurt Fischbeck, M.D. Dr. Fischbeck received A.B. and A.M. degrees from Harvard University and an M.D. degree from Johns Hopkins. After a medical internship at Case Western Reserve University and a neurology residency at the University of California in San Francisco, he did postdoctoral research on muscular dystrophy at the University of Pennsylvania. In 1982 he joined the faculty in the Neurology Department at the University of Pennsylvania Medical School. In 1998 he came to the NINDS as Chief of the Neurogenetics Branch. Dr. Fischbeck received the Cotzias Award from the American Academy of Neurology and the Jacoby Award from the American Neurological Association and was elected to the Institute of Medicine of the National Academy of Sciences. His laboratory is studying the mechanisms of hereditary neurological and neuromuscular disorders, with the goal of developing effective treatment for these diseases.



Christopher M. 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.

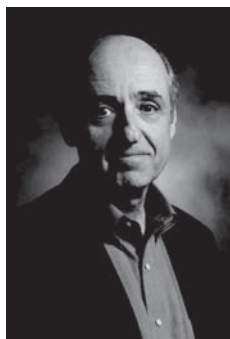


Joel Gottesfeld, Ph.D.

I received my undergraduate training in biochemistry at the University of California, Berkeley (1971), and spent one year as a Fulbright Scholar at Oxford University prior to pursuing graduate studies in biochemistry at the California Institute of Technology, in Pasadena. After finishing research for my Ph.D. in 1975, I then spent three years at the Medical Research Council Laboratory of Molecular Biology, in Cambridge, England, doing postdoctoral research on gene expression. I came to The Scripps Research Institute in 1978 as an assistant professor, and was promoted to full professor of Molecular Biology in 1994. I am also an Associate Editor of the *Journal of Biological Chemistry*.

Research in our laboratory focuses on the interplay between chromosome structure and gene regulation, and our current research concerns the development of small molecules to regulate gene expression. We are using both DNA binding molecules and other small molecules to regulate the expression of clinically significant genes. Recent efforts have focused on histone deacetylase (HDAC) inhibitors as potential therapeutics for neurodegenerative diseases, such as Friedreich's ataxia, spinal muscular atrophy, Huntington's disease, and myotonic dystrophy. These molecules can alter gene expression in cells, and in animal models for disease. We discovered a class of HDAC inhibitors that reactivate the silenced frataxin gene in both cell-based and animal models for Friedreich's ataxia. These molecules also show efficacy in a Huntington's disease mouse model. Most recently, we have explored the utility of HDAC inhibitors in cystic fibrosis, and find that molecules of this class may be potential therapeutics for this disease as well.

Biographies



Sidney Hecht, Ph.D.

Dr. Sidney Hecht is Director of the Center for BioEnergetics in the Biodesign Institute, and Professor of Chemistry at Arizona State University. He obtained his Ph.D. in Chemistry at the University of Illinois and studied as an NIH Postdoctoral Fellow in Molecular Biology at the University of Wisconsin. Over a period of almost four decades, he has held faculty appointments at MIT, the University of Virginia and ASU. From 1981-87 he held concurrent appointments at Smith Kline & French Laboratories, first as Vice President Preclinical R&D, then as Vice President Chemical R&D. At Smith Kline, he identified DNA topoisomerase I as the cellular target of the alkaloid camptothecin and participated in the discovery and development of the camptothecin analogue topotecan, now marketed under the tradename Hycamtin for the treatment of ovarian and small cell lung cancers. He founded the Center for BioEnergetics in 2006 to study the chemistry of the mitochondrial electron transport chain with the goal of devising therapeutic strategies to treat mitochondrial dysfunction. The present focus involves the study of coenzyme Q analogues designed to stabilize single electron transfers, and which can potentially be used to blunt multiple effects of redox stress in partially dysfunctional mitochondria. Another research interest involves the bleomycin group antitumor antibiotics; the current emphasis involves understanding the molecular mechanism of tumor recognition, and the mechanism of DNA and RNA degradation under conditions that model those in a therapeutic setting. Other research interests have included the use of misacylated transfer RNA's in cell free protein biosynthesizing systems for the elaboration *in vitro* of peptides and proteins containing synthetic amino acids at defined positions.



