



Please direct correspondence to:

 National Ataxia Foundation  
2600 Fernbrook Lane, Suite 119  
Minneapolis, MN 55447-4752

Phone: (763) 553-0020  
FAX: (763) 553-0167  
Internet: www.ataxia.org  
E-mail: naf@ataxia.org

Generations Staff:

Julie Braun ..... *Financial Director*  
Sue Hagen ..... *Patient Services Director*  
Joan Jensen ... *Outreach Coordinator & Generations Editor*  
Mike Parent ..... *Executive Director*  
Lori Shogren..... *Special Projects Coordinator*  
Design, Production and Printing ..... *Leader Printing*

Generations is published by the National Ataxia Foundation, Inc., Minneapolis, MN.

**Copyright 2014 by the National Ataxia Foundation, Inc. All rights reserved.**

Please contact us for permission to reprint or post on the Internet any article from Generations.

**Disclaimer**

The National Ataxia Foundation does not endorse products, therapies, services, or manufacturers. Those that are mentioned in **Generations** are included only for your information. The NAF assumes no liability whatsoever for the use or contents of any product or service mentioned in the newsletter.

**Table of Contents**

**Annual Membership Meeting (AMM)**

Progression of Ataxia ..... 27-33  
NAF's Travel Grant Program  
Needs Your Support ..... 34  
2015 AMM Announcement & Overview .... 35-39  
Planning Your Trip to Denver? ..... 39  
AMM Exhibitors and Sponsors Wanted ..... 39

**IAAD, Events & Fundraisers**

International Ataxia Awareness Day ..... 33

**Research/Research Opportunities**

CoRDS & NAF Celebrate One Year  
of International Ataxia Registry ..... 4  
Pioneer SCA Translational Grant  
Award Summaries ..... 5-8  
Young Investigator Award Summaries ..... 9-12  
Patients with SCA1, SCA2, SCA3, SCA6  
and MSA-C Needed for MRI Study ..... 9  
Research Grant Award Summaries ..... 13-17  
Seeking Patients with SCA for Study ..... 14

**Research/Research Opportunities (cont.)**

Young Investigator for SCA Research  
Award Summaries ..... 18-21  
Post-Doc Fellowship Award Summaries .... 21-26  
Brain Donation Program ..... 24  
Fragile X Gene (FMR1) Study ..... 50  
Friedreich's Ataxia Study ..... 54

**Membership Topics**

FDA's Patient Representative Program ..... 26  
From the Desk of the Executive Director ..... 34  
Chapter and Support Group News ..... 40-44  
NAF Merchandise ..... 45  
NAF Directory of Chapters, Support  
Groups and Ambassadors..... 46-50  
Calendar of Events ..... 51-54  
Remembering NAF in Your Will ..... 52  
Memorials and In Your Honor ..... 55

**Personal Stories & Poems**

Ataxia Research Funding Moves Forward ..... 3  
Who Was Your Hero in Your Ancestry ..... 11

*The deadline for the upcoming Winter issue of  
Generations is Friday, October 31, 2014*

*Ataxia Research Drive...*  
*Continued from page 1*

---

best science in the world to find answers in ending ataxia.

Currently there are no effective treatments or cure for the ataxias, however, each research study funded is driven to this ultimate goal. Each of us has an opportunity to further the important work of ataxia scientists from around the world by supporting the 2014 NAF Annual Ataxia Research Drive.

After an extensive multi-level scientific peer review process and a full assessment made by the National Ataxia Foundation's Board of Directors, the most promising ataxia research studies will be funded in late December 2014. The number of these studies and the dollar amounts depends greatly upon you and your generous donations.

Together we are making a difference. Together we can end ataxia. Please give to the 2014 NAF Annual Ataxia Research Drive.

Thank you. ❖

---

# Ataxia Research Funding Moves Forward

*Submitted by Desmond Harvey*

Not many people know the word ataxia, hence what it means.

“Ataxia is a symptom of many disorders,” states the National Ataxia Foundation (NAF), “there are no cures for most of the ataxias,” and “there is no specific treatment to delay or halt” ataxia.

Why no cure or treatment? Two main reasons, namely:

- Ataxia is a rare disorder (“less than 150,000” in the U.S. according to NAF, although the true prevalence of the degenerative ataxias is unknown); and,
- Lack of biomarkers that can indicate a clinically meaningful change in a therapeutic intervention

Thus, research activities need to continue.

The unmet need for therapies for those who are affected with ataxia should be brought to the attention of the National Institutes of Health and the like, so that more research funding, more attention, could be steered towards ataxia research.

In no way should this revelation diminish the

gratefulness for the research on ataxia which is being carried out, reflect on the kindness of the donors for ataxia research, reduce the love of families and friends of ataxia persons, or shrink the value of ataxia support groups. Most of all, and in every way, this note is to support and increase the courage within persons with ataxia.

Dear reader, please always remember that there is someone worse off than you. Look for ways that you can bring ataxia awareness and be a positive influence in your community. ❖

## **CFC Number**

The National Ataxia Foundation's Combined Federal Campaign (CFC) number is 10752.

This program, the world's largest and most successful annual workplace charity campaign, provides a convenient way to donate to the Foundation.

Please give as generously as you can and please ask your co-workers to also give to the National Ataxia Foundation.

# CoRDS & NAF Celebrate One Year of International Ataxia Registry

*This update was recently published in the CoRDS Summer Newsletter.*

In July 2013, the CoRDS Registry and the National Ataxia Foundation (NAF) partnered to launch a disease-specific registry for ataxia.

“One of the NAF’s core goals is to advance promising research, and this partnership is a vehicle to continue moving forward in that area,” said Michael Parent, NAF’s Executive Director. “The NAF is honored to be part of a comprehensive registry that aims to understand and develop treatments for rare diseases.”

As of July 2014, over 568 participants have completed the disease-specific questionnaire by participants from 14 countries and 43 U.S. states. Spinocerebellar Ataxia Type 3 (SCA-3) and Friedreich Ataxia are the disease subtypes reported most frequently in the CoRDS-NAF registry.

\*\*\*\*\*

With your help, support and enrollment in the CoRDS ataxia patient registry, ataxia research will be able to move forward to find answers to end ataxia. By enrolling in CoRDS, participants who qualify will be notified of any opportunities

to participate in clinical trials as well as other research studies.

Participants who are diagnosed with ataxia or who are at-risk for ataxia may enroll in this registry by completing a brief questionnaire. The questionnaire includes common data elements recommended by the NIH Office of Rare Disease Research as well as data elements specific for ataxia that were developed by the NAF Medical Research Advisory Board.

We are pleased to announce that the numbers of registrants in the CoRDS ataxia registry continues to grow. If you have not already enrolled in the registry, you are encouraged to enroll and ask family members who have ataxia or those who are at risk for ataxia to enroll also.

Thank you. ❖

*If you have any questions about the ataxia registry within CoRDS patient registry, you may contact Sue Hagen, NAF Patient Services Director at (763) 553-0020 or [susan@ataxia.org](mailto:susan@ataxia.org). If you have questions about CoRDS or how to register, you may contact CoRDS personnel at (605) 312-6413, 1-877-658-9192 or [CoRDS@sanfordhealth.org](mailto:CoRDS@sanfordhealth.org).*

CoRDS Registry

Coordination of Rare Diseases  
at Sanford

 National Ataxia  
Foundation  
[www.ataxia.org](http://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](http://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](http://www.sanfordresearch.org/cords) or call (605) 312-6413 or 1-877-658-9192. Thank you for participating in this important research tool.

## Pioneer SCA Translational Grant Award

# VEGF as Therapy for SCA1

By **Puneet Opal, MD, PhD**

Northwestern University, Chicago, IL

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

SCA1 is a dominantly inherited neurodegenerative disorder characterized by progressive motor incoordination. This disease, caused by a CAG trinucleotide repeat expansion in the *SCA1* gene, results in a glutamine repeat expansion in the encoded protein, ataxin-1 (the normal length of repeats ranges from 6-40). In SCA1, cerebellar Purkinje cells are the first neurons to succumb and cerebellar degeneration ensues. Eventually, other neuronal groups are affected. Patients eventually die from brainstem dysfunction with aspiration and respiratory complications.

Our grant focused on exploring the potential of using VEGF as a therapeutic agent in SCA1. The reason for studying VEGF was that we had earlier found that VEGF was transcriptionally downregulated in the cerebella of SCA1 mice.

We have made important progress. Our first studies on VEGF resulted in a manuscript describing the mechanism by which VEGF is transcriptionally reduced (Cvetanovic et al., *Nature Medicine* 2011). We have found that the most benefit of VEGF is on the cerebellar symptoms of VEGF. We have begun testing the improvement of VEGF later in the disease in SCA1 knock-in mice at 6 months of age. This corresponds to the late symptomatic phase of the disease. We have found that with just two weeks of recombinant VEGF delivery by ICV pump, we find a significant improvement of the cerebellar phenotype as assayed by the rotarod test. Similarly, we also see an improvement of cerebellar phenotype by digital treadmill analysis (Digi-gait©). We are currently in the process of analyzing histopathological changes in these mice and investigating the molecular mechanisms of VEGF on cerebellar phenotypes.

SCA1 knock-in mouse model demonstrates both motor and cognitive deficits as seen in SCA1 juvenile cases (Watase et al., 2002). Since VEGF has been shown to increase learning and memory in mice by upregulating neurogenesis, we are also comprehensively exploring the therapeutic role of VEGF in the mouse SCA1 knock-in focusing on non-cerebellar symptoms.

Specifically, we are performing Morris Water Maze assays on SCA1 knock-in mice that genetically expressed human VEGF transgenes. For these assays, we are using genetic VEGF delivery, since we were concerned that



**Dr. Puneet Opal**

water may be a source of infection from the ICV pump. We hope to publish this work in the coming year. In addition, we are performing mechanistic experiments to determine how VEGF is exerting its beneficial effects in SCA1.

In addition to using recombinant VEGF, we are also beginning to explore novel ways by which VEGF can be delivered. Specifically, we are collaborating with Dr. Sam Stupp, a nano-engineer to develop a novel reagent: VEGF-PA (VEGF-Peptide Amphiphile). The molecular design of VEGF-PA involves covalent attachment of a previously described 15-amino acid VEGF mimetic

*Continued on page 6*

## VEGF as Therapy for SCA1

Continued from page 5

sequence (KLTWQELYQLKYKGI) to a peptide amphiphile (PA) (D'Andrea et al., 2005; Webber et al., 2011). In the aqueous tissue environment, the PA induces hydrophobic collapse, while the peptide backbone of the PA promotes intermolecular  $\beta$ -sheet hydrogen bonding. Together these effects promote the self-assembly of the VEGF-PA molecules into cylindrical nanostructures (~10 nm in diameter) that present the VEGF mimetic sequences on the surface at a high density.

A therapeutic strategy using VEGF-PA offers several potential advantages over both the VEGF mimetic peptide and native recombinant VEGF protein: First, VEGF-PA has a better pharmacokinetic profile for long-term delivery. This is because of (a) *a long half-life*: VEGF-PA are retained in tissues even four weeks after delivery, eventually being biodegradable by design (recombinant VEGF itself only lasts a few hours *in vivo*, and the 15 residue peptide itself is even less stable); (b) *slow release*: VEGF-PA form small filamentous structures that break apart slowly; (c) *polyvalent nature*: each nanoparticle presents VEGF mimetic peptides at a high valency; this promotes receptor dimerization and sustained activation; (d) *hydration and spread in vivo*: VEGF-PA are highly hydrated, a feature that also promotes efficient and potent receptor binding and signaling. Overall these properties allow for continuous and sustained signaling. VEGF-PA also has other important qualities: they do not cause an immune response, and they are a relatively inexpensive reagent, an important feature given the high cost of using recombinant VEGF for a slowly debilitating disease like SCA1. Finally, this nanoreagent is already in an advanced stage of development (Webber et al., 2011).

For all these reasons, we are excited to test its potential in SCA1; indeed this would be the first application of this technology for any neurodegenerative diseases. In our preliminary experiments we have found that these nanoparticles

spread rapidly in the brain once injected (we have used fluorescently tagged PA to show their spread in the substantia nigra as an example. We hope that in the near future we will be able to completely validate the efficacy and safety profile of VEGF-PA.

Finally, we have been intrigued by the possibility of treating SCA1 by reversing transcriptional alterations in gene expression using a more global approach rather than targeting individual genes such as VEGF. In principle, a large-scale reversal of transcriptional aberrations induced by ATXN1 might result in even greater beneficial effect than that achieved by correcting the down-regulation of a few specific genes. In a recent study, we tested the potential for improving the SCA1 phenotype by decreasing the levels of HDAC3, a histone deacetylase (HDAC) that is an important regulator of gene expression and binds ATXN1 as part of a complex. We hypothesized that by recruiting the HDAC3 complex, mutant ATXN1 causes pathogenic transcriptional repression, resulting in gene expression changes relevant to SCA1.

We have tested this hypothesis in a recent study (Venkatraman et al. 2014). In this study, we designed our experiments to genetically test the role of HDAC3 in the context of SCA1. Since, HDAC3 null mice are embryonic-lethal, we used for our analyses a combination of HDAC3 haplo-insufficient and Purkinje cell-specific HDAC3 null mice. We found that deleting a single allele of HDAC3 in the context of SCA1 was insufficient to improve cerebellar and cognitive deficits of the disease. In fact, a complete loss of Purkinje cell HDAC3 was highly deleterious both behaviorally and pathologically. Our results therefore suggest that long term suppression of HDAC3 could have toxic consequences which should be looked for carefully in any future HDAC inhibitor trials.

We thank the NAF for funding our work since without this funding we would not have been able to make progress on these translational projects. ❖

Pioneer SCA Translational Grant Award

# Small Molecule Inhibitors of PKA: A Therapeutic Strategy for SCA1

By **Harry T. Orr, PhD**

*University of Minnesota, Minneapolis, MN*

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

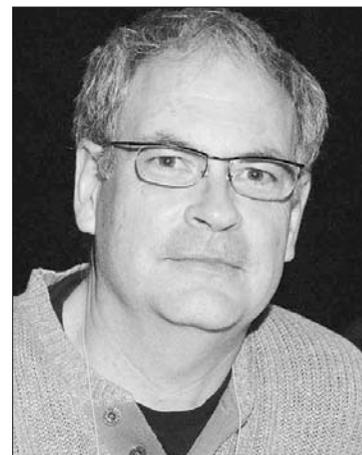
Spinocerebellar ataxia-type 1 (SCA1) is a progressive lethal neurodegenerative disorder. In SCA1, the protein affected, Ataxin-1, has a region within it that typically contains 35 or fewer glutamine residues in individuals not affected with SCA1. Individuals with SCA1 have a genetic mutation that leads to ataxin-1 protein containing greater than 40 (and as many as 83) glutamine amino acid residues. As a result of this mutation, the normal function of the protein is compromised. Ataxin-1 is expressed in the Purkinje neurons of the cerebellum, the brain cells that coordinate balance and movement. Expression of mutant Ataxin-1 leads to degeneration of Purkinje neurons, and subsequent loss of motor and balance coordination.

An amino acid residue found in both normal and mutant Ataxin-1, designated serine 776, was found to undergo a chemical modification termed phosphorylation that is crucial for disease progression. In transgenic mice studies, phosphorylation of serine 776 was found to be necessary for mice to get the disease (Duvick et al., 2010; Emamian et al., 2003). Mice expressing a continuously phosphorylated form of normal Ataxin-1 showed the same pathological and behavioral symptoms as mice expressing the mutant Ataxin-1 protein.

These studies indicate that molecules that inhibit phosphorylation at serine 776 have therapeutic potential for SCA1. To begin to find potential inhibitors, we screened several compounds to find inhibitors of Ataxin-1 serine 776 phosphorylation in a cell-free assay. The

molecules discovered in these initial screens were analyzed in progressively more complex model systems in an effort to find a small molecule inhibitor of Ataxin-1 serine 776 phosphorylation with potential of becoming an effective drug.

In this NAF Pioneer Award we focused on a series of hit compounds that resulted from a kinase drug screen performed by pharmaceutical companies Pfizer or GSK. These compounds were either provided by each company or were synthesized by us based on published methods. We have analyzed nine such compounds using our cerebellar lysate assay and found four that inhibited Ataxin-1 S776 phosphorylation in the  $\mu\text{M}/\text{nM}$  range. During the past funding period assessed several compounds for their ability to inhibit Ataxin-1 S776 phosphorylation in the DAOY cell and cerebellar slice culture assays. The goal of the work was to develop from these hit compounds a high-quality small molecule with validated biology. The compound, GSK690693, was identified for further modification towards development of an effective small molecule drug.



**Dr. Harry T. Orr**



Pioneer SCA Translational Grant Award

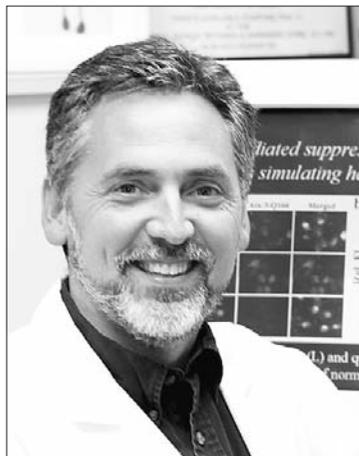
# Novel Cell-Based Screens for Therapeutic Compounds in SCA3

By Henry Paulson, MD, PhD

University of Michigan, Ann Arbor, MI

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

No preventive treatment exists for any of the dominantly inherited forms of ataxia including Spinocerebellar Ataxia Type 3 (SCA3). In this grant, we used cell-based assays to seek potential drugs that might be developed into preventive therapy for SCA3. We created two complementary cell-based assays designed to identify compounds that either a) reduce levels of the SCA3 disease protein, ataxin-3, or b) inhibit its aggregation. The disease protein, and aggregates formed by it, both represent attractive therapeutic targets because they reside very early in the molecular cascade occurring in disease. In other words, targeting them does not require a detailed understanding of downstream disease mechanisms, which still remain elusive.



**Dr. Henry Paulson**

In both assays we screened small molecule libraries comprising over 4,000 compounds, including FDA-approved drugs and modulators of protein quality control, to identify molecules that reduce levels of ataxin-3 or oligomers. The first screen, which provides a read-out of ataxin-3 levels, proved to be a more robust assay with better signal-to-noise ratio. Accordingly, it became the focus of most of our subsequent work. A series of secondary screens in independent cell

lines and dose-response analyses winnowed down promising targets to 14 compounds that consistently reduced cellular levels of expanded ataxin-3. These compounds were then tested in a final secondary screen employing brain slice cultures derived from mice engineered to express the full human disease protein. This brain slice culture method allows us to gauge, relatively quickly and economically, whether a compound is active in actual brain tissue expressing the ultimate target: the human SCA3 disease protein. In this slice culture screen, at least six compounds showed some reduction in ataxin-3 levels in brain. One of these, an FDA-approved anti-psychotic, is active in the brain and widely used as a medication; a second promising compound is a cannabinoid receptor antagonist. Next, we delivered these two promising drugs separately into living mice expressing the human SCA3 disease gene; mice were injected daily for 10 days and levels of brain ataxin-3 were then assessed. This analysis showed that the anti-psychotic drug did indeed reduce high molecular weight aggregates formed by ataxin-3 and, to a lesser extent, levels of the disease protein. Finally, this confirmatory in vivo screen encouraged us to perform pharmacokinetics analysis of the anti-psychotic drug in mice, in order that we could then design an appropriate preclinical trial to test whether this drug effectively reduces levels of the SCA3 disease protein in mice and corrects aspects of the disease phenotype. If this ongoing preclinical trial succeeds, we will be encouraged to advance this particular drug into human studies in the ataxia clinic. ❖

## Young Investigator Award

# Development of New Mouse Models of Friedreich's Ataxia

By **Christopher P. Baines, PhD**

University of Missouri-Columbia, Kansas, MO

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

Mouse models of Friedreich's Ataxia (FA) have provided important test systems for the development of new therapeutic agents for treatment of this disease. However, the FA-like disease that develops in these animals is either too severe or too mild. Moreover, the genetic changes required to generate and maintain these mice are very complex thus making their use very time-consuming and laborious. Our studies, therefore, proposed to generate a new mouse model of FA that will ideally would fall somewhere in between these existing systems, both in terms of disease progression and in ease of use.

