Welcome to the Second Ataxia Investigator's Meeting

Sponsored by the National Ataxia Foundation

In conjunction with funding support from:

Athena Diagnostics, Inc.
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Friedreich's Ataxia Research Alliance (FARA)
Friedreich's Ataxia Research Association (FARA Australia)
Gordon and Marilyn Macklin Foundation
Chesapeake Chapter - NAF
National Institute of Neurological Disorders and Stroke (NINDS)
Santhera Pharmaceuticals
Muscular Dystrophy Association

March 25-28, 2008 ✹ Flamingo Hotel and Casino ✹Las Vegas, NV

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Ataxia Investigator's Meeting General Overview

Tuesday	Event	Location
4:00 - 7:00 PM	Registration	Laughlin Foyer
4:00 - 7:00 PM	Poster Session I (Posters on Display through Thursday)	Reno Foyer
7:00 - 8:00 PM	Keynote Speaker: Mary E. Hatten, PhD	Laughlin 1 & 2
8:00 - 9:30 PM	Dinner	Laughlin 1 & 2
WEDNESDAY	**Event**	**Location**
7:30 - 8:30 AM	Breakfast	Laughlin 1 & 2
8:30 - 10:30 AM	Session 1: Development	Laughlin 1 & 2
10:00 -10:30 AM	Discussion	Laughlin 1 & 2
10:45 - 12:15 PM	Session 2: Abnormal DNA and RNA Processing	Laughlin 1 & 2
12:15 - 12:45 PM	Discussion	Laughlin 1 & 2
12:45 - 2:00 PM	Lunch & Poster Session II	Reno Foyer
2:00 - 3:05 PM	Session 3: Protein mediated Ataxia Pathogenesis	Laughlin 1 & 2
3:05 - 3:50 PM	Discussion	Laughlin 1 & 2
4:00 - 9:00 PM	Social Event: Lake Mead Boat Ride & Dinner	Lake Mead
Thursday	**Event**	**Location**
7:30 - 8:30 AM	Breakfast	Laughlin 1 & 2
8:30 - 10:00 AM	Session 4: Cell Biological Mechanisms of Ataxia	Laughlin 1 & 2
9:30 - 10:00 AM	Discussion	Laughlin 1 & 2
10:30 - 12:30 PM	Session 5: Mitochondrial Dysfunction	Laughlin 1 &2
11:45 - 12:30 PM	Discussion	Laughlin 1 & 2
12:30 - 2 PM	Lunch and Poster Session III	Reno Foyer
2:00 - 3:30 PM	Session 6A: Therapeutic and Assessment Methods - I	Laughlin 1 & 2
4:00 - 5:30 PM	Session 6B: Therapeutics and Assessment Methods - II	Laughlin 1 & 2
4:45 - 5:30 PM	Discussion (I & II)	Laughlin 1 & 2
6:00 - 8:30 PM	Dinner	Scenic Room
7:30 - 8:30 PM	Session 7: Breaking News / Young Investigator Awards	Scenic Room
FRIDAY	**Event**	**Location**
7:30 - 8:30 AM	Breakfast	Twilight Room
8:30 - 10:45 AM	Session 8: Program Development	Laughlin 1 & 2
10:15 - 10:30 AM	Discussion	Laughlin 1 & 2
11:00 - 12:00 PM	Session 9: Summary Session	Laughlin 1 &2
11:00 - 12 :30	Lunch	Laughlin 1 & 2
2:00 - 5:00 PM	Birds of a Feather: Interaction of investigators with patients and families	Various Meeting Rooms

Tuesday, March 25, 2008

Time	Event	Room
4:00 - 7:00 PM	**Registration**	**Laughlin Foyer**
4:00 - 7:00 PM	**Poster Session I** **(Posters on Display through Thursday)**	**Reno Foyer**
7:00 - 8:00 PM	**Opening Keynote Speaker: Mary E. Hatten, PhD, The Rockefeller University** "Cerebellar Development, Migration and Differentiation?"	**Laughlin 1 & 2**
8:00 - 9:30 PM	**Dinner**	**Laughlin 1 & 2**

Thank you!

The National Ataxia Foundation would like to take this opportunity to thank each AIM participant for taking time from their busy schedule to attend the Second Ataxia Investigator's Meeting.
On behalf of the ataxia families we serve, Thank You.

Thank You to the AIM Steering Committee:
Alexis Brice, MD
Harry Orr, PhD
Henry Paulson, MD, PhD
Huda Zoghbi, MD
Rob Wilson, MD, PhD
Stephan Pulst, MD
Thomas Klockgether, MD
A Special Thank You to
John Day, MD for heading up the
AIM Steering Committee
The National Ataxia Foundation's
Board of Directors

The National Ataxia Foundation has Research Funding Available!

The National Ataxia Foundation (NAF) began direct funding of ataxia research studies in 1978. Over the past nine years the Foundation has funded one-hundred seventeen ataxia research studies in ten countries.

The National Ataxia Foundation has four (4) ataxia research programs including: 1) NAF Research Program, 2) NAF Research Fellowship Award, 3) NAF Young Investigator Award, and 4) NAF Friedreich's Ataxia Special Projects Fund.

Currently, the NAF Young Investigator Award's funding emphasis is on two forms of ataxia: Friedreich's ataxia, and SCA3.

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

Wednesday March 26, 2008

Time	Event	Room
7:30 - 8:30 AM	**Breakfast**	**Laughlin 1 & 2**
8:30 -10:30 AM	**Session 1: Development** **Chair: M. Hatten, PhD**	**Laughlin 1 & 2**
8:30 AM	**Special Lecture: Nathaniel Heintz, The Rockefeller University** "Cerebellar Cellular Interaction"	
9:15 AM	**Dandy-Walker: K. Millen, PhD**	
9:30 AM	**X-linked Microcephaly with Cerebellar Hypoplasia: W. Dobyns, MD, PhD**	
9:45 AM	**Joubert: E.M. Valente, MD, PhD**	
10:00 AM	**Discussion: Crawford, H. Orr, PhD, and H. Zoghbi, MD**	
10:30 - 10:45 AM	**Break**	
10:45 - 12:45 PM	**Session 2: Abnormal DNA and RNA Processing** **Chair: L. Ranum, PhD**	**Laughlin 1 & 2**
10:45 AM	**Special Lecture: Yosef Shiloh, PhD, Tel Aviv University** "Ataxia Telangiectasia: Closer to Unraveling the Mystery….?"	
11:30 AM	**AOA-1: M Koenig, MD**	
11:45 AM	**SCA-10: T Ashizawa, MD, PhD**	
12:00 PM	**AOA2: M. Lavin, PhD (CIC)**	
12:15 PM	**Discussion: C. Pearson, PhD, Crawford, R. Margolis, MD**	
12:45 - 2:00 PM	**Lunch and Poster Session II**	**Reno Foyer**
2:00 - 4:00 PM	**Session 3: Protein Mediated Pathogenesis** **Chair: S. Pulst, MD**	**Laughlin 1 & 2**
2:00 PM	**SCA 1: H. Orr, PhD**	
2:10 PM	**SCA 1: H. Zoghbi, MD**	
2:20 PM	**SCA 3: U. Wüllner, MD, PhD**	
2:35 PM	**ARSACS: A. Richter, PhD**	
2:50 PM	**DRPLA: S. Tsuji, MD, PhD**	
3:05 PM	**Discussion: H. Paulson, MD, PhD, A. Brice, MD, A. LaSpada, MD, PhD**	
4:00 - 9:00 PM	**Social Event: Lake Mead Boat Ride & Dinner**	**Lake Mead**

Thank You!

