

Generations

EXPANDED EDITION



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_____ *Spring 2014*

Annual Membership Drive Begins in Mid-May

The National Ataxia Foundation's Annual Membership Drive will begin in mid-May 2014. Membership support is essential in providing important programs and services to ataxia families. NAF offers various levels of membership support and welcomes renewing, new, and recurring gift members.

Membership support:

Provides current and accurate information about ataxia through numerous publications on different forms of ataxia, genetics and gene testing, and symptom management, medications, and research opportunities.

Brings together ataxia families through local support groups and chapters, social networks and nationally through the annual membership meetings.

Brings world leading ataxia scientists together through the Ataxia Investigators Meetings (AIM) and fostering ataxia research through hosting, partnering, and sponsoring additional ataxia research conferences and initiatives.

Allows an extensive and comprehensive web site on ataxia, a 48-page quarterly ataxia news publication, *Generations*, neurological resource list, and much more.

Creates awareness about ataxia through promoting International Ataxia Awareness Day, staffing information booths at Abilities Expos and medical conferences, press releases, and events.

Membership support significantly helps in providing important programs and services, but also provides you with a free subscription to NAF's quarterly news publication, *Generations*, as well as registration discounts in attending the NAF Annual Membership Meeting.


Please become or renew your membership and please ask others to also become a NAF member. Thank you.



Inside This Issue

- Over **\$1,000,000 for Ataxia Research**. See pages 12-32.
- **Want to get Involved with IAAD?** See the information on page 30.
- Connect with others: **Personal Stories** appear on pages 34-41
- **Walk n' Roll for Ataxia** Information and events are on page 42
- Check the **Calendar of Event** on pages 52-54 to see what's going on near you

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Table of Contents

Annual Membership Meeting (AMM)

2014 AMM Recap	3
Dr. Brian Brooks Answers	
Questions from 2014 AMM	4
2015 AMM Announcement	5
2014 AMM Recordings Order Form	6

Articles

Social Security Made Easier	32
Caring for a Seriously Ill Child	51

International Ataxia Awareness Day

International Ataxia Awareness Day	30
Walk n' Roll for Ataxia	42

Membership Topics

Annual Membership Drive	1
NAF Merchandise	7
From the Desk of the Executive Director	33
Chapter and Support Group News	43
NAF Directory of Chapters, Support Groups and Ambassadors	47
Calendar of Events	52
Memorials and In Your Honor	55

Personal Stories & Poems

Mark Breland	34
Auburn Chef Earns Food Network Nod	35
New Method of 'Live Looping' Helps Musician Play On	36
I'll Do the Driving, Thank You	37
Our Story in a Nutshell	38
My Journey	40
Don't Bite the Hand that Feeds You!	41

Research/Research Opportunities

Final Research Summaries	8-10
Brain Donation Program	11
Friedreich Ataxia Tissue Donation	11
Second Year NAF Awards Over \$1,000,000 for Ataxia Research	12
New Research Grant Awards	12-32
CoRDS/NAF Ataxia Patient Registry	17
Fragile X Gene Study	18
Early Symptoms of Friedreich's Ataxia Study ..	22
MRI Study at University of Minnesota	26
Transcranial Magnetic Stimulation Clinical Trial	31
New Medication Trial for FRDA	41

— *The deadline for the Summer issue of Generations is May 16, 2014* —

National Ataxia Foundation 57th Annual Membership Meeting “Betting on Ataxia Research”

The 2014 Annual Membership Meeting (AMM) was hosted by NAF’s Ataxia Support Groups in the Western Region. It was a pleasure working with the Support Group Leaders of the Western Region. These support groups did an outstanding job coordinating this event and the National Ataxia Foundation would like to congratulate them on hosting such a successful meeting! Be sure to check out the pictures that will be posted on NAF’s website and in the Summer issue of *Generations* showing the fun Las Vegas themed banquet, the colorful meeting t-shirts, and the happy faces of the attendees.

The Foundation would like to extend a special thank you to all the attendees, speakers, facilitators, exhibitors and the numerous volunteers of the NAF 2014 “Betting On Ataxia Research” AMM held in Las Vegas, NV. This conference would not have been possible without the time, contributions, and efforts given by so many. Thank you so much for the wealth of information and knowledge that was brought to the conference by all the speakers, facilitators and exhibitors. The information and skills taken away from this conference by the attendees is invaluable. We would like to thank this year’s MC’s (Sherry McLaughlin, Angela Li, and Mary-Lisa



Garcia

Orth), Lora Morn for volunteering as our on-site nurse at the conference, David Garcia for taking such memorable pictures of this year’s event, Frank Fiorito for the great dancing music and entertainment at the Saturday evening banquet, Mike and Michelle Wolfson for the great job they did as this year’s Volunteer Coordinators, Heather Evans for facilitating the activities in the Activity Room again this year,

the DeMint & Furton families for the Las Vegas themed banquet décor and t-shirts, and to the time and talents given in the registration area and to the silent auction by Char Danielson, Camille Daglio, Nettie Jones, Bailey Vernon, Gerry Mahler, John Mauro, Greg Rooks, Mary Fuchs, Dave Reedstrom, Renae Parent, Sam Kirton, Cathy and Joe DeCrescenzo.



Members of Western Region support groups at the 2014 NAF Annual Membership Meeting

We would also like to thank this year’s sponsors Athena Diagnostics, the FA Project, MetLife Center for Special Needs Planning, and California Analytical Instruments. Thank you to the Las Vegas Convention and Visitors Bureau for the welcome bags and local information provided for this year’s meeting. Thank you to Official Security for donating their services and for the exceptional job they did in keeping the silent auction items secure. Thank you to Bally’s and Encore Productions for their service and hospitality throughout this event.

Thank you to all the organizers, participants, and supporters of the fundraisers held to help offset the cost of the 2014 AMM. ❖



Dr. Brian Brooks Answers Questions from 2014 AMM

Dr. Brian Brooks is the chief of the Unit of Genetic and Developmental Eye Disease at the National Eye Institute (NEI), National Institutes of Health (NIH). We had the privilege of hearing Dr. Brooks at NAF's Annual Membership Meeting in March. He described the upcoming clinical study that will take place in SCA7 at NIH. SCA 7 differs from most other forms of spinocerebellar ataxia in that visual problems, rather than poor coordination are generally the earliest signs of the disease. Because of travel obligations, Dr. Brooks was unable to remain for the Q & A session, but provided answers to questions asked at the meeting. Not only are his answers helpful for those with SCA7, but may address vision problems in other forms of ataxia.

Lights seem to “bounce” at night. What can be done to improve it?

I am not completely sure why lights would bounce at night, but here are a couple of thoughts: When it is dark outside, we may lose some of the “visual clues” from our surroundings that keep our eyes straight, pointed in the right direction. It is possible that when you are in such a situation, a natural tendency for your eyes to drift apart may appear. (This same kind of tendency can occur when we are tired or sick.) The sense that a light is moving in this case might be that your eyes are drifting and you are unconsciously trying to bring back an image to

make it one. Another possibility is that, as a result of your ataxia, there is a tendency for your eyes to involuntarily move (nystagmus) that might be brought out when you are tired and it is dark. Really, to get to the bottom of this, you would have to have an ophthalmologist – perhaps even a neuro-ophthalmologist – examine you.

Is there anything to improve pursuit in my vision?

Pursuit can be a problem for a couple of different reasons. One, in folks with SCA7, it is possible that there are “holes” in the visual field of vision. These holes may make it hard to pursue an object. In pretty much all forms of SCA, there can be problems with the brain's wiring of the eye muscles in a coordinated fashion. This can make moving your eyes to the correct target quickly or following an object steadily difficult. Unfortunately, there is not any specific treatment for either of these. Perhaps the most important thing is awareness of the problem, recognizing what limitations it might present, and then taking necessary precautions to make sure it does not affect your safety.

Are eye changes detectable in other SCAs?

To my knowledge, the only form of SCA that affects the light sensing cells (the photoreceptors) in the retina is SCA7. However, in some patients with other forms of hereditary ataxia (for example, Friedreich ataxia, some of the other SCAs), there may be some slow damage to cells in the retina that form the “cables” connecting the eye to the brain. These are the retinal ganglion cells that form the optic nerve. In general, however, this problem in the optic nerve does not affect the ability to read the eye chart. Many forms of ataxia can produce abnormal eye movements such that the eyes move involuntarily and/or that the brain has problems targeting/pursuing an object.



Vehicle Donation

Donation of your vehicle to the National Ataxia Foundation will help support the important work of the Foundation. To donate your car, truck, motorcycle or motor home, please call 1-800-240-0160 or visit www.donateacar.com.

*The NAF Board of Directors along with the
North Central Regional Support Groups would like to invite you to attend the*

National Ataxia Foundation 58th Annual Membership Meeting March 13-15, 2015



Join us in Denver for the Annual Membership Meeting!

— RESERVATION INFORMATION WILL BE ANNOUNCED SOON —

For the latest information on conference registration, program schedule, and area information, keep checking the National Ataxia Foundation website, www.ataxia.org.

2015 NAF AMM “Support Our Conference” Campaign

<https://naf.myetap.org/fundraiser/15AMM/>

For more information on Denver, visit www.denver.org.



Share Your AMM Stories & Photos

Personal stories and photos from our readers are a valuable part of *Generations*.

Do you have a story or photo from the 2014 AMM that you would like to share?

Please submit them to joan@ataxia.org to be considered for publication. The deadline for the upcoming summer issue is May 16.

NAF SESSION RECORDINGS



National Ataxia Foundation 57th Annual Membership Meeting Betting on Ataxia Research

Bally's Las Vegas Hotel and Casino, March 21-23, 2014



Visit Us Online at: www.dcprovidersonline.com/naf/

SPECIAL A limited number of FREE recorded sessions from this year's Conference will be available after the Annual Meeting online at: www.dcprovidersonline.com/naf/

The free sessions are available for "view only" while purchased sessions are downloadable.

SESSIONS OFFERED FOR FREE:

- Friday :** NAF Update, Ataxia Patient Registry, Why Should I Be Seen at an Ataxia Clinic?
Saturday : AIM Overview and Highlights, Research Review & Tissue Donation Program, Transitioning into Adult Healthcare
Sunday : Wrap-up: What We Have Learned

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NAF Merchandise

BOOKS

Healing Wounded Doctor-Patient Relationships
by Linda Hanner with contributions by John J. Witek, MD \$10

Living with Ataxia: An Information and Resource Guide by Martha Nance, MD (2nd ed. 2003) \$14

Managing Speech and Swallowing Problems: A Guidebook for People with Ataxia
by G.N. Rangamani, PhD with contributions from Douglas E. Fox, MS (2nd ed. updated 2006) \$7.50

Ten Years to Live
by Henry J. Schut \$8.75

There's Nothing Wrong with Asking for a Little Help ... and Other Myths
by Dave Lewis \$15.95

Recipes and Recollections
by Kathryn Hoefler Smith \$10

Cooking for a Cause
by Julie Karjalanti for FRDA research \$12

VIDEO/CD

Ballads of a Family Man CD \$5
Together There is Understanding VHS \$20 **DVD** \$25

SHIRTS/MISCELLANEOUS

Original NAF IAAD T-Shirt \$10
NAF Baseball Cap (White or Blue) \$10

International Ataxia Awareness Day T-Shirt \$10
Available in youth L, and adult S to XXXL (XXL out).

Past Annual Membership Meeting (AMM) T-Shirts
\$1 each while supplies last!

NAF Polo Shirts \$25
Mens – Royal blue w/white NAF logo in M to XXXL.
Womens – Light blue w/ navy NAF logo in S to XXL.

NAF Denim Shirt w/ white embr. NAF logo. \$27.50

“Ataxia is Not a Foreign Cab” T-Shirt \$10
White. New design. Sizes S to XXXL.

“Ataxia is Not a Foreign Cab” Long-Sleeve T-Shirt
Blue. Sizes S to XXXL. \$15

“Ataxia is Not a Foreign Cab” Sweatshirt
White. Sizes S to XXXL. \$20

NAF 50th Anniversary Coffee Mug \$3

“Ataxia is Not a Foreign Cab” Magnet \$1

Window Cling or Bumper Sticker \$1 ea. or 6/\$5

NAF Ataxia Awareness Band Blue One size. \$2

NAF Ataxia Awareness Ribbon Magnet \$4

Reusable Grocery Bag w/ NAF and Cab Logos \$5

“Know Ataxia” Backpack 20”x16” \$5

NAF Grip n’ Sip Water Mug \$5

NAF Lapel Pin \$5

Magnetic Power Clip \$3

NAF Shoulder Bag \$10

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Research Grant Award

Identifying Proteins Regulated by AFG3L2 by Quantitative Proteomics

By David Chan, MD, PhD

Howard Hughes Medical Institute and California Institute of Technology, Pasadena, CA

The following is a research summary of a grant funded by NAF for fiscal year 2012.

SCA28 is an early-onset form of SCA caused by mutations in a protein called AFG3L2. AFG3L2 resides within mitochondria, the sites of cellular energy production, and it belongs to a family of proteins whose function is to degrade specific “substrate” proteins. Through this activity, AFG3L2 is thought to maintain cellular health by removing damaged proteins or by allowing the mitochondria to adapt to cellular demands. The aim of this project was to identify molecules that underlie the pathogenesis of spinocerebellar ataxia type 28 (SCA28).



Dr. David Chan

Mutations in SCA28 patients disrupt the ability of AFG3L2 to degrade proteins, but information regarding the identities of its substrates in human cells remains largely unknown. Identifying these substrates however, has important implications for understanding SCA28, and could potentially illuminate new therapeutic strategies. To achieve this goal, we used quantitative mass spectrometry to comprehensively analyze the change in protein composition of mitochondria upon depletion of AFG3L2, and found that many proteins important for forming energy-producing complexes within mitochondria are selectively depleted. This observation demonstrated that diminished energy production from mitochondria is a consequence of loss of AFG3L2 activity. Furthermore, we found a number of proteins that were selectively enriched upon loss of AFG3L2 activities. This group of proteins likely includes direct AFG3L2 substrates, and their unregulated levels

may be especially relevant to SCA28. Indeed, among this group of proteins, we found a candidate protein that has known role in suppressing the expression of key components of the mitochondrial energy-producing complexes. We are currently investigating the regulation of this candidate by AFG3L2, and its role in causing the various defects observed in AFG3L2-depleted cells. ❖

CFC Number

The National Ataxia Foundation's Combined Federal Campaign (CFC) number is 10752. This program provides a convenient way to donate to the Foundation, and provides great benefit to those with ataxia. Please give generously. Thank you.

Matching Gifts

Many employers will match your gift to the National Ataxia Foundation through a Matching Gifts Program. This valuable benefit will allow you to have twice the impact on the lives of families affected by ataxia when you make a donation to NAF.

Research Fellowship Award

Studying the Cellular Mechanisms of Spinocerebellar Ataxia Type-13

By Dr. Fadi A. Issa

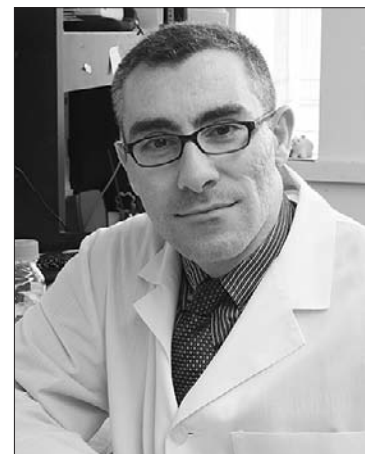
Department of Physiology, David Geffen School of Medicine, University of California, Los Angeles

The following is a research summary of a grant funded by NAF for fiscal year 2012.

