



# National Ataxia Foundation

# GENERATIONS

THE OFFICIAL PUBLICATION OF THE NATIONAL ATAXIA FOUNDATION

## SCA Global Launches

Cutting-Edge World-Wide Ataxia Research

Pg. 22

## 2019 Annual Ataxia Conference

Start Planning your trip!

Pg. 30

## Pioneering Discoveries

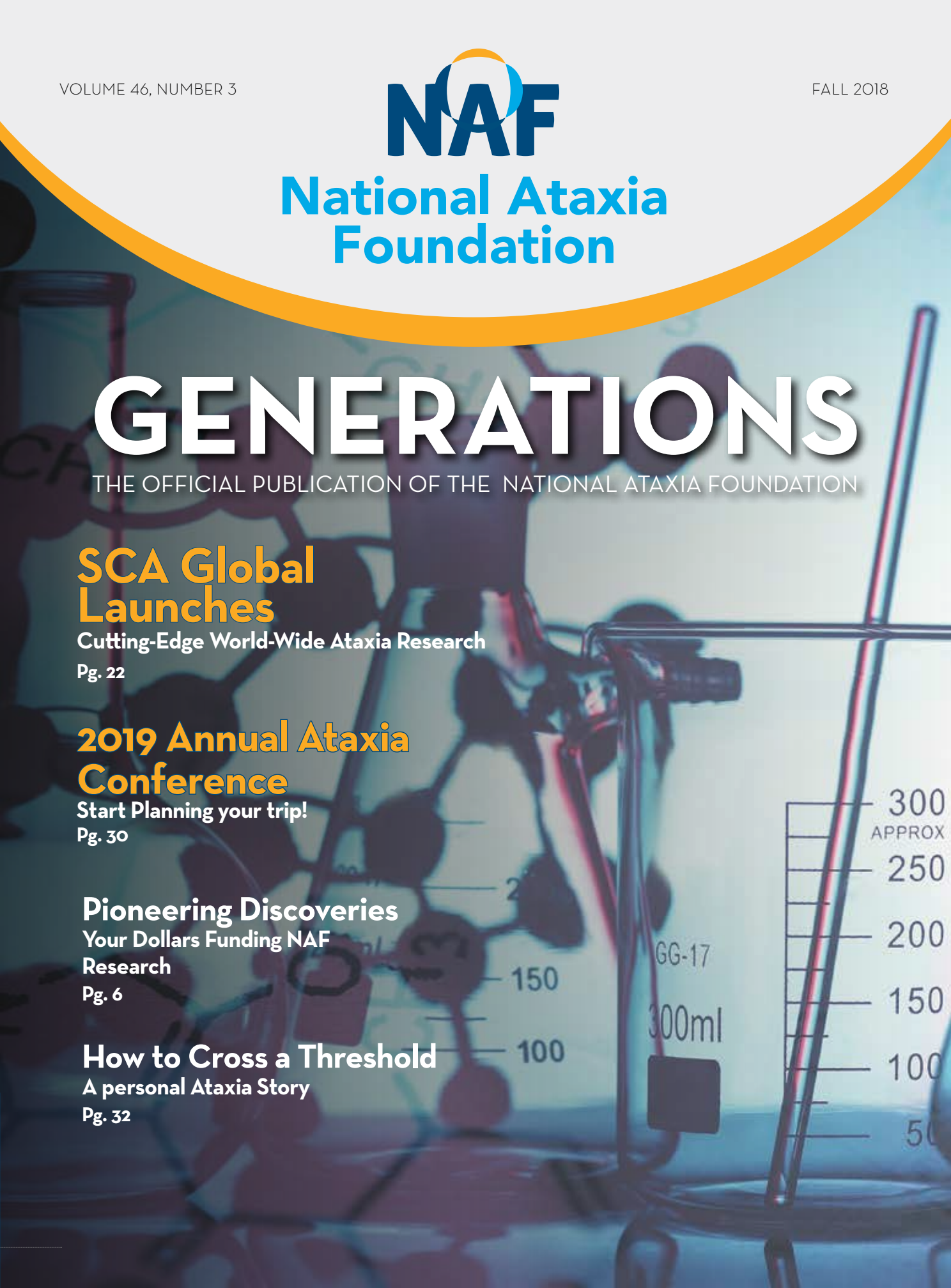
Your Dollars Funding NAF  
Research

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## How to Cross a Threshold

A personal Ataxia Story

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**Deadline to submit materials for the Winter issue of *Generations* is November 5, 2018**

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### Generations Schedule:

Issue	Spring	Summer	Fall	Winter
Mail Date	April	July	October	January

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# Letter from the Director of Development



Joel Sutherland

## **“Money buys research, research finds answers.”**

This is a mantra we all came to embrace just a couple of years ago. Since then we have come together to increase our local grassroots fundraising efforts across the country. While the number of Walk n’ Rolls remain NAF’s largest fundraising campaign, the dollar amounts continue to grow. This is fantastic! Our initial “60 for 60” campaign in 2017 has become our “Go the Extra Mile” campaign and dozens of people are doing what they can to raise funds through their own personal networks. Absolutely phenomenal! In fact, support groups in Sacramento and Wisconsin tripled their initial fundraising goals due to the success they achieved.

Where we see a real increase in activity is in our “Passion Fundraising” programming. Here people work within their own circles of friends, family and colleagues to raise funds by conducting events they enjoy. The result has been golf tournaments, tailgate parties, rock n’ roll events, tea times for a cure, hot air balloon events and many, many more. For those of you who have participated in Walk n’ Rolls, Go the Extra Mile, IAAD or any of the many passion fundraisers, our community says, THANK YOU! It is your efforts that are going to get us closer to the answers we seek. If you have yet to get involved, it is never too late. Heck, last year a long-time member of our community got involved with a “60 for 60” walk, a couple weeks into his fundraising efforts he told me, “I have asked 17 people if they would donate to my walk. They have all said yes!” He went on to raise more than \$15,000 and had so much fun along the way that he is doing his own “Go the Extra Mile” walk this year.

So, thank you, thank you, thank you to all those who have helped raise funds, build awareness and energize our community. For those who have yet to get involved, trust me, it is never too late but why not start today. You see, money buys research and research find answers and we all want these answers sooner rather than later.

All the best to all of you!

A handwritten signature in blue ink that reads "Joel Sutherland".

**SHARE YOUR IDEAS FOR GENERATIONS –  
DEADLINE FOR WINTER ISSUE NOVEMBER 5, 2018**

# Mechanism Behind Rare Ataxia Disease Found by NIEHS Scientists

By Robin Arnette from the National Institute of Environmental Health Sciences

People born with a rare disorder known as ataxia with oculomotor apraxia 1 (AOA1) have problems coordinating body movements and difficulty walking. They are also unable to move their eyes side-to-side and must use their peripheral vision to see by turning their head. No specific treatment exists for this debilitating condition, but NIEHS researchers have now reported the mechanism behind it.



(Left to right): Williams is deputy chief of the NIEHS Genome Integrity and Structural Biology Laboratory and head of the Structural Cell Biology Group. (Photo courtesy of Steve McCaw)

Tumbale is an Intramural Research Training Award (IRTA) fellow in the Williams group. (Photo courtesy of Steve McCaw)



“The DNA damage involves bulky lesions that develop on the ends of DNA,” Williams said. “The wedging mechanism allows aprataxin to expose damaged DNA ends and make it possible for other DNA repair proteins to gain access to these ends.”

## Living with AOA1

When aprataxin does not work properly, it gives rise to AOA1, a rare disorder that affects 1 in 100,000

The results, published June 22 in the European Molecular Biology Organization (EMBO) Journal, are a continuation of their aprataxin research that appeared in the journal Nature

A team led by NIEHS researcher Scott Williams, Ph.D., and NIEHS fellow Percy Tumbale, Ph.D., have shown how the protein aprataxin recognizes DNA damage, or lesions, and how in those with AOA1, aprataxin is inactivated.

Using biochemical and structural studies, they found that in AOA1, aprataxin mutations affect protein stability, how repair proteins bind to DNA, and the rate of the chemical reaction, which is known as catalysis. The team also uncovered a dominant mutation that affects the enzymatic reaction that normally reverses DNA damage. “If aprataxin is inactive and not doing its job, it will impact DNA repair function in cells and cause AOA1,” Tumbale said. “Our characterization of AOA1 mutants may help explain why there is a range of symptoms in these patients, from early onset through late onset.”

## Wedging to repair damage

Williams said when aprataxin is working properly, it uses a wedge-pivot-cut process to recognize DNA damage and create a kink in the DNA. Like a woodworker that uses a triangle wedge to pry open a piece of wood, aprataxin wedges into the DNA damage, kinks it 90 degrees, then cuts out the lesion.

people. Williams and Tumbale said that as people with AOA1 age, they will experience a degeneration of the area of the brain known as the cerebellum, which is characteristic of a lot of DNA repair disorders.

The researchers do not understand exactly how this brain deterioration happens, but over time – generally by adolescence – AOA1 patients are wheelchair bound.

Tumbale received a research grant from the National Ataxia Foundation a few years ago and was fortunate to travel to some of the organization’s meetings. She said meeting AOA1 patients and their families changed her perspective from just doing experiments at the bench. She realized what she was working on could lead to positive changes in their lives.

“They really appreciated that someone was out there trying to understand their disease, and it motivated me to work harder,” Tumbale said. “Hopefully, these findings will lead to a treatment to help them one day.”

Citation: Tumbale P, Schellenberg MJ, Mueller GA, Fairweather E, Watson M, Little JN, Krahn J, Waddell I, London RE, Williams RS. 2018. Mechanism of APTX nicked DNA sensing and pleiotropic inactivation in neurodegenerative disease. EMBO J 37(14):e98875.

# A Look at NAF Funded Research

With the support of our generous donors, NAF funds Ataxia research each year. Researchers submit their grant applications to NAF, which are then put through a rigorous review process by our Medical Research Advisory Board. The highest rated and most promising Ataxia research projects are funded. Many of these projects are deeply scientific and hard to understand to those not in the medical field. However, we think it is important to provide our supporters with insight into the projects that were funded each year. The following are lay summaries directly from the principal investigator for each Ataxia research project funded by NAF for the fiscal year 2017.