Karl Herrup, Ph.D.

Karl Herrup is a Professor and Chair of the Department of Neuroscience and Cell Biology at Rutgers University, a position he has held since 2006. He received his bachelor's degree from Brandeis University in Waltham, MA and his Ph.D. in Neuroscience from Stanford University in 1974. After two postdoctoral fellowships – in Neurogenetics at Children's Hospital/Harvard Medical School, and in Neuropharmacology at the Biozentrum in Basel Switzerland – he joined the faculty of the Human Genetics Department of Yale Medical School in 1978. He became Director of the Division of Developmental Neurobiology at the E. K. Shriver Center in Waltham, MA in 1988. In 1992 he moved to Case Western Reserve University Medical School and University Hospitals of Cleveland where he directed the Alzheimer's Center from 1999 through 2005. The prime focus of his current research is the relationship between the loss of cell cycle control in the mature 'post-mitotic' neuron and the process of cell death. He explores this linkage in human neurodegenerative diseases such as Alzheimer's disease and ataxia-telangiectasia and in their mouse models. The result has been a fresh perspective on the biology of neuronal cell death and on the role of cell cycle suppression in healthy adult neurons.



Ellen Hess, Ph.D.

Ellen J. Hess is a Professor in the Departments of Pharmacology and Neurology at Emory University School of Medicine. She received her B.A. in Psychobiology from Wellesley College and Ph.D. in Neuroscience from University of California at San Diego with postdoctoral training at The Scripps Research Institute. Before joining the faculty at Emory University in 2008, she held academic appointments at Johns Hopkins University School of Medicine and Pennsylvania State University College of Medicine. Dr. Hess research uses molecular genetic and pharmacological approaches to understand the roles of the basal ganglia and cerebellum in movement disorders, including dystonia and ataxia. Her work on episodic ataxia type-2 is providing insight into the factors that trigger ataxia with a focus on preventing nervous system dysfunction to build a foundation for the development of novel therapeutics.

Biographies



Grazia Isaya, M.D., Ph.D.

Grazia Isaya graduated *summa cum laude* from the University of Padova School of Medicine (Italy) in 1982 and enrolled in the neurology residency program there. After completion of her residency Dr. Isaya completed a research doctorate (Ph.D.) in Developmental Sciences. In 1987, she joined the laboratory of Dr. Leon Rosenberg in the Department of Genetics at Yale University. She obtained a faculty position at Yale in 1994, and joined the Mayo Clinic faculty in 1998. She is currently a Professor of Pediatrics and Biochemistry and Molecular Biology and the Director of the Mayo Medical Scientist Training Program. Her research laboratory focuses on mitochondrial iron balance and the biochemical basis of Friedreich's ataxia.



Joanna 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 rare disorders affecting balance and eye movement control in neurodevelopment and neurodegeneration. She has received funding support from private foundations and government agencies, including the National Eye Institute, the National Institute of Deafness and Communication Disorders, and the National Institute of Neurological Diseases and Stroke. She is an investigator in a multi-center Consortium for Clinical Investigations of Neurological Channelopathies (CINCH) headed by Dr. Robert Griggs at the University of Rochester. The ongoing natural history clinical study on episodic ataxia and other channelopathies will serve as a resource for future clinical trials.



Kamran Khodakhah, Ph.D.

Dr. Kamran Khodakhah obtained his bachelors (1989, Pharmacology, Kings College, University of London) and Ph.D. (1992, Pharmacology, University College, London) in the United Kingdom. His Ph.D. thesis research focused on examining the properties of InsP₃-mediated calcium release in cerebellar Purkinje cells. He then performed a post-doctoral fellowship with Dr. Clay Armstrong at the University of Pennsylvania, and subsequently assumed his first independent academic position in the department of Physiology and Biophysics at the University of Colorado Health Sciences Center in 1998. Dr. Khodakhah moved to Albert Einstein College of Medicine in 2001 where he currently holds the rank of Professor of Neuroscience. His interests revolve around understanding the role of the cerebellum and basal ganglia in motor coordination and movement disorders. His laboratory combines a multitude of techniques from electrophysiology and optophysiology in vitro and in vivo, to behavioral examination of transgenic animal models.



Thomas Klockgether, M.D.

