Welcome to the First Ataxia Investigator's Meeting

Sponsored by the National Ataxia Foundation

In conjunction with funding support from:

Athena Diagnostics, Inc.

Friedreich's Ataxia Research Alliance (FARA)

Outback Steakhouse®

March 2 - 4, 2005 ✹ Embassy Suites/USF Near Busch Gardens ✹ Tampa, Florida

Biographies

George (Chip) Wilmot, MD, PhD
Dr. Wilmot received his medical and graduate training in Neuroscience at the University of Michigan, and completed a neurology residency at Emory University School of Medicine in 1997. He is currently Assistant Professor at Emory and Director of the Emory Ataxia Center. Dr Wilmot has been one of the leaders and the principle organizer of the Cooperative Ataxia Group, a group of ataxia investigators interested in developing clinical trials for 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.

Dr. Wilson 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 signs out cases in the Molecular Pathology Laboratory of the Hospital of the University of Pennsylvania. He also teaches general pathology to medical students and molecular pathology to residents and fellows.

Huda Zoghbi, PhD
Dr. Zoghbi's interest is in using the tools of modern genetics to understand the proper development of the brain as well as what goes awry in specific neurodegenerative conditions. She has published seminal work regarding the molecular basis of late-onset neurodegenerative diseases. She also discovered that mutations in the methyl-CpG-binding-protein (*MECP2*) cause the neurodevelopmental disorder Rett syndrome as well as a broad spectrum of disorders ranging from mild mental retardation to autism.

Huda Zoghbi is Professor of Pediatrics, Neurology, Neuroscience, and Molecular and Human Genetics at Baylor College of Medicine and serves as an Investigator with the Howard Hughes Medical Institute. Dr. Zoghbi received her medical degree from Meharry Medical College in Nashville, Tennessee. Her postgraduate training included residencies in Pediatrics and Pediatric Neurology, and a research fellowship in Molecular and Human Genetics at Baylor College of Medicine.

Dr. Zoghbi is a member of several professional organizations and serves on the editorial boards of a number of prominent journals. Among Dr. Zoghbi's honors are the Sidney Carter Award from the American Academy of Neurology, the Derek Denny-Brown Neurological Scholar Award from the American Neurological Association, E. Mead Johnson Award from Society of Pediatric Research, Bernard Sachs Award from the Child Neurology Society, and the Javits Award from the National Institutes of Health. She is also a member of the Institute of Medicine, a fellow of American Association for the Advancement of Science, and a member of the National Academy of Sciences.

Biographies

Stefan Pulst, MD
Stefan M. Pulst, MD, is Director of the Division of Neurology at Cedars-Sinai Medical Center and holder of the Carmen and Louis Warschaw Chair in Neurology. Dr. Pulst is also Medical Director of the American Parkinson Disease Information and Referral Center and Co-Director of the Neuromuscular Center at Cedars-Sinai. In addition, he is Professor of Medicine and Neurobiology at the University of California, Los Angeles (UCLA), School of Medicine.

Dr. Pulst holds several national offices. He is the Founding Chair of the Section on Neurogenetics of the American Academy of Neurology and is the Scientific Director of the National Ataxia Foundation. Dr. Pulst is internationally known for his work on identifying genes that cause neurologic diseases. He has written more than 100 research papers and book chapters, edited two neurology textbooks and is Editor-in-Chief of the international journal Current Genomics. Dr. Pulst received his neurologic training in Germany and at Harvard Medical School. He completed a postdoctoral fellowship in neurobiology at the University of California, San Francisco (UCSF), where he worked with Dr. Earl Mayeri on peptidergic neurotransmission in aplysia.

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 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, the Huntington's Disease Society of America and the National Institutes of Health.