## SEED MONEY GRANTS



**Margit Burmeister, PhD**  
**University of Michigan**  
**Ann Arbor, MI**

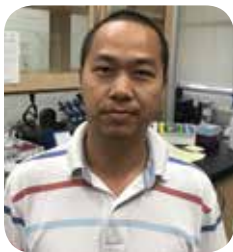
### Role of VPS13D in Ataxia

Before this grant, we had identified a large nuclear family with adult onset recessive ataxia in which we had identified two mutations (one nonsense, one missense) in VPS13D. Mutations in similar genes VPS13A, VPS13B and VPS13C are involved in other neurological disorders.

When only a single family with mutations in a rare gene in ataxia is available, much work is needed to demonstrate that they don't carry these mutations by chance and another gene actually causes the disease. However, within one week of the NAF newsletter publishing the title and lay summary of our grant, we were contacted independently by two different investigators from Europe who had patients with ataxia, spastic paraplegia or combinations such as spastic ataxia, as well as children with early developmental delay and severe movement disorder. Hence, the NAF newsletter had resulted in bringing people together!

We had already identified mitochondrial defects in the flies, and moved then to demonstrating mitochondrial defects in fibroblasts of patients, which were quite striking. Friedreich's ataxia had long known to be a mitochondrial disorder. At the same time, we also heard from another group that also had patients, mostly children, with a variety of movement disorders and VPS13D mutation. Both our group in collaboration with the two European groups and the other group's data have been published and are available online (1, 2). With more than a dozen cases/families, VPS13D can now be recognized as an ataxia/spastic paraplegia gene. Future work is needed on two fronts: 1) to better understand the spectrum of mutations and disorders, as currently, mutations in this gene can lead to a fairly late onset ataxia without cognitive impairment, pure spastic paraplegia, or early onset severe movement disorder with or without intellectual impairment. 2) to identify drugs that help keep the mitochondria in better shape, which may help the patients with this disorder. The patient in our family were helped with N-acetyl cysteine (NAC), as was reported online (3), and after our publication, we are aware that some other patients with VPS13D mutations were advised by their neurologist to try this drug.

## NAF Funded Research (continued)



**Fang He, PhD**  
**Texas A&M University**  
**Kingsville, TX**

### Development of *Drosophila* Model for Spinocerebellar Ataxia Type 36 (SCA36)

Expanded DNA repeat sequences are a common cause of cerebellar ataxia. These repeats can elicit cerebellar degeneration either as RNA or as protein, but the exact mechanisms vary depending on the relative environment of the repeat expansion. Spinocerebellar ataxia type 36 (SCA36) is a recently identified subtype of cerebellar ataxia that is caused by hexanucleotide GGCCTG repeat expansion which is found in different ethnic groups globally. Although the accumulation of expanded repeat-containing RNAs has been observed, how these repeat-containing RNAs contribute to the neuronal toxicity is largely unknown. To examine the potential pathogenic mechanisms that lead to SCA36, we generated constructs containing 100 GGCCTG repeats with various contexts. Evaluation of flies expressing these constructs in the eyes revealed that the expanded GGCCTG repeats are toxic and the toxicity could be independent of the host gene *Nop56* in SCA36, indicating a possible gain-of-function mechanism in the pathogenesis of SCA36 disease.



**Martin Lavin, PhD**  
**University of Queensland**  
**Centre for Clinical Research**  
**(UQCCR)**  
**Brisbane City, Australia**

### Assessing the Role of Senataxin in Cellular Inflammation, Gene Regulation, and Innate Immunity in *Setx*<sup>-/-</sup> Mice and a Human Neuronla Model

The project was designed to investigate the role of senataxin in cellular inflammation, gene regulation,

and innate immunity in a mouse model (*Setx*<sup>-/-</sup>) of the human neurodegenerative disorder, ataxia oculomotor apraxia type 2 (AOA2), a progressive form of cerebellar ataxia. This is a neglected rare neurological disorder that develops mostly in late adolescence to early teens. AOA2 occurs from mutations in the *SETX* gene. The *SETX* gene encodes the senataxin protein, which plays a key role in the response to DNA damage by resolving lesions in DNA called R-loops to regulate gene expression. We had previously shown that senataxin was involved in RNA metabolism including termination of transcription and the splicing of specific forms of mRNA. More recently in collaboration with Dr Ivan Marazzi, Columbia University, we described an unanticipated role for senataxin in controlling innate immunity, the part of the immune system that responds immediately when a toxin or other foreign substance appears in the body. The involvement of senataxin in innate immunity offers new insight into a possible link between neurodegenerative disorders and inflammation. This was the first description of immunodeficiency associated with AOA2. As an approach to understanding the role of senataxin in neuroinflammation we employed an animal model of AOA2, *Setx*<sup>-/-</sup> to identify disease-related mechanisms of pathogenesis by examining patterns of gene expression. We employed RNA sequencing (RNA-Seq) in collaboration with Dr. Peter McKinnon, St Jude's Children's Research Hospital to investigate not only expression of genes but also to determine whether there might be an abnormal pattern of gene splicing in the mutant mouse. A comparison was made between samples from *Setx*<sup>-/-</sup> mice and those from wild-type controls (*Setx*<sup>+/+</sup> mice). This approach confirmed the knockout of *Setx* expression in the *Setx*<sup>-/-</sup> mouse model. A statistical model and computer program, replicate MATS (rMATS) was employed for the detection of differential alternative splicing from replicate RNA-Seq data which detected 255 separate events in splicing of genes. While there

were some differences in individual splicing of genes between the Setx mutant and wild-type mice there was no overall abnormalities in gene splicing in *Atm*<sup>-/-</sup> mice. It was also of interest that only a few genes were either upregulated or downregulated in *Setx*<sup>-/-</sup> compared to *Atm*<sup>+/+</sup> mice. Among these were genes relevant to the neurodegenerative phenotype in AOA2. Future studies will involve the validation of these results and investigations into how these genes function in protecting the brain.



**Sandra de Macedo Ribeiro,  
PhD**  
**Instituto de Biologia  
Molecular e Celular  
Porto, Portugal**

#### New Therapeutic

#### Approaches for Machado-Joseph Disease: Chaperoning Protein Self-Assembly

Spinocerebellar Ataxia type-3 (SCA3) also known as Machado-Joseph disease is a neurodegenerative disease caused by the expansion of a CAG trinucleotide repeat, codifying for amino acid glutamine, in the gene associated to the disease -Ataxin-3 (*Atx3*) - leading to the expansion of the polyglutamine (polyQ) tract within the translated protein. This mutation increases *Atx3* propensity to aggregate leading to its accumulation as intracellular inclusions in specific neuronal cells leading to severe neurodegeneration.

Despite the extensive research in the field, no effective MJD therapies have been developed so far and treatment is mostly symptomatic. One of the reasons for this condition is the lack of information on the three-dimensional structure of full-length *Atx3*, mostly due to its high conformational flexibility and aggregation propensity, delaying the development of new drugs/molecules specifically designed to bind and block *Atx3* aggregation in neuronal cells, preventing MJD. With the financial support

of NAF, we completely characterized a set of specific *Atx3*-binding proteins that can be used as crystallization chaperones and that additionally could be further developed for applications as anti- aggregation agents. The results obtained were crucial for continuing this research and applying for further funding to explore the therapeutic potential of one particular molecule that selectively delays aggregation of the disease related protein. These molecules represent novel tools to improve our understanding of MJD pathogenesis and develop novel targeted therapies.



**Ana Teresa Antunes Simões,  
PharmD, PhD**  
**Center for Neuroscience  
and Cell Biology of Coimbra,  
University of Coimbra  
Coimbra, Portugal**

#### Calpain-Mediated Proteolysis in Machado- Joseph Disease

Machado-Joseph disease, also known as Spinocerebellar Ataxia type 3 (MJD/SCA3), is the most frequent worldwide autosomal dominantly-inherited Ataxia.

In MJD, a mutation leads to a polyglutamine stretch bigger than normal at ataxin-3 protein, a biological molecule that is important for cellular quality control. It is believed that ataxin-3 is cut into smaller fragments, being these considered the toxic species. We and others have recently shown that the molecules responsible for ataxin-3 break down are called calpains, which mediate the formation of toxic fragments, ataxin-3 translocation to the nucleus and neurodegeneration (Haacke et al., 2007, Koch et al., 2011, Simões et al., 2012, Hübener et al., 2013, Simões et al., 2014, Weber et al., 2017). However, their specific modes of activation and posterior action have not been investigated.

## NAF Funded Research (continued)

The aim of this project was to understand in vivo the calpains contribution to MJD pathogenesis. For that, we evaluated in which places is ataxin-3 cut, which cleavage fragment is more toxic and whether its decrease and the deletion of the amino acids responsible for cleavage could be envisioned as a therapeutic strategy for MJD.

In this sense, we: a) identified the putative calpain cleavage sites for both wild-type and mutant ataxin-3, b) confirmed that the mutation of these sites led to the abrogation of the toxic fragments formation, c) identified the 26 kDa fragment as the major contributor for striatal degeneration, and d) observed a neuroprotective effect upon calpain cleavage sites mutagenesis.

In conclusion, these findings suggest that the calpain system should be considered for MJD therapeutic intervention and also for other neurodegenerative diseases vulnerable to calcium deregulation. Furthermore, the identification of the calpain cleavage sites will allow the design of specific drugs or the use of tools for genome editing at a specific location.



**Clara Van Karnebeek, MD, PhD**  
**University of British Columbia**  
**Vancouver, BC, Canada**

### Whole Exome Sequencing in the Diagnosis and Management of Atypical Childhood Hereditary Ataxia Conditions

The National Ataxia Foundation (NAF) was formed by John and Henry Schut in 1957 to promote awareness of the degenerative ataxias, support research into the causes and cure of the disease and provide support and comfort to ataxia patients and their families. We are resolved to advance these primary objectives through our project “WES in Atypical Childhood Ataxia.”

New technologies have drastically changed the way we diagnose rare diseases. The human genome contains about 3 billion bases or letters. For over a decade, researchers have had the ability to read a person’s entire genome through a process called whole genome sequencing (WGS). Now, through testing known as whole exome sequencing (WES), we can now focus in on the “coding” portion of the genome, called the exons, that provides instructions for making proteins. WES methods allow the body’s entire set of instructions—or exome—to be examined as a single laboratory test—rather than having to individually analyze all 20,000 genes that make up our exome. Through WES, we are able to look for “spelling mistakes” (known as pathogenic mutations) in gene(s) and then determine if these changes are the cause for the person’s illness.