To do this we expressed something called a short hairpin RNA in the hearts of mice to reduce levels of a protein called frataxin. In humans, FA is caused by a severe reduction in frataxin protein levels. Our goal was to reduce frataxin levels to the same extent as that seen in patients with FA, thereby causing the heart failure seen during FA. We generated several lines of mice. In some mice the frataxin levels were unaltered. In some they were decreased by a small amount. However, in two of the lines frataxin protein was reduced by 40-50%, and we still have further lines we are analyzing where hopefully the levels are reduced to an even greater extent.

We have examined how well the heart is functioning in some of these mouse lines. So far we have seen no signs of the heart failure, which occurs during FA, in these mice. That being said we are still in the process of analyzing all of the lines. Moreover, we will take the lines that have the 50% reduction in frataxin and breed them together so that hopefully we will get an even greater reduction of frataxin in the heart and thereby cause heart failure. Should this approach be successful, this new mouse model would represent a significant addition to our arsenal in the study of the cellular mechanisms that cause FA and the development of therapeutics to treat this disease clinically. ❖



**Dr. Christopher Baines**



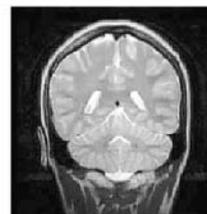
**PATIENTS with  
SCA1, SCA2, SCA3, SCA6 and  
MSA-C  
needed for an MRI study  
at the University of Minnesota, Minneapolis**

Travel expenses reimbursed.

**Contact: Diane Hutter**

(612) 625-2350

hutte019@umn.edu



## Young Investigator Award

# Diagnosis of Rare and Novel Genetic Cerebellar Ataxias Using Next-Generation Sequencing

By Brent L. Fogel, MD, PhD

David Geffen School of Medicine at UCLA, Los Angeles, CA

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

My laboratory studies the causes of inherited (or “genetic”) forms of ataxia. These diseases are caused by changes in a person’s DNA, or genetic blueprint. All the DNA a person has is referred to as their “genome” and includes approximately 21,000 separate “genes” which together combine to make each person a unique individual.



**Dr. Brent L. Fogel**

Changes in some of these genes can lead to diseases like cerebellar ataxia. To better diagnose genetic ataxias and identify novel disease genes, my lab uses a technology called next-generation sequencing, which allows rapid and efficient examination of all the genes in a person’s genome (“the exome”). Because there are at least 60 different known genes that cause cerebellar ataxia as a primary feature, and hundreds of other diseases which may include cerebellar ataxia as an associated symptom, this simplifies the challenge of finding the problem gene. Although approximately 50% of inherited cases are due to mutations in just a few genes (SCA1, SCA2, SCA3, SCA6, SCA7, and Friedreich ataxia), the remaining 50% of genetic causes account for 1% or less of all cases

worldwide each, so exome sequencing provides a rapid and cost-effective means of viewing all ataxia genes at once, thereby streamlining and expediting diagnosis.

In the first part of this project, I proposed to use next-generation sequencing to test for ataxia mutations in families with a “dominant” pattern, where disease passes from generation to generation. We have now sequenced exomes from over two dozen families and are currently analyzing this data. We have already begun to identify potential new disease genes and are testing them in different ways to confirm that they truly cause disease.

The second aim of this project has focused on the evaluation of families with suspected “recessive” inheritance, where disease is seen in multiple children but not the parents. We have now sequenced over a dozen such families and our results can be summarized by four categories. First, in several cases we have identified novel variants in known ataxia genes. Second, in other families we have identified mutations in genes known to cause recessive disorders not typically thought of as ataxia. Third, a few of our cases do not have apparent mutations in any known disease genes and we are investigating these families for novel ataxia genes as described above. Fourth and finally, the last group likely possesses new types of mutations in known ataxia genes and we are using various methods to describe these mutations, which will help doctors diagnose them easier in the future. ▶▶

The third aim of this project was devoted to looking in our patient population for mutations in recently identified disease genes. We currently have one major project underway involving a rare ataxia gene and several others in the preliminary stages.

Overall, the work begun during this project will ultimately provide new clinically relevant information for the ataxia community. The validation of new strategies for the identification of novel mutations and ataxia genes will provide

physicians and researchers with new tools for diagnosing patients and, hopefully, uncover new insights into the causes of cerebellar ataxia. Together we will further advance our knowledge of how the cerebellum works, what types of damage cause ataxia, and how we, as physicians and researchers, can partner with patients and their families to find cures for these progressively debilitating illnesses someday. I thank the National Ataxia Foundation for helping me contribute to this important mission. ❖

## Who Was Your Hero in Your Ancestry?

*Submitted by Bob Amos*

When I was diagnosed with SCA 6 in 1999, the doctor informed me that a French origin was suspected. I inherited the SCA gene from my father (paternal). I retired in 2004 and being unable to physically engage in past hobbies, I spent time on other things, one of those things being genealogy research (searching for my ancestors). I am trying to trace my roots back to France. My paternal great-grandfather was born in England in about 1839. My paternal great-grandmother was born in England in about 1851. She and my great-grandfather immigrated to the U.S. in 1871, landing in New Orleans, LA. I know where they were sailing to but I don't yet know where their port of departure was when they left England or where or when they married. My paternal great-grandmother was only about 20. I do know their son, my paternal grandfather, was born in Newark, New Jersey in 1878. His name was Waldo Adams Amos ... where did the 'Adams' come from?

I know there is a lot of ataxia research projects going on that are in various stages. These ataxia research projects all require funding. I propose conducting a family genealogy research project. I do most of my genealogy research on the internet with my computer. Although my name is Robert, I generally go by Bob. You do not have to use a computer – just ask your parents, grandparents, aunts, uncles, cousins – everyone. What do they remember? Find a gravestone. Churches have records of christenings, marriages, funerals and burials. It's interesting and a way to provide a topic for family communication or even be a family project tailored to one's ability.

In the title, I asked 'Who Was Your Hero in Your Ancestry?' My paternal great-grandmother left England to sail to America when she was only 20. She was a hero.

If you would like more information, or to contact me, my email address is [robertjamos@comcast.net](mailto:robertjamos@comcast.net). Happy hunting! ❖



**Bob Amos and friend Rose**

Young Investigator Award

# RNA-based Therapy for Spinocerebellar Ataxia 3 and Dentatorubral-pallidoluysian Atrophy

By Jiaxin Hu, PhD

University of Texas, Southwestern Medical Center at Dallas, Dallas, TX

The following is a research summary of a grant funded by NAF for fiscal year 2013.

Nucleic acids are known to bind specifically to their target genes and reduce the protein production. Nucleic acids and their analog drugs are being actively developed to treat a variety of diseases. Huntington's disease (HD), Spinocerebellar ataxia 3 (SCA3) and Dentatorubral pallidoluysian atrophy (DRPLA) are different inherited neurodegenerative disorders. These diseases share common features: they are caused



**Dr. Jiaxin Hu**

by the expanded CAG repeats on their mutant genes. Most patients have two copies of gene, one is mutant, containing the prolonged CAG repeats. The other is normal gene with much smaller number of repeats. Specifically reducing the disease-causing mutant gene expression without affecting the normal one may provide a promising strategy for treatment. Previously, we have developed a novel strategy to selectively inhibiting the mutant huntingtin (HTT) production for HD. We use nucleic acids or duplex RNAs targeting the expanded CAG repeat and selectively inhibiting the mutant HTT with little influence on the wild-type protein.

In this study, we applied the anti-CAG strategy

for SCA3 and DRPLA. We tested a large variety of duplex RNAs with different chemical modifications, including 2'-O-Me RNA, abasic RNA, and Unlocked Nucleic Acids. After screening in the SCA3 patient-derived cells, we identified a pool of potent and selective inhibitors. This will become a reservoir for testing compounds in animals to identify agents with optimal *in vivo* properties. We also tested singlestranded silencing RNAs (ss-siRNAs). This kind of compounds has a lot of advantages and is ready to be used in animals. We found many potent and selective ss-siRNA inhibitors. We also explored into the inhibition mechanism and discovered that the ss-siRNAs could modulate RNA splicing and produced a new ataxin-3 isoform. This strategy may provide a new way for future development of treatment for SCA3.

DRPLA is a relatively rare neurological disease. Little work has been done to explore a potential treatment. For the first time, we developed a reliable assay for evaluation of antisense reduction of atrophin-1 expression. We demonstrated that mutant atrophin-1 expression could be selectively reduced by a large pool of anti-CAG agents including duplex RNAs, ASOs and ssiRNAs. Some selective inhibitors are active for HD, SCA3, and DRPLA.

Continuing study on this subject will not only expand our knowledge on selective inhibition, but may also lead to development of a single drug for treatment of several different CAG expansion diseases. ❖

## Research Grant Award

# Modifying Machado-Joseph Disease Progression by Caffeine Blockage of Adenosine A<sub>2A</sub> Receptors

By Luís Pereira de Almeida, PhD  
University of Coimbra, Portugal

The following is a research summary of a grant funded by NAF for fiscal year 2013.

Machado-Joseph disease (MJD) was originally described in people of Portuguese descent and is presently considered the most common dominantly-inherited cerebellar ataxia worldwide. It is characterised by progressive movement abnormalities involving malfunction of multiple motor-related brain systems associated with an unstable CAG expansion, which translates into an expanded polyglutamine tract within ataxin-3.

We investigated whether blockage of the adenosine A<sub>2A</sub>R receptors (A<sub>2A</sub>R) by caffeine administration or genetic silencing would alleviate MJD. We found that both chronic consumption of caffeine or the genetic elimination of A<sub>2A</sub>R alleviated neuropathological modifications and motor behaviour impairments. In particular,

administration of caffeine to genetic mouse models of MJD prevented the loss of synaptic markers, astrogliosis and microglia activation, and reduced neurodegeneration. Moreover caffeine, administered through the drinking water to transgenic mice with a severe ataxic behaviour, prevented progressive loss of motor function, balance and grip strength, in parallel with cerebellar morphology preservation.

This study (partially published in Gonçalves et al 2013, *Annals of Neurology*) suggests that A<sub>2A</sub>R blockage may be a promising therapeutic target to manage MJD. Further studies are necessary to translate these studies into patient therapy.

**References:** Gonçalves N, Simões AT, Cunha RA, Pereira de Almeida L. Caffeine and Adenosine A<sub>2A</sub> receptor inactivation decrease striatal neuropathology in a lentiviral-based model of Machado-Joseph disease. *Annals of Neurology*; Volume 73, Issue 5, May 2013, Pages: 655–666, first published online 26 APR 2013, DOI: 10.1002/ana.23866.



Dr. Luis Pereira de Almeida

## GoodSearch

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 and follow the easy steps to make NAF your charity of choice. Then use GoodSearch as you would any other search engine. This simple change will make a difference in the lives of those with ataxia!



Research Grant Award

# Generation of Peripheral Sensory Neurons from Friedreich's Ataxia Human iPS Cell Lines

By Satyan Chintawar, PhD  
 Université Libre de Bruxelles, Brussels, Belgium

The following is a research summary of a grant funded by NAF for fiscal year 2013.



Dr. Satyan Chintawar

Friedreich's ataxia (FRDA) is a rare inherited neurological disorder, which affects the nervous system leading to impaired muscle coordination along with heart disease. It is one of the most common forms of hereditary ataxia affecting 1/40,000 individuals. Discovering the inherited gene mutation was a milestone in FRDA research and led to the identification and role of encoded protein frataxin in physiological and pathophysiological processes. FRDA research, like many other neurological diseases, has been hampered by the availability of appropriate model systems to unravel disease mechanism. Routinely used human cellular types, such as fibroblasts and engineered cell lines, are not disease-affected cell types and animal models do not fully recapitulate human disease. Induced pluripotent stem cell technology has now offered an unprecedented opportunity to generate any cell type from the patient's blood or skin cells and is of the same genetical identity to

that of the patient. We have generated disease-affected brain cells and identified biochemical and physiological defect in them and the detailed investigation is ongoing to understand why certain cell types are most vulnerable by this disease.



**SEEKING PATIENTS WITH SCA (ANY TYPE)  
 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  
 Seth Wakefield at 617-667-0209 or email [swakefie@bidmc.harvard.edu](mailto:swakefie@bidmc.harvard.edu)

Research Grant Award

# Development of Knock-in Mouse Models for Early- and Late-Onset SCA13

By Diane M. Papazian, PhD

David Geffen School of Medicine at UCLA, Los Angeles, CA

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

Spinocerebellar ataxia (SCA13) is a rare autosomal dominant disease characterized by substantial atrophy of the cerebellum and locomotor deficits. SCA13 is caused by mutations in the KCNC3 gene, which encodes the voltage-gated Kv3.3 K<sup>+</sup> channel. This channel plays a central role in controlling the electrical excitability of neurons, including several types of cerebellar neurons. The mutations alter channel function, making it likely that SCA13 is a disease that results from altered excitability in neurons.

SCA13 presents in two clinical forms depending on the causative mutation. An early-onset form of the disease is evident in infancy or early childhood with motor delay, persistent motor deficits, severe cerebellar atrophy, and intellectual disability. A late-onset form of the disease emerges in adulthood with progressive ataxia and progressive loss of cerebellar volume. The infant- and adult-onset phenotypes are strongly correlated with the causative mutation in unrelated families, indicating that they do not reflect differences in genetic background. How different mutations in the same gene give rise to such distinct clinical phenotypes is unknown.

Animal models are essential for determining how changes in excitability lead to downstream pathogenic changes in SCA13, and how differential changes in excitability lead to distinct clinical forms of the disease. The goal of this project is to generate two lines of knock-in mice

with point mutations in the endogenous *Kcnc3* gene analogous to those that cause early- or late-onset SCA13 in humans. These knock-in mice will be the first genetically accurate models of SCA13 in a mammalian model system. The mice will be used to identify mechanisms that contribute to pathogenesis in SCA13, to determine how different mutations in the same gene give rise to distinct clinical phenotypes, to investigate whether the etiology of SCA13 overlaps with that of other hereditary ataxias, and to translate the results of basic research into new therapeutic approaches.



**Dr. Diane M. Papazian**

With support from the National Ataxia Foundation, we have been able to generate essential reagents needed to produce SCA13 knock-in mice, starting with targeting vectors to replace the wild-type Kv3.3 gene in mouse embryonic stem cells with alleles encoding mutations that cause early- or late-onset SCA13.

Production of the mice is now continuing with grant support recently obtained from the NIH. ❖

## Research Grant Award

# Exome Sequencing to Identify New Ataxia Genes

By Andrew Singleton, PhD

National Institute of Aging, Bethesda, MD

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

The ataxias comprise a wide spectrum of progressive neurodegenerative disorders with ataxia as the leading symptom. Classifications distinguish between hereditary and sporadic ataxia. Hereditary ataxias are further classified as autosomal dominant, autosomal recessive, X-linked and mitochondrial. Autosomal recessive cerebellar ataxias (ARCA) are heterogeneous and complex with more than 20 different forms currently identified. Symptoms start in childhood and include balance abnormalities, incoordination and dysarthria. Friedreich ataxia, the most common



**Dr. Andrew Singleton**

ARCA, was first described in 1863 and is now seen worldwide. In the last few years, several other ARCA have been recognized but genes remain unidentified for most recessive ataxias.

The aim of our study was to identify novel genes responsible for ataxia using whole exome sequencing. Our cohort comprised 112 Tunisian individuals diagnosed with ataxia with unknown origin including familial and sporadic cases. The Tunisian population was chosen for its high rate of consanguinity and large pedigrees which facilitate genotype-phenotype correlations in genetic diseases.

We identified novel mutations in known

recessive ataxia genes (SACS, SETX) as well as in recessive spastic paraplegia genes (SPG11, SPG20) expanding our knowledge about the disease phenotype and mechanisms.

More interestingly, we identified GBA2 as a new gene responsible for ataxia (Hammer et al., 2013). Three mutations were found in four unrelated families; two nonsense mutations (c.1542C>T [R340X] and c.363C>A [Y121X]) and a missense mutation (c.3142G>A [R873]). These variants segregated with the disease within the families and were absent in neurologically normal controls. They affect amino acids highly conserved across species. Clinically, ataxia was the presenting feature, but later, spasticity became very pronounced involving initially the lower limbs and later also the upper limbs. The mutation mechanism is likely to be loss of function which, based on mouse work, will lead to an accumulation of glucosylceramide and subsequently the clinical phenotype. ❖

## Vehicle Donation

The donation of your vehicle to the National Ataxia Foundation will help support the important work that is being done on behalf of all who are affected by ataxia.

To donate your car, truck or motor home, call 1-800-240-0160 or visit [www.donateacar.com](http://www.donateacar.com). Your vehicle will be picked up at your home, office or other place that you designate. Be sure to have the certificate of title with the vehicle.

Young Investigator for SCA Research Award

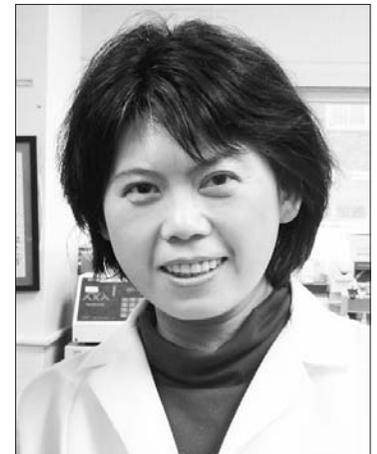
# Pathogenesis and Disease Course of Spinocerebellar Ataxia Type 14 in a Mouse Model

By Dong-Hui Chen, MD, PhD  
University of Washington, Seattle, WA

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

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 (PKC $\gamma$ ) 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 PKC $\gamma$  lead to neuronal death and how the disease develops is unknown. An animal model of SCA14 would be invaluable for such investigations. We have already created

mouse lines that have normal and mutated forms of the SCA14 gene. The mice with the mutant human PKC $\gamma$  develop abnormalities in their brains, including aggregations of the mutant protein, but do not have overt neurologic phenotypes, for example ataxia gait. To achieve a mouse model that recapitulates SCA14, we hypothesize that reducing endogenous PKC $\gamma$  may enhance the toxic effect of mutant transgenic PKC $\gamma$ .