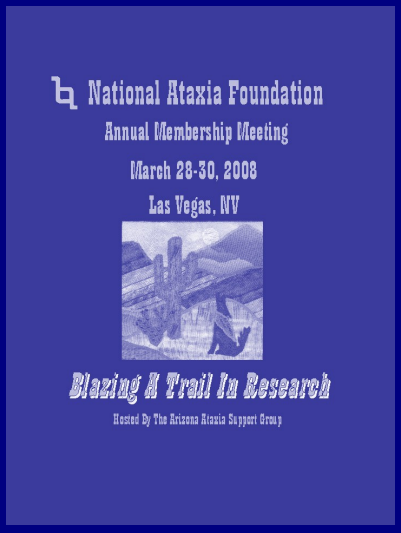
The National Ataxia Foundation wishes to extend a very special thank you to the organizers and supporters of this meeting. Thank you!

Thursday, March 27, 2008

Time	Event	Room
7:30 - 8:30 AM	Breakfast	Laughlin 1 & 2
8:30 - 10:00 AM	Session 4: Cell Biology Mechanisms of Ataxia Chair: A. LaSpada, MD, PhD	Laughlin 1 & 2
8:30 AM	SCA 5: L. Ranum, PhD	
8:45 AM	EA6: J. Jen, MD, PhD	
9:00 AM	SCA 13: S. Pulst, MD	
9:15 AM	ARCA-1: N. Dupré, MD, MSc, MPH, FCRP(C)	
9:30 AM	Discussion: G. Lopez, MD, C. Gomez, MD, PhD, Durr, MD, PhD	
10:00 - 10:30 AM	Break	
10:30 - 12:30 PM	Session 5: Mitochondrial Dysfunction Chair: M. Pandolfo, MD	Laughlin 1 & 2
10:30 AM	FA Cellular: G. Isaya, MD, PhD	
10:45 AM	FA Models: H. Puccio, MD, PhD	
11:00 AM	FA GAA Instability: S.I. Bidichandani, MBBS, PhD	
11:15 AM	Infantile onset spinocerebellar ataxia: K. Nikali, MD, PhD	
11:30 AM	Cayman Ataxia: M. Burmeister, PhD	
11:45 AM	Discussion: K. Fishbeck, MD, M. Koenig, MD	
12:30 - 2:00 PM	Lunch and Poster Session III	Reno Foyer
2:00 - 3:30 PM	Session 6A: Therapeutic and Assessment Methods - I Chair: K. Fischbeck, PhD	Laughlin 1 & 2
2:00 PM	Special Lecture: Don Cleveland, PhD, USCD "Non-Cell Autonomous Death of Motor Neurons in ALS: RNA Silencing Therapy Neurodegenerative Disease"	
2:45 PM	HDACi: J. Gottesfeld, PhD	
3:00 PM	PolyQ Screen: P. Muchowski, PhD	
3:15 PM	FA High Throughput Screen: R. Wilson, MD, PhD	
3:30 - 4:00 PM	Break	
4:00 - 5:30 PM	Session 6B: Therapeutic and Assessment Methods - II Chair: H. Paulson, MD, PhD	Laughlin 1 & 2
4:00 PM	MRI Quantitative methods: J. Schmahmann, MD	
4:15 PM	MRS: G. Oz, PhD	
4:30 PM	Transcriptome Markers: O. Riess, MD, PhD	
4:45 PM	Discussion (I & II): G. Lopez, MD, T. Klockgether, MD, C. Pearson, PhD	
6:00 - 8:30 PM	Dinner	Scenic Room
7:30 - 8:30	Session 7: Breaking News / Young Investigator Awards	
7:30 - 7:50PM	Selected Junior Poster Presentations - 1, 2, & 3	
8:00 PM	Breaking News - SCA 11: N. Wood, PhD, FRCP, FMedSci	
8:10 PM	Breaking News - SCA 15: A. Singleton, PhD	
8:20 PM	Breaking News - SCA 28: F. Taroni, MD	
8:30 PM	Breaking News - Genetics of MSA: S. Tsuji , MD, PhD	

Friday, March 28 2008

Time	Event	Room
7:30 - 8:30 AM	Breakfast	Twilight Room
8:30 - 10:45 AM	Session 8: Program Development Chair: T. Ashizawa, MD, PhD	Laughlin 1 & 2
8:30 AM	Special Lecture: Ira Shoulson, MD, University of Rochester "HD: Evidence-Based Therapeutics and Therapeutic-Based Evidence"	
9:15 AM	CAG: S. Subramony, MD	
9:30 AM	EuroSCA: T. Klockgether, MD & A. Brice, MD	
9:45 AM	Euro FA: M. Pandolfo, MD	
10:00 AM	USFA: D. Lynch, MD	
10:15 AM	Discussion: G. Wilmot, MD, PhD, S. Perlman, MD	
10:30 AM	Break	
11:00 - 12:00 PM	Session 9: Summary Session Chair: J. Day, MD, PhD	Laughlin 1 & 2
11:00 AM	NIH perspective: J. Porter, PhD	
11:30 AM	FARA Perspective: R. Bartek	
11:35 AM	NAF Perspective: H. Orr, PhD	
11:40 AM	A-TCP Perspective: B. Margus	
11:45 AM	MDA Perspective:	
11:50 AM	Patient's Perspective: E. McLaughlin	
11:55 AM	Conclusion: J. Day, MD, PhD	
11:30 - 12:30 PM	Lunch	Laughlin 1 & 2
2:00 - 5:00 PM	"Birds of a Feather" interaction of investigators with patients and families	Various



We hope that you are planning on joining us at the NAF Annual Membership Meeting.

Please remember to register in the Reno I Room and pick up your conference materials if you are extending your stay.

Registration Hours:

Wednesday: 6:00 PM - 9:00 PM
Thursday: 8:00 AM - 9:00 PM
Friday: 7:30 AM - 5:00 PM
Saturday: 7:30 AM - 5:00 PM
Sunday: 7:30 AM - 11:00 AM

Biographies

Tetsuo Ashizawa, MD, PhD

Dr. Tetsuo Ashizawa is Professor and Chairman of the Department of Neurology at the University of Texas Medical Branch at Galveston. Dr. Ashizawa also holds the John Sealy Chair in Neurology, established in 2001 by an endowment from the Sealy & Smith Foundation.