Spinocerebellar Ataxia Type-13 SCA-13 is an autosomal dominant disease that leads to either abnormal development or degeneration of the cerebellum. The manifestation of either of these two pathological conditions depends on the type of mutation of the KCNC3 gene the individual carries. The KCNC3 gene codes for the Kv3.3 voltage-gated potassium channel, which is abundant in the cerebellum and spinal cord motor neurons. The Kv3.3 channel is thought to enhance the electrical excitability of neurons, which is an important aspect in ensuring that neurons communicate their information properly with one another. We have proposed that in SCA-13, perturbing the neurons' electrical excitability is likely to cause them to malfunction and perhaps even develop improperly and eventually die.

To test this hypothesis we studied the effects of the infant-onset forms of SCA-13 (RH and FL mutations: these acronyms refer to the type of the mutation in the potassium channel) on development of cerebellar Purkinje neurons. We expressed each of the mutated Kv3.3 channels in the cerebellum of developing zebrafish larvae. The brain of zebrafish develops rapidly and can be viewed in vivo without any invasive procedures due to the optical clarity of the animals. We found that the Purkinje neurons of embryos expressing the normal form of the Kv3.3 channel developed normally as expected with numerous and extensive dendritic branches. Surprisingly, the Purkinje neurons of larvae expressing the FL infant-onset mutation did not appear to be

affected. Their morphological shape appeared normal with numerous dendritic branches similar to neurons that expressed the normal form of the Kv3.3 channel. However, Purkinje neurons expressing the R4H infant-onset mutation displayed dramatic change in their morphological appearance. These neurons showed reduced number of branches early during development. This result is significant because branches of neurons play a vital role in their ability to navigate their environment during development and to communicate with neighboring neurons. Currently, we are testing the hypothesis that the R4H mutation may have a more severe effect on neuronal function and excitability than FL that results in differences of severity on the morphological development of the Purkinje neurons.



Dr. Fadi A. Issa

To test this hypothesis, we are working on measuring changes in the electrical excitability of the infant-onset mutant expressing neurons by developing a zebrafish transgenic fish line that expresses LCK-GCaMP5 protein in the

Continued on page 10

Studying the Cellular Mechanisms...
Continued from page 9

cerebellar Purkinje neurons. LCK-GCaMP5 is a fluorescent protein marker that binds calcium and fluoresces green when the neurons become active. This approach will enable our continuing research endeavor to monitor the activity of the neurons and investigate how these mutations perturb the excitability of developing neurons and whether these functional changes account for the morphological differences observed during development.

These experiments have provided us with

basic operational information that was missing prior to the acquisition of this Fellowship grant. With the transgenic lines that were developed and ones I continue to develop along with the technical approaches I am utilizing, I'm confident that my ongoing research is optimally positioned to elucidate new insights to the mechanisms of how the SCA-13 mutations exert their effects on the cerebellum. I'm thankful for the National Ataxia Foundation for providing me the funding to conduct this research, and I'm looking forward to continue to provide my contribution to achieve the National Ataxia Foundation's goal of alleviating the symptoms and eventually curing this disease. ❖

Kyle Bryant Translational Research Award

New Ways to Detect and Treat FRDA-Related Heart Disease at its Earliest Stages

By *Subha Raman, MD*

Ohio State University, Columbus, OH

The following is the final research summary of a study that recently concluded. This research grant was jointly funded by NAF and FARA.

Dr. Raman's research generated several important findings that should lead to better heart health for patients with FA: first, her team showed with cardiac MRI that the blood flow to the innermost portion of the heart is reduced before there is a measurable abnormality in heart function; second, they found that there were specific abnormalities in cholesterol levels, also known as features of the "metabolic syndrome," in patients with FA, and the greater these abnormalities the worse the blood flow to the heart; third, they showed that there was measurable scarring of heart muscle, also before heart function was abnormal; and fourth, they showed that increased thickening of the heart worsened with the genetic abnormality captured in minimum number of GAA repeats. Together these findings suggest that cardiac MRI and measurement of metabolic syndrome factors, especially high triglyceride and low HDL or "good cholesterol" levels, can pick up very early changes in patients with FA. Her team hopes to pursue further research building on these results to prove that early treatment using medications that have been proven to benefit reduced blood flow, heart muscle scarring, cardiac thickening and metabolic syndrome can reduce the risk of complications due to heart disease in FA. ❖



Dr. Subha Raman

Brain Donation Program

The following is an excerpt from a presentation by Dr. Christopher Gomez at the 2014 AMM titled "Why Should I be Seen at an Ataxia Clinic."

Many people with ataxia and other diseases have shown an interest in offering their brain tissues for research after they pass away. The NAF, under the guidance of Dr. Laura Ranum, has begun to build out a Brain Donation Program that attempts to simplify the process and details of making this generous offer with little or no cost to the family.

For several reasons the most efficient approach is to make the initial contact or enroll through an ataxia clinic, where a neurologist specializing in ataxia can make the needed contacts. If an ataxia clinic is not available, it is still preferable to be seen at a university clinic or academic medical center.

An academic medical center is preferable because brain donation depends on several things, not only your generosity with donating your brain for research, but also cost. The process is expensive and there are not research dollars to pay for some very necessary steps to the process of brain donation.

1. If the person with ataxia passes away at a location far from a research center, they have to be transported there, and that is a cost that has to be borne by the funeral home which has to be passed on to the family.

2. There is a cost to removing the brain tissues that is about a \$1,000. There are no research dollars to pay for this procedure. However, academic centers have programs that take advantage of the value of ataxia brains for education. Any place that has a medical center, that has a training program for pathology for the professors who study heart, lungs, kidneys and brains, have to maintain their accreditation by having a regular supply of tissues for training residents. Therefore, they make the cost of doing a post-mortem free for anybody who has been seen as a patient and has a medical record at a place that has a training

program. All it takes is having been there one time, to be officially registered as a patient, so that makes it a very useful opportunity. Thus it helps both the training program and the patient and the family to be seen at least one time at a medical center. This should ideally be an ataxia clinic, but any university center where there is an autopsy program. You can contact the physician and say, "I'd like to donate my brain, and I know that you have a training program. Do you have a free autopsy policy like they have in most training programs?"

Thus, there is an additional advantage to be seen in an ataxia clinic because of the free autopsy for the purpose of brain donation, although this only eliminates the brain removal cost, not the cost of transporting the body after death.

If you are unable to find a medical center in your area, but still wish to donate, you may begin the Brain Donation pre-plan process, by contacting the National Ataxia Foundation at susan@ataxia.org or (763) 553-0020 and ask to have the Brain Donation Pre-Plan form mailed, faxed or emailed to you. This form will guide you on the important decisions that need to take place for brain donation to occur. On behalf of the entire ataxia research community, thank you for considering the donation of this important gift. ❖

Friedreich Ataxia Tissue Donation

If you have Friedreich Ataxia and you are interested in the Tissue Donation Program, please contact Dr. Arnulf Koeppen at (518) 626-6377 or arnulf.koeppen@va.gov.

Second Year NAF Awards Over \$1,000,000 for Ataxia Research

The National Ataxia Foundation (NAF) is pleased to report that 24 promising ataxia research studies totaling more than \$1,000,000 from the United States, Italy, Australia, United Kingdom, Portugal, and Germany were awarded funding for fiscal year 2014.

There were 60 reviewers assigned to evaluate 81 research applications this past year. Each research application was assigned two to three independent peer reviewers. In addition, the NAF Scientific Review Panels conducted their own independent reviews of these research applications who then made their recommendations to the NAF Board of Directors who made the final funding decisions.

The funding for these important ataxia research studies were made possible through the generosity of NAF donors and partners who contributed

to the 2013 NAF Annual Ataxia Research Drive, the \$150,000 matching research grant from The Gordon and Marilyn Macklin Foundation, our corporate and foundation friends, the Michael and Patricia Clementz Family Endowment Fund for SCA 3 Research, NAF chapters and support groups, individual and group donations, individuals and families who conducted fund raising events, NAF Walk n' Rolls for Ataxia events, the Bob Allison Ataxia Research Center (BAARC) and our generous anonymous donor who committed \$500,000.

The generosity of our members and others enabled NAF to continue its mission in supporting ataxia research studies that bring us closer in ending ataxia. Thank you! **The research studies that follow are currently being conducted and were funded for FY 2014.** ❖

Pioneer Awards

Translating RNAi Therapy for Spinocerebellar Ataxia 1 (SCA1) to the Clinic

By *Beverly Davidson, PhD*

The Children's Hospital of Philadelphia Research Institute, Philadelphia, PA



Dr. Beverly Davidson

There are currently no therapies that delay onset or progression of spinocerebellar ataxia. In earlier work, we showed that gene silencing approaches had a profound positive impact on disease readouts in two animals models of spinocerebellar ataxia type 1 (SCA1). We have also completed initial pilot studies in monkeys to determine if the delivery paradigm we use for mice can be translated into a primate brain. Importantly, our delivery approach is scalable from mice to primates, and in these initial monkey studies we found efficient gene silencing of the target ataxin-1 gene in critical brain areas affected by disease. Here, we propose to expand this pilot work. In the first phase, we will build the vector that could be used in humans. In the second phase we will test the utility of this vector at several doses, as regards safety and silencing activity in nonhuman primates. ❖

Ataxin-7 Knock-Down Therapy to Treat SCA7: Preclinical Trial Validation in Mice

By **Albert R. LaSpada, MD, PhD**
University of California – San Diego

Spinocerebellar ataxia type 7 (SCA7) is a dominantly inherited neurological disorder. SCA7 patients develop ataxia due to degeneration of the cerebellum and the brainstem, and also suffer from visual difficulties due to a form of retinal degeneration, resulting in blindness in many patients, especially patients who present before the age of 30. SCA7 is caused by a CAG trinucleotide repeat expansion in the ataxin-7 gene. The CAG repeat tract is translated into a series of glutamine amino acid residues that occur in a consecutive row in the ataxin-7 protein. The presence of an expanded tract of glutamines in the ataxin-7 protein causes the protein to adopt an aberrant conformation; hence, production of the mutant ataxin-7 protein is the first step in a pathogenic cascade that culminates in neurological disease and vision loss.

SCA7 is one of nine so-called CAG – polyglutamine repeat diseases, and these disorders belong to a broad category of neurodegenerative diseases involving proteins that misfold. As production of the altered protein is the crux of the pathology in all such diseases, numerous studies have tested the hypothesis that reduced expression of the disease protein can yield improvements in disease pathology and progression. To achieve a reduction in the expression of the mutant protein, a number of strategies have been devised. Many of these strategies target the messenger RNA (mRNA), which provides the template for encoding of the disease protein. One strategy with enormous potential is to chemically synthesize a short sequence of either DNA or single-stranded RNA that matches a sequence of

the mutant messenger RNA. The binding of DNA to the mRNA yields a duplex that is then recognized by the enzyme RNase H. Upon recognition, RNase digests the duplex, resulting in destruction of the mRNA. Binding of RNA to the mRNA can lead to its destruction or can stop it from being translated into protein. Both strategies achieve the same result: they prevent the disease protein from being made.

We have been working to create oligonucleotides directed against ataxin-7, and have validated a set of oligonucleotides that can destroy the ataxin-7 mRNA and thereby reduce the expression of ataxin-7 protein. In this project, we propose to produce oligonucleotides, test



Dr. Albert R. LaSpada

them for toxicity effects in mice, and then use them to treat SCA7 retinal degeneration in mouse models that recapitulate the exact SCA7 retinal disease phenotype. If these “preclinical trials” are successful, we would then be in a position to file an application with the FDA for permission to attempt this therapy in human SCA7 patients. This would also set the stage for using oligonucleotides to treat SCA7 brain disease and other related SCAs caused by CAG repeat expansions that encode polyglutamine proteins. ❖

Preclinical Development of NS13001 for SCA2

By **Ilya Bezprozvanny, PhD**

University of Texas Southwestern Medical Center, Dallas, TX



Dr. Ilya Bezprozvanny

Spinocerebellar ataxia type 2 (SCA2) is an autosomal dominant genetic neurodegenerative disorder caused by a polyglutamine (polyQ) expansion in ataxin-2 (Atxn2) protein. The reason for neuronal degeneration in SCA2 is not known. Our previous results suggested that neuronal cell death in SCA2 and other ataxias may be related to calcium overload and excitotoxicity. In the previous studies we also demonstrated that activators of small conductance calcium-activated potassium channels (SK channels) can stabilize abnormal firing pattern of neurons in SCA2 mice and rescue the pathology. In the present project we will expand on these studies and will attempt to identify the optimal molecular candidate from this class of compounds for clinical trial in ataxia patients. ❖

Pharmacological Activation of Autophagy to Alleviate Machado-Joseph Disease

By **Luis Pereira de Almeida, PhD**

University of Coimbra, Portugal



Dr. Luis Pereira de Almeida

Machado-Joseph disease (MJD) is a hereditary neurodegenerative disorder with severe clinical features, being the most common autosomal dominant spinocerebellar ataxia (SCA) worldwide. This disease is caused by the over repetition of the CAG trinucleotide in the ATXN3/MJD1 gene, which translates into an expanded polyglutamine tract within the affected protein ataxia-3 (Kawaguchi et al., 1994). The mutant ataxia-3 protein becomes prone to misfolding, accumulating as nuclear and intracellular inclusions, and acquires toxic properties which lead to neuronal dysfunction and cell death.

We recently showed that activating the lysosomal-macroautophagy pathway leads to elimination of mutant ataxin-3 and alleviation of neurodegeneration in MJD (Nascimento-Ferreira et al., *Brain* 2011; 2013).

Therefore, modulation of autophagy might be a strategy to attenuate the neuropathology of the disease. Recent evidence suggests that carbamazepine, an anticonvulsant and mood-stabilizing FDA-approved drug, enhances autophagy and clearance of aggregate-prone ►►

proteins in the liver (Hidvegi et al., 2010).

Given these evidences the main goal of this study is to investigate the beneficial effects of carbamazepine in the alleviation of Machado-Joseph disease, mediated by its autophagy-enhancing ability in a lentiviral and a transgenic mouse model of MJD (Nobrega et al., 2013; Nascimento-Ferreira et al., 2013; Simões et al., 2012). Based on our preliminary data we expect that carbamazepine will promote activation of autophagy resulting in decrease of mutant ataxin-3 aggregation, cell injury and degeneration in the mouse brain, providing a new therapeutic approach that can reach MJD patients, as well as other ataxias within a short time frame. ❖

Post-Doc Fellowship Awards

Dendritic Transport Dysfunction in a Novel *Drosophila* Model of SCA5

By Adam Avery, PhD

University of Minnesota, Minneapolis, MN

Spinocerebellar ataxia type 5 (SCA5) is a progressive neurodegenerative disorder that causes disability by loss of coordinated movements and slurred speech. Prominent neuronal targets in SCA5 pathogenesis are Purkinje cells of the cerebellum. In a healthy state cerebellar Purkinje cells extend elaborate dendritic arbors that are critical for proper function of neurons and the brain. In SCA5 pathogenesis, the dendritic arbors of Purkinje cells undergo dramatic atrophy. SCA5 stems from mutations in the SPTBN2 gene that encodes the protein β -III-Spectrin. I am interested in understanding the mechanisms by which β -Spectrin proteins carrying different SCA5 mutations induce SCA5 pathogenesis. In order to gain insight into the mechanisms of SCA5 pathogenesis, I developed a novel SCA5 disease model using the fruit fly *Drosophila melanogaster*, an organism in which sophisticated genetic studies can be easily performed. *Drosophila* contains a population of neurons, called dendritic arborization (da) neurons, which like Purkinje cells, extend elaborate dendritic arbors. Expression of SCA5 β -Spectrin mutant proteins in *Drosophila* da neurons causes dendritic arbor atrophy. My preliminary characterizations of this model together with previous

studies in my mentor's lab support the hypothesis that SCA5 pathogenesis is due to a deficiency in intracellular transport mediated by a motor protein complex termed Dynein and Dynactin. In this proposal I will combine genetic techniques using the novel SCA5 *Drosophila* da neuron model and in vitro methods to test the hypothesis that the SCA5 β -Spectrin mutations induce dendritic arbor atrophy by disrupting Dynein/ Dynactin function in dendritic transport of cargoes required for neuronal expansion and maintenance. This work will shed light on the protein interactions underlying the molecular mechanism of SCA5 pathogenesis. These studies will thus identify possible drug targets and inform the development of novel therapeutic strategies to mitigate the action of disease proteins. ❖



Dr. Adam Avery

CRISPR-Modified ATXN1 in SCA1 Patient-Derived iPS Cells

By *Nissa Mollema, PhD*

University of Minnesota, Minneapolis, MN



Dr. Nissa Mollema

Spinocerebellar Ataxia Type 1 (SCA1) is caused by genetic changes to the ATXN1 gene. ATXN1 has a repetitive stretch of DNA where a three-base “CAG” sequence is repeated over and over again. Normally, this region of DNA has 28 to 35 CAG’s interrupted with other DNA bases. In patients with SCA1, the interruption in the CAG repeat is lost and the CAG stretch expands. There is no cure or present treatment for the disorder. Our group used a variety of model systems to study SCA1 and understand the pathway to disease. Using humanized mouse model systems, we determined that the extended CAG repeat causes the ATXN1 protein to behave differently in the cell. Lowering the levels of the diseased ATXN1 protein may reduce the severity or halt the progression of SCA1.