**Dr. Dong-Hui Chen**

In this NAF funded project, we have modified the mice to carry a null endogenous PKC $\gamma$  gene so that they have reduced endogenous PKC $\gamma$ . The study in these mice have found earlier pathologic abnormalities in Purkinje cells, supporting our hypothesis. But we need to further study them, both behaviorally and pathologically, to determine the onset of brain changes, and progression of neurologic impairments. We will continue 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. ❖

## Deadline

The deadline to submit materials for the upcoming winter issue of *Generations* is Friday, October 31, 2014. Please submit content by e-mail to [joan@ataxia.org](mailto:joan@ataxia.org) or to the address shown on page 2.

Young Investigator for SCA Research Award

## Unraveling the Mitochondrial Conundrum at the Basis of Spinocerebellar Ataxia Type 28 and its Novel Therapeutic Approaches

By Francesca Maltecca, PhD  
 Università Vita-Salute San Raffaele, Milan, Italy

The following is a research summary of a grant funded by NAF for fiscal year 2013.

Spinocerebellar ataxia type 28 (SCA28) is characterized by unbalanced standing, gait incoordination, nystagmus and ophthalmoparesis. Several disease-causing mutations have been identified in the *AFG3L2* gene. The encoded protein, *AFG3L2*, resides in the mitochondrion and is essential for energy production and for the regulation of mitochondrial morphology. We characterized a mouse model of SCA28 that recapitulates the features of patients, showing progressive ataxia due to degeneration and loss of Purkinje cells (PCs). In SCA28 PCs undergo “dark degeneration” (DCD) since they appear shrunk, atrophic and dark. This degeneration, which generally follows increased calcium concentration associated to dysfunction of the glutamatergic system, is quite particular in SCA28, since it originates from mitochondrial dysfunction.



**Dr. Francesca Maltecca**

We demonstrated in cultured PCs that *AFG3L2* mutant mitochondria ineffectively buffer the evoked calcium peaks, thus enhancing cytoplasmic calcium concentration and finally triggering PC-DCD. This defect is caused by both alteration in the metabolic status of mitochondria and impaired transport of these organelles to PC dendrites.

Proving this mechanism, we completely recover the ataxic phenotype of SCA28 mice by genetically reducing the glutamate receptors, and thus decreasing calcium influx in PCs. The same result has been successfully replicated by a pharmacological treatment favoring the synaptic glutamate clearance. This treatment is effective when applied at both pre- and post-symptomatic stages of neurodegeneration, thus representing an immediately accessible therapy for pre-symptomatic carriers of *AFG3L2* mutations and also SCA28 patients with overt symptoms. ❖

### Sign Up for E-mail Blasts

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

Take a moment right now to send your e-mail address to [joan@ataxia.org](mailto:joan@ataxia.org) so you don't miss

out on receiving important information from the Foundation.

Please make sure to add [naf-eblast@ataxia.org](mailto:naf-eblast@ataxia.org) to your address book or “safe sender” list to make sure you get the e-mails, or check your spam folder.

Young Investigator for SCA Research Award

# Isoforms and Polymorphisms of Ataxin-3 as Modifiers of the Pathogenesis in Spinocerebellar Ataxia Type 3

By Thorsten Schmidt, PhD  
University of Tuebingen, Germany

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

Spinocerebellar ataxia type 3 (SCA3) or Machado-Joseph disease (MJD) is a family brain disease leading to movement defects. The cause of the disease lies within the so called ataxin-3 gene. This gene contains an area with multiple repetitions of the three DNA elements C, A, and G. Everybody in the general population has between 12 and 40 repetitions of CAG in one's own ataxin-3 gene. In SCA3 patients, however, the number of such CAG repetitions is increased to more than 62 of these repeats. Everyone has two copies of the ataxin-3 gene, one inherited by the mother, one inherited by the father. The repeat expansion in just one of these two copies causes the disease meaning that the second copy is usually normal.

One part of our project, supported by the National Ataxia Foundation, focused especially on the interplay between both copies of ataxin-3 as we observed that they seem to mutually influence each other. Human beings are and look obviously not identical. This is reflected by approximately four million variances which discriminate any two humans (except for genetically identical twins). Such genetic variances also occur in the ataxin-3 gene. We observed that each patient has a specific combination (a so-called haplotype) of these different variants both in the normal and the expanded copy of ataxin-3. This is of particular importance as it is known that patients with a higher number of CAG repeats develop first symptoms of the disease at an earlier age and patients with less repeats have first

symptoms later in their life. However, this purely statistically correlation does not allow to predict the exact age at onset from just the number of CAG repeats. For example, a SCA3 patient with 71 CAG repeats may get first symptoms as early as with 25 years or not until 60 years of age indicating that additional factors besides the CAG repeat itself contribute to the severity of the disease. In order to improve the prediction when first symptoms may occur, we analyzed whether additional variations of the ataxin-3 gene – besides the CAG repeat – influence the age at onset. Interestingly, 2 % of European patients with one specific haplotype had a much later (five years later) onset of symptoms thereby indicating that different variances of ataxin-3 and their specific combination in each individual impact the disease severity and progression. We, therefore, further focused on the mechanistical background of this observation. We discovered that the variations of ataxin-3 modify both its normal and disease function. Ataxin-3 variances differ in their stability as well



**Dr. Thorsten Schmidt**

---

*Continued on page 20*

*Isoforms and Polymorphisms of Ataxin-3...*  
Continued from page 19

as on their impact on important cellular processes causing the disease including the formation of so called protein aggregates. Interestingly, the non-affected copy of ataxin-3 with the normal CAG repeat seems to modify these processes. The 2 %

of European patients with a later onset of symptoms seem to have variations of the affected protein which specifically work together to protect from its toxic effect. The results of our project will help to understand the processes which lead to SCA3, may lead to a better prediction of the age at onset and may point to novel targets for a possible future therapeutic intervention. ❖

## Young Investigator for SCA Research Award

# Mechanisms of Neuroprotection in SCA3/MJD

By Sokol Todi, PhD

Wayne State University School of Medicine, Detroit, MI

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

Spinocerebellar Ataxia Type 3 (also known as Machado-Joseph Disease; SCA3/MJD) is perhaps the most common dominantly inherited ataxia in the world. SCA3/MJD is a progressive loss of full control of bodily movements. It arises



**Dr. Sokol Todi**

and how the functions of the causative protein, ataxin-3, relate to SCA3/MJD. Currently, there is no cure for SCA3/MJD.

Ataxin-3 appears to be involved in cellular mechanisms that discard abnormal proteins.

from the expansion of a region of the ataxin-3 protein beyond normal levels (12-42) to over 60 repeats of the amino acid glutamine. Such expansions affect several areas of the brain and the spinal cord. It is unknown how SCA3/MJD arises

Importantly, ataxin-3 protects neurons from dying in fruit fly models of SCA3/MJD. How ataxin-3 protects neurons from death is unknown. We had proposed to investigate how ataxin-3 functions relate to SCA3/MJD pathogenesis by using genetics, morphological studies, and physiological assays. We also sought to discover proteins whose function may slow down or stop neurodegeneration in SCA3/MJD models in fruit flies.

The work that we conducted during 2012 and 2013, with great support from the NAF, led to some exciting findings about the normal function of ataxin-3 in intact animals. We discovered that ataxin-3 is quite adept at inhibiting the formation of toxic protein inclusions in flies and that it is through this activity that ataxin-3 rescues neurons from degeneration. We also found a molecular process that sends ataxin-3 for degradation; we hope to exploit this mechanism for therapy. Through other studies, we discovered several enzymes which, when inhibited, suppress degeneration caused by ataxin-3 in fruit flies. Some of this work has been published in ►►

scientific journals, and some is in the process of being evaluated for publication. We are moving forward with genetic and molecular studies to come up with strategies to suppress degeneration in SCA3/MJD and to enhance the clearance of

toxic ataxin-3 protein for therapy. We are also exploring the possibility of utilizing some of the enzymes that we have uncovered as neuro-protectors in this debilitating disease. ❖

### Post-Doc Fellowship Award

## A Role for ADAR and TDP-43 RNA Binding Proteins as Key Mediators of Toxicity in the Autosomal Dominant Spinocerebellar Ataxias

By Clare Louise van Eyk, PhD  
University of Adelaide, Australia

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

The spinocerebellar ataxias (SCAs) are a group of diseases characterized by progressive degeneration, primarily in a region of the brain called the cerebellum; a part of the hind-brain that controls motor coordination, balance and muscle tone. At present, there are 28 known SCAs with an autosomal dominant pattern of inheritance, meaning that they equally affect males and females and that the gene with the disease-causing mutation dominates the unaffected copy of the gene. Of those for which the causative mutation is known, more than half are caused by a special type of mutation involving an increase in the number of repeats of a sequence in the DNA. The pathway leading from the initial mutation to disease symptoms is still not fully understood.

In order to address the gap in understanding of the mechanisms involved in the SCAs and other diseases caused by this type of mutation, we have developed disease models in the fruit fly. In flies, introduction of repeat sequences similar to those found in SCA affected individuals results in morphological and behavioral changes. We then genetically alter these flies and observe the effects on their morphology and behavior and thereby gain information about the steps in the disease

pathway.

In this research project, we investigated the effect of the repeating sequences on the structure of RNA and how this might be involved in disease. RNA is the intermediate molecule which gives a read-out of the DNA code, generally allowing the message it carries to be converted to a working protein. Where there is a repeating sequence in the DNA, this means that the RNA which gets produced also contains a repeat-



**Dr. Clare Louise van Eyk**

ing sequence and it is thought that in the case of diseases such as the SCAs, this repeating sequence causes the RNA to fold up in an incorrect way. This folding results in the RNA interacting inappropriately with other cellular factors. We

*Continued on page 22*

*A Role for ADAR and TDP-43 RNA...*  
Continued from page 21

have identified two candidate proteins which have reduced function or altered localisation in neurons from SCA patients and therefore are excellent candidates for a role in SCA pathology.

We now have several lines of evidence which support a role for these candidate proteins in mediating toxicity caused by misfolded RNA. Using our fly models, we were able to begin mapping out the pathway leading from the repeating sequence in the RNA to cellular dysfunction. We are currently troubleshooting a technique which will allow us to read the RNA code of our flies

and determine how it is altered as a result of changing levels of these candidate proteins. We also now have the tools to examine whether altering levels of these candidate proteins changes where in the cell the RNA is located. We aim to then use human tissue samples donated by individuals with SCA or other expanded repeat diseases to look for these same features and therefore verify their importance in mediating disease.

Funding provided by the National Ataxia Foundation has enabled the continuation of this research which we hope will ultimately result in the identification of key candidates for therapeutic intervention. ❖

## Post-Doc Fellowship Award

# Understanding the Protective Functions of the SCA3/MJD Protein Ataxin-3

*By Wei-Ling Tsou, PhD*

*Wayne State University School of Medicine, Detroit, MI*

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

The most common dominantly inherited ataxia in the world is believed to be Spinocerebellar Ataxia Type 3, which is also known as Machado-Joseph Disease. SCA3/MJD is caused by mutations in the protein ataxin-3. In an effort to understand the pathogenic underpinnings of SCA3/MJD and to find new avenues for therapy, we investigated the function of ataxin-3. Ataxin-3 is involved in cellular processes that dispense of proteins in need of being discarded, either because they have completed their function, or because they are abnormal. In the fruit fly, *Drosophila melanogaster*, ataxin-3 plays a peculiar role by suppressing neurodegeneration caused by toxic versions of itself as well as other proteins that cause ataxia or other types of movement disorders. We decided to investigate how ataxin-3 performs this protective function. Through a combination of genetics and biochemical assays, we discovered partners of ataxin-3 that are critically important for this protein to suppress degeneration. We also found that ataxin-3 has a great capacity to reduce or eliminate protein aggregates, a process this that is tightly linked to its protective function. We are currently conducting



**Dr. Wei-Ling Tsou**

further experiments to describe precisely the functions of ataxin-3 at the molecular level, in order to understand how this activity is perturbed in SCA3/MJD. ❖

Post-Doc Fellowship Award

## SCA5: Molecular Effects of Mutant $\beta$ -III Spectrin

Written by *Laura Ranum, PhD*  
*University of Florida, Gainesville, FL*

*The following is a research summary of a grant funded by NAF, received by Tyisha Hathorn, PhD, Post-doctoral Fellow in the laboratory of Dr. Laura Ranum, for fiscal year 2013.*

### SCA5 Mouse Model

Spinocerebellar ataxia type 5 (SCA5), is a disabling and progressive neurodegenerative disease. In 2006, the Ranum lab discovered that this form of ataxia is caused by mutations in the *SPTBN2* gene on chromosome number 11 (*Nature Genetics* 38:184-190). The *SPTBN2* gene is especially active in the production of its protein product,  $\beta$ -III spectrin, in cells within the brain called “Purkinje” cells. Purkinje cells are located in the coordination center of the brain known as the “cerebellum”. These cells are large important neurons in the brain that are especially vulnerable in SCA5 patients and the death of these cells over time causes people with SCA5 to lose the ability to coordinate their movements. Finding the SCA5 gene was an exciting discovery because it is helpful to diagnose ataxia patients and it meant we could now ask detailed questions to understand how mutations in this gene cause Purkinje neurons to get sick and to die. Understanding in detail how mutations in the *SPTBN2* gene cause ataxia is an important step to developing treatment strategies and a cure.

To begin to answer how mutations in *SPTBN2* cause SCA5, the Ranum lab developed a mouse model of the disease. These mice were developed and characterized by Dr. Karen Armbrust, a former graduate student, and others in the Ranum lab, including Dr. Tyisha Hathorn. To develop these mice we “cloned” or made copies of the *SPTBN2* gene with and without the SCA5-causing mutation and put it into mice. Families of mice with SCA5 were

generated, which allowed us to study the disease in this model in detail. We showed that the expression of mutant but not normal or “wild-type”  $\beta$ -III spectrin causes ataxia in these mice. Additionally, we showed that the SCA5 mutation prevented the spectrin protein from doing one of its normal jobs – anchoring the mGluR1a protein at the very fine tips called “spines” of the Purkinje neurons. These Purkinje cell spines receive signals from other neurons in the brain



**Dr. Tyisha Hathorn**

and mGluR1a is part of the machinery that allows Purkinje cell spines to receive brain signals from other neurons. In collaboration with Dr. Ebner’s laboratory at the University of Minnesota, we showed that SCA5 Purkinje cell neurons lose the ability to adapt to stimulation. Normally, stimulation of a neuron will strengthen the response of that neuron. This is similar to the strengthening of our muscles when we exercise. What we found is that the Purkinje cells in the mice with the SCA5 mutation do not respond to neuronal signals in the same way as the control mice, and the signals in the SCA5

*Continued on page 24*

*SCA5: Molecular Effects...*  
Continued from page 23

Purkinje cells did not get stronger. Overall, this result demonstrates that one important function of  $\beta$ -III spectrin in maintaining functional spines to receive brain signals is lost in SCA5.

In summary, we have developed the first mouse model to test the effects that disease causing SCA5 mutations have on the brain. We show that SCA5 mutations change the way Purkinje cells in the cerebellum receive and respond to signals in the brain and demonstrate that changes in this response are linked to changes in mGluR1 function. Importantly, these mice provide a new tool that can be used to test therapeutic strategies for SCA5. The results of our study were recently published in July of 2014 in the *Journal of Neuroscience* 34:9891-9904.

### **Identification and Characterization of Additional SCA5 Mutations**

A second part of Dr. Hathorn's efforts focused on the identification and characterizations of additional mutations in the *SPTBN2* gene that are found in individual ataxia patients and additional smaller families. For these experiments,

Dr. Hathorn has studied a number of new DNA changes in *SPTBN2* that were identified by Dr. Hathorn, Dr. Damaris Lorenzo (a former graduate student in the Ranum lab) and other collaborators. The first question Dr. Hathorn addressed is which of these variants cause disease and which are simply DNA variants in the population that are not disease causing? To sort this out, Dr. Hathorn has used a combination of approaches. First she has used computer modeling to predict how individual mutations are predicted to affect how the  $\beta$ -III spectrin protein folds – protein folding is very important for protein function. Second, she performed a series of laboratory tests to determine if individual DNA variants change the way that  $\beta$ -III spectrin protein interacts with other proteins. Third, she performed genetic comparisons to determine the frequency that each variant occurs among ataxia patients and if these changes are also found in the general population. We are currently writing a manuscript for publication that describes this work.

Finally, I would like to say that funding from the National Ataxia Foundation in support of this work came at a critical stage of this project and is gratefully acknowledged. ❖

## **Brain Donation Program**

There have been major advances in recent years in understanding ataxia as it affects the brain. Enhancements in brain imaging, gene testing, and rating scales provide a clearer picture of how ataxia affects the brain, but there is also a need to better understand the changes that occur in the brain. A brain donation is a valuable gift to the ataxia research community. Scientific discoveries from studies of postmortem human brain tissue have provided significant contributions to better understand ataxia.

It is important that family members support the decision of the brain donor, as the next-of-kin will become the research participant advocate upon the death of the donor. Brain donation must take place within 24 hours of death, and preferably sooner, so it is essential that a donor

complete the necessary steps well in advance, such as selecting a funeral home or mortuary. If the donor is able to be seen at an ataxia clinic at least once in his or her life, this may eliminate all or most of the costs associated with brain donation.

The National Ataxia Foundation will assist in beginning a pre-plan process for brain donation, however NAF is unable to provide funds for additional funeral expenses that may be incurred. If you have Friedreich Ataxia and would like information, please contact Dr. Arnulf Koeppen at (518) 626-6377. For other forms of ataxia, please e-mail [susan@ataxia.org](mailto:susan@ataxia.org) or call the NAF office at (763) 533-0020 to request a Brain Donation Pre-Plan Form.

Thank you for your interest in this program.

Post-Doc Fellowship Award

# A New Induced Pluripotent Stem Cell Model of SCA2

By Robert Moccia, MD, PhD

Massachusetts General, Harvard University, Cambridge, MA

*The following is a research summary of a grant funded by NAF for fiscal year 2013.*

Spinocerebellar ataxia type 2 (SCA2) is a neurodegenerative disease mainly characterized by impaired coordination, balance, speech, and eye movements. There can also be additional features such as peripheral neuropathy (e.g., loss of sensation in the hands and feet, dulling of reflexes), muscle weakness, and memory problems. Scientists know that this disease results from an increase in the size of a specific region in a protein called ataxin-2. The expanded part of the protein consists of a single repeated amino acid called glutamine. In fact, SCA2 is one of a small number of clinically distinct neurological diseases that share these so-called polyglutamine expansions. While we have learned much about the disease by studying it in other animals like mice, there is still a great deal more we need to know about the normal function of ataxin-2 in human neurons and the consequences of polyglutamine expansion that lead to SCA2.