With this innovative tool, we have achieved our project’s goal and offered WES to 12 children with severe or complex Ataxia and merged our center’s unique expertise in combining deep characterization of a patient’s clinical picture (combined physical, neurological and metabolic symptoms) with these WES data. This enabled us to study the Ataxia patients at a physical, genetic and chemical level which resulted in (1) a diagnostic yield of 67% (n=8 patients) with (pending) publications in medical journals for all of these ; (2) the discovery of new genes that cause Ataxia (GLS, ATP1A1, GOT2, CIAO1, MMS19), (3) the expansion of what we understand about the clinical picture of known human genes that cause ataxia (MTO1, PROSC) (4) make sense of these rare genetic Ataxic conditions at a microscopic “molecular” level so that we can understand, target, treat and potentially cure these disorders: tauroursodeoxycholate as chemical chaperone for the mitochondrial disease MTO1 deficiency (pre-clinical studies in cell-lines ongoing), monosodium glutamate supplementation for GLS deficiency (n-of-1 studies starting fall 2018), Lserine and

pyridoxine supplementation for GOT2 deficiency (n-of-1 trials ongoing), Levo-carbidopamine for PAK3 deficiency). With our focused study of Ataxic cases at our center, we have positively affected the lives of patients and families by providing them a diagnosis (an answer, an end to a diagnostic odyssey, more accurate genetic counselling), while forging the discovery of 5 new genetic Ataxia disorders, along with an expansion of the descriptions and understanding of known conditions, and finally created new treatment opportunities for 4 genetic Ataxia conditions. Thanks to the National Ataxia Foundation we are able to deliver on the of Precision Medicine and create a better future for children and families suffering Ataxia.



**Adam Vogel, PhD**  
**Center for Neuroscience of Speech**  
**Parkville, Victoria, Australia**

### **Intensive Home-Based Speech Rehabilitation for Adults with Degenerative Ataxia**

Speech worsens in Ataxia, yet there are no evidence-based treatments to slow, halt or reverse its progression. With the help of the National Ataxia Foundation, we tested the efficacy of a dysarthria treatment program designed to improve speech. Methods were based on the latest understanding of how the brain produces speech in people with Ataxia and how improvements can be made with practice and the right tasks. We developed a speech treatment program that was completed in the home without face to face contact with a therapist. It was delivered on laptop computers which provided different types of feedback (visual, listening) to users as they progressed through therapy. Participants were required to complete tasks five days a week for a month which focused on improving how clearly, they produced speech and how much control they had of their voice. The preliminary study was run

in Germany and Australia and yielded promising results. Expert listeners blinded to time point and diagnosis rated the speech of participants. They found that 18/20 (90%) participants showed a clinically meaningful response to treatment. The two participants who saw no change had very mild dysarthria at baseline. These positive results have meant the treatment program has been refined and is now being evaluated in a large multisite trial run across Australia, Germany, France and New Zealand.



**Lilians Simões Mendonça, PhD**  
**Harvard University**  
**Boston, MA**

### **The Transplantation of Induced Pluripotent Stem Cells (iPSC)- Derived Neural Stem Cell (NSC) in Machado Joseph Disease (MJD).**

Machado-Joseph disease (MJD) is a very incapacitating neurodegenerative disease caused by a mutation on the ATXN3 gene that originates a mutant ataxin-3 protein. Mutant ataxin-3 is toxic causing neuronal dysfunction and degeneration in specific brain regions. MJD patients exhibit significant motor impairments, such as gait ataxia and speech problems, associated with multiple neuropathological modifications including significant neuronal loss and atrophy of the cerebellum. There is no effective treatment able to stop the progression of this disease. Nevertheless, we have recently demonstrated that transplantation of neural stem cells (NSC) in a MJD-mouse model result in a substantial improvement of the MJD associated neuropathology and motor impairments. However, absence of human NSC sources that escape immune rejection and without associated ethical limitations is a huge obstacle in the implementation of cell therapies for treatment of neurodegenerative diseases as MJD. One way to overcome this problem is through the generation of

## NAF Funded Research (continued)

the required cells by reprogramming patients' skin cells into induced pluripotent stem cells (iPSC), which can be induced into neural progenitor cells to be used for brain transplantation. Therefore, we evaluated whether it is possible to generate patient-specific iPSC-derived neural progenitor cells depleted of the mutation responsible for the disease in order to use these cells to promote functional recovery in MJD mouse models. Our results indicate that iPSC-derived neuroepithelial stem cells (NESCs) obtained by reprogramming fibroblasts of control and MJD patients originate upon in vitro differentiation into glia and functional neurons. Moreover, 2 months upon transplantation of the NESC into the cerebellum of adult mice it was observed that these cells survive, migrate and differentiate into glia and neurons. Presently, we are evaluating the safety of iPSC-derived NESC brain transplantation at longer time points and the impact of the cells transplantation in the MJD-associated neuropathology and motor coordination impairments. It is expected that transplantation of human NESC will alleviate MJD phenotype providing the proof of principle for this approach and open the way for future clinical trials.

## YOUNG INVESTIGATOR AWARDS



**Sathiji Nageshwaran, MD**  
**Harvard University**  
**Boston, MA**

**Epigenetic Editing Using  
CRISPR/Cas Mutant  
Proteins as a Novel Therapy**

### for Frataxin Gene Silencing

Thanks to the funding offered by NAF I am able to conduct a study of the specific epigenetic silencing mechanisms in Friedreich's Ataxia (FA) using CRISPR technologies within Prof George Church's Lab at Harvard Medical School. I have

also been able to develop a tool along with Dr Alejandro Chavez as a means to ease the use of multiple CRISPR gRNAs. The award also permitted my interaction with Prof Schmahmann at Massachusetts General Hospital, where I have presented my results and have integrated into the MGH Ataxia Unit. My research experience in the USA, facilitated by NAF, has also led to my decision to continue on as an Ataxia focused Physician-Scientist in the USA, and I will hopefully be entering Neurology training in the following year. I intend to expand my research scope beyond FA, to other trinucleotide repeat disorders including the SCAs and FXS.

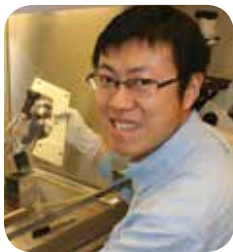
### CRISPR based Epigenetic Engineering

In FA, the abnormal expansion of GAA repeats within the frataxin gene results in pathological silencing of the gene. A majority of patients harbour these mutations and subsequently produce less frataxin RNA and an insufficient quantity of frataxin protein. Despite the presence of the mutation, patients are still able to produce small amounts of frataxin. This fact allows for the possibility of increasing intrinsic frataxin production as a means to therapy. Drugs have been trialed with varying success that can release the silencing of frataxin by inhibiting enzymes that silence repetitive DNA (histone deacetylase inhibitors). However, these drugs are broad in their therapeutic targets and 'treat' all the DNA within a cell. As such, my project has been to specifically target the frataxin gene with proteins that may increase the production of frataxin RNA, without effect elsewhere in the genome. This has now been made possible by the genome engineering tool CRISPR, which the Church Lab were fundamental in the discovery and use of.

I have firstly been able to replicate results by a Canadian group whereby targeting specific promoter regions of the frataxin gene with an 'activator' was able to increase RNA by 1.3 fold. This did not occur when targeting around and within the GAA repeat.

I then used a specific enzyme that is able to add activating marks to DNA around the GAA region, which in diseased cells has silencing marks. A moderate effect (1.5 fold increase) was seen when targeting the region directly before the GAA expansion. The lack of a strong effect is likely due to the multiple layers of silencing involved in FA. As such attempting to combine enzymes that are likely to address the various layers is a sensible next step of investigation (a FA de-repression tool) as well as a single experiment which addresses multiple layers (combinatorial epigenome engineering). I am presently attempting to develop these tools in the lab. This work is being completed in collaboration with Prof Festenstein's group at Imperial College London, where I am mentoring 3 undergraduate students remotely.

As unbiased methods of screening are invaluable in providing new therapeutic targets. I have also been able to create and am currently validating a cell line which contains a fluorescent protein fused to the frataxin gene with a GAA expansion. Such a line does not exist in the field to date and will provide targets that are active in silencing frataxin in an unbiased manner.



**Bing Yao, PhD**  
**Emory University**  
**Atlanta, GA**

**Epigenetic Modulation  
Mediated by RNS-  
Binding Proteins in  
Neurodegeneration**

Deoxyribonucleic acid (DNA) carries our genetic information coded by sequence combinations. DNA conveys this information by transcribing genes into sequence complemented Ribonucleic acid (RNA), which can make proteins for all the proper functions in the body. RNA-binding proteins (RBP) directly associate with RNA molecules and play fundamental roles for many aspects of RNA regulations, such as how long

they exist in the cells and when they can produce proteins. Many RBPs are specifically present in the brains of mammals, including mouse and human, and are crucial for proper neuronal activities. Loss of these RNA-binding proteins in neurons often leads to severe neurodegenerative diseases. Fragile X-associated tremor/ataxia syndrome (FXTAS) is a neurodegenerative disorder that usually develops in the late stage of adulthood. It is caused by a 55 to 200 copy nucleotide "CGG" repeat in the fragile X mental retardation 1 (FMR1) gene whereas a normal individual should have less than 50 copies. In FXTAS, these extra CGG repeats in RNA (rCGG) transcribed from DNA can trap an RNA-binding protein called "Heterogeneous nuclear ribonucleoproteins A2/B1 (hnRNP A2/B1)", which has important functions in neurons. Thus, the restraint of this critical RBP leads to the death of critical motor neurons called Purkinje cells, which causes ataxia. To confirm the primary roles of hnRNP A2/B1 in FXTAS pathogenesis, we re-expressed hnRNP A2/B1 in the Purkinje cells of FXTAS mice where 90 copies of "rCGG" cause Purkinje cell death and Ataxia in the mice. Remarkably, replenishment of hnRNP A2/B1 in Purkinje cells ameliorates the CGG-induced Purkinje cell death in FXTAS model, presenting the direct evidence of hnRNP A2/B1 sequestration in FXTAS pathogenesis in mammals. Interestingly, our recent finding suggests 5-hydroxymethylcytosine (5hmC), an important chemical modification on DNA molecules that participates in controlling gene expression, displays genome-wide alteration in FXTAS mouse brains. In order to determine how the restraint of RNA-binding protein hnRNP A2/B1 leads to DNA 5hmC alteration in FXTAS mouse brains, we propose to explore whether hnRNP A2/B1 can also interact with DNA molecules and influence chemical modifications on DNA. Funded as a National Ataxia Foundation Young Investigator, I find hnRNP A2/B1 indeed physically interacts with DNA molecules in the test tubes. The hnRNP A2/B1 DNA-binding characteristics are

## NAF Funded Research (continued)

further confirmed inside the cell lines cultured in the laboratory. Importantly, when hnRNP A2/B1 level is artificially reduced in the cell lines by a technique called “small RNA knockdown”, a process mimicking hnRNP A2/B1 restriction in FXTAS brains, the global 5hmC is indeed reduced similar to the observation in FXTAS. By using a technique termed fluorescence-activated cell sorting (FACS) that is able to isolate Purkinje cells from mouse brains, we confirmed 5hmC is consistently altered in FXTAS mouse Purkinje cells. Many genes with 5hmC alteration are relevant to FXTAS pathology. My study presents a direct link of RNA-binding protein hnRNP A2/B1 in FXTAS pathogenesis through genome-wide DNA 5hmC regulation to potentially influence key gene expression. These findings shed light on the discovery of therapeutic targets for FXTAS and potentially other ataxia patients.