Dr. Klockgether is currently at the University of Bonn in the Department of Neurology - Bonn, Germany. From April 1981 – April 1982 he obtained clinical training as a Resident in the Dept. of Medicine at Evangelisches Krankenhaus Oldenburg. Furthermore, from May 1982 – April 1983 he served as Resident, in the Dept. of Anaesthesiology at Pius-Hospital, Oldenburg and from April 1987 – Sept. 1991 he served as Resident in the Dept. Of Neurology at the University of Tübingen. Dr. Klockgether has been the Coordinator of the German Collaborative Research Group *Molecular Pathogenesis of SCA 3* since 2001 and is also the Coordinator of the EUROSCA Clinical Project. 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.

Biographies



Michel Koenig, M.D., Ph.D

Dr. Koenig currently is Professor of Human Genetics at the Faculty of Medicine of Strasbourg and is adjunct director of the Genetic Diagnosis Laboratory of the University Hospital. He is team leader in the Department of Neurobiology and Human Genetics at the Institute of Genetics and Molecular and Cell Biology at the University of Strasbourg, France where he received his Ph.D. in 1986 and his M.D. in 1990. From 1986 – 1989, Dr. Koenig worked in the laboratory of Pr LM. Kunkel on the characterisation of the Duchenne Muscular dystrophy gene. Dr. Koenig has been instrumental in the research on Friedreich's Ataxia, Ataxia/Oculomotor Apraxia 1 and 2, and several other recessive ataxias. He has worked with mapping and identification of novel ataxias, disease loci and genes, and construction of mouse and cellular models of Friedreich's ataxia.



Albert LaSpada, M.D., Ph.D., FACMG

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.

Biographies



Martin Lavin, Ph.D.

Martin Lavin is a molecular biologist and biochemist who with a major research interest in DNA damage recognition and maintenance of genome stability. His work has focused on the human genetic disorder ataxia-telangiectasia (A-T) with particular emphasis on cancer predisposition and neurodegeneration. Over the years he has contributed to cell cycle studies in A-T and a member of an international consortium under the leadership of Yosef Shiloh who cloned the A-T gene, ATM. More recently he has identified interacting partner proteins for ATM; cloned the full-length ATM cDNA and demonstrated correction of the radiosensitive cellular phenotype in A-T and described important substrates that mediate ATM signaling. A significant contribution to this area of research was the generation of a knockin *Atm* mutant mouse by his group that mimicked the A-T phenotype but which also revealed a propensity to develop tumours in heterozygote carriers. This provided important support for the observations that A-T carriers are predisposed to breast and other cancers. More recently he has extended his research interests to include a number of other autosomal recessive ataxias and is currently investigating the role of proteins such as aprataxin (defective in AOA1), senataxin (AOA2), CABC1 (ARCA2) in the response to stress with a view to understanding their roles in protecting the brain.

Martin Lavin obtained a BSc (Hons) from the National University of Ireland in Dublin before proceeding to a Ph.D. in Trinity College Dublin. After a post-doctoral stint at Syntex Research, Palo Alto he moved into a second post-doctoral position in the Biochemistry Department, University of Queensland, Brisbane, Australia. The emphasis of his work was on damage to DNA and its repair. During this period he initiated his association with ataxia-telangiectasia (A-T) in an attempt to identify the biochemical basis of the defect involved. He was appointed lecturer in Biochemistry, University of Queensland, promoted to Senior Lecturer and Associate Professor and in 1989 was appointed Professor in the Joint Oncology Program between the Queensland Institute of Medical Research and the University of Queensland. He became Foundation Professor of Molecular Oncology in 1994 as a joint position between these two institutes. In 2002 he was appointed a Senior Principal Research Fellow with the Australian National Health and Medical Research Council. He has published 342 papers in peer reviewed journals.



Xiao-Jiang Li, M.D., Ph.D.