Historically it has been very challenging to directly study living human neurons, but the landscape changed dramatically in 2007 with the production of the first human induced pluripotent stem cells (iPS). As Dr. Clive Svendsen eloquently explained in the Summer 2011 issue of *Generations*, this is a game-changing technology that allows us to take a small skin sample from a person and convert it into a type of cell capable of producing virtually any tissue type in our body, including neurons. These cells can be maintained in the lab for very long periods of time and can even be shared with other research groups. Importantly, when we make these iPS cells from an individual with an inherited ataxia we can

capture the actual mutation that caused their disease. This means that we can now study the effects of disease-causing mutations like polyglutamine expansions directly in human neurons. This is a very powerful complement to the more traditional animal models which often do not tell researchers the whole story of what's happening given that mice and humans have important biological differences.

With the support of the National Ataxia Foundation, we have created a new iPS model of SCA2 and have begun using it to study how the polyglutamine expansion affects neurons. We used a newer method for making the iPS cells that avoids exposing them to viruses that could potentially cause damage to the cell's DNA. The SCA2 iPS cells we made can be converted to neurons very efficiently – indeed, as efficiently as iPS cells made from the patient's father who does not have SCA2. This model system provides a near limitless supply of human SCA2 neurons that can be used to unravel the mysteries of how the polyglutamine expansion leads to disease. We expect it will also be a very valuable resource for screening potential drugs to treat SCA2 in the near future. ❖



**Dr. Robert Moccia**

Post-Doc Fellowship Award

# Determination of the Toxic Species in Fragile X-Associated Tremor Ataxia Syndrome

By Fang He, PhD

University of Michigan Medical School, Ann Arbor, MI

The following is a research summary of a grant funded by NAF for fiscal year 2013.



**Dr. Fang He**

Expanded DNA repeat sequences are a common cause of cerebellar ataxia. These repeats can elicit cerebellar degeneration as either RNA or as protein, but the relative contributions of each toxic species to the development of disease in patients is unknown. Fragile X-associated Tremor Ataxia Syndrome (FXTAS) is an inherited cause of cerebellar ataxia that results from such a repeat. To date, most studies have focused on how the FXTAS repeat as RNA can elicit neurodegeneration. However, our group has now found that the repeat can also be translated into a potentially toxic protein. Using fly models of this disease, we systematically evaluated the relative toxicity of the repeat RNA and the polyglycine protein. We found that expression of repeat RNA and polyglycine protein in a single construct is sufficient to elicit toxicity.

However, expression of the repeat RNA alone is not sufficient to induce significant toxicity in the simple assays of neurodegeneration in flies. In contrast, enhancing polyglycine protein expression from constructs expressing CGG repeats augment toxicity. These results suggest a disease mechanism that requires the presence of both repeat RNA and polyglycine protein to recapitulate FXTAS pathogenesis. ❖

## FDA's Patient Representative Program

The Patient Representative Program is the US Food and Drug Administration's (FDA) method of bringing patient or family caregivers into discussions about new and already approved drugs. Patient Representatives represent the unique perspective of those who are affected by a serious disease.

Because the FDA is committed to making more opportunities for patients to participate in the FDA decision making, they are currently recruiting patient representatives.

If you are interested in being considered for the Patient Representative Program, please follow this link: <http://patientnetwork.fda.gov/about-patient-representative-program>. You will be asked about yourself as a family member or individual who is affected by ataxia.

If you are selected to be a Patient Representative for the FDA, the National Ataxia Foundation would be interested in hearing about your experience in participating in this important program for drug approval.

# Progression of Ataxia

*Speaker: Susan L. Perlman, MD*

*Clinical Professor of Neurology and Director, UCLA Ataxia Clinic*

*Medical Director, National Ataxia Foundation*

*The following was a presentation that Dr. Susan Perlman gave at the 2014 NAF Annual Membership Meeting in Las Vegas, NV and has been edited for publication in Generations.*

Dr. Perlman stated: The following personal financial relationships with commercial interests relevant to this presentation existed during the past 12 months:

- No personal relationships to disclose.
- Research relationships include: Edison Pharmaceuticals, ViroPharma, Teva Pharmaceuticals.

I am currently involved in some drug trials working with pharma. I have no personal benefit from them, just all the excitement of finally getting into treatments for ataxia.

It is always delightful to come to the National Ataxia Foundation's Annual Membership Meetings and see old friends, meet new people, and get new ideas. I revised my presentation three or four times in the course of the past few days because I kept bumping into new information. It's a good opportunity for us all to increase our knowledge and our resources in ultimately finding a cure for all the forms of ataxia.

Many questions that we have been working on with our patients and our mission over the years have been:

- We want to help them get a diagnosis,
- We want to maximize the quality of life,
- We want to help them get involved in research,
- And now, we are working to become a platform for clinical trials.

My experience in the recent Ataxia Investigators Meeting has increased the urgency of that last bullet point item. We need to have sites that are ready to step up after a notice of a clinical trial and get something rolling in three months which

is usually the deadline that you are given. The shorter the better; when a drug company wants to move forward, they don't want to wait for a year while you get your act together.

The aspects that I will present now are the progression of ataxia, focusing on the questions:

- Will it get worse?
- How bad will it get?
- How soon will it get there?

What we know is:

- If it is genetic, there are no cures at this time. The symptoms will progress.
- If it is non-genetic (or minimally genetic), unless a treatable cause is found, the symptoms will progress.
- If a treatable cause is found early enough, symptom progression can be halted and maybe even reversed a little bit or even recovery.

Although I have had only two patients who completely recovered. One was an undiagnosed Vitamin B12 deficiency. I am not sure how that slipped past the primary care doctor. The other was a woman who presented with some dizziness and mild cerebellar symptoms. We worked her up, this was 25 years ago so we really didn't have a lot of things that we could test for, but she came back a year and a half later and was totally normal. In the interim she had been diagnosed with breast cancer and had been successfully treated. It was probably a paraneoplastic set of symptoms that the breast cancer was causing and she fully recovered. So the earlier you can work on something that is treatable, the better you will

*Progression of Ataxia*  
Continued from page 27

do in the future.

- If the ataxia was caused by a one-time damage to the cerebellum, like a stroke or something similar, symptoms may improve slowly over time. I have seen damage of that nature improve over three, four, five years and continue to show slow improvement. But one thing I have noticed with some of my older patients who had a head injury as a youth or as a child, is that once you pass the age of 50 and aging kicks in, the effects can sometimes undermine the recovery that you have achieved and the effects of aging can cause worsening again.

So for anybody who is over 50 with ataxia of almost any cause, aging is going to become part of the equation. And that, for sure, is something that we do not have a cure for, yet.

You do need to be on the lookout for other medical or surgical illnesses, certain medications, poor diet, lack of exercise, sleep disturbances, vitamin deficiencies, toxic exposure, alcohol or recreational drug use, and depression, which can all worsen symptoms of ataxia and may actually speed up progression.

NAF shared some of the concerns and questions that people have expressed:

- Experience of fear and anxiety regarding the progression of the condition.
  - How long can this condition last?
  - What are the symptoms for late-stage ataxia?
  - Do you have any information regarding progression of the disease?
  - Provide me with some input of what the future could hold.

**There have been good efforts made to determine the progression of ataxia.**

Once you have a diagnosis of ataxia, you want to have some sense of what you can do to help yourself and what you can expect so that you can anticipate and prepare. Certainly knowing what to expect can help patients and their families

become more proactive about their treatment and lifestyle. Physician researchers knowing what to expect can design better clinical trials of new treatments.

We know that not all forms of ataxia progress at the same rate. Not all forms of ataxia cause serious complications or problems with speech, swallowing, hand coordination, bowel and bladder function, or memory. But we know that all the ataxias impair the ability to stand and walk and increase the risk of falls. It should be a shared goal with all clinicians treating people with any form of ataxia to keep ataxians safely on their feet for as long as possible.

The EuroSCA group did a study of falls. They looked at their large cohort of individuals in their natural history study and determined that falls are a significant risk. One of the things contributing to the risk of falling wasn't just the ataxia, it was the other non-ataxia symptoms that could be interfering with the ability to compensate and catch your balance. They developed a scale, which was a check list, on the inventory of non-ataxia symptoms. They asked questions about non-ataxia symptoms from the list shown below, such as, "Do you have this...? Do you have that...?"

**Inventory of Non-Ataxia Symptoms**

*Developed by EuroSCA-Jacobi, H., et al. 2013  
More likely to be seen in certain SCAs, FA, and MSA.*

*Less likely to be seen in idiopathic late onset cerebellar ataxia.*

*But can occur in anyone (having ataxia does not make you immune to other diseases).*

*Hyperreflexia, Areflexia, Extensor plantar, Spasticity, Paresis, Muscle atrophy; Fasciculations, Myoclonus, Rigidity, Chorea/ dyskinesia, Dystonia, Resting tremor; Sensory symptoms, Urinary dysfunction, Cognitive dysfunction, Brainstem Oculomotor.*

They found that some combinations of some of these symptoms, like brisk reflexes and loss of reflexes were markers for changes in other ►►

systems. Spasticity, where the muscle tone is increased, weakness in muscle, atrophy of muscle, tremors of various types, (anything outside of the cerebellum), if the symptoms were bothersome enough, they could impair performance and increase the risk of falling. These multiple extra symptoms are more common in certain spinocerebellar ataxias, Friedreich ataxia and multiple system atrophy. They're relatively rare on complicating any of the other ataxias.

Certainly if you have balance problems, and you have been compensating pretty well, so you have done physical therapy, you've got a good exercise program, you've got your walker, and then all of a sudden you're falling with your walker overnight, it's not that we think your disease has suddenly changed course, but we wonder, "Do you have a bladder infection? Have you been constipated for two weeks and it is starting to backup into your system?"

There are certainly other things that could come in that are totally unrelated to your ataxia diagnosis. Just because you have ataxia doesn't mean that you are immune to other problems. You do need to be alert to the possibility.

Exercise always helps. We have heard several people comment on this and we cannot stress it enough.

There have been clinical trials showing that it helps and there are more trials planned to try to select the specific types of exercise that can be most helpful for ataxia. There are an increasing number of clinical trials barreling toward us and so you certainly want to be in the best shape in order to participate. For many clinical trials one of the entry criteria is being able to walk, even if it is with an assistive device like a walker. So it is to everybody's benefit to really focus on keeping your physical condition in as good a shape as possible.

We have focused on rehab and on treatable

causes for the ataxias, but there are a number of medicines that can be tried for symptoms of ataxia. The National Ataxia Foundation has a fact sheet that lists these medications and what symptoms they may treat. If any of your doctors tell you, "You have ataxia and there's nothing that can be done," that could not be further from the truth. Patients have experienced benefits from the rehab options we know about and there are also symptomatic medications that can be of great help. (To download a copy of the Medications fact sheet, go to [www.ataxia.org](http://www.ataxia.org) or e-mail [naf@ataxia.org](mailto:naf@ataxia.org) and request a copy by mail.)

**Natural History Studies** try to show the average changes of ataxia over time. These studies are important because they give us some statistically documented way to predict what is going to happen in planning a clinical trial design. They can provide a baseline against which to compare your own symptoms and give some guidelines as to what the average person with a certain ataxia diagnosis goes through, but they may not provide the whole answer regarding the progression of ataxia.

**Rating scales**, that have been validated and tested at multiple sites in Europe, United States and Australia, are used by ataxia researchers for Natural History Studies. The scales are derived from parts of the neurologic exam that seem to be most sensitive to track changes in ataxia. Below is a list of the ataxia rating scales:

- ICARS (International Cooperative Ataxia Rating Scale)
- SARA (Scale for Assessment and Rating of Ataxia)
- FARS (Friedreich's Ataxia Rating Scale)

**Patient Related Outcome Measures** collect information about how the ataxia patient feels their symptoms are doing. This is required by the



**Dr. Susan Perlman**

*Progression of Ataxia*  
Continued from page 29

FDA and the FDA is very interested in hearing things that a patient may report such as: fatigue, pain levels, and quality of life. When investigating a possible new drug, the FDA does not want to just hear: “Finger tapping was two seconds faster.” They want to hear that your handwriting is better, your ability to use a computer is better, you can place your key in the lock better. We have a number of questionnaires that we give to people to try to help establish which of the patient related reports are going to be the most helpful.

Let’s look at the SARA scale, it has eight components which include: Gait, Stance, Sitting, Speech disturbance, Finger chase, Nose–finger test, Fast alternating hand movements, and Heel–shin slide. So in the Gait portion you can have a score between 0 and 8. Zero points means that you are walking normally, one point means that there is minimal changes and the scale continues up to a score of eight which is being wheelchair bound and unable to walk. So to worsen by one point could mean going from walking down a hall and occasionally touching the wall to walking down a hall and having to use cane.

Worsening by a half-point could mean you developed more noticeable tremor in one hand. So 2.2 points is not a huge decline in performance. It is a scale that we can quantify, we can do this from visit to visit; it’s pretty accurate. However, there are people who when I tell them that they look about the same; they were 21.5 points last time, and they are still 21.5 points this time, they’ll tell me, “But my walking is worse.” So your reports are as important as any of the scales that we use.

In a study of a few 100 people over two years using the SARA scale, below is what the research found:

- SCA1 seemed to worsen by 2.2 points over two years.
- SCA3 by 1.6 points.
- SCA2 by 1.4 points.

- SCA6 by 1.4 points.

Progression seemed to be fastest in the patients with SCA1 and SCA2 who had larger mutations and earlier age of onset. This makes sense from what we know about these disorders. But what can you possibly make out of 2.2 points? What does that mean? Most of the people were starting from a baseline of about 15 points, give or take a few. So worsening by 2.2 points out of 15 to start, that is 15% worse.

So what does that mean for the next two years and the next two years after that? Not everything can be predicted. There are many things that influence the progression of ataxia.

Looking back at your own performance or the performance of other family members may not be an accurate measure of how things are going to go in the future. Many factors may influence the progression of spinocerebellar ataxia:

- Size of genetic mutation – however that may only explain only 50% of progression that we see. Some patients have a relatively large mutation but have unexpectedly slow progression.
- Age of onset of first symptoms – when symptoms first come on at a younger the age, it is expected that the person might get worse more quickly.
- How the disease progressed in other family members.
- How the disease progressed up until now.
- Environmental or lifestyle factors that may help or undermine the brain’s ability to compensate.
- Presence of other medical or surgical conditions.

Similar Natural History studies were calculated for Friedreich’s Ataxia, again in hundreds of patients, which you need in a rare disease to be able to say anything coherent. You can’t look at 10 patients and say, “They were all still walking after 10 years.” Or, “None of them were walking after 10 years.” You have to look at a larger group.

In Friedreich ataxia (FRDA), change was most reliably seen over one to two years of follow ►►

up. Dr. David Lynch did an analysis over the course of two years. The rate of change from year to year for mild to moderate symptoms was about the same. There really weren't that many surprises. Things slowly changed. But once you got out to having pretty noticeable symptoms, probably in a wheelchair, the rate of change slowed down. So that in the later stages of Friedreich ataxia, changes, or rates of progression, tend to be slower. There may be of group of nerve cells that are just resistant to all the stress that is going on from that gene and you just can't knock them off anymore. They are going to be there and they are stable and they're going to maintain some level of baseline performance through the later stages of the disease. The pattern, when he separated it out into people with small, medium or large mutations, was similar. So at least in FRDA, in the later stages, there aren't going to be a lot of surprises of rapid progression.

There were two European studies of Multiple System Atrophy (MSA) done and, again the researchers used a large number of patients, 100 or more. Below is a comparison of the two studies, looking at changes in MSA in the 20 years from 1994 to 2013. The two studies showed:

### 1994 Study

- Average age of onset 53 years old – About half started with Parkinson's and half with ataxia. At some point they developed both sets of symptoms
- Spasticity in 61% – This can be very limiting for walking because muscle tone makes it hard to move or catch yourself if you're falling
- Autonomic symptoms in 97% – Low blood pressure, bowel and bladder problems were ultimately seen in almost everybody with MSA to some extent or another.
- Wheelchair bound by five years – Most of

them were wheelchair bound after five years with the illness. I have seen people use a wheelchair at year three, but I have also seen people go out to nine or 10 years, still able to use a walker however, the average group was five years.

- Median survival 9.5 years – This means that they took all of the 100 people in their study and divided it up at the 50 person mark and put them all in a row – those who passed away here, who passed away later, who passed away the latest, who are still alive. It ended up about half the people had passed away and half the people were still living.

“  
**Not everything  
 can be predicted.  
 There are many  
 things that  
 influence the  
 progression  
 of ataxia.**  
 ”

### 2013 Study

- Average age of onset 56 years old – Over two years patients got worse by 50% over baseline (compared with SCA which worsened by 15%)
- Median survival 9.8 years – The survival rate is just about the same as the 1994 study. So that the cohort of people they studied – half of them had passed away, but half of them were still living, still ambulatory using aids or in wheelchairs.
- Shorter survival with severe Parkinsonism or bladder retention – People who passed away earlier, who had a shorter survival, were associated with more severe Parkinson's type symptoms and that probably relates to more difficulty moving around. Parkinsonism, besides causing shakiness can very early in the disease make you very stiff and very slow and can really impair the ability to walk, so you sit more. The more you sit, the more likely you are to get infections and other complications of sitting a lot and also severe bladder retention which is a site for infection and an infection can become sepsis and you can end up in the hospital.

So over these years, we have a good idea of

*Progression of Ataxia*  
Continued from page 31

---

what MSA is, however, there is a lot of variation.

For other non-genetic ataxias, the rate of progression may give clues to the underlying cause which may be treatable. If your ataxia comes on quickly and progresses quickly you think of certain things; if it comes on slowly and progresses slowly you think of other causes. So often the time course, the progressive course, can give you a clue to what really is the cause.

- **Acute** (*minutes to hours*) – traumatic, vascular, metabolic/toxic, infectious, inflammatory
- **Sub-acute** (*days to weeks*) – post traumatic, metabolic/toxic, infectious, inflammatory, neoplastic, paraneoplastic
- **Slowly progressive** (*months to years*) – congenital, metabolic/toxic, infectious, inflammatory, neoplastic, paraneoplastic, degenerative, genetic
- **Episodic** – vascular, metabolic-toxic, inflammatory, genetic
- **Static** – congenital; residual of trauma, vascular insult, or infection

Research may not have the answers specific for those with non-genetic ataxias. But, we should be monitoring them closely so that we can be proactive in dealing with whatever challenges the ataxia presents. It is important for the patient and family to have some idea what to expect and to know what to watch for, to help them make decisions about long-term care and hospice care.

- **Untreatable rigidity, autonomic failure**, where the blood pressure just bottoms out and can't be brought back up and bulbar symptoms (central or obstructive apneas, stridor, choking/aspiration) can lead to death in under a year if they can't be treated. However, often there are treatment strategies for these and you don't want to ignore them. You want to treat them quickly.
- **Increased falling or becoming chair- or bed-bound** may lead to life-threatening complications. You can get injuries, bedsores,

infections; these are all preventable for the most part and certainly treatable.

- **Dementia, behavioral problems, and depression** make management, compliance, and care much more difficult. We need to identify these things and work with them so they don't become part of the problem.

- **Pain and fatigue** are treatable. We still have a lot more to learn about fatigue in all of the ataxias. In all of the neuromuscular diseases fatigue is a big problem and it's not just because your muscles are weak, so I am hoping to see more research in that area.