Lawrence J. Schut, MD
Dr. Schut is head of the neurology section of CentraCare Clinic in St. Cloud, Minnesota, where he has practiced for the past 7 years. Formerly, he was Medical Director of GRECC (Geriatric, Research, Education and Clinical Center) at the Minneapolis VA Hospital and also served as Medical Director of the United Pain Center in St. Paul. He received his MD at the University of Minnesota where he also completed his neurology residency and fellowship in neurochemistry. He began his practice in 1967 at the Minneapolis Clinic of Neurology in Golden Valley, MN, where he held multiple free clinics for ataxia patients and their families under the auspices of the National Ataxia Foundation. His special interest in neurology has been the inherited ataxias, one of which, SCA 1, has afflicted over 65 members of his extended family. He has also collaborated with Dr. Laura Ranum and Dr. John Day in investigations of SCA 5 and SCA 8.

S. H. Subramony, MD
Dr. S. H. Subramony received his MD from the Delhi University in New Delhi, India in 1974. Between 1979 and 1981, Dr. Subramony served as a staff neurologist at the VA Medical Center in Jackson, Mississippi. From 1979 until 1985, he served as Assistant Professor of Neurology and as Attending Physician in the Department of Neurology at the University of Mississippi Medical Center.

Currently, Dr. Subramony serves as Professor and Vice-Chairman of Neurology at the University of Mississippi Medical Center, the Medical Director at the VA Medical Center and as State Liaison Officer of the Professor Standards Committee of the American Association of Electrodiagnostic Medicine. He also serves as the Medical Director for the Muscular Dystrophy Association Clinic at University Children's Rehabilitation Center in Jackson, MS. Dr. Subramony is also a member of the National Ataxia Foundation's Medical and Research Advisory Board, and served as NAF Research Director and former Chair of the Cooperative Ataxia Group (CAG).

Ataxia Investigator Meeting General Overview

TIME	EVENT	LOCATION
WEDNESDAY		
3:30 - 6:00 PM	Registration & Social Hour	Citrus
3:30 - 6:00 PM	Poster Viewing	Waterfall Area (lower Level)
6:00 - 7:00 PM	Keynote Speaker: Rodolfo Llinas, New York University	Palm
7:00 PM	Dinner	Citrus
Thursday		
7:00 AM	Breakfast	Mangroves Grille
8:00 AM - 12:30 PM	Ataxia Talks I - Dominant Ataxias	Palm
12:00 - 1:30 PM	LUNCH	Citrus
2:00 - 3:00 PM	Invited General Interest Talk: Salvatore (Billi) DiMauro, Columbia University	Palm
3:00 - 5:00 PM	Ataxia Talks II - Friedreich's Ataxia	Palm
5:30 - 6:30 PM	General Discussion: Tetsuo Ashizawa, Stefan Pulst	Palm
8:00 PM	Dinner at Carrabba's Italian Grill. **Meet in the hotel lobby at Embassy Suites at 7:40 PM**	Carrabba's Italian Grill
FRIDAY		
7:00 AM	Breakfast	Mangrove's Grille
8:30 - 9:30 AM	Invited General Interest Talk: Robert Brown, Harvard University	Palm
9:30 AM - 12:00 PM	Ataxia Talks III - Clinical Methods & Investigations	Palm
NOON	Lunch	Citrus
1:30 - 3:30 PM	Posters	Waterfall Area (Lower Level)
3:30 - 4:30 PM	Poster Commentary	Palm
4:30 - 5:00 PM	Patient/Family/Physician Perspective: From Fear of the Unknown to Hope for the Future by Dr. Larry Schut	Palm
5:00 PM	Buffet Dinner	Citrus

Reminder:
Please wear your name badges throughout the entire AIM meeting. Your cooperation is appreciated.

Thank you !

Wednesday, March 2, 2005

Time	Event	Room
3:30 - 6:00 PM	**Registration / Social Hour**	**Citrus**
3:30 - 6:00 PM	**Poster Viewing**	**World Carpet (Lower Level)**
6:00 - 7:00 PM	**Keynote Invited General Interest Speaker:** ***Cerebellar Past, Present & Future*** Rodolfo Llinas, MD, PhD New York University	**Palm**
7:00 PM	**Dinner**	**Citrus**

Thank you!