**Vincenzo Gennarino, PhD**  
**Columbia University**  
**New York, NY**

### **PUMILIO1 Deficiency: Understanding a New Ataxia Gene and its Role in**

### **Cerebellar Dysfunction in Mice and Humans**

Lay Summary: The molecular genetic revolution of the 1990's brought us tremendous knowledge of the genetic mutations that cause many neurological diseases, including many ataxias. Further research into the proteins produced by these genes has revealed that there is another way for a protein to cause havoc in the brain besides being mutated: it might be expressed at levels too low or too high. In the case of several neurodegenerative diseases, including Spinocerebellar Ataxia Type 1 (SCA1) and more common diseases such as Alzheimer's and Parkinson's, it has been shown that too much of even the normal protein (known as “wild-type” among scientists) can cause the same disease.

This led me to search for factors that might control the levels of Ataxin-1 (ATXN1), the protein that is mutated in SCA1. I discovered that ataxin1 levels are regulated by an RNA-binding protein called Pumilio1 (PUM1). Take away PUM1 in a mouse model of SCA1, and ATXN1 returns to normal levels, and the SCA1 mice no longer have ataxia (Gennarino et al, Cell 2015).

We also noticed, however, that mice lacking Pum1 (the mouse version of the protein) developed other symptoms, such as seizures, and they developed ataxia earlier than the SCA1 mice. This led us to suspect that loss of PUM1 function might underlie some childhood ataxia diseases. Thanks to the National Ataxia Foundation, I was able to reach out to medical geneticists to search for such patients. We identified eleven patients who had severe loss of PUM1 function (nine from a deletion of the PUM1 region, and two patients with inactivating mutations in PUM1) who show symptoms that are very similar to those of Pum1 mutant mice. All suffered developmental delay, ataxia and seizures in childhood. We called this disease PADDAS (Pumilio1-associated developmental delay and seizure) syndrome. We also identified a large family with a mild mutation in PUM1 who have an adult-onset, slowly progressive mild ataxia. We refer to the late-onset disorder as PRCA (Pumilio1-Related Cerebellar Ataxia) (Gennarino et al, Cell 2018). After the publication of this paper on the patients, the diseases were entered into the online library known as OMIM (Online Mendelian Inheritance in Man) under the rubric of SCA47.

The NAF's support allowed me to confirm that these mutations in human patients are indeed causative of disease. (We all carry various polymorphisms, and most of them do not cause harm in any obvious way.) We obtained cells from PADDAS and PRCA patients and made cell lines to study how mutant PUM1 was functioning. We found that all the patient cell lines had lower-than-

normal total PUM1 protein levels—and the degree of protein reduction was directly related to the age of onset and severity of symptoms. Patients that had 40% or less of normal PUM1 protein levels developed disease in infancy, whereas patients who retained ~75% of PUM1 levels developed late-onset, mild disease. We are now working on identifying the molecular mechanisms by which PUM1 levels influence neuronal function in human and mouse, with the hope of developing viable therapeutic approaches.



**Ricardo Mouro Pinto, PhD**  
**Harvard Medical School**  
**Boston, MA**

Identification of Genetic Modifiers of Somatic GAA Instability in Friedreich

Ataxia by in vivo CRISPR-Cas9 Genome Editing

In Friedreich's Ataxia (FRDA) the most common mutation consists of an expanded stretch of repetitive DNA in the frataxin gene - a GAA trinucleotide. The longer the GAA repeat, the less frataxin protein is produced. In addition to being expanded in FRDA patients, this repeat has a tendency to further expand, not only in transmissions from parent to child, but also throughout the life of the patient, particularly in organs primarily affected in FRDA. This raises the hypothesis that this continuous expansion process can accelerate the onset and progression of the disease in FRDA patients. To date, we have already learned that genes involved in maintaining the integrity of our genetic makeup throughout the life of a cell (DNA repair genes) are involved in the GAA expansion mechanism. In addition, a recent study in Huntington's Disease (HD), which is also caused by a trinucleotide repeat, revealed that genes involved in various DNA repair tasks are affecting the age of disease onset, therefore supporting the potential therapeutic impact of targeting these genes. However, we still have a very limited understanding of how these DNA

repair genes actually contribute to trinucleotide repeat expansions. Knowing the key players and understanding the mechanism in much more detail is important since it will enable the development of therapeutics that directly target the genetic cause. With this goal in mind, we have developed a CRISPR/Cas9-based experimental platform (in adult mice) that facilitates testing if candidate genes can either slowdown or accelerate the trinucleotide repeat expansion process.

## **POST-DOC FELLOWSHIP AWARDS**



**Collin J. Anderson, PhD**  
**University of Utah**  
**Salt Lake City, UT**

Development and Mechanistic Study of Deep Brain Stimulation of Dentate

Nucleus for the Treatment of Degenerative Ataxia

In this work, we set out to perform experiments that could begin to address the major need for novel therapeutic strategies in the treatment of Degenerative Cerebellar Ataxias (DCAs). In many forms of DCAs, cerebellar Purkinje cells degenerate, and this modifies the inputs received by the deep cerebellar nuclei, which help integrate several forms of inputs in the production of coordinated movement. We determined that one possible way to go about treating this group of disorders would be to target the deep cerebellar nuclei with a novel form of electrical stimulation - like deep brain stimulation for Parkinsonism or essential tremor - aimed at modulating the neural activity in the deep cerebellar nuclei to a state in which they can better contribute to coordinated movement after Purkinje cells have been irreversibly lost.

We conducted a series of experiments using the Wistar Furth shaker rat, a genetic model that

## NAF Funded Research (continued)

closely recapitulates the Purkinje cell degeneration in the cerebellum and consequent ataxia, also presenting with cerebellar tremor and falls that are similarly observed with relatively high frequency in patients. We developed novel methods of quantifying severity of incoordination, tremor, and falling in an automated fashion, allowing the avoidance of rating scales and indirect measures frequently used to evaluate rodent models of Ataxia, and we used these methods to rigorously characterize the model in comparison with age- and breed-matched wild type rats, determining baseline measures from which to measure symptomatic improvement when testing therapeutic strategies. Once the model was fully characterized, we implanted several cohorts of shaker rats and evaluated the effects of electrical stimulation targeted to the dorsal dentate nucleus on motor symptom severity.

Our primary result in these studies is that low-frequency stimulation of 20-30 Hz reduced all three measured symptoms. However, several additional results are of interest. First, high-frequency stimulation of more than 100 Hz, similar to basal ganglia stimulation for Parkinsonism, actually worsened coordination while failing to significantly change the presentation of other symptoms. Since high-frequency and low-frequency stimulation are thought to have opposite effects, these results generate mechanistic insight as to how dysfunction in the differing regions of cerebellum and basal ganglia lead to symptoms in very different ways. Further, very low frequency stimulation - 4 Hz, matching the frequency of tremor - worsened tremor, demonstrating how large-scale neuronal oscillations can encode tremor. The initial set of experiments occurred with stimulation applied during a short duration; however, to further demonstrate feasibility and lasting therapeutic nature, we applied 30 Hz stimulation for a more moderate timeframe, several hours, and showed that symptom alleviation was

still present. Thus, reduction in symptoms is not simply a short-term response to low-frequency electrical stimulation, and electrical stimulation may provide a viable, long-term therapeutic strategy. Finally, we localized implanted electrode locations in all implanted animals and determined that therapeutic benefit required at least one of the two electrodes to be located within a region in which stimulation spread would reach the target. Thus, generalized cerebellar stimulation without dentate stimulation is ineffective, and therapeutic benefit requires precise stimulation.

The completion of the studies described above demonstrated that deep brain stimulation of the dentate nucleus might provide a novel means of treating numerous forms of ataxia that don't currently have a viable treatment method. Further, these results have led us to the point of being able to couple neural recordings with therapeutic strategies. The absence of treatment strategies in most studies has limited observations to those pertaining to neurological changes that occur with symptoms, but do not necessarily cause them. However, with our newfound ability to modify symptom severity, we can determine if certain activities within neurons and neural groups return to those observed in healthy conditions with the application of treatment. Thus, we can determine if these changes covary with symptom severity. Further, we can now use electrical stimulation to encode neural signals thought to underlie motor symptoms directly into wild type rats, and we can directly evaluate related hypotheses. Thus, in our currently ongoing work, we have implanted both shaker and wild type rats with electrode arrays capable of both stimulation and neural recording at the same time, and we are performing exciting work to determine covariance and causation in the cerebellothalamocortical circuitry. These results will not only enrich our understanding of the cerebellum and ataxia, but they will provide new metrics for future therapeutic studies.

This fellowship award from the National Ataxia Foundation has directly led to important results in the ataxia field, and the results of the thus-far completed studies are continuing to improve our ability to conduct important experiments.