#### **My Observations:**

- I believe we will see disease modifying treatments for ataxia in my professional lifetime. There are currently 48 open ataxia studies posted at *ClinicalTrials.gov*. Thirty-two are interventional studies, testing symptomatic and disease modifying therapies which could, with others still in development, be available to us over the next ten years. It is very important that every person with ataxia join in one of the registries and be ready to participate in bringing these treatments into clinical practice.

- In SCA, the first five years are manageable, the next five years may require medical intervention, and after 10 years there may be significant limitations that may cause you to limit activities.

- In Friedreich ataxia, there may be motor plateaus where nothing new happens or changes, followed by falling off the plateau and then stabilizing again. Sometimes exercise can boost you back up onto that plateau, so it's not that steady 15% downhill skateboard slide.

- The literature says things like: Death 10 to 20 years after onset for SCA 1, 2, 3 and for Friedreich ataxia the mean age of death (due to cardiomyopathy) at 38 years old; but the exceptions can be more common than the rule.

#### **Let me tell you about Scott who has SCA2:**

He was not able to attend this meeting so he and his wife asked if I could tell you some of the things that he has been able to do. He had onset of symptoms at age 25 and he is now 46. ►►

He's been symptomatic for 21 years. According to the statistics he should have been dead two years ago. However, he exercises, he gets out on his trike, he plays with his kids, he stays active and stays healthy. He is doing everything he can, proactively.

*"80 percent of success is showing up."*  
 — Woody Allen

**In closing I want to say:**

- Thank you for helping to make this Annual Membership Meeting the success it has been.
- Thank you for supporting each other.
- Thank you for supporting the National Ataxia Foundation.
- Thank you for supporting ataxia research by

signing up for the CoRDS Ataxia Patient Registry (see page 4 for information on the CoRDS registry) and volunteering for clinical trials. This is a huge commitment on your part, even if your travel costs are paid, you still need to give the time and the agony of blood draws or whatever the researcher needs, but it is so important that you step up and help us out with this.

- Thank you for keeping ataxia awareness high.

A big THANK YOU to NAF, and all the groups, individual donors and foundations that have supported our work at UCLA. There are now a lot of people at UCLA who are interested in ataxia.

Thank you.



# International Ataxia Awareness Day

— Thursday, September 25, 2014 —



## How Did You Participate in IAAD?

Tell us how you recognized International Ataxia Awareness Day (IAAD) this year. Share a photo with us to be included in a future issue of *Generations*. Please e-mail your story/photo to [joan@ataxia.org](mailto:joan@ataxia.org) or mail to the National Ataxia Foundation, Attn: *Generations* Editor, 2600 Fernbrook Lane, Suite 119, Minneapolis, MN 55447-4752. Sharing your stories on how the day was recognized could live on in a future issue of *Generations*. Please send your articles, photos, and proclamations so the entire NAF community can relive this historic day. Thank you.



---

## From the Desk of the **Executive Director**

---

*By Michael Parent, NAF Executive Director*

As the autumn colors begin to display their annual brilliant hues of red, yellow, and orange, it is also time for the NAF Annual Ataxia Research Drive to begin.

This year the Annual Ataxia Research Drive will begin on October 15 and each research dollar received until December 15 will be matched dollar for dollar up to \$200,000 by our anonymous donor.

NAF has received 106 applications from scientists in 19 countries, the largest number of applications and the most countries applying for research funding in the history of NAF. To fund the best of those studies, we will need your support.

What does it mean when you support the NAF Annual Ataxia Research Drive? You will be:

- Supporting the best science in the world
- Increasing the number of young investigators working on ataxia
- Doubling each dollar you donate by the generous matching gift of our anonymous donor
- Supporting translational research
- Funding more research ... finding more answers
- Increasing the opportunity for scientists to see an often five- to 10-fold increase from other sources once an NAF grant is awarded
- Developing partnerships with others to help fund additional ataxia research

- Finding additional ataxia genes
- Accelerating effective treatments
- Giving hope for a brighter future

Your research gift is a multiplier. Yes, each dollar you donate to the research drive is doubled. When NAF partners with another research group, your research gift is doubled again. The scientists awarded an NAF research grant many times sees a five- to 10-fold increase from NAF's initial research grant ... so your research gift can multiply many times over. You have a real opportunity to make a significant impact in furthering ataxia research by donating to the NAF Annual Ataxia Research Drive.

We have from October 15 – December 15 to raise \$200,000 for research to reach the \$200,000 challenge grant. Over the past three years the membership has responded brilliantly and generously in meeting the challenge of these research matching grants and because of your donations more crucial research was funded to help find answers in ending ataxia.

I would like to take this opportunity to sincerely thank all of our amazing donors from last year's challenge grant who made it possible for NAF to support over \$1 million dollars in funding 24 ataxia research studies. We humbly ask for your continued support of the 2014 Annual Ataxia Research Drive and to encourage others to also support these most important research efforts. Thank you. ❖

---

## NAF's Travel Grant Program Needs Your Support

For those with ataxia, traveling to the National Ataxia Foundation's Annual Meeting (AMM) may be financially difficult.

Our travel Grant program was created to assist individuals with some of the costs associated with attending the AMM.

You can help an individual attend the AMM by

making a donation to our Travel Grant Program today! Simply designate your donation to the AMM Travel Grant Fund to make an impact.

We thank you for your support and for making the AMM experience possible for an individual affected by ataxia who may not have been able to attend without your help. ❖

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

## **National Ataxia Foundation 58th Annual Membership Meeting**

**★★★★ New Date: March 6-8, 2015 ★★★★★**



***Join us in Denver for the Annual Membership Meeting!***

Sheraton Downtown Denver is pleased to provide the facilities for the 2015 AMM.

### **Standard Room Reservations**

Standard room reservations at the Sheraton can be made online at

<https://www.starwoodmeeting.com/Book/naf2014>

Guests who prefer to phone in their reservation can call Hotel Reservations at 1-888-627-8405 and ask for the National Ataxia Foundation's group rate, which is under the group name "Natl Ataxia Foundation 2015."

### **ADA Room Reservations**

**Please note all ADA rooms must be reserved through the NAF office starting on October 1 at noon CDT by contacting (763) 553-0020 or [lori@ataxia.org](mailto:lori@ataxia.org). Calls or e-mails prior to noon CDT on October 1 to reserve an ADA room cannot be honored. There are a limited number of ADA rooms. Reservations for the ADA rooms will be made on a first-come first-serve basis.**

The NAF special discounted group rate is \$159 + tax. Please note there is limited availability on discounted rate rooms.

### **Meeting Registration**

Registration for the 2015 NAF AMM will open in mid-December. Please make sure you take advantage of the early registration discount rate. In addition, members of the NAF pay a lower registration fee to attend the annual membership meeting. If you are not currently a member of the Foundation go online at [www.ataxia.org](http://www.ataxia.org) or call the NAF office at (763) 553-0020 to become a member or renew your membership. The meeting registration fee includes attendance at all the sessions, light appetizers at the Welcome Reception and a delicious plated meal at the Banquet.

For the latest information on conference registration, program schedule, and area information, keep checking NAF's website, [www.ataxia.org](http://www.ataxia.org).

**2015 NAF Annual Membership Meeting "Support Our Conference" Campaign**

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

For more information on Denver visit [www.denver.org](http://www.denver.org).

## The National Ataxia Foundation 58th Annual Membership Meeting

# “Soaring Mile High for a Cure”

★★★★ **New Date: March 6-8, 2015** ★★★★★

The National Ataxia Foundation (NAF) Board of Directors and the National Ataxia Foundation North Central Region invite you to attend the 58th Annual Membership Meeting (AMM). Please join us at the Sheraton Denver Downtown in Denver, CO to learn, share, network, have fun, and enjoy the sites.

The 2015 AMM will bring together NAF members and their families not only to meet and learn from world-leading ataxia researchers and clinicians, but also to build new friendships and reunite with old friends. Come and be part of the largest ataxia gathering in the world!

NAF continues to strive to offer quality and important programs and services to the ataxia community. One of the centerpieces of the NAF is the annual membership meeting. The 2015 AMM will be held at the Sheraton Downtown Denver.

The hotel contacted the NAF in August 2014 to offer an exceptional meeting space opportunity that was not previously available. The new space will better serve our attendees at the 2015 meeting by providing greater accessibility and a more enjoyable experience. To accept the new space, the meeting date needed to be changed one week earlier to March 6-8, 2015.

The decision to proceed with a new date was not made lightly and was based solely on NAF's continuing efforts to offer a high quality program. We apologize for any inconvenience that the change of dates may have caused.

Thank you for your understanding and support.

When registration opens, you are encouraged to take advantage of the early registration discount rate. In addition, members of the National Ataxia Foundation pay a lower registration fee to attend the Annual Membership Meeting. If you are not currently a member of the Foundation, if your membership renewal is coming soon, or if you are uncertain of your membership status, use this opportunity to go online at [www.ataxia.org](http://www.ataxia.org), call (763) 553-0020, or e-mail Joan at [joan@ataxia.org](mailto:joan@ataxia.org) to become a member or renew your membership. Take time now to confirm your membership status and save money when you register for the 2015 Annual Membership

Meeting. The meeting registration fee includes attendance at all the sessions, light appetizers at the Welcome Reception and a delicious plated meal at the Banquet.

The National Ataxia Foundation is able to offer Travel Grants to help with a portion of the travel costs associated with attending the meeting. Adults or children with ataxia are eligible to apply for a travel grant. Visit the NAF website, [www.ataxia.org](http://www.ataxia.org), to download the application or contact Lori Shogren, [lori@ataxia.org](mailto:lori@ataxia.org), at (763) 553-0020 to request an application by mail.

The complete meeting schedule, events and registration forms will be listed in the winter ►►

2014–2015 issue of *Generations* and on NAF's website when available. The following is a brief program overview. Additional information can be found on the NAF website, [www.ataxia.org](http://www.ataxia.org).

### Pre-Meeting Activities

#### Thursday, March 5

- Registration Opens: 9 a.m. – 8 p.m.
- Leadership Meeting: 1 – 3 p.m.
- Walk n' Roll Meeting: 4 – 5 p.m.

### Program Overview

#### Friday, March 6

- Registration: 8:30 a.m. – 5 p.m.
- General Sessions: 9 a.m. – 12:15 p.m.
- Exhibitors: 8:30 a.m. – 5 p.m. (set-up available at 8 a.m.). Exhibitors will be present from Friday morning through Sunday morning as their schedules permit.

• Silent Auction Bidding: 8:30 a.m. – 5 p.m. All items being donated for the Silent Auction are due in the Silent Auction room by Friday, March 6 at 4 p.m.

• Activity Room: 10 a.m. – 5 p.m. The activity room is open to all ages. Persons under the age of 12 must be accompanied by a parent or guardian who is age 18 or older.

• Birds of a Feather (Group A): 2 – 5 p.m. This year the Birds of a Feather will be offered on Friday and Saturday afternoon. The schedule will be available in the Winter issue of *Generations*. Please review the schedule for your specific session.

• Meet & Greet Reception: 7 p.m. Light appetizers.

#### Saturday, March 7

- Registration: 8 a.m. – 5 p.m.
- Exhibitors: 8 a.m. – 5 p.m. Exhibitors will be present from Friday morning through Sunday morning as their schedules permit.
- General Sessions: 8:30 a.m. – 12 p.m.
- Silent Auction Bidding: 8:30 a.m. – 12:30 p.m. **Winners must pick up and pay for their items from 4 – 7 p.m. on Saturday.**

- Activity Room: 10 a.m. – 5 p.m.

- Birds of a Feather (Group B): 2 – 5 p.m.
- Saturday Evening Banquet: 7 p.m. Delicious plated meal and entertainment.

#### Sunday, March 8

- Registration: 9 – 11 a.m.
- Exhibitors: 9 – 11 a.m.
- General Sessions: 9 a.m. – 12:30 p.m.

### About Denver – “The Mile High City”

Welcome to Denver, where 300 days of sunshine, a *thriving cultural scene*, *diverse neighborhoods*, and natural beauty combine for the world's most spectacular playground. A young, active city at the base of the Colorado Rocky Mountains. Denver's stunning architecture, award-winning dining and unparalleled views are all within the walking distance from the 16th Street pedestrian mall, a mile-long pedestrian promenade of outdoor bistros, microbreweries, shopping and entertainment. The 16th Street Mall offers a *Free and Accessible bus* that travels 16th Street on a regular basis daily. Free and close attractions to the conference hotel are the *Federal Reserve Money Museum* and *Denver U.S. Mint*. Tours of the *Denver U.S. Mint* fill up quickly, so please make your reservation early if you are interested in taking a tour.

Please visit [www.denver.org](http://www.denver.org), for a complete list of attractions, planning, and transportation information.

### Sheraton Denver Downtown

Sheraton Denver Downtown is the official conference hotel of the 2015 NAF Annual Membership Meeting. Sheraton Denver Down-town is located on the *16th Street Mall* and 25 minutes from the Denver International Airport at 1550 Court Place, Denver CO 80202.

For your stay and planning purposes at the Sheraton, the following information is provided. Additional details will be listed in the winter 2014–2015 issue of *Generations* and on the NAF website.

## 2015 AMM Overview

Continued from page 37

- The majority of guest rooms and meeting space being held for NAF is in the Plaza Tower of the hotel. Access with a Room Key is required in the elevators to reach floors with guest rooms. If you require assistance with the elevator to reach your guest room floor please see a Front Desk Agent.

- For room reservation information please refer to the AMM announcement on page 35.

- If you need ADA equipment you are encouraged to bring those items with you or make arrangements to rent equipment locally. NAF is unable to provide ADA equipment however the Sheraton Denver Downtown may have some extra shower chairs, grab bars, or detachable shower heads available. Be sure and request these items when making your reservation if needed. **The width of the bathroom door in the standard Plaza Tower guestrooms is 33 inches.** The hotel is able to take the bathroom room door off if necessary in Plaza Tower guest rooms. **The width of the bathroom door in the standard Tower guest rooms is 21 ½ inches.** The height of the beds in every guest room is 28 inches.

- Parking at the Sheraton Denver Downtown – Valet parking and Self-parking are avail-

able. NAF has discounted parking rates available for AMM attendees. The Sheraton Denver Downtown parking garage is located underground with direct access via elevator to the hotel lobby and has a **clearance of 6'4"**.

- Oversized parking – The Sheraton Denver Downtown Hotel can accommodate a limited number of oversized vehicles at a lot that is nearby, but not directly located at the Hotel. For more information about the oversized parking, contact Towne Park at (303) 352-2454.

- Service Dog Information – The service dog relief area at the Sheraton Denver Downtown is located at the Civic Center Park

- Visit the Sheraton Denver Downtown website, [www.sheratondenverdowntown.com](http://www.sheratondenverdowntown.com), for more information.

The following information can be used as a guide as you plan your stay and transportation needs in Denver. The National Ataxia Foundation does not endorse products, services, or manufacturers. Those mentioned are included for your information only. The NAF assumes no liability for the use or contents of any product or service mentioned.

### Transportation and Getting There

Denver International Airport Ground Transportation – The Ground Transportation ►►

## Planning Your Trip to Denver?

Request a “Denver” visitor guide book provided by Visit Denver, The Convention and Visitors Bureau. The complimentary guide book is your best resource on how to navigate the Mile High City, with highlights, tips, maps and information on attractions, hotels, restaurants and more.

You can view a virtual guide by following this link:

<http://browndigital.bpc.com/publication/?m=17858&l=1>

Download their app to your iPad here:

<https://itunes.apple.com/us/app/official-visitors-guide-to/id467668610?mt=8>

Or by signing up to receive the guide here:

<http://www.denver.org/about-denver/denver-resources/visitors-guide/>

To find out more about the NAF Annual Membership Meeting, visit NAF's website, [www.ataxia.org](http://www.ataxia.org),

Information Counter at the Denver International Airport is located in the central area on Level 5 of Jeppesen Terminal.

Visit [www.flydenver.com/parkinggt](http://www.flydenver.com/parkinggt) or call (303) 342-4059 for a complete listing of ground transportation options to and from the Denver International Airport.

## Denver Area Services and Resources

### Personal Care Attendants (PCA)

If you need a personal care attendant, please make arrangements prior to attending the meeting to have someone accompany you or have a PCA hired before you arrive in Denver. Please note that NAF is unable to provide attendant care services. Due to liabilities and health concerns, NAF staff or volunteers and hotel employees are not able to provide PCA services.

Personal Assistance Services of Colorado (PASCO)

(303) 233-3122

[www.pascohh.com](http://www.pascohh.com)

### Childcare

If you need childcare, please make arrangements prior to attending the meeting. Please note that NAF is unable to provide childcare services. Due to liabilities and health concerns, NAF staff or volunteers and hotel employees are not able to provide childcare services.

Tots on the Spot

(970) 214-8373

[www.totsonthespot.com](http://www.totsonthespot.com)

### Accessible Equipment, Wheelchair, and Scooter Rentals

Major Medical Supply Brighton

7000 N. Broadway

Denver, CO 80221

303-654-0720

[www.majormedicalsupply.com/](http://www.majormedicalsupply.com/)

nu motion

13450 E. Smith Rd.

Aurora, CO 80011

303-781-1474 Toll-free: 1-800-525-8586

[www.numotion.com](http://www.numotion.com)

Penfield's Office Business Center

Sheraton Denver Downtown Hotel

303-626-2547

[www.penfieldsoffice.com](http://www.penfieldsoffice.com)

Scootaround Inc

Toll-free: 1-888-441-7575

<http://scootaround.com/>

### ADA Assistance Office

Denver Office of Disability Rights

201 W. Colfax Avenue, Dept. 1102, 2nd Floor

Denver, CO 80202

(720) 913-8480 / (720) 913-8475 TTY

[www.denvergov.org/disabilities](http://www.denvergov.org/disabilities)



## AMM Exhibitors and Sponsors Wanted

The National Ataxia Foundation is looking for companies or individuals who have products or services that would be helpful for those with ataxia to submit an exhibitor application to exhibit at "Soaring Mile High for a Cure" the National Ataxia Foundation's 58th Annual Membership Meeting (AMM). The 2015 AMM will be held in Denver, CO on March 6-8, 2015. Please email [joan@ataxia.org](mailto:joan@ataxia.org) for an exhibitor application.

NAF is grateful to those organizations that

have provided generous support of the annual membership meeting. Please consider being a sponsor of the 2015 Annual Membership Meeting. For more information on becoming a sponsor please contact Mike Parent at [mike@ataxia.org](mailto:mike@ataxia.org).

If you are affected by ataxia or are a caregiver and know of a product or service that has been helpful for you please let us know by calling (763) 553-0020 or e-mail [joan@ataxia.org](mailto:joan@ataxia.org).

**Chapter and Support Group News**  
*from Around the Country*

**Arizona Ataxia Support Group**

*Submitted by Angela Li*

Our summer meeting was held on Saturday, August 16 and we were so lucky to have two amazing guest speakers! Piper Laird, a music therapy coordinator, joined us for a fun music session. We learned how music can be used to energize and motivate us, for relaxation, and for stress management. Ron Hankins, a speech language pathologist, provided an overview of ataxia and how it affects speech and swallowing. Ron offered ideas for speech exercises and answered so many of our questions!

We have many exciting events coming up in the next few months. Join us for our IAAD Bowl & Roll Celebration at Lucky Strikes Bowling Lanes on Saturday, September 21 from 2-4 p.m. We also have a social event on October 5 at the Chandler Symphony Orchestra performance.