The National Ataxia Foundation would like to take this opportunity to thank each AIM participant for taking the time to attend the First Ataxia Investigator's Meeting. We truly appreciate you taking this time out of your busy lives.

The National Ataxia Foundation's Board of Directors

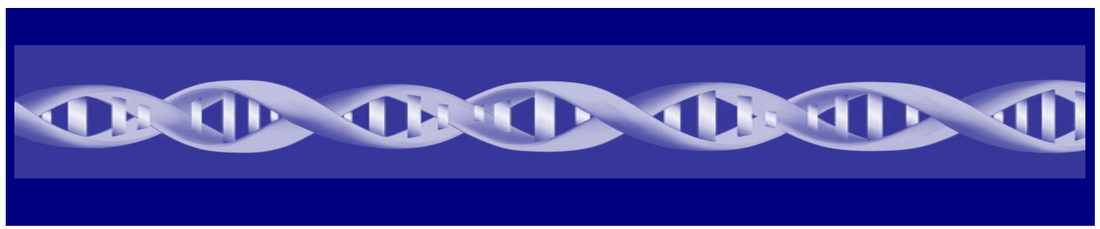
The National Ataxia Foundation has Research Funding Available!

The National Ataxia Foundation (NAF) began direct funding of research of ataxia research studies in 1978. Over the past six years the Foundation has funded sixty-four ataxia research studies in eight countries.

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

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

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



Biographies

Harry Orr, PhD
Harry Orr, PhD is a full professor for the Department of Laboratory Medicine and Pathology at the University of Minnesota Medical School. 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 has published author of more than 120 articles many on the genetics of ataxia. 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.

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.

Susan Perlman, MD
Susan L. Perlman, M.D., is Clinical Professor of Neurology and Director of the Ataxia Center/ Neurogenetics Clinic at the David Geffen School of Medicine at UCLA. She is the principal investigator for a pilot study of the National Ataxia Database and Registry (funded by the NAF) and is a site director for the Friedreich's Ataxia Clinical Outcome Measures study (P.I. Dr. David Lynch at Children's Hospital of Philadelphia, funded by the MDA and FARA). The UCLA Ataxia Center is a multi-disciplinary evaluation and treatment program for patients with inherited and acquired cerebellar disorders, whose activities have been partially supported by the Mariette Monnier bequest.

Helene Puccio, MD
After obtaining a PhD from Harvard University in 1998, Dr. Hélène Puccio joined the group of Professor Michel Koenig (Friedreich's ataxia and other recessive ataxia projects), to work on the Friedriech ataxia research project. She is the permanent scientist (Research Associate INSERM) in charge of the Friedreich ataxia project, which focuses mostly on the use of the mouse models developed by Dr. Puccio for the unravelling of the physiopathology of the disease, the initiation on preclinical therapeutic trials, and the development of cellular models for large scale drug screening. Dr. Puccio has received funding for her important work by the National Ataxia Foundation.

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.

Biographies

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.

Rodolfo Llinas, PhD
Dr. Llinas is the Thomas and Suzanne Murphy Professor in Neuroscience. He is Chairman and Professor at the New York University School of Medicine. He received his MD at the University Javeriana in Bogotá, Colombia in 1959 and his PhD in Neuroscience at the Australian National University, Canberra in 1965. Dr. Llinas is the Chief Editor of Neuroscience and Chairman of NASA/Neurolab Science Working Group.

David Lynch, MD
Dr. Lynch graduated from Yale College with a B. S. in Molecular Biophysics and Biochemistry. He subsequently obtained M.D. and Ph.D. degrees at The Johns Hopkins University School Medicine. His Ph.D. thesis in Neuroscience investigated enzymes involved in the proteolytic processing of enkephalins. Dr Lynch was an Intern in Internal Medicine, resident in Neurology and Postdoctoral fellow at the University of Pennsylvania, He was an Assistant Professor of Neurology and Pediatrics, at University of Pennsylvania School of Medicine from 1995-2004, and was promoted to Associate Professor in 2004.