Our lab identified several drug compounds that are capable of reducing the levels of ATXN1 protein. Drug compounds identified in our

previous studies need additional analysis before they may be considered for potential development into a treatment for SCA1. We wanted to examine the effects of these drugs in a patient-derived neuronal cell system, and to do that we turned to “induced Pluripotent Stem (iPS) cells.” iPS cells are often generated from adult patient skin samples: cells in the skin are collected and infected with a virus that reprograms the cells. iPS cells are unique because they can be prompted to become other cell types, such as neuronal cells. Neuronal cells generated from iPS cells are excellent model systems for studying disease.

We determined that ATXN1 is expressed in neuronal cells generated from SCA1 patient iPS cells. Most of the SCA1 patients in our studies have around 40 CAG repeats in their diseased copy of ATXN1 and 30 CAG repeats in their normal copy of ATXN1. The sizes of the normal and diseased ATXN1 proteins are too close to separate them using our available methods. In order to study the diseased ATXN1 protein in detail, we propose to increase the size of the protein using a genetic tag. Patient iPS cells with a tagged ATXN1 protein will allow us to screen drug compounds and identify differential effects on the diseased versus the normal ATXN1. Tagged iPS cells will also allow us to determine when ATXN1 turns “on” during the transition from iPS cell to neuron.

These studies are important because ATXN1 has not been studied in a patient-derived neuronal system. We will learn critical details about the function of ATXN1 in human neuronal cells, and be able to test potential therapeutic compounds to inhibit the detrimental activity of the diseased ATXN1. ❖

Deadline

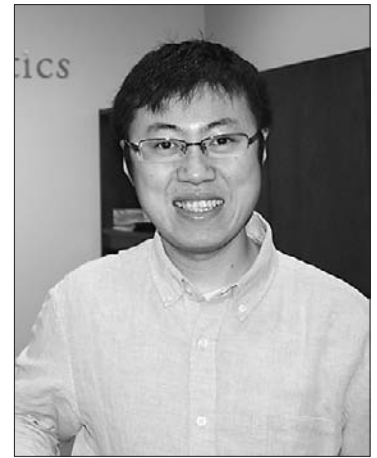
The deadline to submit materials for the summer issue of *Generations* is May 16. Submit content by e-mail to joan@ataxia.org or to the postal address listed on page 2.

5hmC-Mediated Epigenetic Modulation in rCGG-Mediated Neurodegeneration Associated with FXTAS

By **Bing Yao, PhD**

Emory University, Atlanta, GA

Fragile X-associated tremor/ataxia syndrome (FXTAS) is a late-onset neurodegenerative disorder in which patients carry excessive alleles of 55–200 CGG DNA nucleotide repeats in the FMR1 gene. The detailed molecular mechanisms underlying the development and progression of this disease are not clear. In the proposed study, we will utilize cutting-edge next generation high-throughput sequencing technology to systematically profile a specific DNA modification, termed 5-hydroxymethylcytosine (5hmC), using a FXTAS disease mouse model. It has been recently suggested that 5hmC plays critical roles in neuronal cells, and its aberrant distribution may be linked to neurodegeneration. We will profile 5hmC patterns and distribution genome-wide in Purkinje neurons from FXTAS mice. Purkinje neurons have been suggested to be the key loci for FXTAS pathogenesis. We will correlate the 5hmC alteration to gene expression change in Purkinje neurons and draw a direct link between this particular DNA modification and its impact on gene expression during FXTAS disease progression. We will also use genetic tools to knockout the ten-eleven translocation (TET) family of proteins, which is responsible for generating 5hmC on the genome, to investigate their roles in FXTAS pathogenesis. The proposed work, together with preliminary data, is completely novel and will open up a new avenue to facilitate our understanding of the contribution of altered 5hmC-mediated epigenetic modulation to the pathogenesis of FXTAS. Ultimately, this research may identify therapeutic interventions to alleviate or reverse the progression of neurological deficits in FXTAS patients or even prevent the FXTAS onset. ❖



Dr. Bing Yao

CoRDS Registry

Coordination of Rare Diseases
at Sanford

 **National Ataxia
Foundation**
www.ataxia.org

Everyone who has any form of ataxia or who is at risk for ataxia is encouraged to enroll in the CoRDS/NAF ataxia patient registry. To register in the CoRDS ataxia patient registry, go to www.ataxia.org and click on “**Ataxia Patient Registry**.” If you prefer to enroll by postal mail, please contact CoRDS personnel.

For more information on CoRDS and/or enrollment, visit www.sanfordresearch.org/cords or call (605) 312-6423. Thank you for participating in this important research tool.

Regulation of Organismal Proteostasis by Integrated Stress-Response Networks

By *Jian Li, PhD*

Northwestern University, Evanston, IL



Dr. Jian Li

Underlying the pathologies of late-onset neurodegenerative diseases such as multiple types of spinocerebellar ataxia (SCA) is the age-associated collapse of protein homeostasis (proteostasis). Many SCAs are polyglutamine diseases in which the disease-associated proteins contain polyglutamine expansions (polyQ) that are highly prone to aggregation. Stress response transcription factors have been identified as prominent suppressors of Ataxia-associated protein aggregation and toxicity. My research plan is to characterize the protective mechanisms mediated by factors controlling responses to different stresses (such as heat-shock, oxidative stress and metabolic stress) and understand how neurons sense the toxic aggregation and communicate with neighboring tissues to achieve an organismal protection. ❖

Studies of Brain and Behavior in Individuals with Premutations of the Fragile X gene (FMR1)

We currently are seeking individuals who carry a premutation allele of the Fragile X gene, FMR1. Some individuals with this premutation allele may show signs of Fragile X-associated Tremor/Ataxia Syndrome (FXTAS) which is characterized by motor and cognitive issues. This study aims to better understand these issues and their bases in the brain in individuals with FXTAS.

Eligible participants will be asked to complete the following:

- questions about family medical history and behavior
- tests of thinking abilities
- testing of sensory processing and movement control
- brain activity recording and imaging
- genetics testing

Testing will be conducted at the Center for Autism and Developmental Disabilities at UT Southwestern. Participants will be compensated for their time.

For more information, please contact us by phone at **214-648-5155** or by e-mail at **fragilex@utsouthwestern.edu**

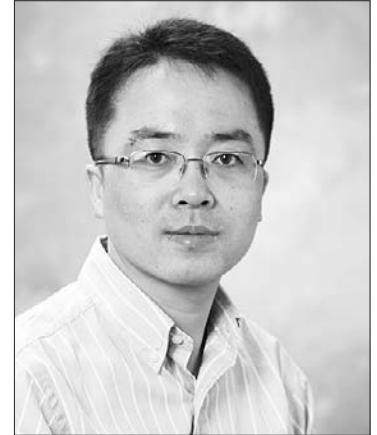
Young Investigator – SCA Research Awards

Molecular Pathogenesis Studies of Spinocerebellar Ataxia Type 1

By **Janghoo Lim, PhD**

Yale University, New Haven, CT

The human inherited cerebellar ataxias are a genetically heterogeneous but clinically similar group of disorders that share many neurological and pathological features, such as loss of balance and coordination, as well as cerebellar Purkinje cell degeneration. We have utilized spinocerebellar ataxia type 1 (SCA1) as a prototype of dominantly inherited cerebellar ataxias. By investigating the fundamental mechanisms of SCA1 pathogenesis, we hope to gain insight into the common key features of this and several other neurodegenerative diseases. SCA1 is caused by a polyglutamine expansion in the protein ATAXIN1 (ATXN1). Building on our studies of SCA1 and ATXN1, we have recently found that activation of Wnt- β -catenin signaling is significantly upregulated within the Purkinje cells in SCA1 transgenic mice. In this proposal, we will test the hypothesis that SCA1 activates Wnt- β -catenin signaling in the Purkinje cells, which leads to Purkinje cell degeneration in SCA1. To test this idea, we will perform a combination of biochemical and cell biological studies, as well as genetic studies in mice. We believe that this study will lead us to better understand the pathogenic mechanisms of SCA1 and several other inherited ataxias, which we hope will open the possibility of future therapies. ❖



Dr. Janghoo Lim

Analysis of Transcriptional Changes in Machado-Joseph Disease Using Isogenic Patient-derived Neurons

By **Philipp Koch, MD**

University of Bonn, Germany

Machado-Joseph Disease (Spinocerebellar Ataxia type 3) is an inherited and devastating neurodegenerative disorder clinically characterized by progressive ataxia. It is caused by a CAG repeat expansion mutation in the MJD1 gene, which results in an expanded stretch of the amino acid glutamine in the protein ataxin-3 (ATXN3). Currently, no effective treatment or cure is

known for this disorder. During the course of the disease, the protein ATXN3 accumulates and forms large aggregates in the nuclei of affected neurons. These aggregates are believed to induce molecular changes in the neurons, which eventually result in neurodegeneration. However, the

Continued on page 20

Analysis of Transcriptional Changes ...
Continued from page 19

exact mechanism for how expanded ATXN3 induces neuronal degeneration remains elusive.



Dr. Philipp Koch

In this context, ATXN3 has been shown to bind directly to specific genes and to modify gene expression. Recently, a new

In the past, researchers concentrated on the idea, that ATXN3 induces cell death via a toxic “gain of function.” Other lines of evidence suggest that the expansion of ATXN3 interferes with the physiological function of

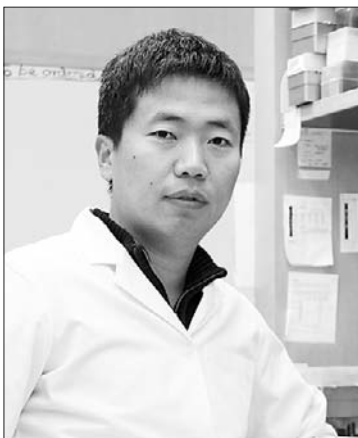
technique has been described which enables the generation of patient specific neurons in cell culture by reprogramming and subsequent differentiation of skin cells derived from affected patients. This technique allows for the first time to investigate changes in gene expression directly in living human neurons. However, considering the genetic variations among human individuals, the best control for analysis of differential gene expression is to use the same, isogenic cell without the disease-causing genetic mutation. In the project, we will generate gene-corrected cells from the patient cells and identify genes bound and regulated by ATXN3.

The direct comparison of the neuronal gene expression patterns in neurons harboring the mutation with those not harboring the mutation will allow for the first time to identify changes in gene expression directly in human neuronal cells and may help to develop potential new and effective strategies to treat the disease. ❖

The Role of Ataxin-2 in the Regulation of Circadian Behavior and Sleep in Mice

By Yong Zhang, PhD

University of Massachusetts Medical School, Worcester, MA



Dr. Yong Zhang

tensing and other movement incoordination symptoms, as well as sleep disorders. Sleep is reg-

ulated by circadian clocks, which drive 24-hour physiological and behavioral rhythms in human and most animals. Interestingly, we have found that Ataxin-2 is a crucial regulator of circadian clocks in the fruit fly *Drosophila melanogaster* (Zhang et al., 2013, *Science*), which is a powerful model organism to understand circadian rhythms in animals, including humans. Since Ataxin-2 proteins and the mechanisms controlling circadian behavior are well conserved between fruit flies and humans, we hypothesize that Ataxin-2 also regulates circadian behavior and sleep in mammals.

Spinocerebellar Ataxia Type 2 (SCA2) is an inherited neurodegenerative disease caused by an elongated stretch of glutamine amino acids in the Ataxin-2 protein. SCA2 patients usually have uncontrolled muscle

I will focus on two aims. In my first aim, I will study the role of Ataxin-2 in the control of ►►

circadian behavior using Ataxin-2 deficiency mice. In addition, I will determine whether a gene closely related to Ataxin-2, named Ataxin-2 like, also contributes to circadian rhythms. In my second aim, I will study sleep in the Ataxin-2 deficiency mice. Since sleep symptoms have an earlier onset than movement disorders in SCA2 patients, I will test sleep behavior in young

Ataxin-2 deficiency mice. I will then determine in which brain regions Ataxin-2 regulates sleep. In conclusion, my work should elucidate the role of the Ataxin-2 family of proteins in the control of circadian behavior and sleep in mice to establish a direct link between circadian dysfunction, sleep disturbances and SCA2 disease. ❖

Pathogenesis and Disease Course of Spinocerebellar Ataxia Type 14 in a Mouse Model

By Dong Hui Chen, MD, PhD

University of Washington, Seattle, WA

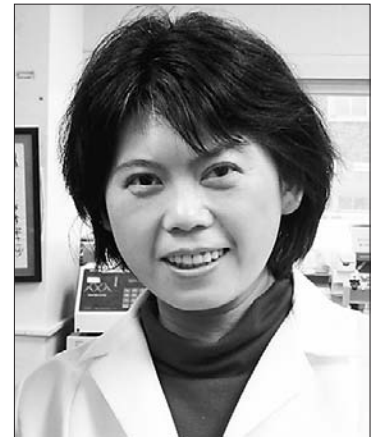
Spinocerebellar ataxia (SCA) is a group of hereditary neurologic disorders caused by degeneration or malfunction of the cerebellum and the spinal cord. Poor coordination of movement is a common symptom of all the SCAs.

At least 30 genetically different forms of autosomal dominant SCA have been described, including SCA14. SCA14 shares many clinical and pathologic features with other autosomal dominant SCAs, but may have additional phenotypes of myoclonus (spasmodic jerky contraction of muscles) and a variety of cognitive impairments.

We previously discovered that SCA14 is caused by mutations in the protein kinase C gamma (PKCg) gene. The gene belongs to a serine/threonine kinase family and plays a role in diverse processes such as signal transduction, cell proliferation and differentiation. The mechanism of how the mutations in PKCg lead to neuronal death and how the disease develops is unknown. An animal model of SCA14 would be invaluable for such investigations. We have generated mouse lines that have normal and mutated forms of the SCA14 gene. The mice with the mutant human

PKCg develop abnormalities in their brains, including aggregations of the mutant protein, but do not have overt neurologic phenotypes, for example ataxic gait.

To produce mouse models that develop the same incoordination and neuronal loss as seen in people with SCA14 we have altered the relative number of copies of the human and mouse PKCg genes. We will study them both behaviorally and pathologically to determine the onset of brain changes, the onset and progression of neurologic impairments, and whether these abnormalities are temporally related. We will investigate the functions and effects of the mutant proteins with the long-range goal of eventually discovering therapeutic interventions that may slow down or reverse the neurologic deterioration. ❖



Dr. Dong Hui Chen

Research Grants

The Effect of Cerebellar Abnormality on Hearing in Individuals with Spinocerebellar Ataxia (SCA) and Friedreich Ataxia (FRDA)

By Gary Rance, PhD

University of Melbourne, Parkville, Victoria, Australia



Dr. Gary Rance

The cerebellum plays a significant role in the timing and coordination of our movements. Recently the cerebellum has also been shown to play a part in the timing and coordination of language and thinking. Some inherited conditions such as Friedreich ataxia (FRDA) and Spinocerebellar ataxia type 1 (SCA1) directly impact on the cerebellum as apparent by slow, uncoordinated movement and occasional slowed thinking processes.