**Laura C. Bott, PhD**  
**Northwestern University**  
**Evanston, IL**

### Transcellular Regulation of the Proteostasis Network in SCA3

Spinocerebellar Ataxia type 3 (SCA3), also known as Machado-Joseph disease, is an inherited neurodegenerative disorder that is caused by a mutation in ataxin-3. At present, effective treatment is not available for this disease, and the key mechanisms that underlie neuronal dysfunction are unknown. Ataxin-3 is present in many tissues, including non-neuronal cell types, where it functions in the cellular machinery controlling protein abundance, folding, and transport (protein homeostasis, or proteostasis). Maintenance of protein homeostasis is essential because imbalances in any of these processes can pose a danger to cell function and the health of organisms. In this project, we aim to obtain a better understanding of protein homeostasis in SCA3. We make use of the microscopic worm *Caenorhabditis elegans*, which is ideal to study this process in an intact animal owing to its small size, transparency, and short lifespan. We have developed new tools that allow us to monitor protein homeostasis inside live worms in real-time. This technique can reveal cell populations that are vulnerable to imbalance and it may shed light on the sequence of events in SCA3 before and after symptoms first appear. We are also searching for ways to restore protein homeostasis in worms that have been genetically engineered to have the disease-causing mutant protein in their neurons. Detailed knowledge of cell type-specific events and cross-talk between tissues will not only help to improve our

understanding of the disease but may also lead to new treatments for SCA3.



**Jonathan Chen, PhD**  
**Scripps Florida**  
**Jupiter, FL**

### Rapid Structure-Based Lead Optimization of a Small Molecule Drug that Target r(CAG)<sub>exp</sub>

In Spinocerebellar Ataxias (SCAs), r(CAG) repeat expansions [r(CAG)<sub>exp</sub>] located in the untranslated region (UTR) or coding region form stable structures and cause neuron degeneration through RNA and/or protein gain-of-function mechanisms. These mechanisms include sequestration of splicing proteins by r(CAG)<sub>exp</sub>, which lead to aberrant splicing, and translation of r(CAG)<sub>exp</sub> into polyQ proteins, which lead to formation of aggregates. One strategy for treating these diseases is to target the RNA structures with small molecules that reverse splicing defects or inhibit translation of the RNA. Previously, our group identified a small molecule, D6, that was bioactive against r(CAG)<sub>exp</sub> in Huntington's disease. In this work, we characterized structural interactions between D6 and a single r(CAG) motif. Using computational methods and distance restraints generated by NMR spectroscopy, we determined the structures of an unbound and D6-bound RNA construct containing a r(CAG) motif. In the unbound structure, the r(CAG) motif consists of an AA mismatched base pair stabilized by a single hydrogen bond and closed by canonical GC pairs. In the bound structure, D6 stacks between the adenines of the adenines and closing GC pairs, disrupting the AA base pair. D6 also forms hydrogen bonds with the closing GC pair and sugar phosphate backbone of the RNA. Overall, binding of D6 to the RNA induces distortions in the structure of the RNA that may facilitate these interactions that stabilize the RNA-small molecule complex. This structure

## NAF Funded Research (continued)

may be used to optimize the compound for binding to the RNA. Separately, we determined the structure of an RNA construct with three adjacent r(CAG) motifs using NMR spectroscopy and computational methods. The results indicate that each r(CAG) motif consists of stable AA base pairs flanked by canonical GC pairs, and that the r(CAG) motifs are conformationally dynamic. These results will be useful for understanding the structure and dynamics of r(CAG) repeats and further aid in the development of therapeutics, such as dimeric compounds, which simultaneously target two r(CAG) motifs with enhanced selectivity and bioactivity against r(CAG)<sub>exp</sub> in SCAs.



**Stephanie Seminara, MD**  
**Massachusetts General**  
**Hospital**  
**Boston, MA**

### Ataxia with Hypogonadotropic Hypogonadism Due to Ubiquitin Ligase Dysregulation

Gordon Holmes syndrome is a clinical disorder characterized by dementia, difficulty in coordination and loss of reproductive function. This neurodegenerative disorder, which affects the cerebellum, hypothalamus and pituitary, progresses inexorably to death, but little is known about what causes this devastating disease. Our team identified mutations in genes (RNF216 and OTUD4) that work in the ubiquitination pathways. RNF216 tags proteins for degradation in the cell. OTUD4, a deubiquitinating enzyme, can reverse these effects by cleaving the bond between ubiquitin and its substrate protein. Although the protein targets of RNF216 and OTUD4 are not fully known, RNF216 appears to play multiple biologic roles, including as a modulator of inflammation and a regulator synaptic strength.

Our aim was to focus on elucidating the biology underlying RNF216. First, we sought to understand the location of the analogous protein, Rnf216, in the brains of normal mice. We demonstrated that Rnf216 is expressed at low levels, but widely, throughout the brain. We then studied genetically engineered mice in which gene encoding Rnf216 was inactivated or “knocked out.” We uncovered abnormalities in the male knockout mice that parallel clinical findings in human patients with Gordon Holmes Syndrome, including both reproductive and neurologic deficits. Mutant male mice have small testes and are infertile. They also have neurologic impairments. Surprisingly, female mutant mice did not demonstrate any reproductive or neurologic abnormalities.

Thus, this mouse model suggests that the actions of RNF216/Rnf216 are concordant across mammalian species. Just like patients with Gordon Holmes syndrome who carry mutations in RNF216, mice with deletions in the analogous gene, Rnf216, have both reproductive and neurologic defects. As human patients include individuals of both sexes, the gender discordance between male and female mice is unclear. The possibility that sex hormones may influence the presentation or severity of the disease deserves further study



**Ravi Chopra, PhD**  
**University of Michigan**  
**Ann Arbor, MI**

### Identifying Dendro- Protective Ion Channels in Cerebellar Ataxia

In the Cerebellar Ataxias including Spinocerebellar Ataxias type 1 and type 2 (SCA1 and SCA2), there are progressive and consistent changes in the shape and appearance of nerve cells prior to their loss. The basis for why there is a reduction in size of the cerebellar Purkinje cells,

defects in which are the major cause of motor impairment in Ataxia, in Cerebellar Ataxia is poorly understood. Purkinje cell dendrites, which receive electrical signals from other parts of the brain, are particularly vulnerable to degeneration in ataxia. We examined whether problems in generating electrical signals in Purkinje cell dendrites contributes to degeneration in cerebellar ataxia. Persistent increases in Purkinje cell dendrite electrical excitability are present in mouse models of ataxia. Remarkably, improving the excessive dendrite excitability also preserves dendrite health, and improves motor impairment in mouse models. Signaling pathways that have been implicated in other spinocerebellar ataxias such as SCA14 also converge on increased electrical excitability in the dendrite, suggesting that addressing excessive dendrite excitability is a shared pathway of neuroprotection across multiple causes of Ataxia.



**Austin Ferro, PhD Candidate**  
**University of Minnesota**  
**Minneapolis, MN**

### **The Role of Astrocytes in SCA1 Via F-kB Pathway**

Spinocerebellar Ataxia 1

(SCA1) is an incurable and genetically inherited disease characterized by progressive impairment in movement and cognition as well as premature death. Our previous results in a mouse model of SCA1 showed that astrocytes—support cells in the brain required for the normal functions of neurons—may play an important role in SCA1. We have found that during the early, presymptomatic stages of SCA1, astrocytes may play a neuroprotective role while they become harmful during the late stages of SCA1. Using NAF funding we have now performed experiments that suggest that regulation of neuronal function may be the mechanism by which astrocytes can be neuroprotective and harmful at different stages of SCA1.



**James Orenge, MD, PhD**  
**Baylor College of Medicine**  
**Houston, TX**

### **Unraveling the Mechanisms of Motor Neuron Degeneration in**

### **Spinocerebellar Ataxia, Type 1**

SCA1 is a genetic disorder caused by numerous repeats of an amino acid (glutamine) within the protein called Ataxin1. In SCA1 neurodegeneration occurs in a region of the brain called the cerebellum. The cerebellum is critical for coordinating movements and fine-tuning balance; therefore the hallmark feature of SCA1 is progressive and debilitating incoordination. However, later in life individuals with SCA1 will also develop progressive muscle weakness, a symptom not associated with the cerebellum. The muscles especially affected are those that support safe breathing and swallowing. In fact individuals of SCA1 often pass away prematurely due to complications of aspiration pneumonia, which are directly related to the weakening of these muscles.

Progressive weakness of the muscles supporting breathing and swallowing is a common symptom in diseases with degeneration of motor neurons. The most common disease within this class is amyotrophic lateral sclerosis (ALS) or Lou Gehrig's disease. Motor neurons are responsible for triggering the voluntary contraction of their target muscles. When motor neurons die they leave behind orphan muscles that subsequently, under the lack of direction, shrink and become weak. Given that SCA1 patients ultimately succumb to symptoms common in diseases involving degeneration of motor neurons, I came up with the hypothesis that motor neuron degeneration occurs in SCA1 and drives premature death. Indeed, previously reported autopsy examinations in SCA1 patients demonstrated that they do have decreased numbers of motor neurons and those that are present are sick looking. Armed with this human data I turned my attention to a mouse model of SCA1, made by Huda Zoghbi's lab, to see if it recapitulates these findings.

## NAF Funded Research (continued)

Over different time-points in the life span of SCA1 mice I measured their breathing function and found that the SCA1 mice over time take in shallower and shallower breaths, and at the same time ramp up their respiratory rate in an attempt to compensate for the decrease in air exchange in their lungs. This together with other parameters studied tell us that the SCA1 mice have a progressive respiratory dysfunction and it is due to weakness of the muscles supporting breathing. I next analyzed the main muscle supporting respiration, the diaphragm and found that the muscle shows features of having lost motor neuron input. I then moved my way up to the spinal cord where the motor neurons that direct the contraction of the diaphragm live. Here I also saw that over time these neurons degenerate, and neuronal scarring takes place. Taken all together this data and other data not discussed here support my hypothesis that the SCA1 mice pass away prematurely from respiratory dysfunction and that this correlates with motor neuron degeneration. This work was published in Disease Models and Mechanisms in February 2018.

## YOUNG INVESTIGATOR – SCA



**Manu Ben-Johny, PhD**  
**Johns Hopkins University**  
**Baltimore, MD**

**Aberrant Regulation of Voltage-Gated Na Channels in the Pathophysiology of Spinocerebellar Ataxia 27**

Spinocerebellar Ataxia 27 (SCA27) is a recently identified class of ataxia with patients experiencing tremors, difficulty with gait, and often performing poorly in cognitive tests. The genetic basis for this disorder are mutations in a small protein called Fibroblast growth factor homologous factor 4 (FHF4), which are found in neurons. Our work here addresses how the family of FHF molecules

modifies the function of voltage-gated sodium channels - molecules that are essential for generation and propagation of electrical impulses in neurons. Specifically, recent work showed that FHF molecules interface with sodium channels via an interface that also dock calmodulin, a protein that typically senses levels of intracellular calcium ions and reduces sodium channel activity. Therefore, we tested whether FHF molecules might alter calmodulin's ability to modify sodium channels and whether this effect plays a role in how SCA27 develops and progresses.