Dr. Lynch's research includes clinical and translational work on genetic neurologic disorders. This includes collaborative studies on NMDA receptor subtypes in brain of individuals with tuberous sclerosis that derive directly from his basic research on NMDA receptors. In addition, Dr. Lynch has performed and collaborated on studies of biological and clinical markers in genetic neurodegenerative diseases, particularly those associated with reactive oxygen species production such as Friedreich ataxia, and Parkinson's Disease. Dr. Lynch has held grant support from the Muscular Dystrophy Association and the Friedreich Ataxia Research Alliance to support his clinical research.

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.

Thursday, March 3, 2005

Time	Event	Room
7:00 - 8:00 AM	**Breakfast**	**Mangrove's Grille**
8:00 -10:00 AM	**Ataxia Talks I - *Polyglutamine Dominantly Inherited Ataxias*** Chair: S. H. Subramony, MD, University of Mississippi	**Palm**
8:10 AM	***Update on SCA 1*** - Harry Orr, PhD University of Minnesota	
8:30 AM	***Pathogenic Studies of SCA 1*** - Huda Zoghbi, MD, PhD, Baylor College of Medicine	
8:50 AM	***Update on SCA 2*** - Stefan Pulst, MD University of California, Los Angeles & Cedars Sinai Hospital	
9:10 AM	***Update on SCA 3*** - Hank Paulson, MD, PhD University of Iowa	
9:30 AM	***Update on SCA 7*** - Al LaSpada, MD, PhD University of Washington	
9:50 AM	***Discussion of Polyglutamine Ataxias (All participants)***	
10:20 - 10:40 AM	**Break (coffee, rolls, fruit)**	**Citrus**
10:40 - 12:30 PM	**Ataxia Talks II - *Non-Polyglutamine Dominant Ataxias*** Chair: John Day, MD, PhD	**Palm**
10:40 AM	***Update on SCA10*** - Tetsuo Ashizawa, MD University of Texas Medical Branch, Galveston	
11:00 AM	***Update on SCA 6*** - Christopher Gomez, MD, PhD University of Minnesota	
11:20 AM	***Update on SCA 12*** - Russell Margolis, MD Johns Hopkins University School of Medicine	
11:40 AM	***Update on SCA 8*** - Laura Ranum, PhD University of Minnesota	
12:00 PM	***Discussion on Non-Polyglutamine Dominant Ataxias (All Participants)***	
12:30 - 2:00 PM	**Lunch**	**Citrus**

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 3, 2005 cont.

Time	Event	Room
2:00 - 3:00 PM	**Invited General Interest Talk 2:** ***Ataxia and Mitochondrial Diseases*** Salvatore (Billi) DiMauro, MD Lucy G. Moses Professor of Neurology Columbia University Medical Center - New York, NY	**Palm**
3:00 - 5:00 PM	**Ataxia Talks II - *Friedreich's Ataxia* -** Chair: Christopher Gomez, MD, PhD	**Palm**
3:00 PM	***Update on Friedreich's Ataxia* -** Massimo Pandolfo, MD University Libre de Bruxelles Belgium	
3:20 PM	***Molecular Basis of Friedreich's* -** Grazia Isaya, MD Mayo Clinic	
3:40 PM	***Pathophysiology of Friedreich's Ataxia* -** Rob Wilson, PhD University of Pennsylvania	
4:00 PM	***Friedreich's Mouse Models* -** Helene Puccio, MD, PhD University of Strausberg, France	
4:20 PM	***Discussion of Friedreich's Ataxia* (All Participants)**	
5:00 - 5:30 PM	**Break (wine & cheese)**	**Citrus**
5:30 - 6:30 PM	***General Discussion:*** Tetsuo Ashizawa, MD & Stefan Pulst, MD	**Palm**
8:00 - 10:00 PM	**Dinner offsite at Carrabba's Italian Grill** ****Meet in Lobby at Embassy Suites at 7:40 PM****	**Carrabba's Italian Grill**

National Ataxia Foundation
2005 Annual Membership Meeting

Ray of Hope

March 3-6, 2005, Tampa, Florida

We hope that you are planning on joining us at the NAF Membership Meeting. Please remember to register in the Magnolia Room and pick up your conference materials if you are extending your stay.