Xiao-Jiang Li, M.D., Ph.D. is a Professor of Human Genetics at Emory University School of Medicine. He obtained his M.D. in China in 1982 and Ph.D. at Oregon Health Science University in Portland, OR, in 1991. He then did his postdoctoral training in Dr. Solomon Snyder's lab in the Department of Neuroscience at Johns Hopkins University from 1991 to 1995. Since 1996, he became a faculty member in the Department of Human Genetics at Emory University. The main interest of the Li Lab is to understand the molecular mechanisms of inherited neurodegeneration caused by a CAG repeat expansion in the disease genes. Currently, his lab focuses on Huntington's disease (HD) and SCA17, which are autosomal dominant genetic diseases caused by CAG repeat expansion in the disease proteins. Dr. Li's lab is using a variety of approaches, including genetic manipulation of animal models, molecular and cell biological analysis of protein transport, and biochemical study of protein-protein interactions, to investigate the relationship between gene mutation and disease phenotypes.

Biographies



Harry Orr, Ph.D.

Harry Orr, is the Director of the Institute of Human Genetics 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 and was appointed as NAF's Research Director on June 14, 2006.



Gulin Öz, Ph.D.

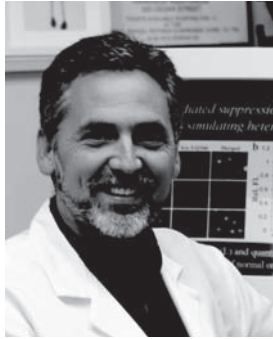
Dr. Öz is a brain imaging scientist who specializes in magnetic resonance spectroscopy (MRS) in degenerative brain diseases with special interest in spinocerebellar ataxias. She graduated from Bosphorus University in Istanbul, Turkey with B.S. degrees in Physics and Chemistry and obtained her Ph.D. in 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.



Massimo Pandolfo, M.D.

Dr. Pandolfo received his M.D. at the University of Milan, Italy in 1980, where he also trained as a neurologist. He did a post doctorate in molecular genetics at the University of California, Irvine. From 1988 to 1993, he worked in the Division of Biochemistry and Genetics of the Nervous System at the National Neurological Institute in Milan, Italy. From 1994 to 1996, he served as Assistant Professor of Neurology at Baylor College of Medicine in Houston, Texas. From 1996 to 2001, he has served as an Associate Professor of Medicine at the University of Montreal and as Adjunct Professor at McGill University's Department of Neurology and Neurosurgery in Montreal, Canada. Since 2001 he is the Head of the Department of Neurology at the Brussels Free University (ULB) Hospital in Brussels, Belgium. Dr. Pandolfo, working in collaboration with other researchers, discovered the Friedreich Ataxia gene in 1996. Dr. Pandolfo is also a member of the National Ataxia Foundation's Medical and Research Advisory Board.

Biographies



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.



Stefan Pulst, M.D.

Dr. Pulst is Professor and Chair of Neurology at the University of Utah in Salt Lake City. His research focuses on inherited diseases of the nervous system with an emphasis on spinocerebellar ataxias and Parkinson disease. Another interest relates to tumor suppressor genes controlling proliferation of Schwann cells. Recently, his work has also branched out into understanding the genetic structure of human visual attention. Dr. Pulst was founding chair of the Section on Neurogenetics and of the Basic Science Subcommittee of the American Academy of Neurology and currently serves as chair of the AAN Science Committee. From 1999 to 2006, he was Scientific Director of the National Ataxia Foundation and continues to serve on NAF's Medical Research Advisory Board.



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. Dr. Ranum joined the faculty at the University of Minnesota in 1994 where she is now a Professor of Genetics, Cell Biology and Development, Research Director of the Paul and Sheila Wellstone Muscular Dystrophy Center and a member of the Institute of Human Genetics and Institute for Translational Neuroscience. 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.

Biographies



Norio Sakai, M.D., Ph.D.

Dr. Norio Sakai is Professor of Department of Molecular and Pharmacological Neuroscience at the Graduate School of Biomedical Sciences, Hiroshima University, Japan. He graduated from Kobe University School of Medicine and obtained M.D. in 1986. After 3-year neurology residency, he entered Graduate School of Medical Sciences, Kobe University and earned his Ph.D. at the Department of Pharmacology in 1993. He trained further at Biosignal Research Center, Kobe University and moved to present position in 2002. One of his research concerns is the function of protein kinase C (PKC), a phospholipids and calcium –activated serine/ threonine kinase. He developed the experimental system, which enable us to monitor the movement of PKC translocation in living cells, using PKC fused with GFP (PKC-GFP). The part of this work is now introduced in the world-wide textbook of the Molecular Biology of the Cells, fifth edition. Using this method, he clarified the temporal and locoregional significance of PKC in cellular functions. Since gamma PKC was identified as a causal gene for SCA14 in 2002, his research interest has been focused on elucidating the pathogenesis of spinocerebellar ataxia. He also applied live imaging method of PKC for this purpose. To date, his lab have clarified that causal mutant gamma PKC found in SCA14 family has a prone to be aggregated and aberrantly-regulated kinase activity.