In this project, we uncovered an interplay between FHF and calmodulin in modifying sodium channel function. Specifically, we found that while calmodulin dynamically reduces activity of sodium channels in response to increases in cellular calcium levels, FHF hinders this process allowing the sodium channels to maintain normal level of activity regardless of calcium levels. Moreover, mutations in FHF disrupt his function, causing enhanced suppression of sodium channel activity. Last, we also found that FHF binding alters baseline activity of sodium channels. To understand these complex and diverse functional effects, we also undertook mathematical modeling to dissect how this novel interplay between calmodulin and sodium channels alter action potentials in the cerebellum, a brain region critical for coordinating movement. Overall, this project helps advance our fundamental understanding of molecular interactions that sculpt neuronal function and coordination of movement and identifies new targets for development of therapies for Ataxia.



**Marija Cvetanovic, PhD**  
**University of Minnesota**  
**Minneapolis, MN**

**Role of Astrocyte Calcium Signaling in the Pathogenesis of SCA1**

Astrocytes are brain cells that are critical for the normal function of neurons. For example, astrocytes provide nutrients, promote neuronal survival, and maintain extracellular environment required for normal neuronal function. Calcium regulates most of these functions of astrocytes. Astrocytes undergo morphological and functional change early in SCA1, indicating that they may contribute to neuronal dysfunction. We have used mouse genetic approach to modulate astrocytic calcium in order to test how it contributes to disease. We have found that SCA1 mice, in which we have altered calcium in astrocytes, perform better on movement and balance test compared to SCA1 mice with unaltered astrocytic calcium. We are currently examining how is cerebellar pathology altered in these mice. These results indicate that modulating astrocytic calcium may delay onset of motor symptoms and ameliorate severity of SCA1.



**Vikram Khurana, MD, PhD**  
**Brigham and Women's**  
**Hospital and Harvard stem**  
**Cell Institute**  
**Boston, MA**

### Systematic edgotyping of ataxin proteins in cellular systems from yeast to patient neurons.

Spinocerebellar Ataxia type 7 (SCA-7) is an inherited neurodegenerative disorder in which CAG polyglutamine (polyQ) repeat expansions in the ataxin-7 (ATXN7) gene lead to retinal and cerebellar degeneration in affected human patients. The gold standard to study human disease such as SCA-7 is generating in vivo mouse models of the disease that capture the human disease pathology. Mouse models of the disease have shown dysfunction of the cell's powerhouse (mitochondria) in neurons in the brain, and photoreceptor (cells responsible for vision) loss in the retina. However, these models cannot always recapitulate the disease mechanism observed in

humans. As a result, human induced pluripotent stem (iPS) cells derived from patients suffering from the disease are becoming the new gold standard. Data from human in vitro SCA-7 iPS cell disease models to date have been limited. We have generated iPS cells from tissue samples of two SCA-7 (abnormal 42 polyQ) patients and an unaffected family member (normal 10 polyQ). Robust protocols to differentiate these cells into neurons and photoreceptors to model the disease in vitro have been established in the lab successfully through funds provided by the current grant. A CRISPR/Cas9 gene editing approach has also been developed to correct the pathogenic 42 polyQ expansion in the iPS cell lines from the patients. Since our SCA-7 patient lines have a 42 polyQ expansion and poly Q expansions of up to 35 repeats are considered normal, it is likely that the in vitro pathological phenotypic differences between control and SCA-7 will be subtle. As a result, we have used the same genetic engineering approach to introduce a 113 polyQ expansion to exacerbate the pathological differences in vitro and be able to study the disease mechanism with ease. This genetic engineering approach has been piloted in a simpler Parkinson's disease iPS cell line in the lab and it has been successful. We are looking to correct the ATXN7 polyQ expansions by the end of the year. We have in addition made progress on other aspect of modelling the disease. It is understood that the pathological polyQ expansions can lead to the aggregation of the ATXN7 protein and block its natural functions, leading to SCA-7. Through this granting mechanism, we have built on our previous published work with the Parkinson's protein alpha-synuclein to devise an approach to map the differences in interactions of normal and abnormal polyQ-expanded ATXN7 protein in living neurons. In sum, this grant has allowed us to build critical tools to better understand the biology of SCA7 in human patient-derived cellular models.

## **PIONEER SCA** **TRANSLATIONAL** **RESEARCH GRANTS**



**Harry Orr, PhD**  
**University of Minnesota**  
**Minneapolis, MN**

**Towards an ASO Therapy  
for Spinocerebellar Ataxia  
Type 1**

Funds were used to complete the pre-clinical investigation of whether an antisense oligonucleotide (ASO) targeting mouse Ataxin-1 in *Atxn1154Q/2Q* knockin mice that manifest with motor deficits and premature lethality. Following a single ASO treatment at 5 weeks of age, mice demonstrated rescue of the disease-associated phenotypes. In addition, RNA-seq on vehicle-treated *Atxn1154Q/2Q* and ASO-treated *Atxn1154Q/2Q* mice were used to demonstrate molecular differences between SCA1 pathogenesis in the cerebellum that underlies Ataxia with disease in the medulla associated with lethality. Using high field MRS, we will evaluate the use of cerebellar and brainstem MRS as biomarkers of therapeutic effectiveness. Select neurochemical abnormalities detected by MRS in vehicle-treated *Atxn1154Q/2Q* were reversed fully or partially in the cerebellum and brainstem (a volume covering the pons and the medulla) of ASO-treated *Atxn1154Q/2Q* mice. Together, these findings support the efficacy and therapeutic importance of directly targeting ATXN1 expression as a strategy for treating both motor deficits and lethality in SCA1. The results of this work are being written-up for submission to *The Journal of Clinical Investigation*.

B) Funds were used to expedite completion of a study demonstrating that the ATXN1-CIC

interaction in vivo drives cerebellar toxicity in SCA1. Importantly, funds were used to obtain using iPSC-derived neurons from SCA1 patients in support of a pathogenic mechanism whereby gain-of-function of the ATXN1-CIC complex is the major driver of toxicity. These data were seminal to acceptance of the manuscript by the journal *Neuron*.



**Willeke M.C. van Roon-Mom, PhD**  
**Leiden University Medical Center**  
**Leiden, Netherlands**

**Advancing the Therapeutic  
Potential of Exon Skipping for Spinocerebellar  
Ataxia Type 3**

Spinocerebellar Ataxia type 3 (SCA3) is a disease of the brain caused by a mistake in the ATXN3 gene. The mistake consists of an abnormal expansion in three letters in the DNA code, namely a CAG repeat. This repeat is translated into a repeat in the ataxin-3 protein, and it is this faulty ataxin-3 protein that causes detrimental effects to brain cells. These effects cause cellular stress and loss of cells mainly in the part of the brain that is called cerebellum and the brainstem of SCA3 patients. Over time this leads to SCA3 symptoms. If the abnormally long repeat in the faulty ataxin-3 protein is removed, we could prevent damage to the brain cells.

Antisense oligonucleotide-based therapy  
In the research described here, we made use of antisense oligonucleotides (AONs) as a potential therapy for SCA3. AONs are a therapeutic tool that can be used to remove a specific region from a single protein. In the case of SCA3, we designed and tested AONs capable of removing the harmful repeat from the ataxin-3 protein. We have previously published promising results with

this strategy, where we showed that the AON treatment resulted in removal of the harmful repeat from the ataxin-3 protein in the brain of SCA3 mice. In this NAF pioneer research additional AONs were tested, to assure that we can proceed with the most optimal AON for potential future clinical application. Furthermore, we tested potential side effects of the AON on the ataxin-3 protein function.

#### Investigating ataxin-3 protein function

Removal of the harmful region from the ataxin-3 protein can prevent damage to the nerve cells but may inadvertently interfere with its function in the cell. A number of cellular functions and protein interactions have been described for the ataxin-3 protein. It is known that ataxin-3 can bind and cleave so called ubiquitin chains. These are important protein markers that are used to degrade proteins within cells and are important for the health of the cells. Using artificially produced ataxin-3 proteins, either containing or lacking the harmful repeat, we established that removal of the harmful region did not interfere with the ubiquitin binding or cleavage function of ataxin-3. Secondly, ataxin-3 is known to interact with a protein called VCP. We were able to experimentally prove that

removal of the harmful region from ataxin-3 does not interfere with this interaction. Together, these experiments provide good evidence that AON treatment does not interfere with these aspects of ataxin-3 protein function, suggesting a good safety profile of the therapeutic strategy.

#### Testing antisense oligonucleotides

We tested 18 AONs targeting ataxin-3. The AONs were first tested in patient derived fibroblast cells, and the AONs capable of inducing the most efficient effect on ataxin-3 were then further tested in SCA3 mice. To this end, 6 AONs were injected in the brain of SCA3 mice. Brain tissue of these mice was investigated 1 week later for molecular analysis. We were able to establish that the AONs indeed removed the toxic region from the ataxin-3 protein in the mouse brain. Hence, we have successfully identified AONs to use for further development. Currently planned experiments will continue to test the AONs in the SCA3 mouse, to see if the onset of ataxic symptoms can be prevented with AON treatment. These experiments should provide the last proof of principle for further clinical development of this treatment strategy.



**JOHNS HOPKINS**  
M E D I C I N E

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**Call (410) 502-4664** to learn more and see if you qualify. Confidential.



## International Collaboration Launches

SCA Global is a consortium of Ataxia researchers from around the world who will collaborate on clinical research on the Spinocerebellar Ataxias (SCAs). NAF will co-facilitate and co-manage the SCA Global International Conference that is being organized by Dr. Ashizawa and Dr. Klockgether. This first ever conference will assemble an international roster of scientific and clinical investigators to address the challenges of clinical research and therapeutic trials in SCAs. The meeting will be held Wednesday March 27, 2019 - Friday March 29, 2019 at the Flamingo Hotel in Las Vegas, NV, immediately preceding NAF's 2019 Annual Ataxia Conference. The Conference will focus on the most recent scientific advances, translational approaches and clinical trials, with the following primary goals:

- Refine our understanding of clinical and genetic characteristics of SCAs in different

- racial, ethnic, social and regional populations;
- Define and agree on common standards for clinical assessment, brain imaging and biosampling;
- Facilitate development of robust clinical trials in SCAs;
- Establish future leaders of ataxia research by facilitating the involvement of young investigators;
- Bring trainees into contact with SCA patients and their families.