We have previously shown that hearing, perception of speech, and hence, general communication are also be affected by FRDA and SCA1. The mechanisms underlying these changes are, however, yet to be determined. The first aim of this study is to establish the nature and extent of hearing impairment in groups of people with FRDA and SCA1. A sub-group will then undergo imaging of the brain in order to identify changes in the structure, function or connectivity of the cerebellum that may contribute to hearing changes. The results of this study will provide information that can be used as a basis for intervention in individuals with FRDA and SCA as well as improving our general understanding of the function of the cerebellum and the role it plays in the processing of sound. ❖

PATIENTS with EARLY SYMPTOMS of FRIEDREICH'S ATAXIA

age 10 and above needed for an MRI study to evaluate the chemistry and connectivity of the brain and spinal cord in Friedreich's ataxia

at the Center for Magnetic Resonance Research at University of Minnesota

You will lie in the scanner for ~1.5 hour while listening to the music of your choice. Reimbursement for travel expenses is available and you will be compensated for your time.

Please note that we cannot scan you if you have Harrington rods, and we cannot scan people with diabetes at this time.

If you are interested or have questions, please call

Diane Hutter @ (612) 625-2350 or e-mail hutte019@umn.edu.



Episodic Ataxias: Looking for New Genes by Next Generation Sequencing Approach

By *Elide Mantuano, Doctor of Biology*

National Research Council of Italy, Institute of Translational Pharmacology, Rome, Italy

The Episodic Ataxias (EAs) is a group of rare autosomal dominant neurodegenerative diseases with heterogeneity both in clinical symptoms (migraine, vertigo, ataxia, epilepsy) and genetic background. The disease respond to acetazolamide treatment, although adverse effects are reported in literature and alternative treatment seems to be needed.

So far, seven distinct genetic form of EA (EA1-7) and four causative genes (KCN1A-CACNA1A-CACNB4 and SLC1A3) are identified. Among these, EA1 (KCN1A) and EA2 (CACNA1A) have been well documented in multiple families accounting for 20-30% of the mutations associated to EA, while mutations in the other known genes are rare, accounting only for about 2-3% of the cases. This picture suggests a relevant part of genetic heterogeneity for the disease and leaves many patients waiting for molecular diagnosis.

At present, search for new genes causing EA phenotype achieved only sporadic positive results. Recently two new genes (PRRT2 and DARS2) have been associated to EA phenotype, although only occasionally.

The main goal of the project is devoted to identify new genes involved in the episodic ataxia (EA) syndrome.

To attain this goal, we propose to use next-generation sequencing in a series of extended multi-generational EA pedigrees referred to our lab, in which mutations in known genes were excluded.

We will select two patients for family to perform a whole exome sequencing. The first criteria to select the pairs of relatives will be based on the number of meiosis that separate them, in order to decrease the number of the genetic variants to be validate, and to increase the value of those that will be identified in each pairs (identical by descend, IBD).

The achievement of the specific aim will allow to:

- increase the rate of molecularly diagnosed EA patients,
- expand the comprehension of molecular mechanism of the disease, and
- indicate new perspective to search for therapies. ❖



Dr. Elide Mantuano

GoodSearch Is Good for NAF

Did you know that donating money to the National Ataxia Foundation can be as easy as changing your Internet search engine?

GoodSearch.com donates 50 percent of its revenue to the charities designated by its users.

To get started, simply go to the site's home page and follow the easy steps to make NAF your charity of choice. Then use GoodSearch as you would any other search engine. You'll make a difference in the lives of those with ataxia!

National Ataxia Database

By **Susan Perlman, MD**

University of California – Los Angeles



Dr. Susan Perlman

The Natural History Study of and Genetic Modifiers in Spinocerebellar Ataxias (*ClinicalTrials.gov* Identifier: NCT01060371), under the direction of Dr. Tetsuo Ashizawa at the University of Florida – Gainesville, continues to recruit subjects and monitor changes in their neurologic examinations, despite being currently unfunded. The participating 12 sites see and examine subjects and enter data into the National Ataxia Database (housed at UCLA under the direction of Dr. Jeanette Papp in the Department of Genetics). The Database has served as a valuable repository for data collected in this important collaborative project, which has enrolled close to 400 individuals with Spinocerebellar Ataxia types 1, 2, 3, and 6. Our first summary paper “Clinical characteristics of patients with spinocerebellar ataxias 1, 2, 3 and 6 in the U.S.; a prospective observational study” has been published in the

Orphanet Journal of Rare Diseases this year.

Dr. Ashizawa has submitted an NIH U54 grant for the upcoming year. Should it be funded, the NIH does provide database access which we will be required to use (the National Ataxia Database serving as backup and continuing to maintain all data past and future for this study – and possibly other studies in the future). ❖

Unraveling Expression Profiles in Human SCA3 Brain Pathology: A Pilot Study

By **Henry Houlden, PhD**

Institute of Neurology, University College London, United Kingdom



Dr. Henry Houlden

Spinocerebellar ataxia type 3 (SCA3), also known as Machado-Joseph disease, is a neurological disorder affecting the brain and the spinal cord of patients. The first symptoms usually start around the age of 40 years and include difficulty walking, but in some cases the disease appears much earlier (four years) or much later (70 years). Other complications, such as problems with vision, speech, and swallowing, among others, may also occur and range in severity. The disease is progressive and many patients ultimately need a wheelchair or become bedridden, as there is no treatment currently available. This is a genetic disorder that can be transmitted from one generation to another. The genetic defect that will determine if a person will have the disease is already known, and having just one copy of this defect is enough to cause the disease. ►►

However, it cannot predict when the disease will start or how severe it will be. Thus, there must be other defects contributing to the differences observed between patients, and these need to be investigated.

We propose a study using post-mortem brain tissues of SCA3 patients (the damaged part of the body in SCA3). We will investigate how genes make products that are needed for cells, and which variations those products show that can be related to the disease. This study will help us to decipher a series of chemical reactions that occur in the brains of patients but not in the brains of people not suffering from SCA3. A better understanding of those chemical reactions may help the future development of therapy. Besides being useful for SCA3 patients, such knowledge may also be important for other patients, namely those suffering from other spinocerebellar ataxias.

We will produce valuable data that we will make publicly available, so that other researchers can also use them, in order to facilitate the understanding of this disease. ❖

The Role of Ataxin-2 in Machado-Joseph Disease: A Molecular Therapy Approach with Viral Vectors

By **Clevio Nobrega, PhD**

University of Coimbra, Portugal

Machado-Joseph disease (MJD) or spinocerebellar ataxia type 3 (SCA3) is a dominantly inherited disorder originally described in people of Portuguese descent, and associated with the expansion of a (CAG)_n tract in the coding region of the causative gene *MJD1*. The abnormal over-repetition of the CAG-trinucleotide is translated into an expanded polyglutamine tract within ataxin-3, a protein whose physiological function has recently been linked to ubiquitin-mediated proteolysis, resulting in severe clinical features and leading to death. There is no therapy for this fatal disease.

Ataxin-2 (ATAX2) is a protein whose function has been linked to translational regulation. There is evidence that ATAX2 interacts with poly-a-binding protein (PABPC1, which is required for poly (A) shortening and translation initiation) in polyribosomes, thus reducing protein translation. Recent studies suggested a role of ATAX2 in other neurodegenerative disorders such as SCA1 or Amyotrophic lateral sclerosis. In MJD several evidences also point to a potential link with ATAX2, as this protein is detected in mutant ataxin-3 nuclear inclusions. Nevertheless, it is not known how these two proteins interact and lead to disease pathology. In this project we hypothesize that the trapping of ATAX2 in MJD, reduces soluble ATAX2 levels freeing PABPC1 to an overactive protein translation, particularly of ataxin-3, which may contribute decisively to the pathology.

This study will contribute to sort out the pathogenic mechanism of MJD, to validate the therapeutic target – ATAX2-mediated translational inhibition – and hopefully for the development of a therapy for MJD. ❖



Dr. Clevio Nobrega

Development of a Brief Ataxia Rating Scale for Children (BARS-c)

By **Jeremy Schmahmann, MD**

Massachusetts General Hospital, Boston, MA



Dr. Jeremy Schmahmann

Children develop cerebellar ataxia as a consequence of a range of diseases including tumors, infections, and degenerative and inherited disorders. The ability to measure the severity of ataxia is a key component of the clinical evaluation, and it is also important in detecting improvements or worsening of the condition with time, and with therapeutic intervention currently available and on the horizon.

The rating scales of ataxia currently in use have been developed in adults. These include the original and modified versions of the International Cooperative Ataxia Rating Scale (ICARS), the Scale for Assessment and Rating of Ataxia, the Friedreich's Ataxia Rating Scale, and the Brief Ataxia Rating Scale (BARS). The problem with using rating scales in

children that have been developed and normed on adults is that the nervous system of the young child is still developing, and age-dependent achievement of fine motor skills, coordination, concentration, and muscle force can influence the interpretation of rating scale scores in a manner unrelated to the actual disease itself, or the response of the illness to therapy.

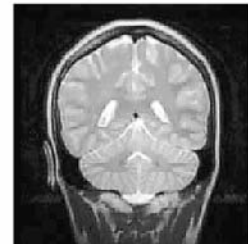
Indeed, recent studies show that the ICARS scores improve with age in the young, healthy child, because the very young child cannot complete all the components of the larger battery of tests, and the young child with an immature nervous system does not yet have a fully coordinated motor system. These age dependent findings are consistent with neurodevelopmental studies showing that complex motor behaviors continue to improve through puberty. Using measures of ataxia in children that have been developed in adults is therefore not optimal.

In this study we will develop a children's BARS (BARS-c) that will include pediatric reference values, and will be quick, easily applicable, and reliable. There is an urgent need for such a scale, and this study aims to fill that need. ❖

❖



**PATIENTS with
SCA1, SCA2, SCA3, SCA6 and
MSA-C
needed for an MRI study**



at the University of Minnesota, Minneapolis

Travel expenses reimbursed.

Contact: Diane Hutter

(612) 625-2350

hutte019@umn.edu

Transplantation of Neural Stem Cells as a New Therapeutic Strategy for Machado-Joseph Disease

By *Liliana Simões Mendonça, PhD*
University of Coimbra, Portugal

Machado-Joseph disease (MJD) is a progressive neurodegenerative disease, originally described in people of Portuguese ancestry, and caused by a mutation on the ATXN3 gene translating into a mutant ataxin-3 protein. Mutant ataxin-3 protein is toxic causing neuronal dysfunction and degeneration in specific brain regions and leading to motor symptoms (incoordination, spasticity, scanning speech, dysphagia and ocular complaints) and non-motor symptoms (minor cognitive and behavioral abnormalities, verbal and memory deficits and chronic pain), with large variability in severity and type of symptoms between patients. There is no specific and effective treatment able to delay and stop the progression of this disease. However, in symptomatic patients, already with extensive neuronal loss, cell replacement strategies, such as the transplantation of neural stem cells (NSC), are very promising therapeutic strategies holding the promise of neuroregeneration. In fact, transplantation of NSC has been successfully used in animal models of several neurodegenerative diseases. It has been demonstrated that after brain transplantation these cells exert a substantial beneficial and therapeutic effect through differentiation into neural cells, production of neurotrophic factors and reduction of neuroinflammation which rescue/improve the damaged neuronal network. Moreover, our preliminary data suggest that NSC transplantation decreases MJD-associated neuropathology and motor coordination impairments in MJD transgenic mice. Thus, we speculate that NSC transplantation can be used for neuroregeneration of brain lesions of MJD patients promoting functional recovery. Therefore,

NSC transplantation is a promising approach that could be brought to patients in a short time frame. Thus, this work aims to evaluate if the transplantation of NSC into the cerebellum of MJD transgenic mice leads to a decrease in MJD-associated neuropathology and motor coordination impairments. Moreover, we aim to assess if the transplantation of NSC leads to modifications in the levels of neurotrophic factors and inflammatory mediators. ❖



**Dr. Liliana Simões
Mendonça**

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iSearchiGive.com is totally free, with no hidden fees and provides valuable support for the important work of NAF.

Please sign up today and indicate that the National Ataxia Foundation is your favorite cause.

Silencing Machado Joseph-Disease/ Spinocerebellar Ataxia Type 3 Through the Systemic Route

By *Rui Jorge Nobre, PhD*
University of Coimbra, Portugal

Machado-Joseph disease (MJD) or spinocerebellar ataxia type 3 is a dominantly-inherited neurodegenerative disease initially described in Portugal. It affects mostly the cerebellum and is the most common dominantly inherited ataxia worldwide. MJD is associated with the expansion of a (CAG)_n tract in the coding region of the causative gene MJD1. The abnormal over-repetition of the CAG trinucleotide is translated into an expanded polyglutamine (polyQ) tract within



Dr. Rui Jorge Nobre

ataxin-3, conferring toxic properties to this protein, resulting in severe clinical features and leading to death. There is no therapy for this fatal disease. Our group has recently showed that intracranial injection of lentiviral vectors encoding silencing sequences targeting ataxin-3 significantly decreased the severity of the neuropathological abnormalities in rodent models of MJD (Alves et al., 2008, 2010, Nóbrega et al., 2013). Despite these recent successes of siRNA-based treatments, several hurdles and limitations need to be addressed. The aforementioned experiments involved craniotomy, general anesthesia and in situ injection of viral vectors in the brain parenchyma. Therefore, there is a need to develop alternative systems, able to cross the blood-brain barrier (BBB) by less invasive routes of administration,

such as injection in the systemic circulation, and that may diffuse over larger areas of the brain.

Adeno-associated virus serotype 9 (AAV9) injected intravenously have recently been shown to bypass the BBB and efficiently transduce neurons and astrocytes in the central nervous system (CNS) of mice, cats and non-human primates (Duque et al., 2009; Foust et al., 2009; Foust et al., 2010). Based on that, we propose to develop an AAV9-mediated system that will allow delivery of RNA interference-based treatments to the MJD brain by intravenous injection. This strategy will allow a less invasive procedure without the need for a craniotomy, general anesthesia and the risks associated with an injection in the brain parenchyma. It is also expected that this system restrict the transgene expression to the CNS, reducing possible toxic effects in peripheral organs.

In the present project, the silencing sequences that we have previously shown to efficiently abrogate MJD (Alves et al., 2008, 2010; Nóbrega et al., 2013) will be cloned into AAV9 vectors. This therapeutic approach will be validated at the neuropathological and behavioral level in a transgenic mouse model of MJD already available in our animal facility.