Ana Solodkin, Ph.D.

Dr. Solodkin is a Research Associate, Assistant Professor in the Department of Neurology at the University of Chicago. She earned her Ph.D. in Biophysics at the “Center for Research and Advanced Studies of the National Polytechnic Institute” in Mexico City and at the National Institutes of Health in Bethesda, MD. After finishing this initial preparation, she completed her post-doctoral training joining the Cognitive Neurology group under the direction of Dr. Antonio Damasio and receiving direct training with Dr. Gary van Hoesen in the area of human neuroanatomy. From her initial work in animal model system addressing issues of plasticity to the study of human cortical anatomy in illness, her active and long-term research has focused on the relationship between basic neurobiology and neurological disease as assessed with mathematical network modeling.

Recently she started working on autosomal dominant spinocerebellar ataxias (type 1 (SCA1) and type 6 (SCA6)), bringing together her combined interests in motor system physiology, degenerative diseases, and connectivity analysis. Although the genetic advances in SCAs have served as the staging platform for the earliest steps to develop therapy, they have also radically outstripped the progress and preparation in the development of useful outcome measures to gauge the success of experimental therapies. Thus, the identification and validation of additional disease biomarkers can both, provide more sensitive surrogate measures of disease progression and enable design of more efficient trials. In the current SCA study Dr. Solodkin applies a combination of functional and structural brain imaging to characterize the state of cerebellar connectivity.



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 currently an Associate 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, sign-

Biographies

ing 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.

Session Co-Chairs



Alexandra Durr, M.D., Ph.D

Alexandra Durr works as a consultant in Neurogenetics in the Genetic Department at the Salpêtrière Hospital in Paris, France in the group of Alexis Brice. She trained in Germany and France in Neurology and Genetics. After her medical degree obtained in 1992 she joined the Neurological Department and the INSERM research laboratory on experimental therapy in neurodegenerative disorders in Paris. Following her neurological degree, she obtained her Ph.D. in medical genetics in 1998. Her research interest is the phenotypical expression of genetic disorders focussed on cerebellar ataxias, spastic paraplegias and inherited conditions of other movement disorders. She opened the first presymptomatic testing clinic for Huntington disease in France and is a member of scientific committee of lay organisations devoted to cerebellar ataxias, spastic paraplegias and Huntington disease. She coordinates a European research network called SPATAX including 27 groups to reinforce the links of laboratories and clinical centers. She is a member of the executive committee of the Ataxia Study Group (www.ataxia-study-group.net).



Wendy R. 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.



Russell Margolis, M.D.

Dr. Russell L. Margolis received his undergraduate degree from Princeton University and M.D. degree from Johns Hopkins University. After a residency in psychiatry at Johns Hopkins, he did a neurogenetics fellowship at the NIH. In 1992, he joined the faculty of the Johns Hopkins Department of Psychiatry. He is now Professor of Psychiatry and Neurology and Director of the Laboratory of Genetic Neurobiology and the Johns Hopkins Schizophrenia Program. Work in his laboratory focuses on the neurogenetics of brain disorders, including ataxias, Huntington's disease and related

disorders, and schizophrenia. He is an attending psychiatrist for the Johns Hopkins Schizophrenia inpatient unit, the Johns Hopkins Bayview Early Psychosis Intervention Clinic, and the Johns Hopkins Affective Disorders Consultation Clinic, and he also provides psychiatric evaluation for patients with movement disorders.

Biographies



Jeremy Schmahmann, M.D.

Jeremy D. Schmahmann received his medical degree in 1980 from 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/Behavioral Neurology Unit, Neurology Clerkship Director, and Director of the Laboratory for Neuroanatomy and Cerebellar Neurobiology. Dr. Schmahmann is Professor of Neurology at Harvard Medical School, and a Scholar in the Academy at Harvard.