Registration Hours:

Thursday:	Noon - 2 PM & 6 - 8 PM
Friday:	9 AM - Noon & 4 - 6 PM
Saturday:	7:30 - 11 AM & 2 - 4 PM
Sunday:	7:30 - 11:00 AM

Biographies

Christopher Gomez, MD, PhD
Dr. Gomez received 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. Dr. Gomez currently serves as Professor of Neurology and Associate Head for Research in the Department of Neurology. He is Director of the University of Minnesota Ataxia Clinic. Prior to this, he served as Clinical Instructor, Ataxia Clinic, in the Neurology Department at the University of California, Los Angeles. Dr. Gomez currently serves as the Associate Research Director for the National Ataxia Foundation, and has received funding from the NAF for his important ataxia research efforts.

Grazia Isaya, MD, PhD
Dr. Isaya obtained her M.D. at University of Padova, Padova, Italy and in 1991, she obtained her PhD in Developmental Sciences. From 1987 through 1998, Dr. Isaya held the titles of Postdoctoral Associate, Associate Research Scientist and Assistant Professor at Yale University School of Medicine, Department of Genetics, New Haven, CT. From 1998 – 2003, She Was the Senior Associate Consultant, Department of Pediatric & Adolescent Medicine at the Mayo Clinic in Rochester, MN. From 2003 to present, she is a Consultant for the Department of Pediatric & Adolescent Medicine, Mayo Clinic College of Medicine, Rochester, MN.

Dr. Mark Hallett, MD
Dr. Hallett obtained his M.D. at Harvard University and trained in Neurology at Massachusetts General Hospital. He had fellowships in Neurophysiology at the National Institutes of Health and at the Institute of Psychiatry in London. From 1976 to 1984, Dr. Hallett was Chief of the Clinical Neurophysiology Laboratory at the Brigham and Women's Hospital and Associate Professor of Neurology at Harvard Medical School.

From 1984, he has been at the National Institute of Neurological Disorders and Stroke where he serves as Chief of the Human Movement Disorders and other problems of Motor Control He also served as Clinical Director of NINDS until July 2000. He is past President of the American Association of Electrodiagnostic Medicine and the Movement Disorder Society. He is currently Vice-President of the American Academy of Neurology, and Editor-in-Chief of *Clinical Neurophysiology*. His interests in Motor Control are wide-ranging, and include brain plasticity and its relevance to neurological disorders and the pathophysiology of dystonia, parkinsonism, and ataxia.

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 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.

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.

Biographies

Tetsuo Ashizawa, MD, PhD
Dr. Ashizawa is Professor and Chair of the Department of Neurology at the University of Texas Medical Branch in Galveston, Texas. He has been involved in research of Friedreich's ataxia and various autosomal dominant cerebellar ataxias, including SCA2, SCA6, SCA7, SCA8 and SCA10. In 1998, he received a grant from the Oxnard Foundation through the National Ataxia Foundation, which led to the identification of an expansion mutation of an ATTCT pentanucleotide repeat in SCA10. His research interests involve both basic science and clinical studies. His basic science projects include investigations of repeat expansion mechanisms, population genetics, and molecular biology of spinocerebellar ataxias. On the clinical side, he has been engaged in characterization of clinical manifestations, genotype-phenotype correlations, development of ataxia rating scales and planning of therapeutic interventions for inherited ataxias.