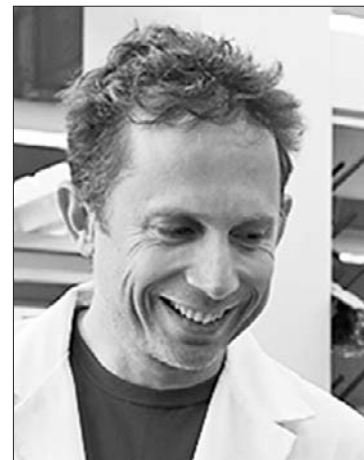
It is anticipated that a single systemic administration of the AAV9 system will be able to transverse the BBB, to transduce the brain and to permanently silence mutant ataxin-3 and the resultant neuropathology. This project is expected to enable the development of a non-invasive gene delivery system for the treatment of MJD and may provide a proof of concept of gene silencing by a non-invasive route for therapy of other ataxias. ❖

Annotation and Analysis of Pre-mRNA Splicing Elements in Ataxia Genes

By *William G. Fairbrother, PhD*
Brown University, Providence, RI

The ataxic disorders reflect a heterogeneous group of progressive cerebellar degenerative disorders characterized by a broad set of symptoms relating to a lack of coordination of voluntary movements. More than 50 different types of hereditary degenerative ataxias have been identified, each with a distinct genetic cause. Nonetheless, for a number of patients with symptoms of an ataxic disorder, the causal gene has not been identified. We are entering the age of precision medicine in which genomes will be sequenced as part of routine medical care. The techniques and methods of precision medicine are especially appropriate for the study of rare disorders, such as hereditary ataxias. However, sequencing studies conducted as part of precision medicine typically indicate hundreds of genomic positions at which the patient's sequence differs from the human reference sequence. Thus, determining which mutation or mutations are in fact causal becomes a daunting task. A huge bottleneck in workflow lies in the analysis of these variants. Many clinical groups may limit their analyses to mutations that are likely to lead to defects in the encoded protein. However, results from our lab suggest that at least one-third of hereditary disease mutations affect genetic processes that occur before the protein is generated. One such process is the "splicing" of pieces of genetic information that will encode for a protein. Our lab has expertise in analyzing the effects of genetic variants on the splicing process. Our methods include the use of a combination of computational and experimental approaches. For example, high-throughput splicing assays are used to map gene elements involved in splicing and to identify mutations that might affect splicing. This mapping information is then

incorporated into mutation analysis programs used to analyze sequence data and predict the effects of genetic variants on the processing of a gene. We propose to apply these methods to the analysis of genes considered causal of hereditary ataxic disorders. Furthermore, results from our studies will be integrated into a software package we intend to develop for predicting causal variants of ataxic disorders based on deep sequencing data. This tool will aid physicians in classifying genetic variants as deleterious or benign, thereby potentially facilitating identification of causal mutations and earlier implementation of appropriate treatment regimes. ❖



Dr. William G. Fairbrother

IAAD Apparel

Create ataxia awareness by wearing an "I am the Strength behind ataxia" T-shirt or other ataxia awareness apparel. You can order ataxia awareness apparel online by visiting NAF's web site, www.ataxia.org, and clicking on "Store," or by using the order form on page 7 of this issue.

International Ataxia Awareness Day (IAAD) on September 25 is just around the corner, so order your ataxia awareness apparel today!

IPSC-derived Neurons from Friedreich's Ataxia (FA) Patients as a Model to Characterize Pathological Mechanisms and Devise Neuroprotective Strategies

By Franca Codazzi, PhD

San Raffaele Scientific Institute and University, Milano, Italy



Dr. Franca Codazzi

Friedreich's ataxia (FA) is a rare autosomal recessive hereditary disorder, characterized by progressive neurological disability, hypertrophic cardiomyopathy and increased risk of diabetes mellitus. FA is caused by a reduced expression of frataxin, a mitochondrial protein involved in important biochemical pathways, including the respiratory chain. As a consequence, frataxin deficit results in severe alterations that greatly affect the activity and integrity of mitochondria and thus not only specific cellular functions but also the ability of the cell to sustain oxidative insults. The molecular mechanisms responsible for neurodegeneration, in both the central and the peripheral nervous systems, are still poorly known, mainly because of the absence of appropriate human neuronal models. Aim of this study is to characterize neuronal cells obtained by the genetic reprogramming (performed in the lab of Professor M. Pandolfo) of human skin fibroblasts obtained from patients or healthy subjects. The use of a neuronal model of human origin is expected to represent an important step forward, with respect to the use of cell lines or murine animal models, in the understanding of the molecular basis of FA. We plan to investigate a number of potential alterations, including cellular iron redistribution, cellular/mitochondrial susceptibility to oxidative stress, mitochondrial changes (loss of membrane potential, morphological alteration etc.) and modifications in neuronal network connectivity. As a further outcome, this study will provide a thorough characterization and validation of a human neuronal model that has all the potential to be used for the screening of molecules with neuroprotective potential in FA. ❖

International Ataxia Awareness Day (IAAD)

Thursday, September 25, 2014

"International Ataxia Awareness Day" (IAAD) is an international effort from Ataxia Organizations around the world to dedicate September 25 as International Ataxia Awareness Day. IAAD has grown over the years, with more ideas and more people getting involved. To find out how you can get involved, please download the IAAD Kit on NAF's website, www.ataxia.org, on the IAAD page under the Event Section. On the site you will also find all the IAAD events near you on the Event Calendar under the Event Section as they become available. Let NAF know about your IAAD event by contacting Lori Shogren at lori@ataxia.org or (763) 553-0020.



Young Investigator Awards

Genomic and Epigenomic Analyses in an Induced Pluripotent Stem Cell Model of ADCA-DN

By *Alexander E. Urban, PhD*
Stanford University, Palo Alto, CA

Autosomal dominant cerebellar ataxia, deafness and narcolepsy (ADCA-DN) is a disease of the nerve system characterized by late onset (in patients that are 30–40 years old). The symptoms are ataxia (loss of control over movements), deafness, narcolepsy-cataplexy (sleep attacks), several additional problems with the nerve system and dementia.

We have found the gene mutation that causes this disease. The mutation is in a gene called DNMT1 which is a gene of great importance to the normal development and functioning of all organs in the human body. However, it is unknown “how” this mutation leads to the disease ADCA-DN, i.e. how does the mutated gene behave differently inside the nerve cells compared to the same gene without mutation?

Here we propose a series of experiments to lay the foundation of understanding the actions of the mutated DNMT1 gene.

First, we will create stem cell like cells from skin cells of the patients. These cells will automatically carry the mutated DNMT1 gene. We will then turn these stem cell like cells into neurons and during this process we will study the actions of the mutated DNMT1 gene.

DNMT1 is a gene that is responsible for leaving control marks on the DNA sequence. We will study how these marks change in the cells with the mutated DNMT1 gene. And we will analyze how these changes affect other genes in their activity and also whether the DNA in each cell in general becomes destabilized as a result of the DNMT1 mutation. ❖



Dr. Alexander E. Urban



SEEKING PATIENTS WITH SCA (1, 2, 3, 6, 7)
FOR A CLINICAL TRIAL USING TRANSCRANIAL MAGNETIC STIMULATION
TO IMPROVE GAIT, POSTURE, AND MOBILITY

at the Berenson-Allen Center for Non-invasive Brain Stimulation at
Beth Israel Deaconess Medical Center, Boston MA

You will be asked to come in for daily treatments (M-F) for 4 weeks, 30 minutes a session.

You will be compensated for your time.

If you are interested or would like more information, please contact

Natasha Atkinson at 617-667-0258 or email natkinso@bidmc.harvard.edu



Cellular Signaling Mechanisms Underlying EA1 in Cerebellum

By Jason Christie, PhD

Max Planck Florida Institute, Jupiter, FL



Dr. Jason Christie

Episodic ataxia type-1 (EA1) is neurological disorder characterized by recurrent bouts of uncontrolled motor movements induced by physical or emotional stress. Loss of motor control in EA1 patients results from mutations in a potassium channel subtype whose role in the cerebellum is not fully understood. Treatment of EA1 stands to benefit if the underlying link between mutations in these potassium channels and the resulting functional deficit in the cerebellum can be identified. In this proposal, we aim to resolve the locus and functional consequence of EA1 potassium channel mutations in signaling properties of cerebellar neurons. ❖

Social Security Made Easier

The following was posted February 2014 at www.socialsecurity.gov/thirdparty/whatsnew.

I have great news! Social Security has made it even easier for people to apply online for disability benefits. We have joined the Social Security disability benefits application, and the adult disability report in “electronic matrimony.” It used to be that you had to complete both of these forms separately when applying for disability benefits. Our new streamlined process puts both forms together all in one place. Please refer to www.socialsecurity.gov/applyfordisability for more information.

Please be advised that this application is for Social Security Disability Insurance benefits only. To find out about applying for Supplemental Security Income benefits, please visit

www.socialsecurity.gov/pgm/ssi.htm.

Beginning August 2014, we will no longer issue Social Security number printouts in our field offices. Individuals, who need proof of their Social Security number and cannot find their card, will need to apply for a replacement card.

In addition, beginning October 2014, our field offices will stop providing benefit verification letters, except in emergency situations. Benefit verifications are available online, and can be obtained anytime by registering for a “my Social Security” account located at: www.socialsecurity.gov/myaccount, or requested through our national toll-free number: 1-800-772-1213.

Stay connected with Social Security. ❖

Remembering NAF in Your Will

Each year we are reminded by the kindness of others who have named the National Ataxia Foundation as a beneficiary in their wills. These planned gifts have made a profound impact on

NAF’s ability to fund important research and programs and are felt for years after they are gone.

Please consider naming NAF in your will. Your gift will truly impact the lives of those with ataxia.

From the Desk of the **Executive Director**

By Michael Parent, NAF Executive Director

The National Ataxia Foundation's 57th Annual Membership Meeting, "Betting On Ataxia Research," concluded on Sunday, March 23. The meeting was the second largest attended NAF annual membership meeting, with over 630 registrants. A special thank you to the Western Region host support groups; Orange County Area Ataxia Support Groups, Los Angeles Area Ataxia Support Group, Northern California Area Ataxia Support Group, Phoenix Area Ataxia Support Group, Willamette Valley Ataxia Support Group, Utah Ataxia Support Group and the Seattle Area Ataxia Support Group for bringing us this exceptional conference. Thank you!

We are also truly grateful to all who supported the conference including our dedicated volunteers, board and staff, our exceptional speakers, our generous donors and sponsors, exhibitors, and outstanding MCs. A special thank you also goes to the ataxia support groups outside the Western Region who also supported the 2014 Annual Membership Meeting including the Detroit Area Ataxia Support Group, the Greater Atlanta Ataxia Support Group, the Central Massachusetts Ataxia Support Group, and the Northeast Florida Ataxia Support Group. A special thank you to The FA Project for their continued support of the AMM travel grant program. Through their support and others, NAF was able to provide 19 travel grants to persons with ataxia to help them attend this year's conference.

The NAF annual membership meetings represent the largest gathering of the ataxia community in the world. This year's conference attendees came from 45 states and nine other

countries, including Australia, Canada, France, Germany, the United Kingdom, Denmark, Mexico, Japan, and Ireland. We thank all of you for being part of this important ataxia conference.

The annual membership meetings offer the latest information on ataxia research, clinical trials, and important topics which impact the ataxia community. This year's conference had 234 "First Timers." Many commented that the conference offered a real sense of community and family. Along with the "First Timers," NAF

saw one couple who has attended 30 consecutive annual membership meetings. Congratulations and thank you for your support.

We are truly grateful to all who contributed to the planning of this conference, and to all who participated, contributed, and attended. A special thank you also to the AMM Co-Chairs, Arnie Gruetzmacher and Camille Daglio. Thank you!

Plans have already begun for the 58th NAF Annual Membership Meeting which will be held on

March 13-15, 2015 in Denver, Colorado at the Sheraton Denver Downtown. The 2015 conference will be hosted by the North Central Region, which includes the states of Colorado, Illinois, Iowa, Kansas, Minnesota, Missouri, Montana, Nebraska, North Dakota, South Dakota, Wisconsin, and Wyoming. We look forward to working with ataxia support groups in these areas. More details about the 2015 NAF Annual Membership Meeting will be available in future issues of *Generations*, on NAF's web site, www.ataxia.org, social media, and in E-Blasts. We look forward in seeing you next



Michael Parent

*From the Desk of the Executive Director
Continued from page 33*

year in Colorado.

Just prior to the NAF Annual Membership Meeting was the 5th Ataxia Investigators Meeting (AIM). This year's AIM had 150 participants from 16 countries. The AIM 2014 was the largest attended AIM and focused on bringing in world leading ataxia scientists as well as the most promising young investigators from around the world to help accelerate world-wide ataxia research, foster collaboration, and facilitate the push towards therapies.

A special thank you to all the AIM 2014

sponsors, who included An Anonymous Donor, ApoPharma, Ataxia UK, Ataxia Ireland, Ataxion, Athena Diagnostics Inc., A-T Children's Project, BioMarin Pharmaceutical Inc., Bob Allison Ataxia Research Center, Friedreich's Ataxia Research Alliance (FARA), Gordon and Marilyn Macklin Foundation, National Institute of Neurological Disorders and Stroke (NINDS), and The Foundation of Ataxia Charlevoix-Saguenay.

Thank you also to the AIM 2014 Co-Directors, Dr. Harry T. Orr and Dr. David Lynch, as well as the AIM 2014 Steering Committee members Dr. Henry Paulson, Dr. Helene Puccio, and Dr. Robert Wilson. ❖

Mark Breland

October 10, 1932 – February 3, 2014

Submitted by Camille Daglio and Peter Hanks on behalf of the Mississippi Chapter

The Mississippi Chapter recently lost one of its founding members, Mark Breland.

Beginning in December 1988 until May 1991, Mark and his wife Linda started and ran a bingo operation that donated over \$100,000 to ataxia research. Mark worked tirelessly transporting a four-wheeler and selling raffle tickets for seven years at wildlife and gun shows to help the Mississippi Chapter make a \$50,000 donation at the 50th anniversary of the National Ataxia Foundation's annual membership meeting. Several years he hosted our annual picnic at his home and usually presented a new fundraising idea each year.

Even though he did not have ataxia and his children were not at risk, he kept his entire extended family involved with all the events of

the Chapter.

Mark was also a member of the Board of Directors of the National Ataxia Foundation from 1986-2003. He was very instrumental in the financial stability of the Foundation in the early years.

Mark served in the Air Force from 1952-1972. He served his country honorably and he was part of and participated in The Korean Conflict.

Up until the end of his life, Mark was a hero and a champion for both the Mississippi Chapter and the National Ataxia Foundation. He even participated in the drawing of our fund raising raffle

in November before his death in February.

We have lost a dear friend whom we will miss greatly. ❖



Mark Breland

Auburn Chef Earns Food Network Nod

Originally posted on the Auburn, MA Daily website on February 4

Local chef and owner of Simply Delicious Catering, John Mauro, was recently chosen as a finalist for the popular cable show, Next Food Network Star.

Mauro was nominated by colleagues, unbeknownst to him. “There was a casting call in Rhode Island” says Mauro. “Denise [Kapulka] basically picked me up and said ‘Here’s what you’re doing today!’”

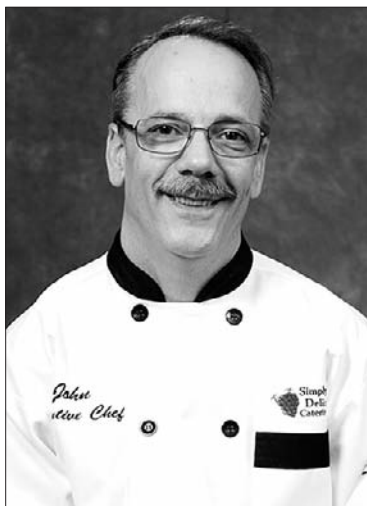
Mauro described the initial interview as “a lot of questions about anything except food,” which he apparently aced, as he was chosen to move on to the telephone interview portion of the selection process. “The producers based on New York called, I had a phone interview with them, the next thing I know I was invited to New York City” says Mauro.

The purpose of the NYC trip was to run through “mock shows” where the contestants simulate actual filming conditions. Unfortunately for Mauro, who lives with ataxia, a degenerative neurological condition, the television set proved to be challenging. “The sets were hard to navigate” says Mauro. “There were wires on the floor, things changed all the time, I couldn’t foresee what would be where and I couldn’t just move things.”

Ultimately, Mauro contacted Food Network producers and gratefully bowed out of Next Food Network Star, but countered with his own idea. “I proposed to producers a cooking show featuring chefs with disabilities” says Mauro. “I suggested they could focus on large format

displays.” By way of example, Mauro points out the enormous gingerbread Christmas tree he and his crew built. “It was huge, it lit up, played music and it was all made of edible material.”

Mauro’s idea is that the show could work with corporate sponsors and have the chefs build these large format displays to coincide with events with which the Food Network or show sponsors are involved. “They [displays] could be for trade shows, sporting events, large national events like that” Mauro adds.