We plan to have a Popcornopolis Fundraiser on November 13 in Sun Lakes. Join our Facebook group, "Arizona Ataxia Support Group," at <https://www.facebook.com/groups/arizonaataxia/> or you can contact Angela at [angelali1010@gmail.com](mailto:angelali1010@gmail.com)

*gmail.com* or Mary at [mary11115@msn.com](mailto:mary11115@msn.com) for details on any upcoming events.

**Central New York Ataxia Support Group**

*Submitted by Mary Jane Damiano*

Central New York Ataxia Support Group met on Saturday, July 26. Five members were present. We discussed the pet service dog fundraisers, ataxia patient registry, our meeting with the general manager of Destiny about wheelchair accessibility problems and solutions, and our support group brochure. Our next meeting will be on Saturday, September 20 to celebrate International Ataxia Awareness Day.

**Central PA Ataxia Support Group**

*Submitted by Chris Rakshys*

Our summer meeting was held on Saturday, July 26. We had over 20 people attend, our largest audience to date! We want to thank our guest speaker, Dr. Joe Savitt, for sharing his medical knowledge and expertise with us. He covered a very broad range of topics with us, including medications, genetics, research, medical exams, therapy and much more. ▶▶



**Members of the Arizona Ataxia Support Group**

We also touched upon our plans for a casual picnic for the group in September, date and location TBD. We will have our final meeting for the year on Saturday, October 25 at the Muhlenberg Community Library in Laureldale.

**Denver Ataxia Support Group**

*Submitted by Charlotte DePew*

The July meeting attendance was light. After our usual potluck lunch, the topic and speakers were appealing to all ages. Two members from Denver’s Adaptive Adventures spoke about their exciting outdoor programs available to all handicapped and immediate family at very low cost. The primary and most compelling speaker, a paraplegic himself, had helped found the Denver-Chicago organization.

The upcoming Run, Walk n’ Roll on September 7 was discussed as well as the 2015 Denver Annual Membership Meeting at the downtown Sheraton.

**Greater Atlanta Ataxia Support Group**

*Submitted by Dave Zilles*

We held our annual picnic at Lake Lanier on June 14. It was fun with hotdogs, hamburgers, all the fixings, and wonderful side dishes members brought to share. The day was beautiful and



**Members of the Greater Atlanta Support Group’s at their annual picnic at Lake Lanier**

it was great to just have some time to catch up with everyone to learn what they had planned for the summer.

September 20 is when we will hold our Fourth Annual Atlanta Walk n’ Roll for Ataxia for International Ataxia Awareness Day. We also have a proclamation signing with Georgia’s Governor Deal on September 23.

**New Jersey Ataxia Support Group**

*Submitted by Priya Mansukhani*

The support group of New Jersey came about after I organized a fundraiser in the memory of my dad Arun, who was diagnosed with ataxia. We raised close to \$1,000 from the event called “Bowl for Arun.” We raised awareness about Ataxia and that was the start to the creation of the idea of a support group in New Jersey for ataxians.

The first support group meeting in New Jersey was held at the Manville Library in Manville, NJ, earlier this year in April. We got off to a great start and had Dr. Johnson, a neurologist at Robert Wood Johnson Hospital, come speak to the group and share his knowledge about ataxia.

The second support group meeting was held more recently in August. We had the pleasure of having Jordan Taylor, an assistant professor at Princeton University who conducts research on

*Continued on page 42*



**Members of the New Jersey Ataxia Support Group**

Chapter and Support Group News  
Continued from page 41

ataxia at the Princeton lab. He discussed how the brain functions and how ataxia alters life's daily activities.

So far, the support group has been very successful! The ataxians of New Jersey finally get the chance to come together. Hopefully, we continue to grow and flourish as a group!

**Northern California  
Ataxia Support Group**

Submitted by Alan Acacia

At our July meeting, with the leadership of Joanne Loveland, we created a fresh set of objectives to reach out to people with ataxia all over the expansive region of Northern California.

Several objectives were already complete by the July meeting. We published our first-ever newsletter for members, some of whom live far from the Bay Area. We revised our roster to reflect membership in five different regions of our chapter. In the future we hope to find leaders for local area contact groups.

We introduced a volunteer coordinator to recruit current members to assist with our new activities. One of those activities is set for our next meeting, when "Living with Ataxia" discussion groups will become a regular feature of Northern California Support Group meetings.

Two prominent researchers from UCSF described a program that assesses changes in physical and mental abilities over time (our



**Research coordinator Joe Miner, Neurologist Dr. Alexandra Nelson and Joanne Loveland**

newsletter has the details).

If you are looking to connect with a community of people with ataxia (which includes family, friends, and caretakers), for the building of mutual support, check us out (for newsletter copies, e-mail [ace@cds1.net](mailto:ace@cds1.net)). Consider coming to the fall meeting on Saturday, October 11, where we will try to take a fabulous group photo for the winter issue of *Generations*!

**Tarheel Ataxia Support Group**

Submitted by Ron Smith

We had a meeting Saturday, August 16 in the Winston-Salem area and adopted recommendations concerning how we can operate in the future. Here are the highlights:

- The statewide support group will meet twice per year, once in the spring and again in the fall. NC is a very large, wide state. From the coast to the mountains is almost 600 miles, so we felt that even having central meetings (100-plus miles one way) put a heavy burden ►►



**Members of the Tar Heel Ataxia Support Group**

on people with disabilities.

- One of the two biannual meetings will be held in the Raleigh area and the other more in the Winston-Salem area. That way if people can only come to one meeting, it would not require an overnight. The biannual meetings will be more “formal” in that we expect to have a speaker.

- The main responsibility of a Support Group is the emotional support from others. Therefore, we divided the state into five regions. The regional groups are designed to be strictly social and can meet as often as they want. For example, they may want to get together for coffee in a home or meet at a restaurant. They are not expected to have a speaker but just to share with each other.

.....

**Tri-State Ataxia Support Group**

*Submitted by Kathy Gingerelli*

We had a large group at our May meeting, a new member and a few people returning.

Our speaker for the night was Pablo Ambrosia, a certified trainer, who shared some information on exercise. Pablo spoke of the need to dig deep and grab the motivation to get yourself to do what you don’t want to do, including pushing through the discomfort and the “I can’t” voice to improve your discipline. The “I can’t” voice is important but it will keep us from exercising so you need to figure out the real “why” you need to do something. The goal should be to move everyday and Pablo gave us some proprioception exercises, they are:

- 1) Spell the alphabet with foot, then with just toes and then both feet.
- 2) Do an isometric hold by holding each leg out and tracing the alphabet.
- 3) Trace alphabet with foot with hands on table, hands on sides and hands up in the air.
- 4) Do all exercises while standing and holding onto table, remember to keep belly flexed or in seated position.
- 5) Chair squat – try to sit above chair ... when

you are tired, sit.

6) Upper body – hold weight up in chair and hold on one leg and then the other.

7) Push-ups – can be done with arms on wall, chair or table ... make sure to be on toes and support weight in arms.

After demonstrating and showing everyone that “you can” exercise, Pablo finished his discussion with the group by answering all questions.

July’s meeting was a special “Back to Basics” discussion on ataxia, starting with Dr. Ann Hunt, DO, specializing in neurology and movement disorders. Dr. Hunt started with two questions:

**1. What is the cerebellum?**

- a. Structure
  - i. Back lower part of the brain
  - ii. Has folded cortex (bark) that contains the nerve cells, connections called white matter and deep nuclei (that also contain nerve cells)
  - iii. Composed of different types of cells and connections.
  - iv. Connections
    1. Input from spinal cord (proprioception, touch pressure, etc.) inner ear (vestibular system), brainstem nuclei (inferior olive, cerebral cortex.
    2. Output (comes from the deep muscle nuclei) to the thalamus, (brainstem nuclei)
    3. Chemical Connections
      - a. Nerve cells communicate with each other by releasing chemicals called neurotransmitters.
      - b. Cerebellum
        - i. Excitatory
        - ii. Inhibitory
        - iii. Also innervation of info coming in Acetylcholine, monoadrenergic
        - iv. Complicating all this is that these neurotransmitters have different families of receptors.

Chapter and Support Group News  
Continued from page 43

**2. What does it do?**

Function – major function is to coordinate and stabilize movement which is important in the maintenance of balance.

- a. Eye movements
- b. Speech
- c. “Motor coordination” including trunk movements and limb movements.

Next we heard from Jordan Taylor, an Assistant Professor of Psychology at Princeton University, who spoke of the early tests done to help figure out what the cerebellum does. The cerebellum is connected to motor areas but is not needed for movement. In WWI helmets did not cover the base of the brain so there were a lot of cerebellum injuries. These injuries were not fatal but a change in movement was always present. Most of the early tests were performed on animals, including lesion studies. Jordan also explained that because the cerebellum is connected to the rest of the brain, the number one goal is to get other areas of the brain to compensate for the lost function in the cerebellum. Is it possible? It is also important to find out if other areas of the

brain are affected as well.

There is many testing equipment to be used and if you are willing to participate please feel free to contact Jordan at [jordanat@princeton.edu](mailto:jordanat@princeton.edu).

Lastly, we heard from Dr. Sheng-Han Kuo, a Movement Disorder Fellow from Columbia University who quickly spoke of new studies being done for ataxia. Tests are being done on MSA brains looking at CoQ levels.

Overall, we had a jam-packed night of information about the cerebellum.

.....

**Western PA Ataxia Support Group**

*Submitted by Donna Eiben*

We had our second meeting and discussed our Awareness Day at the mall. We attained another new member the morning of the meeting!

Our newest member has several affected in her family with SCA7. She asked if I knew of anyone who would be willing to help fund her with some things her children need that she cannot afford.

She is looking for funding for equipment that helps him see better: binoculars, hand-held magnifiers for documents, and a larger one for school books and adaptive software. We are also trying to get a grant for therapies to staunch the progression of muscle and brain degeneration by hupkido a form of martial arts and piano playing to help rewire his brain/nerves.

Any ideas? If so, please contact me at [dawn.eiben@verizon.net](mailto:dawn.eiben@verizon.net). ❖

**iSearchiGive Is Good for NAF**

*iSearchiGive.com* is a search engine powered by Yahoo! Search and *iGive.com*. It is the Internet's first online shopping mall where a portion of each purchase is donated to a charity of your choice. Use it to search the web, and your favorite cause receives money for every qualified search.

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

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



**Members of the Western PA Ataxia Support Group**

# 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** S & XXXL only \$10
- NAF Baseball Cap (White or Blue)** \$10

- IAAD T-Shirt** Sizes S to XXXL \$10
- Past Annual Membership Meeting (AMM) T-Shirts**  
\$1 each while supplies last!
- NAF Polo Shirts** \$25  
Mens – Royal blue w/ white NAF logo S, M & XXXL  
Light blue w/ royal blue NAF logo in L to XXXL  
Womens – Light blue w/ navy NAF logo in S to XXL
- NAF Denim Shirt** w/ white NAF logo \$27.50
- "Ataxia is Not a Foreign Cab" T-Shirt** \$10  
White sizes S to XXXL
- "Ataxia is Not a Foreign Cab" Long-Sleeve T-Shirt**  
Light 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 for \$5
- NAF Ataxia Awareness Band Blue**  
One size \$2 ea. or 3 for \$5
- 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

To place your order, call (763) 553-0020, fax (763) 553-0167, mail a copy of this form to National Ataxia Foundation, 2600 Fernbrook Lane, Suite 119, Minneapolis, MN 55447 or visit <http://tinyurl.com/nafstore>

## ORDER FORM

| Description                   | Qty. | Size | Each               | Total |
|-------------------------------|------|------|--------------------|-------|
|                               |      |      |                    |       |
|                               |      |      |                    |       |
|                               |      |      |                    |       |
|                               |      |      |                    |       |
|                               |      |      |                    |       |
| <b>SUBTOTAL:</b>              |      |      |                    |       |
| <b>Shipping within U.S.:</b>  |      |      | <b>Add \$5.00</b>  |       |
| <b>Shipping outside U.S.:</b> |      |      | <b>Add \$15.00</b> |       |
| <b>ORDER TOTAL:</b>           |      |      |                    |       |

**NAME:** \_\_\_\_\_

**ADDRESS:** \_\_\_\_\_

**CITY** \_\_\_\_\_ **STATE:** \_\_\_\_\_ **ZIP:** \_\_\_\_\_

**PHONE:** \_\_\_\_\_

**E-MAIL:** \_\_\_\_\_

*For credit card orders, please fill out the following information (you must include phone number and signature):*

**PLEASE CIRCLE ONE:** Visa Mastercard Discover

**NAME ON CARD:** \_\_\_\_\_

**CARD #:** \_\_\_\_\_

**EXP DATE:** \_\_\_\_\_ **CVV #:** \_\_\_\_\_

**SIGNATURE:** \_\_\_\_\_

**PLEASE ALLOW 4-6 WEEKS FOR DELIVERY**

# 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](mailto: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](http://www.ataxia.org/forum/toast.asp)

### NAF CHAT ROOM

**Moderator – Della** ([ddpokernut@yahoo.com](mailto:ddpokernut@yahoo.com))

[www.ataxia.org/connect/chat-rooms.aspx](http://www.ataxia.org/connect/chat-rooms.aspx)

### NAF FACEBOOK GROUP

[www.facebook.com/group.php?gid=93226257641](http://www.facebook.com/group.php?gid=93226257641)

### NAF FACEBOOK FANS

[www.facebook.com/lshogren?ref=profile#!/pages/National-Ataxia-Foundation/227766109304](http://www.facebook.com/lshogren?ref=profile#!/pages/National-Ataxia-Foundation/227766109304)

### NAF YOUTUBE CHANNEL

[www.youtube.com/user/NatlAtaxiaFound?feature=mhum](http://www.youtube.com/user/NatlAtaxiaFound?feature=mhum)

## Chapters, Support Groups and Ambassadors

### – ALABAMA –

#### ALABAMA SUPPORT GROUP LEADER

**Becky Donnelly** – Hover, AL

(205) 987-2883

E-mail: [donnely6132b@aol.com](mailto:donnely6132b@aol.com)

[www.ataxia.org/chapters/Birmingham/default.aspx](http://www.ataxia.org/chapters/Birmingham/default.aspx)

#### AMBASSADOR

**Dianne Blain Williamson** – Huntsville, AL

(256) 429-9092 or (256) 520-4858

E-mail: [diannebw@aol.com](mailto:diannebw@aol.com)

[www.ataxia.org/chapters/DianneWilliamson/default.aspx](http://www.ataxia.org/chapters/DianneWilliamson/default.aspx)

### – ARIZONA –

#### PHOENIX AREA SUPPORT GROUP LEADERS

**Angela Li** – Peoria, AZ

(847) 505-4325

E-mail: [angelali1010@gmail.com](mailto:angelali1010@gmail.com)

**Mary Fuchs** – Sun Lakes, AZ

(480) 212-6425

E-mail: [mary11115@msn.com](mailto:mary11115@msn.com) **Facebook Group:**

<https://www.facebook.com/groups/arizonaataxia/>

[www.ataxia.org/chapters/Phoenix/default.aspx](http://www.ataxia.org/chapters/Phoenix/default.aspx)

#### AMBASSADOR

**Bart Beck** – Tucson, AZ

(520) 885-8326

E-mail: [bbeck15@cox.net](mailto:bbeck15@cox.net)

[www.ataxia.org/chapters/Tucson/default.aspx](http://www.ataxia.org/chapters/Tucson/default.aspx)

### – ARKANSAS –

#### AMBASSADORS

**Judy and David King** – Hot Springs Village, AR

E-mail: [drkingpd@suddenlink.net](mailto:drkingpd@suddenlink.net)

[www.ataxia.org/chapters/JudyKing/default.aspx](http://www.ataxia.org/chapters/JudyKing/default.aspx)

### – CALIFORNIA –

#### LOS ANGELES AREA SUPPORT GROUP LEADERS

**Bonnie Hasegawa** – Hacienda Heights, CA

(626) 840-6005

E-mail: [bahasegawa@verizon.net](mailto:bahasegawa@verizon.net)

**Lora Morn** – Santa Monica, CA

(310) 664-8808

E-mail: [loramorn@gmail.com](mailto:loramorn@gmail.com)

Web: <http://laasg-ca.info>

[www.ataxia.org/chapters/LosAngeles/default.aspx](http://www.ataxia.org/chapters/LosAngeles/default.aspx)

#### N. CALIFORNIA AREA SUPPORT GROUP LEADER

**Joanne Loveland** – Danville, CA

E-mail: [joanneloveland@gmail.com](mailto:joanneloveland@gmail.com)

[www.ataxia.org/chapters/NorthernCalifornia/default.aspx](http://www.ataxia.org/chapters/NorthernCalifornia/default.aspx)

#### ORANGE COUNTY AREA SUPPORT GROUP LEADER

**Cindy DeMint** – Yorba Linda, CA

(714) 970-1191

E-mail: [cindyocataxia@gmail.com](mailto:cindyocataxia@gmail.com)

**Daniel Navar** – Montebello, CA

(323) 788-7751

E-mail: [danieln27@gmail.com](mailto:danieln27@gmail.com)

[www.ataxia.org/chapters/OrangeCounty/default.aspx](http://www.ataxia.org/chapters/OrangeCounty/default.aspx)

#### AMBASSADORS

**Barbara Bynum** – Merced, CA

(209) 383-1275

E-mail: [bjb@vtlnet.com](mailto:bjb@vtlnet.com)

[www.ataxia.org/chapters/BarbaraBynum/default.aspx](http://www.ataxia.org/chapters/BarbaraBynum/default.aspx)

**Deborah Omictin** – Hayward, CA

(510) 783-3190

E-mail: [rsisbig@aol.com](mailto:rsisbig@aol.com)

[www.ataxia.org/chapters/DeborahO/default.aspx](http://www.ataxia.org/chapters/DeborahO/default.aspx)

**Martha Elliott** – Camarillo, CA

(805) 987-2490

E-mail: [DOElliott268@gmail.com](mailto:DOElliott268@gmail.com)

[www.ataxia.org/chapters/Camarillo/default.aspx](http://www.ataxia.org/chapters/Camarillo/default.aspx)



**— COLORADO —****DENVER AREA SUPPORT GROUP LEADER**

**Charlotte DePew** – Aurora, CO  
 (720) 379-6887  
 E-mail: cldepew77@comcast.net  
[www.ataxia.org/chapters/Denver/default.aspx](http://www.ataxia.org/chapters/Denver/default.aspx)

**— CONNECTICUT —****TRI-STATE SUPPORT GROUP LEADER**

**Denise Mitchell** – Bronxville, NY  
 (212) 720-2179  
 E-mail: markmeghan@aol.com  
[www.ataxia.org/chapters/Tri-State/default.aspx](http://www.ataxia.org/chapters/Tri-State/default.aspx)

**Kathy Gingerelli** – Parsippany, NY  
 (973) 334-2242  
 E-mail: kgingerelli@msn.com

**AMBASSADOR**

**Terre Di Placito** – Torrington, CT  
 (860) 489-5092  
[www.ataxia.org/chapters/TerreDiPlacito/default.aspx](http://www.ataxia.org/chapters/TerreDiPlacito/default.aspx)