Dr. Ashizawa received his medical degree from the Keio University School of Medicine in Tokyo in 1973. Dr. Ashizawa moved to Houston in 1975 to begin his residency at Baylor College of Medicine, completing fellowships in neuromuscular diseases and neurochemistry, and where he continued his clinical practice, teaching and research until coming to UTMB in July of 2002. As a researcher, he is well known within the neuroscience and human genetics communities for his molecular genetics research on neurodegenerative disorders, such as Huntington's disease, Parkinson's disease and cerebellar ataxias. He has published over 170 papers in leading scientific and clinical journals and books.

Under his leadership, the Department of Neurology has been focusing on Alzheimer's Disease research. With external funding of more than 15 million dollars acquired in the past 3 years, the Mitchell Center for Alzheimer's Disease Research has become one of the major translational research centers in US. Dr. Ashizawa's plan for the next 3 years is to expand the Center to one of the premier centers for neurodegenerative diseases in the world to develop new diagnosis and treatments.

Ronald J. Bartek

Co-founder and President of the Friedreich's Ataxia Research Alliance (FARA), a non-profit organization supporting medical research; 4-year member, NIH National Advisory Neurological Disorders and Stroke Council; former partner and president of a business and technology development, consulting, and government affairs firm; twenty years of federal executive branch and legislative branch service in defense, foreign policy and intelligence: including six years at the State Department's Bureau of Politico-Military Affairs, including a year as a negotiator on the U.S. Delegation to the Intermediate-Range Nuclear Forces (INF) Treaty talks in Geneva; six years as a CIA analyst of political-military aspects of the East-West balance, including a year as an Intelligence Community representative to the interagency groups charges with US arms control policy; former Director, American Friends of the Czech Republic; following graduation from the United States Military Academy at West Point, fours years as an Army Officer, serving as a company commander in Korea and an Infantry and Military Intelligence Officer in Vietnam; Master's degree in Russian Area Studies from Georgetown University.

Sanjay I. Bidichandi, MBBS, PhD

Dr. Bidichandani received a medical degree from the University of Poona (India; 1990), and won a competitive scholarship to obtain a Ph.D. degree in Medical Genetics from the University of Glasgow (Scotland, U.K.; 1994). He received a fellowship from the Muscular Dystrophy Association for his post-doctoral training at the Baylor College of Medicine. He also completed the Master Teacher Fellowship Program while at Baylor College of Medicine. A Scientist Development Grant from the American Heart Association facilitated his transition into an independent, tenure-track, faculty position at the University of Oklahoma Health Sciences Center in the department of Biochemistry & Molecular Biology in 2000. He is presently a tenured Associate Professor in the department. The research in his lab is focused on the mechanisms underlying triplet-repeat instability, which is funded by grants from the National Institutes of Health (NIH / NINDS), Muscular Dystrophy Association, and Friedreich Ataxia Research Alliance (FARA). He serves on the editorial board of Mutation Research, serves on study sections for NIH and DoD, and is a member of the Medical Advisory Committee of the MDA. He also directs the Medical Biochemistry and Molecular Genetics course for first-year students in the College of Medicine, and has received numerous teaching awards.

Alexis Brice, MD

Alexis Brice is a neurologist trained in medical genetics whose research focuses on the molecular basis of neurodegenerative disorders. He coordinates the Department of Genetics at the Pitié-Salpêtrière Hospital in Paris, France and the National Reference Centre for rare neurogenetic diseases. His research team has gained expertise in the field of spinocerebellar degenerations (cerebellar ataxias and spastic paraplegias), Parkinson's disease and fronto-temporal dementias. As a clinician he developed clinical trials and established genetic diagnosis for spinocerebellar degenerations at the hospital-based Genetic Department. As researcher, his INSERM group mapped

and contributed to the cloning of several genes responsible for spinocerebellar degenerations. They also approach the physiopathology of polyglutamine disorders using SCA7 as a model.

Margit Burmeister, PhD

Dr. Burmeister received her Diplom (equiv. to Master) in Biochemistry from the Free University Berlin in conjunction with her thesis work at the Weizman Institute, Rehovot, Israel. She received her Ph.D. in Biology from the Ruprecht Karls University of Heidelberg for her thesis work at the European Molecular Biology Laboratory (with Hans Lehrach) on the genetics of Duchenne muscular dystrophy. After postdoctoral work at the University of California in San Francisco (with Richard Myers and David Cox) on the map of chromosome 21 and Alzheimer's disease, she joined the faculty of the Mental Health Research Institute, now renamed Molecular & Behavioral Neuroscience Institute, the Department of Psychiatry and the Department of Human Genetics of the University of Michigan in 1991. She is also affiliated with the Neuroscience program and co-directs the Bioinformatics interdisciplinary graduate program.
Her laboratory investigates how genes influence behavior and risk for neurological and psychiatric disorders, including bipolar disorder, depression and alcoholism. She has identified several deafness and ataxia genes in humans and mice. More recently, she is combining genetic with functional studies to identify novel neurological genes, including ataxia genes.

Don Cleveland, PhD

Dr. Don Cleveland, Professor of Medicine and Neuroscience and Head of the Laboratory for ALS Research at the University of California at San Diego, has made discoveries into the causes and treatment of motor neuron disease. He showed that disorganization of neurofilaments causes selective failure of motor neurons in mice and humans. He then demonstrated that similar disease could also arise by a toxicity of mutant superoxide dismutase unrelated to its normal activity, thereby uncovering the mechanism underlying the major genetic form of Amyotrophic Lateral Sclerosis (ALS). Most importantly, he also showed that motor neuron death in inherited ALS is non-cell autonomous, requiring mutant damage to both motor neurons and the neighboring supporting cells. These findings have redirected efforts at stem cell replacement therapies in ALS. Cleveland's team has also developed a gene silencing therapy for ALS that will enter clinical trial in 2008.

Thomas Crawford, MD

Johns Hopkins Hospital

John Day, MD, PhD

John W. Day, M.D., Ph.D., is Professor of Neurology and Pediatrics at the University of Minnesota. He received his M.D. from the University of Minnesota, Ph.D. in Neuroscience from the Albert Einstein College of Medicine, and Neurology training at the University of California, San Francisco before retuning to Minnesota, where he established and continues to direct the Paul and Sheila Wellstone Muscular Dystrophy Center and where he maintains active ataxia and neuromuscular research programs. He also cares for pediatric ataxia and neuro-muscular patients at the Gillette Pediatric Specialty Heathcare Clinic in St. Paul. Dr. Day helped identify and characterize SCA5 and SCA8, as well as other forms of ataxia, and serves as Medical-Research Liaison for the National Ataxia Foundation.

William Dobyns, MD, PhD

Dr. Dobyns is Professor of Human Genetics, Neurology and Pediatrics at The University of Chicago. He works in the field of Developmental Neurogenetics, focusing on the nature and genetic basis of human neurodevelopmental disorders including cerebellar and neocortical malformations, as well as infantile epilepsies and autism.

Nicholas Dupre, MD, MSc, MPH, FCRP©

Dr. Nicolas Dupré did his medical training at McGill University and his neurological training at the Montreal Neurological Institute. He did his post-doctoral studies at both McGill and Harvard to subspecialize in

Biographies

the field of neurogenetics. He has completed a masters degree in neurobiology, and more recently in epidemiology. He is presently assistant professor at the Faculty of Medicine of Laval University and associate member of the Centre d'étude en neuromique de l'université de Montréal (CENUM). He is an expert in the characterization of inherited neurological diseases in founder populations, particularly the French-Canadian population.