John Mauro

The show would also add diversity to the network’s programming. “There isn’t really anything else like it on any cable network” says Mauro. “This would also be a great way to bring more awareness to the National Ataxia Foundation” he adds.

Mauro has been deeply involved with the Ataxia Awareness since being diagnosed with the disorder in 2007. Instead of sitting by waiting for the disorder to progress, Mauro became an advocate. He started the “Ride For John – Walk and Roll for Ataxia” fundraiser in 2007. Mauro was recently nominated to sit on the National Ataxia Foundation board of directors, and expects to be officially appointed at the NAF Annual Conference in Las Vegas in late March.

Currently, Mauro’s television concept has passed the producers’ initial muster, and has moved to phase two. “They [the producers] are figuring out the show formatting and trying

Continued on page 36

Auburn Chef Earns Food Network Nod
Continued from page 35

to line up sponsors” says Mauro. Any money Mauro earns from the show, should it become a reality, he plans to donate 50% back to the National Ataxia Foundation.

On the topic of a “missed opportunity” with Next Food Network Star and the prospect of becoming a celebrity chef with this new concept, Mauro states, “This is really about driving awareness and fundraising for research. That’s the most important thing to me right now.” ❖

New Method of ‘Live Looping’ Helps Musician Play On

Submitted by Ian C. Bouras

I am a guitarist and music producer from New York who was diagnosed with ataxia. I thought my guitar-playing days were over.

I originally lived in California when I was diagnosed in 2010. It was not confirmed until I moved back to New York in late 2011 that I took it more seriously. Unfortunately I do not know what type of ataxia I have. There is an ataxia support group that meets every two months in New York and I attend sometimes, but I was shocked that over half the group does not know what type they have.

As my condition progressed, it seemed that I might not be able to play the music that I loved much longer. It started to affect my guitar playing in 2011, so I had figure out a different approach to playing. That is why I started experimenting with a system called live looping.

Live looping means that I can attach a guitar synthesizer to my guitar and play every element from a live band, including bass and piano as well as the guitar. I run everything through a delay pedal, a phase shifter, and I have at least one other pedal. Then I manipulate the sounds, live, so that

they loop together to create a full band. In essence, I am the composer, musician and audio technician all at the same time, something I know that I am fortunate enough to be able to do. I can still record, but playing live is an issue now because my fingers won’t go where I want them to.

I am not thrilled about having ataxia, but I am happy to be forging my own path, and am the first person, that I know of, to be doing what I’m doing. Because of my ataxia, playing with a band is out of the question, which is why I’m working on the live looping, it allows me to create, and perform live. People do live looping, but not like this. As people say, necessity is the mother of invention, so maybe that’s why I’m the first to do what I do, no one else has had to do it!

I want people to see, that although not the ideal situation, it can push us to create in new ways. Ataxia makes us extraordinary people since we have to come up with new ways to do things, that no one ever thought of.

All of Ian’s music, as well as some free downloads, can be accessed on his website, <http://www.sdmprecords.com>. ❖



Ian C. Bouras

I'll Do the Driving, Thank You

Submitted by Jason Wolfer

Ataxia showed up in my life like a loud, obnoxious party crasher. It barged its way in, interrupting the peaceful proceedings, and appearing to be already half-drunk, like it was just gracing my festivities after having previously been at a different event. It was an uninvited presence, not just as a guest that can eventually be pushed out the door, but it brought with it all its baggage and I knew then that this was a permanent situation. I was not consulted, was never asked if it would be okay if ataxia came over, and not involved in any long-range planning meetings on the subject.

No, my life was not perfect, but it was, however, without serious physical challenges. I never had great balance or the adventurous spirit that drives many people. But I was happy. I was married (just recently celebrated our 25th anniversary), and had three great kids. And then suddenly one day, there it was, ataxia. The general atmosphere of the whole party changed at that moment because the baggage that ataxia brought with it was all negative. The diagnosis was negative, the outlook was negative, and to my recollection, the diagnosing neurologist was negative. When it showed up in my life it also brought lots of stress, anxieties, and doubts. Mostly because I had never heard of ataxia, didn't know anyone with ataxia, and didn't foresee a very bright future.

What I have come to learn in the 11 years since that day is that a diagnosis is not the end of the journey, but rather the beginning. I continued to live my life and did not give up. The neurologist made some predictions based on statistics about my future that I refused to allow free reign in my

life, one of which I am convinced would have come to pass had I simply used their words as an excuse and given up. I have also reached out and through a local support group and social sites on the Internet, made some very good friendships and been able to find a deep level of support.

My encouragement for a person who is just beginning to go down this path is to realize you are not alone on this sometimes overwhelming, dark, and scary journey. Reach out. Find a support group or individual you can share with. Find support by joining NAF and participating with the community on Facebook, or by starting your own support group if there isn't one in your area. You can also access through the NAF a list of people you can correspond with

as a pen pal.

The point is, ataxia will not stop you if you do not let it. We all get depressed from time to time but the key is to not stay there. My friends, do not let yourselves be driven by ataxia but instead YOU do the driving. ❖



Jason Wolfer

Sign up for E-mail Blasts

E-mail Blasts from the National Ataxia Foundation are sent out periodically on ataxia research, events and other timely issues of interest regarding ataxia.

Please e-mail your e-mail address to joan@ataxia.org so you don't miss out on receiving important information.

Our Story in a Nutshell

Submitted by Cathy DeCrescenzo

Joe and I have been married 37 years ... that in itself is an amazing feat! Our blessings are bountiful. We have two beautiful daughters and memories that are endless. We found joy in raising our girls and were very protective of them, as any parent would be. Before our eyes they matured into strong, confident young women. They are grown now with families of their own. We welcomed two wonderful men, our “sons”-in-law, into our family and we are the doting grandparents of three very special granddaughters; our oldest is 10, and the twins are four.

The diagnosis of Spinocerebellar Ataxia-2, or SCA2, came eight years ago for my husband Joe. With slurred speech, our physician ordered an MRI for fear of a possible stroke, but that MRI led to testing for ataxia, which was confirmed. A few years later, our youngest daughter tested positive for SCA-2; she is asymptomatic thus far. The most difficult aspect of our journey is knowing our child is affected with a progressive, degenerative disorder, and, unlike when she was a little girl with a skinned knee, there is nothing we can do to fix it – it is heart wrenching. Though the diagnosis was upsetting, we continue on. In fact, it has made us stronger and changed our outlook on life – it has given us the opportunity to be advocates on behalf of all who are affected by this disorder.

Our daughter is a model of strength and courage, knowing she eventually will be symptomatic; she fights every day to stay active and strong. After extensive genetic counseling, they

consulted a fertility facility, and after months of testing, injections, and emotional and physical impact, her eggs were retrieved and tested, and two disease-free eggs were re-implanted, thus, the twins were born, and are ataxia-free.

Though life can be challenging, some days more than others, we haven't stopped enjoying life. We have our ups and downs but, we push through the obstacles, as best we can. At times, patience is hard to come by – deep breaths help, and occasionally we have to address the issues at hand. We cannot sugar-coat the fact that, in the future, daily life will become more difficult, but, in the meantime, there is no stopping us! A dream of ours was to see Italy, we made that happen. I encourage you to live your dreams, DO NOT put them off.



Cathy and Joe DeCrescenzo

Joe has participated in numerous research studies, both at Johns Hopkins and the University of Minnesota. It doesn't hurt – it can only help in finding a cure or treatments! We also participated on a panel at a Nurses Conference helping the

nurses understand the challenges of our daily lives. They were grateful for our insight and thanked us for being open and honest.

Searching for a neurologist who you are comfortable with can be cumbersome. In our experience, this took time. After two neurologists, the third was the charm, and we've been with him for five years with no intention to “ever” leave. You will know when you “click” with your doctor and are able to trust his or her recommendations/decisions. Do not be afraid to ask questions or address your concerns. ►►

Education is crucial. You can go the NAF Website where an abundance of information is available. The NAF Annual Membership Meeting (AMM) is an amazing event where you'll learn from the researchers and professionals. (See page 5 of this issue for information on the 2015 AMM.)

Staying active socially and physically is freeing. We have a full calendar and an abundance of dear friends ... we're always on the go! Joe joined a gym where he works out two-three times a week, utilizing the treadmill and leg/arm strengthening machines, and he walks with a cane ... imagine that!

Our family, friends, our faith, our pastor, and our church family offer a wealth of support – we are very fortunate. They are our strength to continue on. NAF is a wonderful source of support, information, and knowledge. I recommend that you make use of what is at your fingertips. If your area has a Support Group, attend it. If not, start one. Joe started the DE Support Group by contacting NAF who helped us through the process. Our Support Group organized a Walk for Ataxia event two years ago raising over \$21,000 – truly overwhelming! The end result far outweighed the sweat and tears.

Our family is extremely proud of Joe and his accomplishments. He is our inspiration. Joe is the NAF's Delaware Ambassador, Chair of the DE Support Group, he is a member of the NAF Board of Directors, and is a recipient of the "I Am the Strength behind Ataxia" award.

As a caregiver, I have found something I love ... country line dancing! It is great exercise and a terrific outlet. You, as caregivers, need to take care of yourselves and find something you are passionate about. Ask for help, if necessary.

If I may speak to our friends with ataxia for a moment – your loved ones/caregivers do the best they can. It can be very stressful for the caregiver, so I ask you to treat them with respect, love, and kindness, while you are on this journey together.

OUR MESSAGE:

- Make every minute count.
- Count your blessings – Be thankful.
- Cherish your family and friends – Let them know they are appreciated.
- Do NOT loathe in self-pity – It will get you nowhere.
- Stay active – You will feel more empowered.
- Laugh every day – It will brighten your outlook and lift your spirit.
- Accept what you cannot change – But, work to find ways to make it easier.
- Live your dreams now – Do not wait.
- Join or start a Support Group – GET INVOLVED.
- Volunteer within the ataxia community – Ataxia awareness is crucial.
- Scope out a Neurologist you are comfortable with – Communication is essential.
- Educate yourself – Knowledge helps us move forward.
- Caregivers – Take care of yourselves.
- Friends with Ataxia – Respect your loved ones/caregivers – Work together.
- Attend the Annual Membership Meeting (AMM) – you will leave there a different person.
- Be an inspiration to others.

Together, we can do this! We hope and pray a cure is found soon ... In the meantime, we will keep our dignity and spirit...we will move forward and celebrate each and every day.

We hope our story helps even one person realize they are not alone. We, the ataxia community, are a strong, vibrant group – we are here for one another through the more challenging times but also the good times.

If you would like more information please contact Cathy at cdecrenzenzo@comcast.net. ❖

My Journey

Submitted by Keith Pate

At the age of 29, in December 2010, I suffered a mid-line cerebellum infarct (stroke), which is quite rare. That morning I woke in a daze. My kids were in the bed with me and my daughter recognized something was wrong and was able to get my brother. He and his wife called an ambulance right away.

I was certainly not prepared for what would come in the following days. While at the North Mississippi Medical Center in Tupelo, I was diagnosed with a hole in my heart that doctors were able to repair. In most people the hole closes at about age 10, but mine remained opened and allowed a blood clot to pass to my cerebellum causing the stroke. During this time, I was heavily sedated and remembered very little. I only know what people have told me. I was placed in the stroke unit and had to have emergency brain surgery to release the pressure that was building and remove the part of the cerebellum that was damaged in the stroke.

While in the hospital, I went from 215 to 160 pounds. I had a feeding and breathing tube. Once I was stabilized, my dad and I flew to Atlanta, where, I was admitted for about three months and assigned a team of doctors, therapists, and nurses. At the Shepard Center we got to work on rehabbing.

After the stroke, with the help of others, I had to learn to do everything again. Things that were once reactive were impossible. My mind seemed to be doing well but my body couldn't do things it once could. I developed tremors that I'm still fighting. The tremors greatly impact my speech, eyesight, etc.

Before I left Atlanta, I had my breathing and

feeding tubes removed. I also had a central line (an IV that went straight to my heart). They removed that too, and I was like a new man. It had been about four months since my stroke at this point.

Afterwards, I was transported via ambulance to the Charlie Norwood VA Hospital in Augusta, Georgia. This took about two hours and I would reside there for the next 14 months as a patient.

I was angry upon my arrival. I did not know why I had to be moved and I took my anger out on some of the staff and my family. They wanted

to put me in a covered bed, and I was not having it. I was placed on the ADRU (active duty rehab unit) ward. Once I met most of my new doctors, therapists, etc., I calmed down. It turned out that this unit was the only one of its kind in the country and the doctors and staff I worked with were professional and very helpful in my recovery. We wore our military uniforms most of the time and had therapy everyday 8-4 p.m. and weekends off.

The next several months were filled with rehab, doctor appointments, and preparing my medical packet for the military board. I was assigned a primary physician, psychiatrist, physician assistant, physical therapist, occupational therapist, speech therapist, vision therapist, recreational therapist, counselor and a nurse case manager. They kept me busy and they all did a wonderful job.

What can I say about the other patients I met? I learned so much from each one of them. Each one of them personifies the values this country was built on. I am thankful to have met each of them. I also learned no matter how bad my ►►



Keith Pate in Iraq

situation is it could always be worse. I met patients from all walks of life: from Hawaii, from New York all the way to Puerto Rico. Some came from Afghanistan and some from Iraq. All were military, active duty, and we all had something in common: we were struggling against something we had very little control over. Some had to make very important decisions at a very young age. Whatever their reasons for being there, most had good attitudes. This certainly was very hard to do considering what some were facing.

Once a week, we'd go with the recreation therapist on outings to events and military programs. Sometimes it would be fun things like rock climbing or kayaking. We bonded during these trips and became like a family.

In the meantime, my doctors and other staff were collecting paperwork to discharge me from the army. The military was all I had ever done, and I didn't know if I would ever be able to do anything else. By May 2012, I was able to be discharged from the facility in Georgia and move back to Mississippi with my brother. This meant I was able to be there that fall for my kids' first day of school, which I was grateful for.

Two years and one day after my stroke, December 2012, I was medically retired from the military. At that time, I was in a wheelchair, then a rolling walker, and now I am currently on forearm crutches. I have double vision and a severe speech impediment. I have come a long way and still have a long way to go. I thank God I'm still here.

October 2013, I visited the Mayo clinic in Jacksonville, Florida to see a neurologist. There I learned I was suffering from ataxia and not tremors.

I know without a doubt that I would not be here if it was not for the many prayers and support from friends, family, staff, and people unknown – so, thanks to all.

If you would like more information please contact Keith at keithpate81@yahoo.com. ❖

Don't Bite the Hand that Feeds You!

(especially if it's your own)

— Submitted by Pete Meyerhoff —

Your fingers pick up a morsel
of food, say it's a grape.

You open your mouth and
deposit the grape inside.

After releasing it, you withdraw
your fingers and bite.

Simple?

Not if you have ataxia!

You are likely to bite your own fingers.

Funny?

Not really, considering that your jaw muscles
are among the most powerful in your body.

This action requires a lot of coordination

Which as an ataxian you don't have.

Worth a laugh?

Why not!

New Medication Trial for FRDA

A trial of an investigational medication (VP 20629) for patients with Friedreich Ataxia (FRDA) is being conducted.

This is Phase 1 trial of an antioxidant medication.

A total of 56 subjects will be enrolled in this nation-wide trial.

Eligibility:

- Ages Eligible for Study: 18 to 45 years
- Genders Eligible for Study: Both

For more information, please follow this link: <http://clinicaltrials.gov/ct2/show/NCT01898884?term=Friedreich+ataxia&rank=2>

Walk n' Roll for Ataxia

The ultimate finish line ... a cure for ataxia

What is Walk n' Roll for Ataxia?

The Walk n' Roll for Ataxia program is the National Ataxia Foundation's largest national grassroots fundraising event. Since its inception in 2007, Walk n' Roll for Ataxia has raised more than \$900,000 thanks to the support and tireless commitment of walkers, rollers, runners, volunteers, donors, and sponsors.