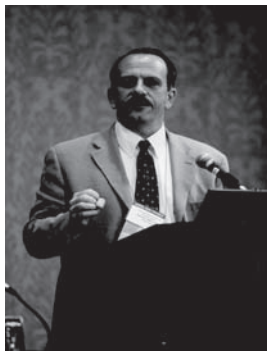
He was awarded the Norman Geschwind Prize in 2000 for research in behavioral neurology from the American Academy of Neurology and the Behavioral Neurology Society, recognizing his contribution to establishing the field of science that explores the contribution of the cerebellum to cognition and emotion. Dr. Schmahmann received 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, a member of the Medical Research Advisory Board of the National Ataxia Foundation, and he has been cited in *The Best Doctors in America* since 1998. Dr. Schmahmann's work on the clinical neurology, neuroanatomy and connections of the cerebellum in humans and monkey has been published in peer reviewed journals and academic texts. His published 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) that was awarded the 2006 Medical Science Award for best book in the category of medical publications by the American Association of Publishers.



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 recognized 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.



George "Chip" Wilmot, M.D., Ph.D.

Dr. Wilmot received his M.D., Ph.D. from the University of Michigan, did a neurology residency at Emory University, and then remained at Emory as faculty in the Department of Neurology. Although trained in basic science and initially focusing his research on mechanisms of axonal stability and regeneration, Dr. Wilmot is currently most active in clinical research in ataxia. He is a member and past leader of the Cooperative Ataxia Group, a member of the Collaborative Clinical Research network in Friedreich's Ataxia, and started the Cooperative Ataxia Registry, a patient-based registry for those afflicted with ataxia.

Biographies



Sarah Ying, M.D.

Dr. Sarah Ying is an Assistant Professor of Neurology with a secondary appointment in Ophthalmology. 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 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, and developing neurophysiology and neuroimaging biomarkers to elucidate systems-level control of movement, balance, and cognition. The Cerebellar Imaging and Eye-movement Laboratory (CIEL) is currently 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.

Junior Lecturers



Mirella Dottori, Ph.D.

Dr Mirella Dottori completed her Ph.D. studies at Walter and Eliza Hall Institute, University of Melbourne, Australia. She then went to The Salk Institute for Biological Studies, in California, USA, where she studied the development of the spinal cord. She returned to Australia as a Howard Florey Fellow and joined Professor Martin Pera's lab at Monash University, working on human embryonic stem cells and their differentiation to neurons. In 2007 Dr. Dottori established a Stem Cell Lab at the Centre for Neuroscience, University of Melbourne, where she is pursuing her research in the use of stem cells for developing treatments for neurodegenerative disorders, including Friedreich Ataxia and Parkinson's Disease



Isabelle Iltis, Ph.D.

Dr. Iltis graduated from the Université de la Méditerranée, Aix-Marseille II in 2005. After a master's thesis in developmental genetics in 2001, she did her Ph.D. thesis in the Centre de Résonance Magnétique Biologique et Médicale (CRMBM) in Marseille, where she studied myocardial blood flow using non-invasive MRI methods. She came to the University of Minnesota in April 2005 and started working at the Center for Magnetic Resonance Research (CMRR) first as a post-doc, now as an associate professor. She studies mainly brain metabolism in rodent models and human using ^1H - and ^{13}C -MRS methods developed at CMRR. Because of her background in genetics and current studies on brain metabolism, research on ataxias has been of particular interest to her.

Biographies



Paula Ladd, Ph.D.

Dr. Ladd received her Ph.D. from the Department of Biochemistry and Molecular Biology at the Indiana University School of Medicine under the direction of Dr. David G. Skalnik. In 2004, she joined the Fred Hutchinson Cancer Research Center as a postdoctoral fellow in the human biology division, where she was co-mentored by Drs. Galina Filippova and Stephan J. Tapscott. She accepted a senior fellow position at the University of Washington in 2008 in the laboratory of Dr. Albert R. La Spada. In 2009, Dr. Ladd was appointed to Assistant Project Scientist at the University of California, San Diego. Her research interests are in trinucleotide repeat expansion disorders, with a particular interest in SCA7 and transcriptional regulation of the human Ataxin-7 gene.