Alexandra Durr, MD, PhD

Alexandra Durr works as a consultant in Neurogenetics at the Salpêtrière Hospital in Paris 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 PhD 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 to reinforce the links of laboratories and clinical centers. She is a member of the executive committee of the recently founded Ataxia Study Group.

Kenneth H. Fischbeck, MD, Senior Investigator

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 newly created Neurogenetics Branch. Dr. Fischbeck received the Cotzias Award from the American Academy of Neurology 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, particularly the polyglutamine expansion neurodegenerative diseases.

Christopher Gomez, MD, PhD

Dr. Gomezreceived his medical degree from the Pritzker School of Medicine in Chicago, Illinois, and his PhD 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 and 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. Dr. Gomez also serves on the Medical Research Advisory Board for the National Ataxia Foundation, and has received funding from the NAF for his important ataxia research efforts.

Joel M. Gottesfeld, PhD

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, and was promoted to full professor of Molecular Biology in 1994.
Research in our laboratory concerns the development of small molecules to regulate gene expression. We are using both DNA binding molecules and enzyme inhibitors to regulate the expression of clinically significant genes. In the first approach, small molecules called polyamides can be programmed by chemical synthesis to recognize a wide range of DNA sequences with affinities and specificities that are comparable to human regulatory proteins. We have shown that polyamides bind their cognate DNA sequences in the context of cellular chromatin, both in simple model systems and in the nucleus of cultured

cells. We have used these molecules as either activators or repressors of gene expression. More recently, our efforts have focused on histone deacetylase inhibitors as potential therapeutics for neurodegenerative diseases, such as Friedreich's ataxia, spinal muscular atrophy, Huntington's disease, and myotonic dystrophy.

Mary E. Hatten, PhD

Dr. Hatten was graduated from Hollins College in Roanoke, Virginia in 1971 with a bachelor's degree in chemistry. She received a Ph.D. in Biochemical Sciences from Princeton University in 1975, followed by postdoctoral research in Neuroscience with Richard Sidman, at Harvard Medical School. She was appointed Assistant Professor of Pharmacology at NYU Medical School in 1978 and promoted to Associate Professor in 1984. In 1987, she moved to the College of Physicians and Surgeons where she was Professor of Pathology in the Neuroscience Program. Dr. Hatten was appointed Professor and Head of the Laboratory of Developmental Neurobiology in 1992, (the first female HOL). In 2,000 she was named Frederick P. Rose Professor at the Rockefeller.

Dr. Hatten was awarded a Westinghouse National Science Talent Search Award in 1967, the Irma T. Hirschl Career Scientist Award (1980), the Pew Neuroscience Award in 1988, the McKnight Neuroscience Development Award (1991), a Javitts Neuroscience Investigator Award (1991), and the Faculty Award for Women Scientists and Engineers from the National Science Foundation (1991), Weill Award, American Assoc. of Neuropathology (1996), Ph.D. Honoris causa Hollins College (1998), Treasurer, American Society for Neuroscience (1998), Wersma Professorship, Caltech (2005), Fellow of the American Association for the Advancement of Science (2002), Chair Section on Neuroscience, AAAS, 2007.

Dr. Hatten currently serves on the Scientific Advisory Board of the Max Planck Institute, Gottingen; the NY State Spinal Cord Injury Research Program Advisory Board (Founding Member); the Scientific Advisory Committee of the March of Dimes Birth Defects Foundation; the Board of Scientific Overseers of the Jackson Laboratory; the PEW Scholars National Advisory Committee; and the John Merck Scholars Board.

Nathaniel Heintz, PhD

The Rockefeller University

Grazia Isaya, MD, PhD

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 (PhD) 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 at Mayo Clinic, College of Medicine, and directs a research laboratory focusing on mitochondrial iron balance and the biochemical basis of Friedreich ataxia.

Joanna Jen, MD, PhD

Dr. Jen is an Associate Professor at UCLA Neurology. She obtained her medical and graduate degrees from Yale University, completed neurology residency at UCLA, and pursued fellowship training in neurology, physiology, and human genetics, also at UCLA. Her clinical interest in balance is complemented by research performed in her laboratory on the genetic, physiological, and functional anatomical bases of a broad range of disorders including episodic ataxia and other ataxia syndromes that affect balance, coordination, and eye movement control.

Thomas Klockgether, MD

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 Tubingen. Dr. Klockgether has been the Coordinator of the German Collaborative Research Group *Molecular Pathogenesis of SCA 3* since 2001 and is also the Co-

ordinator 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.

Michel Koenig, MD

Dr. Koenig currently is an Associate Professor in the Department of Molecular Pathology at the Institute of Genetics and Molecular Cell Biology at the University of Louis Pasteur in Strasbourg, France where he received his MD in 1990. From 1981 – 1983, Dr. Koenig obtained his Maitrise and DEA de Biologie Moleculaire (equivalent to B.Sc. and M.Sc.) in Faculty for Life Sciences at the Louis Pasteur in Strasbourg, France. In 1986, Dr. Koenig was a graduate student in Biology (Molecular Genetics) and his Thesis project was "Analysis of molecular probes of the human X and Y chromosomes. He received his PhD in 1986. Dr. Koenig has been instrumental in the research of Freidreich's Ataxia, Ataxia/Oculomotor Apraxia 1 and 2, and has worked with mapping and identification of cells.

Albert La Spada, MD, PhD

Albert La Spada graduated *Summa Cum Laude* from the University of Pennsylvania with a B.A. in Biology in 1986. As a recipient of a Medical Scientist Training program award, he then 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 (Kennedy's disease). While a graduate student in the laboratory of Dr. Kenneth Fischbeck, La Spada identified the cause of X-linked spinal and bulbar muscular atrophy as an expansion of a trinucleotide repeat in the androgen receptor gene. After completing his M.D. - Ph.D. training in 1993, Dr. La Spada became a Laboratory Medicine resident at the Univ. of Washington Medical Center and then a Clinical Genetics fellow in the Division of Medical Genetics. He also 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 is currently an Associate Professor.

Dr. La Spada has been the recipient of grants and awards from the National Institutes of Health (N.I.H.), Howard Hughes Medical Institute, Muscular Dystrophy Association, and American Federation for Aging Research. He was a recipient of a N.I.H. Research Award for Clinical Trainees. Among his other funding awards is the prestigious Paul Beeson Physician Faculty Scholar Aging Research Award. Dr. La Spada sits on a variety of editorial boards and grant review committees. Last year, he was appointed Director of the newly created Center for Neurogenetics & Neurotherapeutics at the Univ. of Washington.