Why Walk or Roll?

Thousands of families, friends, co-workers, neighbors, and communities come together each year to support NAF's fight to improve the lives of people affected by ataxia and their families.

How Can I Participate?

For more information, or to start a Walk n' Roll in your community, please contact Lori Shogren, NAF Special Projects Coordinator, at (763) 553-0020 or lori@ataxia.org.

Current 2014 Walk n' Roll Events:

- Liverpool, NY – May 24
Marc Alessi – pianoman345@hotmail.com
- Denver, CO – September 7
Charlotte DePew – cldepew77@comcast.net
- St. Louis Park, MN – September 13
Terry Sweeney – mnataxiawalk@yahoo.com
- Auburn, MA – September 20
John Mauro – johnmauro62@me.com
- Detroit, MI – September 20
Tanta Tunstull – tinyt48221@yahoo.com
- Duluth, GA – September 20
Greg Rooks – atlantaataxia@gmail.com
- Jacksonville, FL – September 20
Cory Hannan – coryhannan@hotmail.com
- Lafayette, CA – September 27
Joanne Loveland – joaneloveland@gmail.com
- Long Beach, CA – September 27
Daniel Navar – danieln27@gmail.com
- San Diego, CA – September 27
Earl McLaughlin – emclough@cox.net

For more information please visit
http://www.ataxia.org/events/walk_n_roll.aspx ❖



Chapter and Support Group News

from Around the Country

Willamette Valley Support Group

Submitted by Jason Wolfer

The Oregon support group which meets in Albany, had their annual Christmas party on December 18. The group, led by Ivy Stilwell, celebrated by having a potluck and a white elephant gift exchange. Most of the regular attendees were present.



Members of the Willamette Valley (Oregon) Support Group at their Christmas party

Greater Atlanta Support Group Meeting

Submitted by Dave Zilles

The Greater Atlanta Ataxia Support group held their annual Holiday Party at Grace Life Church in Woodstock, GA. The support group provided turkey, ham, plates, napkins and eating utensils. Members brought their favorite side dishes, desserts and drinks to share. Fun was had by all. Santa and Mrs. Claus appeared thanks to the LaRoches.

Everyone that could was asked to bring a small gift worth between \$5-15 and we held a gift exchange that required individuals to pick numbers and then select a

gift which or they could steal a gift that had already been open. Everyone had a great time.

This was a new location this year because we had out grown the previous place. Several came dressed in holiday attire as you can see in this picture.



Stephen Cheatwood

Tri-State Ataxia Support Group Meeting

Submitted by Kathy Gingerelli

Here we go again ... 2014! A great time was had by all at our annual "potluck" dinner to kick-off the New Year on January 9. The night started with introductions all around. I talked about how positive it is to become a member of the NAF, signing up for the new ataxia patient registry (CoRDS) and about NAF's Annual Membership Meeting "Betting on Ataxia Research."



Members of the Tri-State Ataxia Support Group at their annual potluck dinner

Chapter and Support Group News
Continued from page 43

Then we got down to the fun part of the night ... the food! Overall the night was great and a terrific way to start off another year.

Arizona Ataxia Support Group Meeting

Submitted by Mary Fuchs

Back in November, the Arizona Ataxia Support Group had a fundraiser with Popcornopolis. Matia Troutt, Mary Fuchs' granddaughter, was our helper/mascot. The fundraiser was easy and very successful.



Matia Troutt at the Popcornopolis fundraiser held by the Arizona Ataxia Support Group

Submitted by David Garcia

The Arizona Ataxia Support Group gathered on February 8. Dr. Larry Schut joined us as our featured speaker as he has for the past seven years for our February meetings. It helps that he and his lovely wife, Loretta, spend a portion of their winter in the wonderful Arizona warmth. This year, Dr. Schut shared with our group the latest information on ataxia related research and answered questions our members had regarding



Members of the Arizona Ataxia Support Group at their February meeting

ataxia and living with ataxia. This meeting was one of our most well attended with many new members joining us. Because there were so many new faces, we started the meeting going around the room with introductions.

In addition to Dr. Larry Schut's talks and a Q & A session, we devoted time during the meeting discussing the upcoming NAF AMM meeting, which our support group is co-sponsoring with the western region support groups. There have been so many activities coordinated by members of our group related to fundraising for the AMM, and so it was good to talk about the successes and the various silent auction items donated. It was also fun to learn who was planning on attending the AMM with the "old-timers" sharing about the many educational and social opportunities we will have at the AMM with the new members, who are looking forward to it. We closed the meeting with a group photo. Afterwards, many of our members got-together at a nearby restaurant to continue the sharing and socializing over a meal.

Our next meeting is tentatively scheduled for May. Please contact Mary, Angela or Rita for details.

Denver Area Ataxia Support Group

Submitted by Charlotte DePew

At our quarterly meeting on January 18, 35 members were impressed and very grateful for the manner in which the topics of genetics and genetic counseling were presented. The speakers were from Athena Diagnostics: Khalida Liaquat, Genetic Counselor and Rosi Rosenberg, Account Executive – Colorado. One member ►►

stated, "This is the first time I actually understood a genetics talk." Others agreed and many questions, to include cost and payment, were answered.

Updated information was presented on planning the 2014 Run, Walk 'n Roll scheduled for September 7. Two additional volunteers were added to the committee.

Northeast Florida Ataxia Support Group

Submitted by Mac Kelso

The Northeast Florida Ataxia Support Group met at Baptist South Hospital on February 22 and had 14 attendees. Group Leader Cory Hannan opened the meeting by announcing that he will be taking a four-month sabbatical from April through July. He plans to go to Argentina taking time to reflect on his life and to volunteer for the Association civil de Ataxias de Argentina (ATARS), an organization similar to NAF. He recapped the group's successes in 2013 starting with Mayor Brown's proclamation declaring September National Ataxia Awareness Month in Jacksonville; then he touted the huge success of the group's first Rock, Roll n' Bowl at Bowl America Mandarin in support of IAAD, donating over \$4,500 to NAF; and finally mentioned the contributions made to the million dollar grant given to NAF. Cory laid out plans for our group events in 2014, starting with Rare Disease Day and ending with the Rock, Roll n' Bowl on September 20 at Bowl America Mandarin.

Moving on, Cory presented some interesting topics in the news, citing sources from *Generations*: A new medication that slows the progression of Friedrick Ataxia that has been approved by FDA and SCA 7 clusters in humans have been found in fruit flies, meaning researcher will start using fruit flies to discovery a cure.

Cory's final comments before closing were directed toward ataxians and caregivers. He reiterated the purpose of the group is to support each other during difficult times, so you don't feel so isolated and alone. He also stated support

does not have to be related to just this group, it can be with anyone, in any group; that one special person or persons you may connect with for your support.

In closing, the group decided to have the off-site dining out on May 24 at Corky Bell's in Palatka in lieu of the regular scheduled meeting. Our next regular scheduled meeting will be at Baptist South on August 23.



Members of the Northeast Florida Ataxia Support Group at their February meeting

Happy Hoosiers Ataxia Support Group

Submitted by Cheri Bearman

We had a very nice time together at Joan's lovely home in Columbia City. Two of Joan's daughters prepared a yummy brunch and her husband Carl and her son Bill helped serve and host. Joan's youngest sister, Donna, Pam's husband, Don, my oldest sister, Sandy, who is blind and has ataxia, attended.

We enjoyed brunch together and then had a casual meeting, sharing general info about ourselves, our families, and our experiences thus far with ataxia.

I shared some TV exercise programs on PBS I do each morning. I highly recommended movement every day and these programs are excellent to keep us moving – they can be adapted to be done sitting on a chair: 7:30 a.m.

Chapter and Support Group News
Continued from page 45

“Body Electric” with Margaret Richards (muscle and bone strengthening, non-aerobic); 8 a.m. “Functional Fitness” with Suzanne Andrews (focusses on a specific topic each day); and 8:30 a.m. “Sit and Be Fit” with Mary Ann Wilson, RN.

I also shared a few quotes from a presentation I attended last year at the NAF Annual Membership Meeting entitled “Cultivating Well-Being.”

Before closing our meeting, we talked briefly about meeting twice a year. I will plan our next meeting for sometime this fall.



Attending the Happy Hoosiers first meeting were (left to right) Cheri Bearman, Sandy Miller, Joan Baermann and Pam Bishop

Rhode Island Ataxia Support Group

Submitted by John Mauro

Rhode Island’s support group held its first meeting on Saturday, March 29. Anabela has been working hard getting the word out on the new group. There were 28 members that showed up. four of them from other groups.

One of the highlights was the Girl Scouts and the Robotic Club showed up with 40,000 tabs from cans they had been saving for the group. I looked around at the members and they just loved it and felt welcomed in the community!

The group had many great talks and found many new friends. Anabela took charge of the

group. The group voiced a strong opinion that they would like to meet more than every three months, wanted to get together and even meet for pizza. I will be working with the group over the next six months. At this time the New England Chapter has 134 members; two years ago we had maybe 40. We are working on getting a resolution for the state of Rhode Island in place for September. Great job Anabela!



Girl Scouts present some of the 40,000 tabs from cans at the Rhode Island Ataxia Support Group’s first meeting on March 29

Rare Disease Day Event

Submitted by Cory Hannan

The NE Florida Support Group staffed an information table at the Rare Disease Day event on February 28. There were a total of six different organizations representing their disease, which allowed us to network for our Walk Roll n’ Bowl.

The garage sale to benefit NAF on March 1 in honor of RDD raised \$465. ❖



This Rare Disease Day Event raised nearly \$500 to benefit NAF

NAF Directory of Chapters, Support Groups and Ambassadors

The National Ataxia Foundation has a large network of volunteers who serve as support group leaders, chapter presidents, and ambassadors for our organization. These volunteers help identify important local resources and professional care for people with ataxia and their families.

If you or a family member or friend has been newly diagnosed with ataxia, please contact the NAF leader nearest you. If there is not a group in your area, we encourage you to visit our online social networks. You may also consider starting a support group in your area or becoming an NAF ambassador. If you are interested in these volunteer positions please contact Lori Shogren at lori@ataxia.org or (763) 553-0020.

The use of these names and contact information for any purpose other than requesting information regarding NAF or joining a chapter or support group is strictly prohibited. Thank you.

Social Networks

NAF BULLETIN BOARD

Moderator – Atilla and Bear

www.ataxia.org/forum/toast.asp

NAF CHAT ROOM

Moderator – Della (ddpokernut@yahoo.com)

www.ataxia.org/connect/chat-rooms.aspx

NAF FACEBOOK GROUP

www.facebook.com/group.php?gid=93226257641

NAF FACEBOOK FANS

www.facebook.com/lshogren?ref=profile#!/pages/National-Ataxia-Foundation/227766109304

NAF YOUTUBE CHANNEL

www.youtube.com/user/NatlAtaxiaFound?feature=mhum

Chapters, Support Groups and Ambassadors

– ALABAMA –

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Continued on page 48

*NAF Directory**Continued from page 47*

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— NEW JERSEY —

NEW JERSEY SUPPORT GROUP LEADER

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— NEW YORK —

CENTRAL NEW YORK SUPPORT GROUP LEADER

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— NORTH CAROLINA —

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NAF Directory

Continued from page 49

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— OHIO —

GREATER CINCINNATI AREA SUPPORT GROUP LEADERS**Jennifer Mueller**

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— OREGON —

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Clinical Research Training Fellowship in Ataxia

Co-sponsored by the American Brain
Foundation and the National Ataxia Foundation

**Padmaja Vittal, MD, MS**

Rush University Medical Center

*The Role of Antisense FMR1 in the
Development of Fragile X-associated
Tremor/Ataxia Syndrome*

Mentor: Deborah Hall, MD, PhD

Watch for her Research Summary in
the Summer issue of *Generations*.

— PENNSYLVANIA —

CENTRAL PA SUPPORT GROUP LEADER**Christina Rakshys**

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— RHODE ISLAND —

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— SOUTH CAROLINA —

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— TENNESSEE —

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— TEXAS —

NORTH TEXAS SUPPORT GROUP LEADER**David Henry Jr.**

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— UTAH —**UTAH SUPPORT GROUP LEADERS****Jenny Durrant**

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E-mail: Lisa.ord@hsc.utah.eduwww.ataxia.org/chapters/Utah/default.aspx**— VIRGINIA —****CHESAPEAKE CHAPTER PRESIDENT****Carolyn Davis**

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International Support Groups & Ambassadors

— CANADA —**OTTAWA SUPPORT GROUP LEADER****Prentis Clairmont - Ontario**

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E-mail: sam_ataxiaindia@yahoo.comsamag.india@gmail.comwww.ataxia.org/chapters/Chandu/default.aspxPlease visit our website: www.ataxia.in<http://seekamiracleataxiagroupindia-samagindia.webs.com> ❖

Caring for a Seriously Ill Child

When a child is diagnosed with a serious illness, it can be difficult for the entire family. It's important that your child, your family, and you get the support and care you need during this challenging time. A special type of care called palliative care can help.

Palliative care can ease a child's pain, help manage other symptoms, and provide important emotional support to the child and family. Research has shown that pediatric palliative care services may also improve overall satisfaction with care for patients and their families. Yet many health care providers hesitate to recommend palliative care for young patients, and parents and caregivers are often unaware of its benefits.

"Initiating palliative care conversations is often hard for both providers and families, especially in the pediatric setting," says Dr. Patricia A. Grady, director of NIH's National Institute of Nursing Research. While it may not be an easy conversation, she says, palliative care can greatly improve a patient's experience.

NIH's Palliative Care: Conversations Matter website provides information for health care providers, patients, and their families. The materials, including online videos, emphasize that palliative care works along with other treatments to enhance quality of life for children of any age living with a range of serious illnesses – not only at the end of life.

Visit Palliative Care: Conversations Matter or call (301) 496-0207 to learn more.

This article was originally printed, February 2014, in the NIH News in Health, a monthly newsletter from the National Institute of Health, part of the U.S. Department of Health and Human Services.

To subscribe, please visit <http://newsinhealth.nih.gov/subscribe>.

Calendar of Events

The most current event information is available on the NAF website, www.ataxia.org.

SUPPORT GROUP MEETINGS

– Saturday, May 3, 2014 –

Greater Atlanta Ataxia Support Group Meeting

Time: 1 p.m.

Location: Emory Center for Rehabilitation Medicine, 1441 Clifton Rd., Room 101, Atlanta, GA

Details: For more information call (404) 822-7451 or atlantaataxia@gmail.com.

Denver Area Ataxia Support Group Meeting

Time: 1 – 3:30 p.m.

Location: The Colorado Neurological Institute, 701 E. Hampden Ave., Suite 415. Doors lock at 2 p.m. No potluck meeting.

Details: For more information contact Charlotte DePew at (720) 379-6887 or cldepew77@comcast.net.

– Thursday, May 8, 2014 –

Tri-State Ataxia Support Group Meeting

Time: 6:30 – 8:30 p.m.

Location: Bethel Israel Medical Center, Phillips Ambulatory Care Center (PACC), Second Floor Conference Room, 10 Union Square East, New York, NY 10003

Details: For more information contact Denise Mitchell at markmeghan2@gmail.com or Kathy Gingerelli at kgingerelli@msn.com.

– Saturday, May 10, 2014 –

Central MN Ataxia Support Group Meeting

Time: 9:45 – 11:45 a.m.

Location: Kimball State Bank of St. Augusta, 24952 County Rd. 7, St. Cloud, MN, in the Board Room

Details: For more information contact Marsha Binnebose at (320) 248-9851 or mbinnebose@hotmail.com.

– Wednesday, May 14, 2014 –

Willamette Valley Ataxia Support Group Meeting

Time: 11:30 a.m. – 1 p.m.

Location: Albany General Hospital, 1046 6th Ave. SW, Albany, OR 97321

Details: For more information contact Ivy Stilwell at (541) 812-4162 or at istilwell@samhealth.org.