This is an exciting new initiative that will facilitate international collaboration among investigators and pave the path for successful clinical trial in these disorders. For more information, contact Sue Hagen at [susan@ataxia.org](mailto:susan@ataxia.org).

### Participants Needed for Research in Spinocerebellar Ataxia!

**Do you have Genetic Confirmation of Spinocerebellar Ataxia (SCA)?  
Are you between the ages of 18 and 45?**

As a participant in this study, you would be asked to complete one survey. Your participation would involve approximately 15 minutes of your time.

For more information about this study or to volunteer for this study please go to [https://www.surveymonkey.com/r/SCA\\_survey](https://www.surveymonkey.com/r/SCA_survey) or contact Suzy Cahn at [scahn@emory.edu](mailto:scahn@emory.edu) or 404-778-8536

# 2,019 by 2019

## Power in Numbers

CoRDS is a centralized international patient registry for all rare diseases; it is based at Sanford Research. The goal of the CoRDS registry is to connect as many patients and researchers as possible to help advance treatments and cures for rare diseases. The CoRDS registry is free for patients to enroll and for researchers to access.

Since 2013, NAF has partnered with CoRDS to enroll participants who have a diagnosis of Ataxia or are at-risk for Ataxia. Since that time, several researchers have accessed the Ataxia registry to help recruit research participants for their important studies. NAF has a goal to have 2,019 people enrolled in the Ataxia registry by 2019.

If you have not enrolled yet, you can register today to help meet the “2,019 by 2019” goal to help researchers find more answers to Ataxia and to participate in research studies and clinical trials.



*Enroll at [www.sanfordresearch.org/SpecialPrograms/cords](http://www.sanfordresearch.org/SpecialPrograms/cords)*

**Questions? Contact CoRDS at**  
**[cords@sanfordhealth.org](mailto:cords@sanfordhealth.org) or (877)658-9192**

### REMEMBERING NAF IN YOUR WILL

Throughout the years, individuals have named the National Ataxia Foundation as a beneficiary in their wills. Their thoughtfulness and foresight has enabled NAF to provide more research studies, more services to patients and families and more education and Ataxia awareness to the public. We are grateful for the impact that has been made by these compassionate acts. If this is something you would like to consider, please contact Joel Sutherland at [joel@ataxia.org](mailto:joel@ataxia.org) or call (763) 553-2748.

### THE ATAXIA COMMUNITY IS INTERESTED IN YOUR GREAT IDEAS.

If you have Ataxia Tips or a personal story you would like to share in a future issue of Generations, please submit it to [naf@ataxia.org](mailto:naf@ataxia.org). Those submitting a personal story are asked to please include a photo or two and a brief author bio (1-2 sentences).



# READISCA Sites have Launched

As previously reported, the National Institutes of Health granted funding for 5 years to support clinical trial readiness at 17 sites across the United States and two sites in Europe. The US sites have completed their regulatory requirements and have begun administering natural history studies on participants with early or presymptomatic stages of SCA 1 and SCA 3. Also included in the grant will be MRI studies at four of the US sites. This is a major initiative that will provide industry partners with the data they will need in the future to measure whether a treatment or drug is effective to stop or slow down the progression of the SCAs.

## MAIN GOALS OF THIS STUDY:

- To establish the world's largest group of early stage and symptomless SCA1 and SCA3 individuals
- To validate imaging signs in early stage and symptomless SCA1 and SCA3 individuals
- To adapt recent findings to design clinic trials for spinocerebellar ataxias

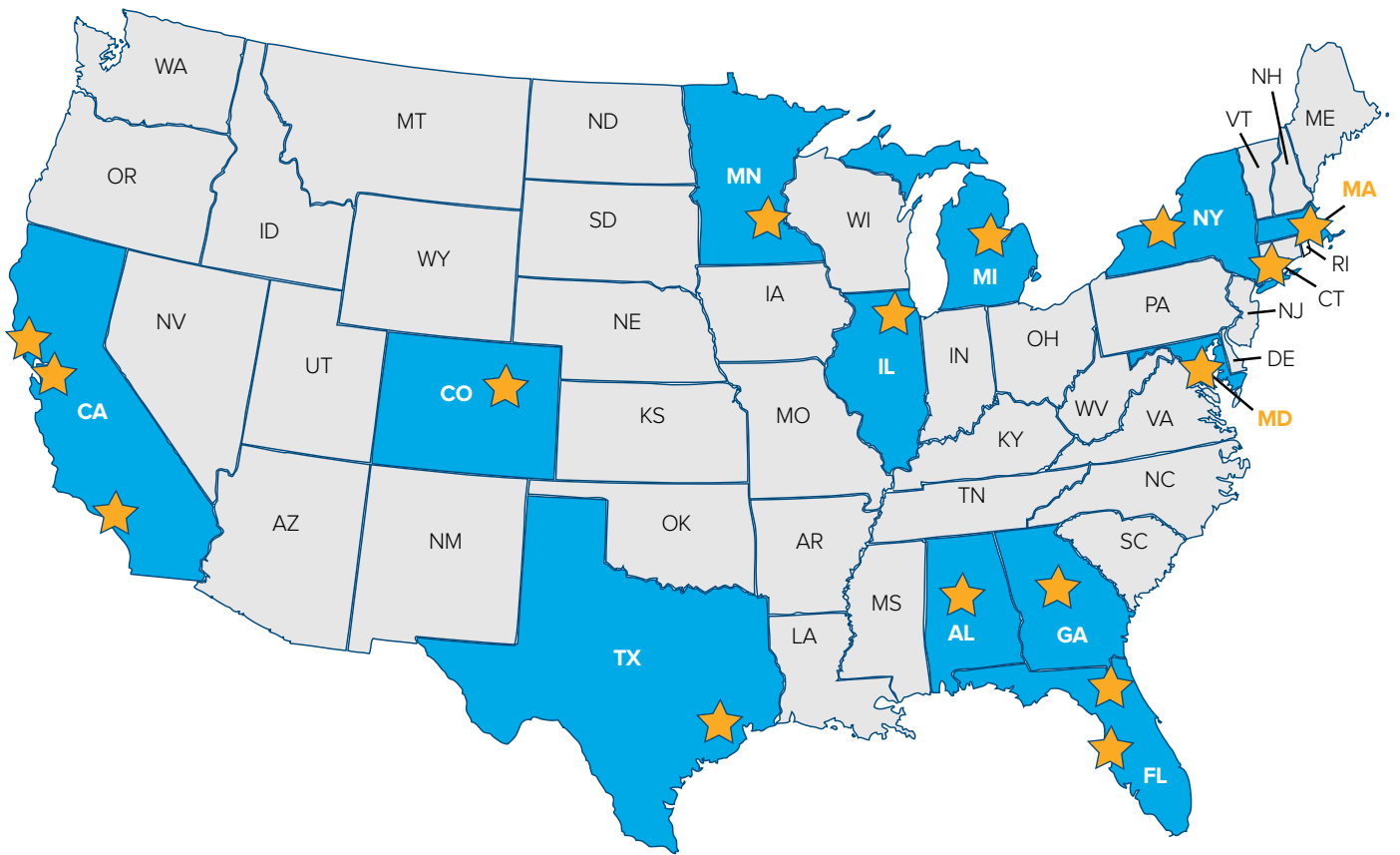
For more information on READISCA contact:

**Houston Methodist Research Institute**

Tetsuo Ashizawa, MD—Contact PI/PD

Phone: 346-238-5021 • Email:U01SCA1&3@houstonmethodist.org





**Columbia University**  
New York, NY

**Emory University**  
Atlanta, GA

**Houston Methodist Hospital**  
Houston, TX

**Johns Hopkins University**  
Baltimore, MD

**Mass General Hospital  
(Harvard)**  
Boston, MA

**Northwestern University**  
Chicago, IL

**Stanford University**  
Palo Alto, CA

**Univ. of Alabama—  
Birmingham**  
Birmingham, AL

**Univ. of California—Los  
Angeles**  
Los Angeles, CA

**Univ. of California—San  
Francisco**  
San Francisco, CA

**Univ. of Chicago**  
Chicago, IL

**Univ. of Colorado—Denver**  
Denver, CO

**Univ. of Florida**  
Gainesville, FL

**Univ. of Michigan**  
Ann Arbor, MI

**Univ. of Minnesota**  
Minneapolis, MN

**Univ. of Rochester**  
Rochester, NY

**Univ. of South Florida**  
Tampa, FL

 Indicates location of  
Clinical Trial Site



**Last Year NAF Funded Over \$1,000,000 In Ataxia Research  
We Need Your Help To Do It Again!**

# 2018 RESEARCH DRIVE



**November 1st - December 31st**

- 🧬 \$200,000 pledge to match your donation**
- 🧬 Mail your donation using the included envelope or donate online starting November 1st**

**w w w . a t a x i a . o r g**

# Ataxia Researchers Agree We Must Move Forward as a Global Effort

Excitement amongst Ataxia experts was tangible at the 2018 Katie Campbell Clinical Trial Readiness Conference hosted by NAF that took place August 13-14. "I'm personally enthusiastic," said Dr. Henry Paulson, meeting facilitator and Ataxia researcher at University of



CRC-SCA Members

Michigan Medical Center, "We're in a position to make a difference." The entire room of researchers, clinicians, and pharmaceutical company representatives seemed to share the sentiment that it is a promising time for Ataxia research. There have been major advances in Ataxia science and treatment trials are coming down the pike. The conference brought together members of the Clinical Research Consortium for the Study of Cerebellar Ataxia (CRC-SCA). Approximately 45 researchers, clinicians, and clinic coordinators spent two days working together to prepare for big things coming with treatment development for Ataxia. Four pharmaceutical companies joined the conference as well, presenting on their current efforts in Ataxia.