Robert Brown, PhD
Dr. Robert H. Brown, Jr. is Director of the Day Neuromuscular Research Laboratory and Muscular Dystrophy Association (MDA) clinic at the Massachusetts General Hospital (MGH) and Professor of Neurology, Harvard Medical School. Dr. Brown graduated from Harvard Medical School and completed his doctoral training in neurophysiology at Oxford University. Dr. Brown trained in Neurology at the Massachusetts General Hospital. In 1984, The Day Neuromuscular Research Laboratory was founded by Dr. Brown to investigate neuromuscular diseases, including Miyoshi myopathy and ALS

John Day, MD, PhD
John W. Day, M.D., Ph.D., is Professor of Neurology at the University of Minnesota, and Medical Director of the National Ataxia Foundation. Dr. Day received his M.D. from the University of Minnesota, and subsequently received a Ph.D. in Neuroscience from the Albert Einstein College of Medicine. He trained in Neurology at the University of California, San Francisco, and has been involved in the diagnosis and care of ataxia patients for 20 years.

Dr. Day is Director of the Paul and Sheila Wellstone Muscular Dystrophy Center at the University of Minnesota, where he has active ataxia and neuromuscular clinics. He also cares for pediatric ataxia and neuromuscular patients at the Gillette Pediatric Specialty Heathcare Clinic in St. Paul. Dr. Day has collaborated for many years with Dr. Laura Ranum, and with Dr. Lawrence Schut, with whom he has worked to identify and characterize SCA5 and SCA8, as well as other forms of ataxia.

Salvatore (Billi) DiMauro, MD, PhD
Throughout his career, Dr. DiMauro has kept a focused interest on inborn errors of energy metabolism, recognizing unusual patients through clinical observation, and using both biochemical and molecular approaches to define disease entities. Thus, although Dr. DiMauro started as a myologist, his studies of mitochondrial diseases have taken him back to the brain. Because the brain is so crucially dependent on oxidative metabolism, mitochondrial dysfunction almost always impairs brain function and often causes mental retardation. His group's interest in the relationship between mitochondrial disorders and mental retardation has been recognized by the National Institute for Child Health and Human Development (NICHD), which supports a Program Project under Dr. DiMauro's direction entitled "Mitochondrial Encephalomyopathies and Mental Retardation".

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.

Friday, March 4, 2005

Time	Event	Room
7:00 - 8:30 AM	**Breakfast**	**Mangrove's Grille**
8:30 - 9:30 AM	**Invited General Interest Talk 2: *ALS* -** Robert Brown, PhD Harvard University	**Palm**
9:30 - 10:30 AM	**Ataxia Talks III – *Clinical Scales and Investigations*** Chair: Chip Wilmot, MD Emory University	**Palm**
9:30 AM	***Clinical Issues in Ataxia* -** Susan Perlman, MD University of California, Los Angeles	
9:50 AM	***Clinical Measures of Friedreich's Ataxia* -** David Lynch, MD University of Pennsylvania	
10:10 AM	***Clinical Methods in Studying Ataxias* -** Thomas Klockgether,MD University of Bonn, Germany	
10:30 - 10:50 AM	**Break (coffee, rolls, fruit)**	**Citrus**
10:50 AM-12:00 PM	**Ataxia Talks III – *Clinical Scales and Investigations (cont)***	**Palm**
10:50 AM	***High Dose Idebenone Treatment for Friedreich's Ataxia*** Kenneth "Kurt" Fischbeck, MD NINDS, NIH Bethesda, MD	
11:10 AM	***Ataxias and Antibodies*** Mark Hallett, MD NINDS, NIH Bethesda, MD	
11:30 AM-12:00 PM	***Discussion Clinical Scales and Investigations*** **(All Participants)**	
12:00 - 1:00 PM	**Lunch**	**Citrus**
1:00 - 3:00 PM	**Posters**	**World Carpet (Lower Level)**
3:00 - 3:30 PM	**Break**	**Citrus**
3:30-4:30 PM	**Poster Commentary** Dr. Harry Orr and Dr. Hank Paulson will select 4 posters for open/directed discussion with primary presenter	**Palm**
4:30-5:00 PM	**Patient/Family/Physician Perspective: From Fear of the Unknown to Hope for the Future** **Commentary by Larry Schut, MD**	**Palm**
5:30-7:30 PM	**Buffet Dinner**	**Citrus**