Martin F. Lavin PhD (CIC)

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. Over the past few years he has extended his research to include several other human genetic disorders characterized by ataxia and oculomotor apraxia. The rationale for this is that the overlapping phenotype might arise as a consequence of a defect in the recognition and/or repair of damage to DNA. This has proved to be the case for AOA1 (aprataxin) and AOA2 (senataxin) where he has described defective responses to oxidative damage. In the last several years Martin Lavin has extended his research to investigate methods for the early detection of prostate cancer and has initiated a major research program in association with QRx Pharma to identify novel therapeutics in the venom of Australian snakes.

Martin Lavin obtained a BSc (Hons) from the National University of Ireland in Dublin before proceeding to a

Biographies

PhD in Trinity College Dublin. The main focus of his research was on a recBC nucleus in *mycobacterium smegmatis*. This was the initiation of a lifelong interest in nucleic acids and proteins that recognise damage in DNA. After a post-doctoral stint at Syntex Research, Palo Alto working on endonucleases that recognise alkylation damage in DNA he moved into a second post-doctoral position in the Biochemistry Department, University of Queensland, Brisbane, Australia. The emphasis of his work was again 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. As described above this provided a very fruitful approach to understanding the recognition of double strand breaks in DNA and how these are signaled to cell cycle checkpoints. He was appointed lecturer in Bicohemistry, 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.

David R. Lynch, MD

Dr. Lynch is an Associate Professor of Neurology and Pediatrics at the University of Pennsylvania. He was educated at Yale College and obtained his MD and PhD degrees at the John Hopkins University School of Medicine. His PhD was performed in the Laboratory of Dr. Solomon H Synder on neuropeptide processing. Dr. Lynch was trained in Neurology at the University of Pennsylvania. His main research interests are in NMDA receptor structure and function and in Friedreich ataxia.

Grisel J. Lopez, MD

Grisel J. Lopez is a Staff Clinician at the National Institute of Neurological Disorders and Stroke (NINDS) and is in charge of the NIH Parkinson Disease Clinic. Before joining NINDS, she was a Staff Clinician at the National Human Genome Research Institute from 2003-2006 working on disease mechanisms and genetic analysis of familial Parkinson Disease and other Movement Disorders. She is a neurologist and is currently Principal Investigator in several clinical protocols relating to the genetics of Parkinson Disease and co-investigator in other clinical protocols relating to the understanding of monogenetic diseases and their association to complex trait disorders. She obtained her medical degree, residency training and post-doctoral training at the University of Kansas Medical Center. Prior to her work at the NIH, she was Assistant Professor at the University of Kansas Medical Center.

Earl McLaughlin

Earl McLaughlin is the San Diego Ataxia Support Group leader, and has served on the NAF Board of Directors since 1988. He was diagnosed with Friedreich's Ataxia in 1981 when he was 23, while serving in the Air Force. Earl graduated from San Diego State University, and recently retired as an accountant. He lives in El Cajon, California with his service dog, "Pip".

Kathleen Millen, PhD

Kathy Millen is a developmental biologist and geneticist who has studied the genes and processes that shape the developing cerebellum for most of her scientific career. As a graduate student in the laboratory of Dr. Alexandra Joyner at the University of Toronto, she demonstrated that the mouse Engrailed-2 gene is required for normal patterning of cerebellar foliation and afferent targeting. These studies resulted in a lifelong passion for cerebellar research. As a post-doctoral fellow in the laboratory Dr. Mary E. Hatten at Rockefeller University, Kathy studied mouse mutants with spontaneous congenital cerebellar malformations using the newly available tools of genomics to positionally clone the underlying causative genes, working on two classical mouse mutants, *weaver* and *dreher*. As an Assistant Professor in the Department of Human Genetics at the University of Chicago and now an Associate Professor, she continues to work on various mouse cerebellar developmental mutants to unravel the fundamental processes that drive cerebellar development. She is particularly interested in the molecular mechanisms of cerebellar cell fate specification during early embryogenesis. She has also extended her analysis of cerebellar malformations to human cerebellar malformations. Working with her colleague, Dr. William Dobyns, together they have estab-

lished the world's largest DNA repository and clinical database for human cerebellar malformations and they are working to categorize human cerebellar malformations and identify the causative genes. Using the power and strengths of both human and mouse genetics, this collaboration has lead to the identification of the first causative loci and genes for Dandy-Walker malformation, the most common congenital malformation of the human cerebellum.

Dr. Russell Margolis, MD

Russell L. Margolis received his undergraduate degree from Princeton University and M.D. from Johns Hopkins University. After a residency in psychiatry at Johns Hopkins, he did a neurogenetics fellowship at the NIMH with De-Maw Chuag and Robert Post, and then joined the faculty at Johns Hopkins, where he is now Associate Professor of Psychiatry and Neurology and Director of the Laboratory of Genetic Neurobiology. Work in his group focuses on three main themes: 1) the genetic etiology and pathogenesis of late-onset movement disorders, 2) the neurogenetics of psychiatric disorders, including schizophrenia and autism, and 3) the neuropsychiatry of movement disorders.

Brad Margus

Brad Margus is a co-founder of Perlegen Sciences, a California-based biotechnology company that identifies and validates clinically relevant genetic variations that predict patient response to treatments and then commercializes high value diagnostic tests to make medicine safer and more effective. Mr. Margus was appointed to the position of Executive Vice Chairman of the company in May 2007 following a seven-year tenure as Chief Executive Officer. After obtaining his M.B.A. from Harvard in 1986, Mr. Margus spent 14 years running a private international agribusiness company. In 1993, after learning that two of his children had the rare disease ataxia-telangiectasia, Mr. Margus formed the A-T Children's Project, a non-profit research organization that has raised over $26 million in donations, recruited first-rate scientists, run scientific conferences, orchestrated numerous biomedical research projects, established a clinical center, sponsored clinical trials, and created tissue banks. In 2000, he entered the biotechnology industry and co-founded Perlegen. Mr. Margus has been a member of various Institutional Review Boards, NIH advisory committees, and corporate boards. He has served on the Advisory Council to the National Institute of Neurological Disorders and Stroke, on the Secretary of Health and Human Sciences' Advisory Committee on Genetics, Health and Society, and on the Board of Genetic Alliance, an umbrella organization representing hundreds of genetic disease organizations. He continues to serve as Chairman and volunteer president of the A-T Children's Project, on the Board of Children's Neurobiological Solutions, an organization aimed at applying brain repair and regeneration to pediatric neurological disorders, and on the Stem Cell Research Oversight Committee at Stanford University School of Medicine.

Paul J. Muchowski, PhD.

Paul Muchowski was born in Montreal, Canada. He obtained his Ph.D in Biological Structure at the University of Washington in Seattle, where his thesis focused on protein folding and molecular chaperones. Subsequently he moved to Martinsried, Germany where he was a post-doctoral fellow with Ulrich Hartl at the Max Planck Institute for Biochemistry. In the Hartl lab he began to characterize the role of molecular chaperones in Huntington's disease, and developed a yeast model to study polyglutamine aggregation. He next returned to Seattle for a second post-doctoral stint with Stanley Fields in the Department of Genome Sciences at the University of Washington. His research in the Fields' lab was centered on understanding basic cellular mechanisms that regulate polyglutamine aggregation using yeast genetics. In 2001 he was appointed as an Assistant Professor in the Department of Pharmacology, also at the University of Washington. In 2005 he moved his laboratory to the Gladstone Institute of Neurological Disease, where he was appointed as an Associate Investigator. He also holds an appointment as an Associate Professor in the departments of Biochemistry and Biophysics, and Neurology, at the University of California, San Francisco. Dr. Muchowski's research program is funded by the National Institute of Neurological Disorders and Stroke, the Hereditary Disease Foundation and the HighQ Foundation.