– Saturday, May 17, 2014 –

Twin Cities Ataxia Support Group Meeting

Time: 10 a.m. on the third Saturday of every month (approximately two hours)

Location: Langton Place in Roseville at 1910 W. County Rd. D, Roseville, MN 55112

Details: For additional information contact Lenore Healey Schultz at (612) 724-3784 or cshultz.lenore@yahoo.com.

– Tuesday, May 20, 2014 –

Cleveland Area Ataxia Support Group Meeting

Time: 6:30 p.m.

Location: Pharma-Snow Library, 2121 Snow Rd., Parma, OH 44134

Details: For additional information contact Carmen Pieragastini at (216) 272-5588 or willowpier@roadrunner.com.

– Saturday, May 24, 2014 –

Alabama Ataxia Support Group Social

Time: To be announced

Location: To be announced

Details: For more information contact Becky Donnelly at (205) 987-2883 or donnelly6132b@aol.com.

New Hampshire Ataxia Support Group Meeting

Time: 10 a.m. – noon

Location: Hannafords at the Bedford Shopping Mall, 5 Colby Ct., Bedford, NH 03110, (603) 625-5431

Details: For more information contact Jill & Ken Porter at (603) 626-0129 or jilleporter@comcast.net.

– Saturday, June 7, 2014 –

Greater Atlanta Ataxia Support Group Spring Picnic

Time: 1 p.m.

Location: Lanier Park, Lake Lanier, Buford, GA

Details: For more information call (404) 822-7451 or atlantaataxia@gmail.com.

– Wednesday, June 11, 2014 –

Willamette Valley Ataxia Support Group Meeting

Time: 11:30 a.m. – 1 p.m.

Location: Albany General Hospital, 1046 Sixth Ave. SW, Albany, OR 97321

Details: For more information contact Ivy Stilwell at (541) 812-4162 or at istilwell@samhealth.org. ▶▶

Saturday, June 14, 2014**Central MN Ataxia Support Group Meeting****Time:** 9:45 – 11:45 a.m.**Location:** Kimball State Bank of St. Augusta, 24952 County Rd. 7, St. Cloud, MN 56301, in the Board Room**Details:** For more information contact Marsha Binnebose at (320) 248-9851 or mbinnebose@hotmail.com.**Kansas City Ataxia Support Group Meeting****Time:** 2 – 4 p.m.**Location:** Northeast Library, 6000 Wilson Rd., Kansas City, MO**Details:** For more information contact Lois Goodman at (816) 257-2428 or Jim Clark at (816) 468-7260 or clarkstone9348@sbcglobal.net.**– Saturday, June 21, 2014 –****Northeast Florida Ataxia Support Group Luncheon****Time:** To be announced**Location:** Corky Bells (Palatka)**Details:** For more information contact Cory Hanna at (904) 314-2061 or coryhannan@hotmail.com.**Twin Cities Ataxia Support Group Meeting****Time:** 10 a.m. on the third Saturday of every month**Location:** Langton Place in Roseville at 1910 W. County Rd. D, Roseville, MN 55112**Details:** For additional information contact Lenore Healey Schultz at (612) 724-3784 or cschultz.lenore@yahoo.com.**– Saturday, June 28, 2014 –****New Hampshire Ataxia Support Group Meeting****Time:** 10 a.m. – noon**Location:** Hannafords at the Bedford Shopping Mall, 5 Colby Ct., Bedford, NH 03110, (603) 625-5431**Details:** For more information contact Jill & Ken Porter at (603) 626-0129 or jilleporter@comcast.net.**– Wednesday, July 9, 2014 –****Willamette Valley Ataxia Support Group Meeting****Time:** 11:30 a.m. – 1 p.m.**Location:** Albany General Hospital, 1046 Sixth Ave. SW, Albany, OR 97321**Details:** For more information contact Ivy Stilwell at (541) 812-4162 or at istilwell@samhealth.org.**– Thursday, July 10, 2014 –****Tri-State Ataxia Support Group Meeting****Time:** 6:30 – 8:30 p.m.**Location:** Bethel Israel Medical Center, Phillips Ambulatory Care Center (PACC), Second Floor Conference Room, 10 Union Square East, New York, NY 10003**Details:** For more information contact Denise Mitchell at markmeghan2@gmail.com or Kathy Gingerelli at kgingerelli@msn.com.**– Saturday, July 12, 2014 –****Central MN Ataxia Support Group Meeting****Time:** 9:45 – 11:45 a.m.**Location:** Kimball State Bank of St. Augusta, 24952 County Rd. 7, St. Cloud, MN 56301, in the Board Room**Details:** For more information contact Marsha Binnebose at (320) 248-9851 or mbinnebose@hotmail.com.**Tampa Bay Ataxia Support Group Meeting****Time:** 12:30 – 3 p.m.**Location:** Morsani Center, 13330 USF Laurel Dr. #1013, Tampa, FL**Details:** For more information contact Nygel Lenz at (727) 451-9165 or nygellenz@gmail.com.**– Saturday, July 19, 2014 –****Denver Area Ataxia Support Group Meeting****Time:** 1 – 4 p.m.**Location:** The Spruce C meeting room, second floor, at the Swedish Medical Center, 501 E. Hampden Ave., Englewood, CO 80113**Details:** Topic to be announced. For more information contact Charlotte DePew at (720) 379-6887 or cldepew77@comcast.net.**Greater Atlanta Ataxia Support Group Meeting****Time:** 1 p.m.**Location:** Emory Center for Rehabilitation Medicine, 1441 Clifton Rd., Room 101, Atlanta, GA**Details:** For more information call (404) 822-7451 or atlantaataxia@gmail.com.**– Saturday, July 19, 2014 –****Twin Cities Ataxia Support Group Meeting****Time:** 10 a.m. on the third Saturday of every month**Location:** Langton Place in Roseville at 1910 W. County Rd. D, Roseville, MN 55112**Details:** For additional information contact Lenore Healey Schultz at (612) 724-3784 or cschultz.lenore@yahoo.com.**– Saturday, July 26, 2014 –****New Hampshire Ataxia Support Group Meeting***Continued on page 54*

Calendar of Events
Continued from page 53

Time: 10 am – noon

Location: Hannafords at the Bedford Shopping Mall, 5 Colby Ct., Bedford, NH 03110, (603) 625-5431

Details: For more information contact Jill & Ken Porter at (603) 626-0129 or jilleporter@comcast.net.

**INFORMATIONAL AND
AWARENESS EVENTS**

– **Friday, May 2-4, 2014** –

NY Metro Abilities Expo

Time: Friday and Saturday 11 a.m. – 5 p.m., Sunday 11 a.m. – 4 p.m.

Location: New Jersey Convention Center

Details: Admission is free. <http://www.abilitiesexpo.com/newyork/index.html>

– **Friday, May 2, 2014** –

5th Annual Chuck n' Duck Dodgeball Tournament

Location: Charleton Heights Elementary School, Ballston Lake, NY

Details: In honor of Jacob Van Buren. Proceeds benefit the National Ataxia Foundation. For more information contact Andrew Haluska at ahaluska@bhbl.org or (518) 399-9141

– **Sunday, May 4, 2014** –

Strike out Ataxia

Time: 4:30 – 7:30 p.m.

Location: Pin Chaser, 4847 N. Armenia Ave., Tampa, FL

Details: This fun filled event includes bowling for all abilities, music, and photo opportunities. Proceeds benefit the National Ataxia Foundation. For more information visit <http://djheadbussa.com/> or call (813) 781-6129.

<https://www.eventbrite.com/e/3rd-annual-strike-out->

ataxia-charity-bowling-event-tickets-11081140991?discount=NAFmembersONLY

– **Sunday, May 18, 2014** –

Fun at the Pub

Time: 3 – 7 p.m.

Location: Christina Pub, 10 W. Main St., Peddlers Village, Christiana, DE 19702

Details: This fun-filled event will include a DJ, dancing, buffet, and raffle. Must be 21 to attend. All proceeds benefit NAF. For more information or to support this event visit the event website, <https://naf.myetap.org/fund-raiser/14pub/>

– **Saturday, May 24, 2014** –

Annual Walk for Dave

Time: 9 a.m. – noon

Location: Liverpool High School Stadium, 4338 Wetzel Rd., Liverpool, NY 13090

Details: This walk is dedicated to the memory of David Alessi. Event registration is \$10. Proceeds benefit the National Ataxia Foundation. For more information or to volunteer for this event please contact Marc Alessi at pianoman345@hotmail.com or (315) 622-3976

– **Friday, June 27-29, 2014** –

Chicago Abilities Expo

Time: Friday and Saturday 11 a.m. – 5 p.m., Sunday 11 a.m. – 4 p.m.

Location: Renaissance Schaumburg Convention Center, Schaumburg, IL

Details: Admission is free. <http://www.abilitiesexpo.com/chicago/index.html>

– **Friday, July 25-27, 2014** –

Houston Abilities Expo

Time: Friday and Saturday 11 a.m. – 5 p.m., Sunday 11 a.m. – 4 p.m.

Location: The Reliant Center, Houston, TX

Details: Admission is free. <http://www.abilitiesexpo.com/houston/index.html> ❖

Attn: Chapter & Support Group Leaders

News and photos covering what is happening with your group and in your area regarding education, support, or ataxia awareness is very important to our readers.

Please submit your stories, events, and re-

ports for inclusion in a future issue of *Generations*. You may e-mail them to joan@ataxia.org or by mail to the NAF office. The deadline for the summer issue is May 16.

Thank you for sharing your news!

Memorials and In Your Honor

The National Ataxia Foundation is grateful to those who have made contributions in memory or in honor of their friends and families whose names are listed below. This list reflects contributions made in November 2013 through February 2014. We are sorry that we cannot separate the memorial contributions from those made in honor of someone, as sometimes the person making the contribution does not let us know if the contribution is a memorial or in honor of their friend or family member.

Debra Adair	Fred Donnelly	Genevieve Jackson	Maury McDonald	Donald Santa Croce
Michael Adinolfi	Rick Donnelly	Jane Jaffe	Linda Meier	Family
Timothy Adkins	Merlene Drake	Lisa Jaffe	Linda Meier Family	Marcella Schiffrin
Ralph Aiello	Pedro Durian	Alton Johnson	Pat Messigian	Roz Schless
George Arruda	Madeline	Kerry Johnson	Dave Mills	Roger Schott
Holly Atlas	Ellingsworth	Terry Johnson	Eileen Monteleone	Dr. & Mrs. Lawrence
Jeffery Barberi	Humberto Fagundes	Marianne Jones	Mark Mooney	Schut
Brandon Barker	Joseph Falcon	Patricia Jozefczyk	Earl Moore	Sophia Sieber-Davis
Ginger Barnes	Katherine Falcon	MD	Elizabeth Moore	Dana Simpson
Bart Beck	Trinity Falk	Kumiko Kamishita	Jack Moore	Monty Sims
Betty Beck	Ferguson Family	Russell Karli	Doris Morgan	Henry Skala Jr.
Claire Beck	Barton Ferris	Lisa Kelso	Adelaide Murphy	Allen Smith
Ted Benson	Field Family	Joshua Kirschbaum	Maureen Murray	Audrey Smith
Saul Berkman	Janice Fileff	Donna Klotz	Hubert Myers	Doyle Smith
Roger Bernier	Father Willard	Matthew Klotz	Hugh Myers	Kathryn Smithers
Arlene Bethelmy	Forman	Mary Komp	Miriam Nagelberg	Jenny Spiller
Stephanie Blake	Olivia Fortino	Richard Korosa	Dwight Nakatsu	Marvin Spillers
Fred Blasberg	Lisa Fountain	Jamie Kosieracki	Marlene Newcomb	Joey Staiger
Carol Brackett	Sharon Franklin	Karin Koski	John Norton	Walt Stangle
Mark Breland	Albert Frei	Cora Belle	Lanea Oakland	John Staton
Markell Breland	John Frey Jr.	Kramer-Bell	John Ober	Darrell Stenseth
Muriel Breland	Ruth Furniss	Marcella Kukelhan	Tom O'Connor	Pearl Straub
Tom Bretthauer	Jeffrey Gibson	Normand Labarre	Elita Omictin	Harry Stricker
Angela Brown	Kathleen Gingerelli	Troy Labarre	Ben Oviatt	John Surabian
Clete Brunnert	Christine Goar	Cynthia LaVallie	Yukiko Parameswar	Mark Swanson
Blanche Buhr	Tanya Goldman	Jennifer Leader	Sandra Parker	Jerry Tait
Liam Cannon	Penny Golminas	William Lee	Paula Partilla	Alice Tapper
Margie Capizzi	Anna Gordon	Johna Leidholt	Cody Peterson	Roger Teske
Karen Carrick	Brenda Graner	Richard Lewis	Eric Peterson	Mary Thyden
Donald Carroll	Lawrence Graner	Mildred Likkel	Erin Peterson	Christine
Peter Castaneda	Jacqueline Gray	William Lilac	Peterson Family	Tossavainen
Paul Cerbatos	Moira Greenway	Sharon Lindberg	Mark Peterson	Buck Turnbull
Clarissa Ching	Sarah Hale	Olie Logan	Family	Phil Turnbull
William Chwee	Stephanie Hales	Lowry Family	Gary Peterson	Paige Turney
Krista Clarke	Jane Haley	Michael Lundquist	John Piccione	Rudy Van't Hoff
Coffey Family	Evelyn Hankins	Howard Lyle	Antonio Pimentel	Antoinette Varron
Joseph Coffey	Jimmy Hankins	Billy Lytle III	Patricia Pisano	Marlea Waddell
Roger Cooley	Ernie Hansel	Jack MacDonald	Ken Porter	Owen Walter
Debra Covington	Susan Harmer	Johanna MacDonald	Rita Powell-Lobascio	David Westrick
Karen Crawford	Owen Hartnett	Michael Markowitz	David Price	Dr. Richard
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Mellisa Davis-Bunds	Higginbotham	Doyle Matteson	Janet Riley	Alissa Wolfson
Charlotte DePew	Susan Home	John Mauro	Florence Rinaldi	Joan Woodward
Connie DiVincintis	Krista Humes	Catherine May	Nathan Robinson	Beverly Wright
Dawn Dizon	Howard Hunnius	Robert May	Mary Rotolo	Sophie Yi
Julie Donahoe	Meghan Hurst	Sue McConnell	Brad Rountree	Kenneth Yousten ❖



National Ataxia Foundation

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Is your address correct? Are you receiving more than one issue of *Generations*? If there are any changes that need to be made, please call NAF at (763) 553-0020 or e-mail joan@ataxia.org. Thank you!

GIFT – HONOR – MEMORIAL

A contribution given in memory of a friend or relative is a thoughtful and lasting tribute, as are gifts to honor your friends or family. A Gift Membership is a wonderful gift to a friend or relative for special occasions like birthdays, graduations, anniversaries, and holidays. NAF will acknowledge your gift without reference to the amount.

Simply fill out this form and mail with your check or credit card information to the National Ataxia Foundation.

Honor/Memorial envelopes are available free of charge by writing or calling NAF.

My contribution is:

- In Memory In Honor Gift Membership

Name _____

Occasion _____

Send Acknowledgment Card to:

Name _____

Address _____

City/State/Zip _____

From:

Name _____

Address _____

City/State/Zip _____

MEMBERSHIP

Yes, I want to help fight ataxia! Enclosed is my membership donation. (*Gifts in US Dollars*)

- Lifetime membership \$500

Annual memberships:

- Patron membership \$100-\$499

- Professional membership \$55

- Individual \$35

- Household \$55

- Addresses outside the U.S. please add \$15

Name _____

Address _____

City/State/Zip _____

Phone _____

E-Mail _____

- Yes, sign me up for NAF e-mails

PAYMENT INFORMATION

Gifts are tax deductible under the fullest extent of the law.

- Check. Please make payable to the
National Ataxia Foundation.

Total Amount Enclosed \$ _____

Credit Card: Visa MasterCard Discover

Name on Card _____

Card # _____

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Phone Number _____