Dr. Ladd has received a Young Investigator Research Grant from the NAF for her ataxia research efforts.

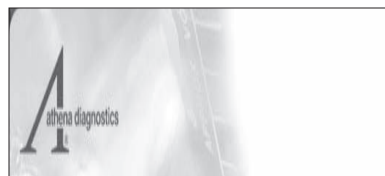


Alain Martelli, Ph.D.

Dr. Martelli received his Ph.D. in Bioorganic chemistry from the University of Grenoble, France, in 2001. After a one year postdoctoral fellow position in the Chemistry department of Yale University, he joined the group of Dr. Jean-Marc Moulis in 2002 at the CEA-Grenoble, developing a cellular and biochemical approach to study iron metabolism. In particular, using *in vivo* and *in vitro* approaches, Dr. Martelli studied the iron-sulfur cluster dependant regulation of mammalian Iron Regulatory Protein 1 activity. In 2006, Dr. Martelli joined the group of Dr. Hélène Puccio to work on Friedreich's ataxia at the IGBMC, Strasbourg, France. He contributed to show that frataxin deficiency was affecting not only mitochondrial iron-sulfur cluster proteins but also cytosolic and

nuclear iron-sulfur cluster proteins, opening new venues on the molecular pathways that might be involved in the pathophysiology. In 2008, Dr. Martelli received the New Investigator Grant from the Friedreich's Ataxia Research Alliance to determine the mechanism of iron dysregulation in frataxin deficient tissues, and to unravel the molecular characteristics of the *de novo* iron-sulfur cluster biosynthesis pathway in which frataxin is involved.

Thank you to the AIM 2010 Sponsors:



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Bob Allison Ataxia
Research Center



Poster Abstracts

AIM 2010 Invited Abstract Presenters

1. Julieto Araujo, MSc: ATXN3 cooperates in the FOXO4-dependent oxidative stress response
University Clinics Bonn, Bonn, Germany
2. Esther Becker, PhD: TRPC3 Signaling in Purkinje Cell Development and Cerebellar Ataxia
University of Oxford, Oxford, UK
3. Sanjay Bidichandani, PhD: Mechanism of epigenetic silencing of the *FXN* gene in Friedreich ataxia
University of Oklahoma Health Sciences Center, Oklahoma City, OK
4. Jana Boy, PhD: Analysing proteasomal impairment in SCA3 by using a conditional mouse model
University of Tuebingen, Tuebingen, Germany
5. Andreia Teixeira de Castro, MSc: Opposing effects of distinct aging-related pathways in a pan-neuronal *C. elegans* model of Machado-Joseph Disease
Univeristy of Minho, Braga, Portugal
6. Dong-Hui Chen, MD, PhD: The effect of PKC mutations on its functions and aggregation in Spinocerebellar Ataxia 14
University of Washington School of Medicine, Seattle, WA
7. G. Giacomo Consalez, MD: On the origin of Purkinje cells and cerebellar nuclei neurons: a genetic fate mapping study
San Raffaele Scientific Institute, Milan, Italy
8. Maria do Carmo Costa, PhD: A cell-based screen for small molecules that reduce steady state levels of ataxin-3
University of Michigan, Ann Arbor, MI
9. Xiaofei Du, MD: Regulatory function of normal and SCA6-expanded *a1A* C termini of P/Q calcium channels in neuronal gene expression and differentiation
University of Chicago, Chicago, IL
10. Uzay Emir, PhD: Detection of Early Neurochemical Changes in a SCA1 Mouse Model by Proton (¹H) Magnetic Resonance Spectroscopy
University of Minnesota, Minneapolis, MN
11. Yuanzheng Gao, PhD: III spectrin is essential for development of dendrites and spines in Purkinje neurons *in vitro*
University of Minnesota, Minneapolis, MN
12. Katherine Hekman, BA: Molecular pathogenesis of SCA26
University of Chicago, Chicago, IL
13. Hong Jiang, MD: SCA32: An Autosomal Dominant Cerebellar Ataxia with Azoospermia Maps to Chromosome 7q32-q33
Central South University, Changsha, Hunan, China