Kaisu Nikali, MD, PhD

Dr. Kaisu Nikali is a human molecular geneticist with main focus on neurodegenerative and mitochondrial diseases, interactions in between mitochondrial and nuclear genomes, and disease gene identification in isolated populations. She started her research career in 1993 in the Department of Human Molecular Genetics (currently that of Molecular Medicine), National Public Health Institute, Helsinki, Finland, being also a third-year medical student in the University of Helsinki at that time. She was introduced to a collection of families, in which a previously uncharacterised recessive ataxia-syndrome segregated, and given a goal to identify the defective gene on the basis of its location in the human genome. Having excluded known ataxia loci, she utilised the assumption of a single founder mutation in the genetic isolate of Finland, and mapped the new ataxia locus by homozygosity mapping/ haplotype sharing –strategy to chromosome 10q24, utilising samples of only four affected individuals in the initial random screen of the human genome. She received her PhD in 1998, with academic dissertation titled 'Molecular genetics of infantile-onset spinocerebellar ataxia', and her MD in 1999. Her further research led to the identification of the founder IOSCA-mutation in a gene coding for mitochondrial replicative helicase Twinkle and splice-variant Twinky, extending the spectrum of hereditary ataxias due to dysfunction of mitochondrial proteins. Twinkle being required for normal bioprocessing of mtDNA, Dr Nikali's discovery of recessive Twinkle-mutations in IOSCA, where both mtDNA and mitochondrial enzyme function stay intact, indicated Twinkle and Twinky have still uncharacterised functions, most important for the specific subpopulations of neural cells degenerating in IOSCA. All this work of hers yielded a series of peer-reviewed original publications, reflecting the logical steps of identifying a novel disease gene by positional cloning/candidate –approach in a genetically isolated population. Having identified the IOSCA-gene and causative mutations, she moved to the MRC Developmental Neurobiology Centre, King's College, London, to study the Twinkle-gene homologue in *Drosophila melanogaster,* as a Post-doctoral Travelling Fellow of first the Academy of Finland and thereafter that of the Wellcome Trust. Currently, she is working as a Post-doctoral Research Fellow in the Galton Laboratory, University College London, within a group with a main interest in the analysis of human genetic variation from an evolutionary perspective and in terms of disease susceptibility in the isolated and admixed populations of South America.

Harry Orr, PhD

Dr. Orr is a full professor for 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. at Washington University, and completed a graduate fellowship at Harvard. He is the Director of the Institute for Human Genetics, and holds the Tulloch Chair in Genetics. Dr. Orr is known as the researcher who, along with Dr. Huda Zoghbi, found the first gene for ataxia, now known as SCA-1. His research is focused on molecular neurogenetics, and he is a published author of more than 120 articles, many on the genetics of ataxia. Dr. Orr has been appointed as NAF's Research Director on June 14, 2006.

Gülin Öz, PhD

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 BS degrees in Physics and Chemistry and obtained her PhD in Biochemistry at the University of Minnesota. She continued with postdoctoral training at the Center for Magnetic Resonance Research at the University of Minnesota where she joined the faculty as assistant professor in 2006. She also serves as the director of the Magnetic Resonance Core of the General Clinical Research Center. 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, MD

Dr. Pandolfo received his M.D. at the University of Milan, Italy in 1980 and his post doctorate in molecular genetics from 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. Since 1996, he has served as an Adjunct Professor at McGill University's Department of Neurology and Neurosurgery in Montreal, Canada.

He also serves as Research Associate Professor in the Department of Medicine. 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.

Henry Paulson, MD, PhD

Dr. Paulson is Associate Professor of Neurology at the University of Iowa Carver College of Medicine in Iowa City, Iowa. He received his MD and PhD degrees in cell biology from Yale University School in 1990. He then completed a Neurology residency at the University of Pennsylvania, followed by a Neurogenetics and Movement Disorders postdoctoral fellowship, also at Penn. In 1997, he joined the Neurology faculty at the University of Iowa, where he was promoted to Associate Professor in 2003.

Dr. Paulson's research and clinical interests concern the causes and treatment of neurodegenerative diseases including the spinocerebellar ataxias. A particular interest is Machado-Joseph disease (also known as SCA3) and related polyglutamine disorders.

Dr. Paulson co-directs the Huntington Disease Center of Excellence at Iowa, and serves on the scientific advisory boards of numerous disease-related national organizations. Among his awards, he is an Ellison Medical Foundation New Scholar in Aging, a semifinalist for the W.M. Keck Foundation Young Scholars in Medical Research, and a recipient of the Paul Beeson Physician Faculty Scholar in Aging Award from the American Federation for Aging Research.

Christopher Pearson, PhD

Dr. Christopher E. Pearson is a Senior Scientist in the Program of Genetics & Genome Biology at The Hospital for Sick Children (Toronto, Canada) and an Associate Professor at the University of Toronto. Dr. Pearson's research program has focused upon elucidating the mechanism(s) of disease-associated repeat instability since 1994. The long-term goal of his lab is to modulate repeat instability with the hope of reducing or eliminating disease severity. Having a background in primate DNA replication, DNA repair and DNA mutagenesis he has made insights into understanding the processes and factors that contribute to trinucleotide repeat instability occurring in spinocerebellar ataxia types 1, 7 and 10, myotonic dystrophy type 1, Huntington's disease and fragile x mental retardation. Dr. Pearson was the first to identify and characterize slipped-strand DNA structures; thought to be mutagenic intermediates of trinucleotide repeat instability. His lab uses various models systems including molecular, cellular, patient tissues, and transgenic mice. Dr. Pearson's current research is focused upon mechanisms of chromosome replication, aberrant DNA repair and epigenetic modification.

Susan Perlman, MD

Susan L. Perlman, M.D., is is a graduate of Cornell University and the State University of NY at Stony Brook School of Medicine. She did her internship in medicine, residency in neurology, and a 2-year fellowship in Friedreich's ataxia at UCLA Medical Center. She has been head of the Ataxia Center at UCLA since 1983.

John Porter, PhD

Disorders and Stroke (NINDS). His program focuses on inherited and acquired peripheral neuropathies, normal biology of and disorders affecting the neuromuscular junction, and neuromuscular disor-

ders affecting skeletal muscle proper, including myotonic dystrophy and Duchenne/Becker, facioscapulohumeral, congenital, limb girdle, oculopharyngeal, and Emery-Dreifus muscular dystrophies. Prior to joining the NIH, Dr. Porter was Professor of Neurology at Case Western Reserve University. Dr. Porter received his undergraduate degree in Biology from the College of William and Mary and his Ph.D. in Anatomy from Medical College of Virginia and completed postdoctoral training in systems neurophysiology at the University of Alabama at Birmingham. His research focused upon extraocular muscle biology in health and disease, including the mechanisms responsible for its novel responses to a variety of neuromuscular disorders. He currently is a Program Director in the Neurogenetics Cluster on a variety of neuromuscular diseases and in the Technology Development Group, where he facilitates large preclinical development projects in the NINDS Translational Research Program.