**— DELAWARE —****DELAWARE SUPPORT GROUP LEADER**

**Joseph DeCrescenzo** – Newark, DE  
 (302) 369-9287  
 E-mail: jdecrc@comcast.net  
[www.ataxia.org/chapters/DeCrescenzo/default.aspx](http://www.ataxia.org/chapters/DeCrescenzo/default.aspx)

**— FLORIDA —****NORTHEAST FLORIDA SUPPORT GROUP LEADERS**

**John Richwine** – Jacksonville, FL  
 (904) 996-0699  
 E-mail: ajrichwine@gmail.com  
[www.ataxia.org/chapters/NortheastFlorida/default.aspx](http://www.ataxia.org/chapters/NortheastFlorida/default.aspx)

**TAMPA BAY SUPPORT GROUP LEADER**

**Nygel Lenz** – Clearwater, FL  
 (727) 451-9165  
 E-mail: nygellenz@gmail.com  
[www.ataxia.org/chapters/TampaBay/default.aspx](http://www.ataxia.org/chapters/TampaBay/default.aspx)

**AMBASSADOR**

**Meghan McBrearty** – Tallahassee, FL  
 (850) 524-9060  
 E-mail: megra10@hotmail.com  
[www.ataxia.org/chapters/McBrearty/default.aspx](http://www.ataxia.org/chapters/McBrearty/default.aspx)

**— GEORGIA —****GREATER ATLANTA SUPPORT GROUP LEADERS**

**Brean Underwood** – Smyrna, GA  
 (678) 314-7198  
 E-mail: breanunderwood@gmail.com

**Dave Zilles** – Atlanta, GA  
 (678) 596-6751  
 E-mail: dzilles@earthlink.net or atlantaataxia@yahoo.com

**Greg Rooks** – Atlanta, GA  
 (404) 822-7451  
 E-mail: rooksgj@yahoo.com

**Lealan LaRoche** – Dunwoody, GA  
 (678) 234-6600  
 E-mail: lealan@mac.com

S.G. e-mail: atlantaataxia@yahoo.com **Facebook Group:**  
<https://www.facebook.com/groups/317380459539/>  
<http://www.ataxia.org/chapters/Atlanta/default.aspx>

**AMBASSADOR**

**Kristie Adams** – Savannah, GA  
 E-mail: opal1011@comcast.net  
[www.ataxia.org/chapters/KristieAdams/default.aspx](http://www.ataxia.org/chapters/KristieAdams/default.aspx)

**— ILLINOIS —****CHI-TOWN FRIENDSHIP GROUP LEADER**

**Jonas Cepkaskas** – Oak Forest, IL  
 (708) 535-0928  
 E-mail: jonas@chi-townataxia.org  
[www.ataxia.org/chapters/Chicago/default.aspx](http://www.ataxia.org/chapters/Chicago/default.aspx)

**METRO AREA CHICAGO SUPPORT GROUP LEADER**

**Christopher (Topher) Marsh** – Chicago, IL  
 (312) 662-1127  
 E-mail: cmarsh34@ameritech.net  
[http://health.groups.yahoo.com/group/u\\_r\\_notalone/](http://health.groups.yahoo.com/group/u_r_notalone/)  
[www.ataxia.org/chapters/ChrisMarsh/default.aspx](http://www.ataxia.org/chapters/ChrisMarsh/default.aspx)

**AMBASSADOR**

**Elaine Darte** – Coffeen, IL  
 (618) 397-3259  
 E-mail: elainedarte@yahoo.com  
[www.ataxia.org/chapters/SouthernIllinois/default.aspx](http://www.ataxia.org/chapters/SouthernIllinois/default.aspx)

**— INDIANA —****HAPPY HOOSIERS INDIANA SUPPORT GROUP LEADER**

**Cheryl (Cheri) Bearman** – Hoagland, IN  
 (260) 452-6231  
 E-mail: cheribearman@gmail.com  
[www.ataxia.org/chapters/Indiana/default.aspx](http://www.ataxia.org/chapters/Indiana/default.aspx)

**— IOWA —****IOWA SUPPORT GROUP LEADER**

**Emily Medina** – West Des Moines, IA  
 (515) 727-8713  
 E-mail: emily061578@yahoo.com **Facebook Group:**  
<https://www.facebook.com/groups/107944351294/>  
[www.ataxia.org/chapters/EmilyMedina/default.aspx](http://www.ataxia.org/chapters/EmilyMedina/default.aspx)

**— KANSAS —****AMBASSADOR**

**Jalean Retzlaff** – Park City, KS  
 (316) 303-2351  
 E-mail: jlrtrolls@yahoo.com  
[www.ataxia.org/chapters/Retzlaff/default.aspx](http://www.ataxia.org/chapters/Retzlaff/default.aspx)

**— KENTUCKY —****AMBASSADOR**

**Janice Johnson** – Brownsville, KY  
 (270) 597-3854  
[www.ataxia.org/chapters/JaniceJohnson/default.aspx](http://www.ataxia.org/chapters/JaniceJohnson/default.aspx)

**— LOUISIANA —****LOUISIANA CHAPTER PRESIDENT**

**Elizabeth Tanner** – Baton Rouge, LA  
 (225) 241-3745  
 E-mail: hammett\_e@hotmail.com  
[www.ataxia.org/chapters/Louisiana/default.aspx](http://www.ataxia.org/chapters/Louisiana/default.aspx)

**— MAINE —****MAINE SUPPORT GROUP LEADER**

**Kelley Rollins** – Bowdoinham, ME  
 E-mail: kelly3902me@yahoo.com  
[www.ataxia.org/chapters/Maine/default.aspx](http://www.ataxia.org/chapters/Maine/default.aspx)

## NAF Directory

Continued from page 47

## — MARYLAND —

**CHESAPEAKE CHAPTER PRESIDENT****Carolyn Davis** – Vienna, VA

(703) 759-2008

E-mail: ccnafpres@gmail.com

[www.ataxia.org/chapters/Chesapeake/default.aspx](http://www.ataxia.org/chapters/Chesapeake/default.aspx)**MID-ATLANTIC SOCIAL SUPPORT GROUP LEADER****Bailey Vernon, Health Educator**

Lutherville, MD

(410) 616-2811

E-mail: bvernon1@jhmi.edu

[www.ataxia.org/chapters/JHASG/default.aspx](http://www.ataxia.org/chapters/JHASG/default.aspx)**AMBASSADOR****Karen DeVito** – Frederick, MD

(301) 682-5386

E-mail: kdrosenberger@comcast.net

[www.ataxia.org/chapters/KarenRosenberger/default.aspx](http://www.ataxia.org/chapters/KarenRosenberger/default.aspx)

## — MASSACHUSETTS —

**BOSTON AREA SUPPORT GROUP LEADERS****Lanie Cantor** – Arlington, MA

E-mail: laniecantor@gmail.com

[www.ataxia.org/chapters/Boston/default.aspx](http://www.ataxia.org/chapters/Boston/default.aspx)**CENTRAL MA SUPPORT GROUP LEADERS****Donna and Richard Gorzela** – Andover, MA

(978) 490-9552

E-mail: donna.gorzela@gmail.com

**John and Dana Mauro** – Auburn, MA

(508) 736-6084

E-mail: ngataxia@outlook.com

E-mail: danamauro63@msn.com

[www.ataxia.org/chapters/CentralMA/default.aspx](http://www.ataxia.org/chapters/CentralMA/default.aspx)

## — MICHIGAN —

**DETROIT AREA SUPPORT GROUP LEADER****Tanya Tunstull** – Detroit, MI

(313) 373-3646

E-mail: tinyt48221@yahoo.com

[www.ataxia.org/chapters/Detroit/default.aspx](http://www.ataxia.org/chapters/Detroit/default.aspx)**WESTERN MICHIGAN SUPPORT GROUP LEADER****Lynn K. Ball** – Grand Rapids, MI

(616) 735-2303

E-mail: lynnkball@aol.com

[www.ataxia.org/chapters/LynnBall/default.aspx](http://www.ataxia.org/chapters/LynnBall/default.aspx)

## — MINNESOTA —

**CENTRAL MN SUPPORT GROUP LEADER****Marsha Binnebose** – St. Cloud, MN

(320) 248-9851

E-mail: mbinnebose@hotmail.com

[www.ataxia.org/chapters/StCloud/default.aspx](http://www.ataxia.org/chapters/StCloud/default.aspx)**TWIN CITIES AREA SUPPORT GROUP LEADER****Lenore Healey Schultz** – Minneapolis, MN

(612) 724-3784

E-mail: schultz.lenore@yahoo.com

[www.ataxia.org/chapters/TwinCities/default.aspx](http://www.ataxia.org/chapters/TwinCities/default.aspx)**AMBASSADORS****Julie Schuur** – Luverne, MN

(507) 283-2555

E-mail: jschuur@wowway.net

[www.ataxia.org/chapters/JulieSchuur/default.aspx](http://www.ataxia.org/chapters/JulieSchuur/default.aspx)**Lori Goetzman**

Rochester, MN

(507) 282-7127

E-mail: logoetz@gmail.com

[www.ataxia.org/chapters/LoriGoetzman/default.aspx](http://www.ataxia.org/chapters/LoriGoetzman/default.aspx)

## — MISSISSIPPI —

**MISSISSIPPI CHAPTER PRESIDENT****Camille Daglio**

Hattiesburg, MS

E-mail: daglio1@bellsouth.net

[www.ataxia.org/chapters/Mississippi/default.aspx](http://www.ataxia.org/chapters/Mississippi/default.aspx)

## — MISSOURI —

**KANSAS CITY SUPPORT GROUP LEADERS****Jim Clark**

Gladstone, MO

(816) 468-7260

E-mail: clarkstone9348@sbcglobal.net

[www.ataxia.org/chapters/KansasCity/default.aspx](http://www.ataxia.org/chapters/KansasCity/default.aspx)**Lois Goodman**

Independence, MO

(816) 257-2428

[www.ataxia.org/chapters/KansasCity/default.aspx](http://www.ataxia.org/chapters/KansasCity/default.aspx)**AMBASSADOR****Roger Cooley**

Columbia, MO

(573) 474-7232 before noon

E-mail: rogercooley@mediacombb.net

[www.ataxia.org/chapters/RogerCooley/default.aspx](http://www.ataxia.org/chapters/RogerCooley/default.aspx)**Sarah “Janeen” Rheinecker** – St. Louis, MO

(417) 379-3799

Email: jrheinecker@yahoo.com

[www.ataxia.org/chapters/Rheinecker/default.aspx](http://www.ataxia.org/chapters/Rheinecker/default.aspx)

## — NEW HAMPSHIRE —

**NEW HAMPSHIRE SUPPORT GROUP LEADER****Jill Porter**

Manchester, NH

(603) 626-0129

E-mail: jilleporter@comcast.net

[www.ataxia.org/chapters/Bedford/default.aspx](http://www.ataxia.org/chapters/Bedford/default.aspx)

## — NEW JERSEY —

**NEW JERSEY SUPPORT GROUP LEADER****Priya Mansukhani**

Bridgewater, NJ

(908) 685-8805

E-mail: priyamans@gmail.com

[www.ataxia.org/chapters/NewJersey/default.aspx](http://www.ataxia.org/chapters/NewJersey/default.aspx)**Tri-State Support Group Leader****Denise Mitchell**

Bronxville, NY

(914) 720-2179

E-mail: markmeghan2@gmail.com

[www.ataxia.org/chapters/Tri-State/default.aspx](http://www.ataxia.org/chapters/Tri-State/default.aspx)**Kathy Gingerelli**

Parsippany, NJ

(973) 334-2242

E-mail: kgingerelli@msn.com



**— NEW YORK —****CENTRAL NEW YORK SUPPORT GROUP LEADER****Mary Jane Damiano** – N. Syracuse, NY**Judy Tarrants** – Fabius, NY

Home: (315) 683-9486 Cell: (315) 706-6555

E-mail: jtarrants@aol.com

[www.ataxia.org/chapters/CentralNewYork/default.aspx](http://www.ataxia.org/chapters/CentralNewYork/default.aspx)**TRI-STATE SUPPORT GROUP LEADER****Denise Mitchell** – Bronxville, NY

(914) 720-2179

E-mail: markmeghan2@gmail.com

[www.ataxia.org/chapters/Tri-State/default.aspx](http://www.ataxia.org/chapters/Tri-State/default.aspx)**Kathy Gingerelli** – Parsippany, NJ

(973) 334-2242

E-mail: kgingerelli@msn.com

**— NORTH CAROLINA —****TARHEEL SUPPORT GROUP LEADERS****Jerry and Tammy Hauser** – Advance, NC

(336) 998-2942

E-mail: deaconwfu@msn.com

**Ron and Dana Smith** – Garner, NC

(919) 779-0414

E-mail: rsmith@sacherokee.com

E-mail: dsmith@sa-pr.com

[www.ataxia.org/chapters/Tarheel/default.aspx](http://www.ataxia.org/chapters/Tarheel/default.aspx)**— OHIO —****GREATER CINCINNATI AREA SUPPORT GROUP LEADERS****Jennifer Mueller** – Cincinnati, OH

(513) 834-7138

E-mail: jenmu@yahoo.com

[www.ataxia.org/chapters/Cincinnati/default.aspx](http://www.ataxia.org/chapters/Cincinnati/default.aspx)**Julia Soriano** – Cincinnati, OH

(513) 899-1195

E-mail: julia@epivision.com

**CLEVELAND AREA SUPPORT GROUP LEADER****Carmen Pieragastini** – Willowick, OH

(216) 272-5588

E-mail: willowpier@roadrunner.com

[www.ataxia.org/chapters/Cleveland/default.aspx](http://www.ataxia.org/chapters/Cleveland/default.aspx)**— OREGON —****WILLAMETTE VALLEY SUPPORT GROUP LEADER****Jason Wolfer** – Gervais, OR

(503) 502-2633

E-mail: wolfer.jason@gmail.com

[www.ataxia.org/chapters/Willamette/default.aspx](http://www.ataxia.org/chapters/Willamette/default.aspx)**— PENNSYLVANIA —****CENTRAL PA SUPPORT GROUP LEADER****Christina Rakshys** – Allentown, PA

(610) 395-6905

E-mail: rakshys@ptd.net

[www.ataxia.org/chapters/Rakshys/default.aspx](http://www.ataxia.org/chapters/Rakshys/default.aspx)**Michael Cammer** – Downingtown, PA

(610) 873-1852

E-mail: michael.cammer62@hotmail.com

[www.ataxia.org/chapters/CentralPA/default.aspx](http://www.ataxia.org/chapters/CentralPA/default.aspx)**POSITIVE PEOPLE IN PA SUPPORT GROUP LEADER****Liz Nussear** – Norristown, PA

(610) 272-1502

E-mail: lizout@aol.com

[www.ataxia.org/chapters/SEPennsylvania/default.aspx](http://www.ataxia.org/chapters/SEPennsylvania/default.aspx)**WESTERN PA SUPPORT GROUP LEADER****Donna Eiben** – South Park, PA

(412) 655-4091

E-mail: dawn.eiben@verizon.net

[www.ataxia.org/chapters/SouthPark/default.aspx](http://www.ataxia.org/chapters/SouthPark/default.aspx)**— RHODE ISLAND —****RHODE ISLAND SUPPORT GROUP LEADER****Anabela Azevedo** – Bristol, AZ

(401) 297-8627

E-mail: azevedo\_anabela@yahoo.com

[www.ataxia.org/chapters/Rhodelsland/default.aspx](http://www.ataxia.org/chapters/Rhodelsland/default.aspx)**— SOUTH CAROLINA —****AMBASSADOR****Brad Forth** – Greenville, SC

(864) 415-8147

E-mail: bradf@photoforth.com

[www.ataxia.org/chapters/Greenville/default.aspx](http://www.ataxia.org/chapters/Greenville/default.aspx)**— TENNESSEE —****MIDDLE TN AREA SUPPORT GROUP LEADER****Vicki Tyler** – Nashville, TN

(615) 646-3024

E-mail: tylerv2@comcast.net

[www.ataxia.org/chapters/TN/default.aspx](http://www.ataxia.org/chapters/TN/default.aspx)**— TEXAS —****NORTH TEXAS SUPPORT GROUP LEADER****David Henry Jr.** – Trophy Club, TX

(817) 739-2886 (contact by e-mail preferred)

E-mail: cheve11e@sbcglobal.net

[www.ataxia.org/chapters/NorthTexas/default.aspx](http://www.ataxia.org/chapters/NorthTexas/default.aspx)**AMBASSADORS****Dana LeBlanc** – Orange, TX

(409) 883-5570

E-mail: tilessal@yahoo.com

Web: <http://ladyd1973.tripod.com/index.html>[www.ataxia.org/chapters/GoldenTriangle/default.aspx](http://www.ataxia.org/chapters/GoldenTriangle/default.aspx)**David Brunnert** – Cypress, TX

(713) 578-0607

E-mail: david.brunnert@sbcglobal.net

[www.ataxia.org/chapters/Brunnert/default.aspx](http://www.ataxia.org/chapters/Brunnert/default.aspx)**Debra Whitcomb** – El Paso, TX

(915) 329-0721

E-mail: debrawhitcomb@hotmail.com

[www.ataxia.org/chapters/Whitcomb/default.aspx](http://www.ataxia.org/chapters/Whitcomb/default.aspx)**— UTAH —****UTAH SUPPORT GROUP LEADERS****Grant Beutler** – Salt Lake City, UT

E-mail: grant.beutler@gmail.com

**Jenny Durrant** – North Ogden, UT

(801) 721-7140

E-mail: jenny@utahataxia.org

**Lisa Ord, PhD, LCSW** – Salt Lake City, UT

(801) 587-3020

E-mail: lisa.ord@hsc.utah.edu

Facebook page: [www.facebook.com/utahataxia](http://www.facebook.com/utahataxia)

NAF Directory  
Continued from page 49

[www.utahataxia.org](http://www.utahataxia.org)  
[www.ataxia.org/chapters/Utah/default.aspx](http://www.ataxia.org/chapters/Utah/default.aspx)

– VIRGINIA –

**CHESAPEAKE CHAPTER PRESIDENT**

**Carolyn Davis** – Vienna, VA  
(703) 759-2008  
E-mail: [ccnafpres@gmail.com](mailto:ccnafpres@gmail.com)  
[www.ataxia.org/chapters/Chesapeake/default.aspx](http://www.ataxia.org/chapters/Chesapeake/default.aspx)

– WASHINGTON –

**SEATTLE AREA SUPPORT GROUP LEADER**

**Milly Lewendon** – Kirkland, WA  
(425) 823-6239  
E-mail: [ataxiaseattle@comcast.net](mailto:ataxiaseattle@comcast.net)  
[www.ataxia.org/chapters/Seattle/default.aspx](http://www.ataxia.org/chapters/Seattle/default.aspx)

**AMBASSADOR**

**Linda Jacoy** – Spokane, WA  
(509) 482-8501  
E-mail: [linda4727@hotmail.com](mailto:linda4727@hotmail.com)  
[www.ataxia.org/chapters/Spokane/default.aspx](http://www.ataxia.org/chapters/Spokane/default.aspx)

– WISCONSIN –

**WISCONSIN SUPPORT GROUP LEADER**

**Jenny Mathison** – Madison, WI  
(608) 285-5285  
E-mail: [mjmathison@att.net](mailto:mjmathison@att.net)  
[www.ataxia.org/chapters/Wisconsin/default.aspx](http://www.ataxia.org/chapters/Wisconsin/default.aspx)

## International Support Groups & Ambassadors

– CANADA –

**OTTAWA SUPPORT GROUP LEADER**

**Prentis Clairmont** – Ottawa, Ontario  
(613) 864-8545  
E-mail: [prentis.clairmont@gmail.com](mailto:prentis.clairmont@gmail.com) **Facebook Group:**  
<https://www.facebook.com/groups/1468963499991380/>  
[www.ataxia.org/chapters/Ottawa/default.aspx](http://www.ataxia.org/chapters/Ottawa/default.aspx)

**AMBASSADOR**

**Terry Greenwood** – Winnipeg, Manitoba  
(204) 488-4155  
E-mail: [wpgmagic@gmail.com](mailto:wpgmagic@gmail.com)  
[www.ataxia.org/chapters/TerryGreenwood/default.aspx](http://www.ataxia.org/chapters/TerryGreenwood/default.aspx)

– INDIA –

**INDIA SUPPORT GROUP LEADER (SAMAG)**

**Chandu Prasad George, CH,**  
Hyderabad, Secunderabad, India  
Mobile: 0091-9949019410 +9885199918  
E-mail: [sam\\_ataxiaindia@yahoo.com](mailto:sam_ataxiaindia@yahoo.com) or  
[samag.india@gmail.com](mailto:samag.india@gmail.com)  
[www.ataxia.org/chapters/Chandu/default.aspx](http://www.ataxia.org/chapters/Chandu/default.aspx)  
Please visit our website: [www.ataxia.in](http://www.ataxia.in)  
<http://seekamiracleataxiagroupindia-samagindia.webs.com> ❖

**Please help us keep your information and schedules  
up-to-date by e-mailing updates to [lori@ataxia.org](mailto:lori@ataxia.org).**

## 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](mailto:fragilex@utsouthwestern.edu)**

# Calendar of Events

The most current event information is available on the NAF website, [www.ataxia.org](http://www.ataxia.org).