Poster Abstracts

14. Chang H. (Brian) Jung, BA: High-resolution magnetic resonance imaging shows a distinct pattern of region-specific cerebellar atrophy in spinocerebellar ataxia type 2

Johns Hopkins University, Baltimore, MD

15. Kevin Kemp, PhD: Stem cell-secreted Superoxide Dismutase protects cerebellar neurons against nitric oxide mediated injury

University of Bristol, Bristol, UK

16. Mee Whi Kim, PhD: Secondary structure of Huntingtin amino-terminal region containing polyglutamine expansion,

University of Texas Southwestern Medical Center, Dallas, TX

17. Mingang Li, PhD: Pathological mutations in the human cytoskeletal protein beta-III spectrin induce neurodegeneration in a *Drosophila* model of Spinocerebellar Ataxia type 5

University of Minnesota, Minneapolis, MN

18. Filip Lim, PhD: Human biopsy-derived olfactory mucosa neural precursors as cell models for Friedreich's ataxia

Universidad Autonoma de Madrid, Madrid, Spain

19. Michele MP Lufino, PhD: Novel genomic DNA-reporter fusion models for the study of Friedreich's ataxia

University of Oxford, Oxford, UK

20. Thorsten Mueller, PhD: Importance of phosphorylation on nuclear localization of ataxin-3

Neurobiology Uniklinikum Bonn, Bonn, Germany

21. Puneet Opal, MD, PhD: VEGF ameliorates the ataxic phenotype in spinocerebellar ataxia type 1 (SCA1) mice

Northwestern University Feinberg School of Medicine, Chicago, IL

22. Alice Pebay, PhD: Generation and Functionality of Induced-Pluripotent Stem Cell Lines from Friedreich Ataxia Patients

The University of Melbourne, Melbourne, Australia

23. Helene Puccio, PhD: Modeling Friedreich ataxia by the development of induced pluripotent stem cells carrying (GAA)_n pathogenic expansions

Institut de Genetique et de Biologie Moleculaire et Cellulaire, Cedex, France

24. Abrar Qurashi, PhD: Fragile X premutation rCGG repeats alter the nuclear export of specific mRNAs

Emory University School of Medicine, Atlanta, GA

25. Myriam Rai, PhD: Two new pimelic diphenylamide HDAC inhibitors induce sustained frataxin upregulation in cells from Friedreich's ataxia patients and in a mouse model.

Université Libre de Bruxelles, Brussels, Belgium

Poster Abstracts

26. Edgardo Rodriguez, PhD: miR-based RNAi targeting of ataxin-3 expression as a potential therapy for SCA3

University of Michigan Medical School, Ann Arbor, MI

27. Francesco Sacca, MD: Epoetin alfa increases frataxin in Friedreich's ataxia without affecting hematocrit

University Federico II, Naples, Italy

28. Joseph P. Sarsero, PhD: Pharmacological screening for the therapy of Friedreich ataxia

The University of Melbourne, Royal Children's Hospital, Victoria, Australia

29. Janine Scholefield, PhD: Gene silencing for SCA7

University of Oxford, Oxford, UK

30. Vikram G. Shakkottai, MD, PhD: Rescue of Purkinje neuron depolarization block improves the motor phenotype in a mouse model of Spinocerebellar Ataxia type 3

The University of Michigan, Ann Arbor, MI

31. Masayoshi Tada, MD, PhD: Detection of Polyglutamine Protein Oligomers in Living Cells Using Protein-fragment Complementation Assays

University of Michigan, Ann Arbor, MI

32. Peter Todd, MD, PhD: Histone Deacetylases suppress CGG repeat-induced neurodegeneration via transcriptional silencing in models of Fragile X Tremor Ataxia Syndrome.

The University of Michigan, Ann Arbor, MI

33. Sokol V. Todi, PhD: The deubiquitinating activity and cellular functions of the SCA3 disease protein ataxin-3 are directly regulated by ubiquitination

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34. Wei L. Tsou, MS: Splice-specific miRNA-based RNAi targeting of the pathogenic CACNA1A transcript

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35. Jamie Weiss, PhD: G-Protein Coupled Receptor Regulation of P/Q-type Ca^{2+} Channels in Ataxia

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