Helene Puccio, MD, PhD

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's 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 Friedreich's ataxia group at the IGBMC. Dr. Puccio laboratory has been mostly focused on the development of mouse models for Friedreich's ataxia to unravel the physiopathology of the disease and test pharmacological compounds in preclinical therapeutic trials. In parallel, her laboratory has developped cellular models deficient for frataxin for large scale drug screening. In 2005, she was the recipient of "Prix 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.

Stefan Pulst, MD

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 related 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.

Laura Ranum, PhD

Dr. Ranum received her PhD from the University of Minnesota in 1989 and did her postdoctoral work with 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. 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 generating mouse models to better understand these diseases and to improve mapping 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.

Andrea Richter, PhD

Dr. Richter is an Associate Professor of Research, Department of Pediatrics, Adjunct professor, Department of Biochemistry, Université de Montréal, Montréal, Québec, Canadaé. Her research group focuses on the study of genes responsible for monogenic diseases frequent in different Canadian populations due to founder effects: autosomal recessive spastic ataxia of Charlevoix-Saguenay (ARSACS) and North American Indian childhood cirrhosis (NAIC) and polygenic congenital malformations such as primary vesicoureteral reflux (pVUR), a common urological disease in children and a frequent heart malformation (left ventricular outflow tract obstruction, LVOTO). Following the cloning of the ARSACS and NAIC genes and the identification of the disease mutations, we are now embarking on the creation cellular and animal models for these diseases and searching for mutations in other populations. For the study of pVUR the collection of the disease cohort was followed by targeted candidate gene search that recently revealed that rare allele of a coding SNP in the receptor tyrosine kinase (*RET)* has highly significant association with disease in Quebec families. The study of genes involved in LVOTO is in genome scan phase. Using the Affymetrix 6.0 SNP genotyping platform, a complete genome scan is underway for 70 multiplex families (over 550 samples). The results of the 1st analyses are expected by the summer 2008.

The current projects of her group are supported by the Canadian Institutes of Health research, while previously we benefited from funding from the National Ataxia Foundation, MRC, MDA-C, and Kidney Foundation of Canada.

Olaf Riess, MD, PhD

Dr. Riess is the head of the Institute of Human Genetics and full professor of Medical Genetics at the University of Tubingen, Germany. The research topic of Dr. Riess is on neurodegenerative diseases. His lab has generated several transgenic mouse and rat models of PD, SCA, HD, and dystonia. These models have been characterized comprehensively and are highly suitable for preclinical treatment studies. Also, these models have been used for deciphering transcriptome pathways. His interest is furthermore to decipher the pathomechanism of these diseases by cell culture models and molecular methods. In humans, his group is searching for mutations causing the respective diseases, and in defining transcriptome biomarkers.

Jeremy D. Schmahmann, MD

Dr Schmahmann received his medical degree at the University of Cape Town Medical School in South Africa. After completing his residency in the Neurological Unit of the Boston City Hospital and his fellowship in the Department of Anatomy and Neurobiology at Boston University School of Medicine, he joined the faculty of the Massachusetts General Hospital in 1989. At MGH he is a Director of the Ataxia Unit, a member of the Cognitive/Behavioral Neurology Unit and the Geriatric Neurobehavior Clinic, and Neurology Clerkship Director. Dr. Schmahmann is an Associate Professor of Neurology at Harvard Medical School, and a Scholar in the Academy at Harvard. In 2000 he was the recipient of the Norman Geschwind Prize for research in the Behavioral Neurology Society, and he has been cited in *The Best Doctors in America* since 1998. He is the medical advisor to the New England Ataxia Support Group, and a member of the Cooperative Ataxia Group of the National Ataxia Foundation.

Dr. Schmahmann's research interests focus on the role of the cerebellum in cognition and emotion. His publications on anatomical and behavioral studies in the monkey investigate the links between the cerebellum and the brain areas critical for complex behaviors. He described the cerebellar cognitive affective syndrome and developed the dysmetria of thought hypothesis. In a series of collaborative projects in his Laboratory of Cerebellar Neurobiology. Dr. Schmahmann is studying cerebellar diseases, and cognitive effects of focal cerebellar lesions in monkeys. Dr. Schmahmann has over 140 scientific publications and 3 books — The Cerebellum and Cognition (Academic Press, 1997), MRI Atlas of the Human Cerebellum (Schmahmann, Doyon, Toga, Petides, Evans, Academic Press, 2000), and Fiber Pathways of the Brain (Schmahmann and Pandya, New York, Oxford University Press, 2006).

He has continued to structure function correlations of the caudate nucleus, thalamus, pons, and cerebral white matter tracts, and is presently funded by the National Institutes of Mental Health to study the cognitive effects of cerebellar lesions in humans following cerebellar stroke.

Yosef Shiloh, PhD

Dr. Yosef Shiloh is Myers Professor of Cancer Genetics in the Department of Human Molecular Genetics and Biochemistry at the Tel Aviv University Sackler School of Medicine. He earned his Ph.D. in Human Genetics at The Hebrew University of Jerusalem, and trained further at Harvard Medical School, the University of Michigan, and the U.S. National Institutes of Health. Most of his work concerns the DNA damage response in mammalian cells. The work stems primarily from his interest in the human genetic disorder, ataxia-telangiectasia. Dr. Shiloh began his quest to understand this disease while working on his doctoral dissertation in the late seventies. After numerous studies on the cellular phenotype of A-T, he turned to positional cloning of the elusive A-T gene, a project that culminated in 1995 in the identification of the *ATM* gene. The Shiloh lab is dedicated to characterizing ATM's function and mode of action. The lab is currently studying the various steps of the DNA damage response and identifying new ATM effectors and genes that are regulated in an ATM-dependent manner. Computational tools are being developed to navigate through the complex DNA damage response network. Special effort is made to understand the role and mode of action of ATM and the DNA damage response in the nervous system, where the lab has recently obtained new insights.

Andrew Singleton, PhD

Dr Singleton received his B.Sc. from the University of Sunderland, UK and his Ph.D. from the University of Newcastle upon tyne, UK. Dr Singleton's research initially focused on genetic determinants of dementia, in particular Alzheimer's disease and dementia with Lewy bodies. His postdoctoral studies were spent at the Mayo Clinic in Jacksonville Florida. Dr Singleton moved to the National Institute on Aging at NIH Bethesda, MD in 2001 and became a principal investigator leading the Molecular Genetics Unit in 2002 and a tenured senior investigator in 2007. Dr. Singleton's laboratory works on the genetic basis of neurological disorders including Parkinson's disease. This research is performed using a variety of methodologies, including family based linkage and positional cloning in addition to genome-wide association. The goal of Dr Singleton's research is to identify genetic variability that causes or contributes to Parkinson's disease in order to facilitate understanding of the molecular processes underlying disease.