## SUPPORT GROUP MEETINGS

– Saturday, October 4, 2014 –

### Rhode Island Ataxia Support Group Meeting

**Time:** 11 a.m. – 2 p.m.

**Location:** Franklin Court, 150-160 Franklin St., Bristol, RI

**Details:** For more information contact Anabela Azevedo at (401) 297-8627 or [azevedo\\_anabela@yahoo.com](mailto:azevedo_anabela@yahoo.com).

– Wednesday, October 8, 2014 –

### Willamette Valley Ataxia Support Group Meeting

**Time:** 11:30 a.m. – 1 p.m. on the second Wednesday of every month

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

**Details:** For more information contact Jason Wolfer at (503) 502-2633 or at [wolfer.jason@gmail.com](mailto:wolfer.jason@gmail.com).

– Saturday, October 11, 2014 –

### Central MN Ataxia Support Group Meeting

**Time:** 9:45 – 11:45 a.m.

**Location:** Kimball State Bank of St. Augusta, in the Board Room, 24952 County Rd. 7, St. Cloud, MN 56301

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

### North Texas Ataxia Support Group Meeting

**Time:** 10 a.m. – noon

**Location:** Las Colinas Cancer Center, 7415 Las Colinas Blvd., Irving, TX

**Details:** For more information contact David Henry at [cheve11e@sbcglobal.net](mailto:cheve11e@sbcglobal.net).

### Northern California

#### Ataxia Support Group Meeting

**Time:** 11 a.m. – Living with Ataxia Groups; noon – General Meeting

**Location:** Our Savior's Lutheran Church, 1035 Carol Ln., Lafayette, CA

**Details:** For more information contact Joanne Loveland at [joanneloveland@gmail.com](mailto:joanneloveland@gmail.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](mailto:nygellenz@gmail.com).

– Saturday, October 18, 2014 –

### Denver Area Ataxia Support Group Meeting

**Time:** 1 – 4 p.m.

**Location:** Swedish Medical Center, Spruce C Meeting Rm, Second Floor, 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](mailto:cldepew77@comcast.net).

### 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 [schultz.lenore@yahoo.com](mailto:schultz.lenore@yahoo.com).

– Saturday, October 25, 2014 –

### Alabama Ataxia Support Group Meeting and Luncheon

**Time:** 10 a.m. – 2 p.m.

**Location:** Covenant Presbyterian Church, Homewood, AL

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

– Saturday, November 1, 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](mailto:atlantaataxia@gmail.com).

– Saturday, November 8, 2014 –

### Central MN Ataxia Support Group Meeting

**Time:** 9:45 – 11:45 a.m.

**Location:** Kimball State Bank of St. Augusta, in the Board Room, 24952 County Rd. 7, St. Cloud, MN 56301

**Details:** For more information contact Marsha

*Continued on page 52*

*Calendar of Events**Continued from page 51*

Binnebose at (320) 248-9851 or [mbinnebose@hotmail.com](mailto:mbinnebose@hotmail.com).

**North Texas Ataxia Support Group Meeting**

**Time:** 10 a.m. – noon

**Location:** Las Colinas Cancer Center, 7415 Las Colinas Blvd., Irving TX

**Details:** For more information contact David Henry at [cheve11e@sbcglobal.net](mailto:cheve11e@sbcglobal.net).

– **Tuesday, November 11, 2014** –

**Utah Ataxia Support Group Meeting**

**Time:** 6 p.m.

**Location:** John A. Moran Eye Center, SLC, UT

**Details:** For additional information contact Jenny Durrant at (801) 721-7140 or [jenny@utahataxia.org](mailto:jenny@utahataxia.org).

– **Wednesday, November 12, 2014** –

**Willamette Valley Ataxia Support Group Meeting**

**Time:** 11:30 a.m. – 1 p.m. on the second Wednesday of every month

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

**Details:** For more information contact Jason Wolfer at (503) 502-2633 or at [wolfer.jason@gmail.com](mailto:wolfer.jason@gmail.com).

– **Thursday, November 13, 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](mailto:markmeghan2@gmail.com) or Kathy Gingerelli at [kgingerelli@msn.com](mailto:kgingerelli@msn.com).

– **Saturday, November 15, 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 [schultz.lenore@yahoo.com](mailto:schultz.lenore@yahoo.com).

– **Sunday, November 16, 2014** –

**Chi-town Ataxia Friendship Group Meeting**

**Time:** 1 p.m. on the third Sunday of odd numbered months.

**Location:** Good Samaritan Hospital, 3815 Highland Ave., Downers Grove, IL 60515

**Details:** For additional information contact Jonas Cepkauskas at (708) 381-5555 or [jonas@chitownataxia.org](mailto:jonas@chitownataxia.org).

– **Saturday, November 22, 2014** –

**Northeast Florida Ataxia Support Group Meeting**

**Time:** 2 – 4 p.m.

**Location:** Baptist South Hospital, Jacksonville, FL. Azelea/Begonia/Camelia meeting rooms

**Details:** For more information contact Cory Hannan at (904) 314-2061 or [coryhannan@hotmail.com](mailto:coryhannan@hotmail.com). ▶▶

## Remembering NAF in Your Will

There have been a number of true heroes over the years that have quietly made a significant impact on the National Ataxia Foundation and the ataxia families it serves. These are people who named NAF as a beneficiary in their will.

Most of the time the Foundation is unaware of the kind acts of these champions until after they are gone, but each time we are deeply touched and honored by their selfless commitment in helping others.

Over the years these individuals, who have chosen NAF as a beneficiary, have given anywhere from a few thousand dollars to nearly one

million dollars. Their forethought and benevolence has enabled the Foundation to support promising research and provide meaningful programs and services to ataxia families. It is because of these quiet heroes that many research studies and programs have been funded.

We are truly thankful for their humanitarian and compassionate acts and we will be eternally grateful for the impact they have made in helping ataxia families. Their legacy lives on in the hope they have given ataxia families.

Perhaps this is the time to consider adding the National Ataxia Foundation in your will.

– **Saturday, December 6, 2014** –

**Alabama Ataxia Support Group Christmas Social**

**Time:** To be announced

**Location:** To be announced

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

**Greater Atlanta**

**Ataxia Support Group Holiday Party**

**Time:** 1 p.m.

**Location:** To be announced

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

– **Tuesday, December 9, 2014** –

**Utah Ataxia Support Group Meeting**

**Time:** 6 p.m.

**Location:** John A. Moran Eye Center, SLC, UT

**Details:** For additional information contact Jenny Durrant at (801) 721-7140 or [jenny@utahataxia.org](mailto:jenny@utahataxia.org).

– **Wednesday, December 10, 2014** –

**Willamette Valley Ataxia Support Group Meeting**

**Time:** 11:30 a.m. – 1 p.m. on the second Wednesday of every month

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

**Details:** For more information contact Jason Wolfer at (503) 502-2633 or at [wolfer.jason@gmail.com](mailto:wolfer.jason@gmail.com).

– **Saturday, December 13, 2014** –

**Central MN Ataxia Support Group Meeting**

**Time:** 9:45 – 11:45 a.m.

**Location:** Kimball State Bank of St. Augusta, in the Board Room, 24952 County Rd. 7, St. Cloud, MN 560301

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

**North Texas Ataxia Support Group Meeting**

**Time:** 10 a.m. – noon

**Location:** Las Colinas Cancer Center, 7415 Las Colinas Blvd., Irving TX

**Details:** For more information contact David Henry at [cheve11e@sbcglobal.net](mailto:cheve11e@sbcglobal.net).

– **Saturday, December 20, 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 [schultz.lenore@yahoo.com](mailto:schultz.lenore@yahoo.com).

– **Saturday, January 10, 2015** –

**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](mailto:nygellenz@gmail.com).

---

**INFORMATIONAL, AWARENESS,  
AND IAAD EVENTS  
AND FUNDRAISERS**

**Global Online Walk n' Roll 2014**

**IAAD Event and Fundraiser**

**Details:** You can create your own team, have a friendly competition and begin creating ataxia awareness and raising funds to support the mission of NAF. For more information please visit the event website. [www.ataxia.org/walk/virtual](http://www.ataxia.org/walk/virtual)

– **Saturday, October 4, 2014** –

**Michigan Walk n' Roll for Ataxia**

**IAAD Event and Fundraiser**

**Time:** Registration 8:30 – 9:00 a.m., Symposium 9 a.m., Luncheon 11:30 a.m.

**Location:** University of Michigan Biomedical Science Research Building (BSRB), 109 Zina Pitcher Place, 5031 BSRB, Ann Arbor, MI 48109

**Details:** Minimum fee of \$25 to receive a t-shirt. All proceeds benefit the National Ataxia Foundation. For more information, please contact Tanya Tundstull at (313) 736-2827 or [tinyt48221@yahoo.com](mailto:tinyt48221@yahoo.com) or Elizabeth Sullivan at (734) 232-6247 or [elizsull@umich.edu](mailto:elizsull@umich.edu). [www.ataxia.org/walk/michigan](http://www.ataxia.org/walk/michigan)

– **Sunday, October 5, 2014** –

**Jack's Run**

**Details:** Ann Nuese is training for the Medtronic Marathon. She will be running for her dad, Jack Moore. All proceeds benefit the National Ataxia Foundation. For more information visit the event website <https://naf.myetap.org/fundraiser/14jacksrun/>.

*Calendar of Events*  
Continued from page 53

– **Saturday, October 11, 2014** –

**Lou Coletti Memorial Golf Tournament  
Fundraiser**

**Time:** Noon

**Location:** World Woods Golf Course, 17590 Ponce De Leon Blvd., Brooksville, FL 34614

**Details:** All proceeds benefit the National Ataxia Foundation. For more information contact Rachel Coletti at [islandrach@gmail.com](mailto:islandrach@gmail.com).

**Tea Time for Ataxia  
Fundraiser**

**Time:** 11:00 a.m. – 1:00 p.m. or 1:30 – 3:30 p.m.

**Location:** Aubrey Rose tea Room, LA Mesa, CA.

**Details:** Cost \$35 per person. All proceeds benefit the National Ataxia Foundation. For more information contact Jane Jaffe at (619) 286-9745 or [sicilianmother@cox.net](mailto:sicilianmother@cox.net).

– **Tuesday, October 21, 2014** –

**Boscov's Friends Helping Friends  
Fundraiser**

**Location:** Boscov's Department Store

**Details:** Friends Helping Friends special shopping day. 25% shopping passes are available for \$5. The shopping pass purchase benefits the National Ataxia Foundation. For more information or to

purchase your shopping pass please contact Cathy DeCrescenzo at (302) 369-9287 or [cdecrescenzo@comcast.net](mailto:cdecrescenzo@comcast.net) or Mike Cammer at (610) 996-5814 or [michael.cammer62@hotmail.com](mailto:michael.cammer62@hotmail.com).

– **Saturday, November 8, 2014** –

**JHU Movement Disorders Symposium**

**Time:** 8 a.m. – 3:30 p.m.

**Location:** BWI Airport Marriott Hotel, 1743 W. Nursery Rd, Linthicum Heights, MD 21090

**Details:** Includes educational seminars, a facilitated lunch for those with ataxia and community resources. Presented by the Johns Hopkins Movement Disorders Center. For additional information or to register please contact Bailey Vernon at (410) 616-2811 or [bvernon1@jhmi.edu](mailto:bvernon1@jhmi.edu) or visit the event's website: <http://events.r20.constantcontact.com/register/event?oeidk=a07e9b5f2qqa242e8de&llr=7gsxstqab>

– **Thursday, November 13, 2014** –

**Popcornopolis  
Fundraiser**

**Time:** 8 a.m. – noon

**Location:** Iron Oaks Clubhouse, 24218 S. Oakwood Dr., Sun Lakes, AZ 85248

**Details:** All proceeds benefit the National Ataxia Foundation. For more information contact Angela at [angelali1010@gmail.com](mailto:angelali1010@gmail.com) or Mary [mary11115@msn.com](mailto:mary11115@msn.com) at for details. ❖

**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](mailto:hutte019@umn.edu).



# 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 June through July 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             | Penny Golminas    | Massanova Family   | Santa Croce Family   |
| Peter Agostini          | Mark Graham       | Willie McDaniel    | Donald Santa Croce   |
| Paul Aiello             | Richard Guerrero  | Robert McMurtry    | Gavin Schenck        |
| JoAnn Aiello-Ciecierski | Teresita Guerrero | Elsie Milliren     | George Schenck       |
| Ana Alarcon             | John Guyon        | Keith Mizutani     | Edward Schlesinger   |
| David Alessi            | Sarah Hale        | Eileen Monteleone  | Derek Semler         |
| Crystal Allsopp         | Grace Haupt       | Jack Moore         | Cheryl Serge         |
| Diane Anderson          | Cloe Hefner       | Kathryn Morgan     | Veronica Silva       |
| Dave Ashley             | Paul Heimann      | Leroy Mueller M.D. | Henry Skala          |
| Sharon Baggett          | Jeffrey Helman    | Keri Naccarato     | Lorenz Snell         |
| Grace Baker             | Helen Henry       | Larry Nichols      | Stafford Family      |
| Jeffery Barberi         | Jody Henry        | Steven Ofenstein   | Joseph Stamer        |
| Teresa Barnes-          | Joseph Henry      | Debbie Omictin     | Edward Steiner, Sr.  |
| Sundquist               | Raymond Hesser    | Darrell Owens      | Mike Strojny         |
| Cheri Bearman           | Alice Hicks       | Willard Peabody    | Grandpa Strojny      |
| Theresa Bent            | Mary Lou Hinman   | Mildred Peabody    | Teresa Sundquist     |
| Frances Berens          | Krista Humes      | Eric Peterson      | Kyle Swier           |
| Sandee Berst            | Howard Hunnius    | Gary Peterson      | Ernest Talarico, Jr. |
| Joseph Black            | Jennifer Jacques  | Ikue Pollak        | Dr. Aymee Torres-    |
| Dr Gary Branch          | Larry Jaffe       | Dominick Pollino   | Michels              |
| Don Britt               | Jane Jaffe        | Ellie Poon         | Kristina Travisano   |
| Ruth Buckley            | Judy Kaiser       | Jeffrey Pracht     | Susan Trippodo       |
| Pauline Caruso          | Dr David Kalamas  | Jack Pyle          | Patty Valiantis      |
| Phyllis Celio           | Marvin Kamen      | Ron Randol         | Jacob Van-Buren      |
| Angelo Cipriani         | Jane Keller       | Charity Ranger     | Janet Veal           |
| Joseph Coffey           | Denis Kelly       | Nate Redman        | Robert Vozar         |
| Raymond Contreras, Sr.  | Grace Kirkwood    | Anne Reed          | David Westrick       |
| Russell Crystal         | Richard Knapp     | Thomas Reese       | Tonia Westberg       |
| Mary Danson             | Jamie Kosieracki  | Jim Richards       | JA White             |
| Kennon Davis            | Kotosky Family    | Janet Riley        | David Whitmer        |
| Page Davis              | Vernon Laible     | Mary Romero        | Emily Young          |
| Mary Davis              | Lealan LaRoche    | Patricia Rymut     | Jon Zilles           |
| Bernadette DeLuca       | Scott Lawrence    |                    |                      |
| Fred Donnelly           | Dwayne LeBlanc    |                    |                      |
| Rick Donnelly           | John Lehto        |                    |                      |
| Denise Drake            | Tony Lewendon     |                    |                      |
| Andrew Egeressy         | Peggy Littlejohn  |                    |                      |
| Lorraine Emanuel        | Adrian Lund       |                    |                      |
| Daniel Eustache         | Michael Lundquist |                    |                      |
| Trinity Falk            | Lee Magnuson      |                    |                      |
| Chie Franklin           | Carly Magnuson    |                    |                      |
| Albert Frei, Sr.        | Patrick Marion    |                    |                      |
| Jerry Frey              | Dave Mason        |                    |                      |
| Gregson Gann            | Susan Mason       |                    |                      |

## 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.



## National Ataxia Foundation

2600 Fernbrook Lane, Suite 119  
Minneapolis, MN 55447-4752  
(763) 553-0020

Non-Profit  
Organization  
U.S. Postage  
**PAID**  
Madison, SD  
Permit No. 32

### 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 \_\_\_\_\_

**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](mailto:joan@ataxia.org).**

### MEMBERSHIP

Yes, I want to help fight ataxia! Enclosed is my membership donation. (*Gifts in U.S. 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

*Recurring Gift Membership Program:*

If you wish to contribute monthly or quarterly, please consider the Recurring Gift Membership Program.

For more information contact the NAF office or visit [www.ataxia.org/giving/default.aspx](http://www.ataxia.org/giving/default.aspx).

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 NAF.

Total Amount Enclosed \$ \_\_\_\_\_

Credit Card:  Visa  MasterCard  Discover

Name on Card \_\_\_\_\_

Card # \_\_\_\_\_

Exp. Date \_\_\_\_\_ CVV # \_\_\_\_\_

Signature \_\_\_\_\_

Phone Number \_\_\_\_\_