Ira Shoulson, MD

Ira Shoulson, MD is the Louis C. Lasagna Professor of Experimental Therapeutics and Professor of Neurology, Pharacology and Medicine at the University of Rochester School of Medicine in Rochester, New York. He received his MD degree (1971) and postdoctoral training in medicine (1971-73) and Neurology (1975-77) at the University of Rochester and in experimental therapeutics at the National Institutes of Health (1973-75). Dr. Shoulson founded the Parkinson Study Group (1985) and the Huntington Study Group (1994), international academic consortia devoted to research and development of treatments for Parkinson's disease, Huntington's disease and related Neurodegenerative and neurologic disorders. He has served as principle investigator of the National Institutes of Health-sponsored trials "deprenyl and Tocopherol Antioxidative Therapy of Parkinsonism" (DATATOP), the "Perspective Huntington At Risk Observational Study" (PHAROS), and more than 25 other multicenter controlled trials. He is the Director of the Experimental Therapeutics Program at the University of Rochester Department of Neurology, the chair of the executive committee of the Huntington Study Group, a consultant for the Food and Drug Administration, Former member of the National Institute of Neurological Disorders and Stroke Council, associate editor of Archives of Neurology, and past-president of the American Society for Experimental NeuroTherapeutics (ASENT). He has authored more than 230 scientific reports.

S. H. Subramony, MD

Dr. S. H. Subramony is currently Professor of Neurology at University of Texas Medical Branch, Galveston, Texas where he directs the EMG laboratory and cares for patients with ataxia. He was previ-

ously Guyton Professor of Neurology at the University of Mississippi Medical Center where he ran an ataxia clinic and directed the Neuromuscular Division. His training was in Neurology and EMG at the Cleveland Clinic Foundation, Cleveland, Ohio. He is active in clinical research in ataxia.

F. Taroni, MD

Dr. Taroni graduated in 1981 at the University of Milan School of Medicine. After his board in Neurology, he carried out postdoctoral studies in the Department of Human Genetics at Yale University. He is now leading the Unit of Genetics of Neurodegenerative and Metabolic Disease at the Department of Experimental Research and Diagnostics, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milan, Italy. He is Professor of Neurogenetics at the Postgraduate School of Medical Genetics, University of Milan School of Medicine, Italy. His research interests are focussed on the molecular bases of neurodegenerative diseases including trinucleotide expansion disorders, hereditary spastic paraplegias, and hereditary ataxias.

Shoji Tsuji, MD, PhD

Dr. Shoji Tsuji is Professor and Chairman at Department of Neurology, University of Tokyo. Applying molecular genetic approches, he has identified genes for Dentatorubural-pallidoluysian atrophy (DRPLA), spinocerebellar ataxia type 2 (SCA2), spinocerebellar ataxia type 17 (SCA17), and early-onset ataxia associated with ocular motor apraxia and hypoabuminemia (EAOH), a rare form of autosomal recessive ataxia. He has also conducted nation-wide surveys in Japan to establish natural histories of patients with spinocerebellar ataxias to provide basis for future clinical trials. Currently his research focus is on development of therapeutic approches for polyglutamine diseases, and elucidation of molecular pathogenesis of sporadic ataxias.

E.M. Valente, MD, PhD

She obtained her Medical Degree at the Catholic University School of Medicine in 1994. She has subsequently trained in Neurology at the same University, attending the services of neurophysiology and movement disorders, and received her certification in Neurology in 1999. She did a PhD in Neurogenetics at the Institute of Neurology, University College of London and obtained her PhD degree in 2003. She has been Head of the Neurogenetics Group at the CSS-Mendel Institute in Rome since 2002 and Associate Professor of Medical Genetics at the University of Messina since 2006. She teaches Human and Medical Genetics to students of Medicine, Pharmacy and Chemistry and to residents in Medical Genetics and Neurology.

Her two major research fields have focused on the clinical and molecular genetic aspects of movement disorders (mainly Parkinson's Disease and dystonic syndromes) and of congenital cerebellar malformations. In these fields, she has attained important results, including the mapping of novel loci and the identification of novel disease genes (PARK6/PINK1; JBTS5/CEP290). Her current research activity focuses on genetic mapping of novel loci, positional cloning of new genes, mutation analysis, genotype-phenotype correlations and functional characterization of the PINK1 gene product. Dr Valente liaises with clinicians from different Institutions in Italy and abroad.

She is author of 77 scientific publications on international peer-reviewed journals (IF: 410.71) and 6 book chapters, is member of genetic and neurological scientific societies in Italy and abroad and is Principal Investigator or co-PI of several national and international research grants focused on Parkinson's disease and congenital ataxias.

George (Chip) Wilmot, MD, PhD

George “Chip” 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.

Robert Wilson, MD, PhD

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 interest is in Friedreich's ataxia. In addition to research, he does clinical sign-out 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 genetics to residents and fellows, and topics related to neurodegenerative disease to neuroscience graduate students.

Nicholas Wood, PhD, FRCP, FMedSci

Dr. Wood qualified in medicine from Birmingham University in 1986 and undertook doctoral research at the University of Cambridge and was awarded a PhD in 1994. He has been a Senior Lecturer, Reader and Professor at the Institute of Neurology, University of London since 1995. He is currently Head of the Department of Molecular Neuroscience and runs the Neurogenetic Laboratory at the National Hospital and the Institute of Neurology. In 2004 he was made a Fellow of the Academy of Medical Sciences. His primary research interest is the genetics of neurological disease with an emphasis on the genetics of Parkinson's disease and the ataxias. He has published over 230 peer reviewed articles and is on the board of several international neuroscience journals.

Ullrich Wüllner, Prof, Dr, Med

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Huda Zoghbi, MD

Dr. Zoghbi is Professor of Pediatrics, Neurology, Neuroscience, and Molecular and Human Genetics at Baylor College of Medicine, and an Investigator with the Howard Hughes Medical Institute. She received her B.S. in Biology from the American University of Beirut and completed her first year of medical school at the American University of Beirut. Due to the war in Lebanon, Dr. Zoghbi transferred and received her M.D. in 1979 from Meharry Medical College in Nashville, Tennessee, and shortly after joined Baylor College of Medicine for training in Pediatrics, Neurology, and Molecular and Human Genetics. Zoghbi's interests range from neurodevelopment to neurodegeneration. Zoghbi's work in neurodevelopment led to the discovery of the gene *Math1*, which governs the development of several components of the proprioceptive, hearing, and vestibular pathways. Zoghbi's group also discovered that mutations in *MECP2* cause the neurodevelopmental disorder Rett syndrome. She and her collaborator, Dr. Harry Orr, have identified the *SCA1* gene and studied the pathogenesis of polyglutamine neurodegenerative disorders.

Dr. Zoghbi serves on NIH panels and several scientific advisory and editorial boards. She is a member of the Institute of Medicine and of the National Academy of Sciences.