## What Is CRC-SCA?

CRC-SCA began as a group of Ataxia researchers from around the country recognizing the need to coordinate to prepare for potential clinical trials in the future. With seed grant funding from NAF, CRC-SCA was formed. Success of a clinical trial relies on standardized measurements to prove efficacy of a treatment and an ability to locate enough research participants to conduct a clinical trial, both issues that we addressed by the CRC-SCA. The group designed a natural history study which documents the natural progression of Ataxia by following patients over a long period of time. Data from this study will become crucial to obtain grants for Ataxia research and as more medications to treat Ataxia reach the clinical trial pipeline.

## Clinical Trial Readiness Conference Highlights

Group discussions were dynamic and covered many topics over the two-day span. Researchers shared

their newest scientific findings and brainstormed together for new studies to pursue. National Institute of Neurological Disorders and Stroke (NINDS) had a representative attend the conference to provide information about clinical trials that involve human subjects. That insight

helps the CRC-SCA design data sets that will be most likely to satisfy NINDS and FDA requirements for clinical trials. Much of the conference involved in-depth science discussions.

Main takeaways from the meeting:

- The Ataxia field is evolving rapidly - we must move forward as a global effort to achieve success faster.
- READISCA is the first large-scale grant that was received from NIH in part because of the work done by the CRC-SCA.
- Patient reported outcomes are essential when the time comes to seek FDA approval for a treatment - it is time to prepare for that.
- More scientific findings are expected by next year's CRC-SCA conference.
- Pharmaceutical industry is hoping for more clinical trial ready sites in the US.
- CRC-SCA will add at least one more clinical trial ready site this year.
- CRC-SCA must begin to plan for having multiple concurrent clinical trials, because it will happen soon.

Overall, the conference made clear that we are no longer in a place to hope for advances in science - we're already there. Now is the time to prepare for action to be taken from those findings. Clinical trials are coming - and they are coming soon. The Ataxia Patient Registry will be a main way for researchers to find participants for future studies. Make sure you're signed up!

Enroll at [www.sanfordresearch.org/cords](http://www.sanfordresearch.org/cords)  
Call or email for questions: (877) 658-9192  
or [cords@sanfordhealth.org](mailto:cords@sanfordhealth.org)

# ATAXIA: A TREATABLE DISEASE

62nd Annual Ataxia Conference  
March 29-30, 2019

Keynote Speaker - **Susan Perlman, MD**



“In the past 50 years, Ataxia has moved from just a symptom to a group of identified genetic and non-genetic disorders with mechanisms that are now understood. In the past 10 years, more and more pharmaceutical companies are finding avenues of research and development that lead to medications that could relieve symptoms and slow down progression. In the next year we could have the first FDA approved drug for Ataxia. Never let anyone tell you that there’s nothing that can be done for Ataxia.”

# Flamingo Hotel and Casino Las Vegas, NV

Conference registration and hotel room  
reservations open November 13, 2018

(Early Registration Discounts Available)

## Conference Highlights

- Ataxia Marketplace
- Research Participation  
Opportunities Onsite
- Ataxia Panel Sessions
- SCA Global - *Lunch and Learn  
Session*
- Birds of a Feather Small Group  
Sessions
- Networking and Social  
Opportunities



**NAF**  
National Ataxia  
Foundation

[www.ataxia.org](http://www.ataxia.org)



# Planning Your Travel to the 2019 AAC

We can't wait to see you at the 62nd Annual Ataxia Conference! Get ready to have fun and be inspired as top Ataxia researchers tell you about new clinical trials in the pipeline. More details on the conference schedule will be available in the Winter issue of *Generations*. Until then, here's the information you'll need to plan your trip:

## Conference Registration

Register online or by mail beginning on November 13, 2018. Registration forms will also be in the Winter issue of *Generations*. **Register before February 1, 2019 for the early registration discount!** Pre-registration closes on March 23, 2019 - but you can still register onsite.

## Registration Cost

<b>Before February 1, 2019</b>	<b>February 1 to March 23, 2019</b>	<b>Onsite</b>
NAF Member - \$125	NAF Member - \$150	NAF Member - \$200
Non-Member - \$180	Non-Member - \$205	Non-Member - \$255

Registration cost covers: All 2019 AAC general sessions, Birds of a Feather sessions, Lunch and Learn sessions, Ataxia Marketplace, Friday night reception, and Saturday night banquet.

## Hotel Reservations

Join us at The Flamingo Hotel and Casino at 3555 Las Vegas Blvd. S, Las Vegas, NV 89109 - don't forget to use our group rate discount! Group rates will be available until March 8, 2019 or until all rooms are booked. **Reservations open on November 13th at 12pm central time.** Standard rooms may be reserved directly through the hotel after November 13, 2018 by calling 1-800-732-2111 and requesting the NAF group rate (extra \$15 fee for phone reservations) or accessing the reservation link on NAF's website (no additional cost). NAF group rates for standard and ADA Rooms are:

### **Sunday - Thursday (per night\*)**

\$137 plus tax\*\*

### **Friday and Saturday (per night\*\*)**

\$177 plus tax \*\*

\*Rate includes \$18 resort fee \*\*Tax rate is 13.38%

## ADA Room Reservations

Need an ADA room? Reservations for ADA rooms are only offered through NAF, and will be offered on a first-come, first-served basis. Requests will be accepted beginning at 12pm central time on November 13, 2018. Call NAF at 763-553-0020 or email [lori@ataxia.org](mailto:lori@ataxia.org) with your ADA room request. The width of the bathroom door in the standard sleeping rooms is 26". NAF is unable to provide ADA equipment. Please make arrangements to rent locally or bring equipment with you.

## Travel Grant Program

Need some help with your travel costs? NAF, with the help of several generous donors, is proud to offer the Travel Grant Program to help with a portion of your travel costs to attend AAC. Adults or children with Ataxia are eligible to apply. Individuals interested in receiving a Travel Grant must apply by January 6, 2019. Applicants will be notified of the status of their application once applications have been reviewed after the deadline. If you would like an application mailed to you, contact Lori Shogren at 763-553-2743 or [lori@ataxia.org](mailto:lori@ataxia.org). Access the application online at <https://ataxia.org/annual-ataxia-conference/>

# Ataxia Tips

## for Ataxians... from Ataxians

For many with Ataxia, everyday tasks can become increasingly difficult. One person's shared tip might just be the help someone needs. See what advice Ataxians are giving to make everyday tasks easier...

- 1** I use a wooden rolling pin to “roll” out leg cramps. It works well on my thighs. (Submitted by Linda Meier)
- 2** I used to spill my morning coffee every day when walking from the coffee maker to the table in my kitchen. That was before I got a Spillnot mug carrier. I learned about this simple but scientific device at the National Conference in Philadelphia this year. It is available for less than \$20 on Amazon.com. I use it every day and it prevents liquid sloshing around and ultimately spilling which was a common occurrence with my ataxia. It has made a big difference in my daily routine. (Submitted by Jesse Diehl)
- 3** I very much recommend the My Cane, pivoting quad base cane, because of its lightness, stability standing alone, light quality... It has saved me many, many times. I found it as highly recommended on Amazon. (Submitted by Gérard D)
- 4** I have discovered that hiking poles help give balance when walking. They are available at most exercise, sporting goods, or camping stores. Although I haven't tried it, the poles are probably available online too. Hiking poles enable you to still enjoy many trails! (Submitted by Melissa Ackley)

**Ataxia Tips must be submitted by 11/5/18 to be eligible for inclusion in the next issue of *Generations*. Submit them via email to [naf@ataxia.org](mailto:naf@ataxia.org).**

## BRAIN TISSUE DONATION PROGRAM

Ataxia researchers have made many discoveries because of donations of brain tissue from those affected with Ataxia. One researcher said the following about brain donation, “This tissue is very precious.” The National Ataxia Foundation’s Brain Donation Program was established to allow those who desire to donate their brain upon death so that researchers can find more answers.

If you are interested in learning more about brain donation, you may contact Sue Hagen, NAF Patient and Research Services Director, at [susan@ataxia.org](mailto:susan@ataxia.org) or (763) 231-2742.

# How to Cross a Threshold

Jay Armstrong

*If you are distressed by anything external, the pain is not due to the thing itself, but to your estimate of it; and this you have the power to revoke at any moment.* -Marcus Aurelius

I saw my neurologist today.

After reviewing a recent MRI of my brain, he informed me that the deterioration that plagued my cerebellum appears to have stopped.

"That can happen?"

"Yes. In some cases, brain atrophy can stop."

"Well, I guess that's good news."

He flashed a smile, leaned back in his chair and said, "That's great news. Four years later...your brain is showing signs of stability."

Like every previous visit, my neurologist put me through a series of tests.

Follow his finger with my eyes. Touch my nose, touch his finger. Open my mouth, stick out my tongue, cluck my tongue. Snap my fingers. Clack my heels on the floor. Stand up, sit down.

He opened the examination room door, turned, "you know the drill," and I stood up and followed him out into the hallway.

I walked to the end of the hall, arms by my side, made a controlled turn—as if vying for my driver's permit— and walked back to him.

"Your gait looks good. You're walking more confidently than you have in years."

"Thanks."

We moved back into his office and sat down. He picked up a microphone that was corded to his computer and began dictating the results of my tests. *Despite extensive cerebellum damage, the patient's gait has shown improvement...*

I commented how when I first meet him, four years ago, he had to scribble down test results and appointment notes by hand.

He smiled, "Yes, this will definitely stave off carpal tunnel for a few more years. But to be honest, I miss the old-fashioned thrill of physical note-taking. But...

things change. Do you have any other questions?"

"I do. This may sound weird...I get a little uneasy around thresholds and doorways. You know, like I'm afraid to transition or something. Is it normal for people with cerebellar damage to have trouble crossing thresholds?"

He leaned back into his seat and crossed his legs, "The brain is a wonderful mystery. Even a healthy brain can find thresholds problematic. It's something primitive. Like the fear the primitive man must have felt while standing barefoot on some rocky ledge, looking for someplace to go. Crossing from room to room, from one plane to the next has always troubled people. Evolution has ingrained it in our psyche. We're simply afraid of transitions."

Of course, it wasn't intentional, but he just conducted an unauthorized, in-office autopsy on my life.

"Do you have any advice on how to cross a threshold?"

"Crossing a threshold is often mental. The initial fear of just transitioning from one place to the next often prevents us from progression. But when you find the nerve to finally cross, you realize there was nothing to fear at all."

I stood up, shook his hand, said I was looking forward to seeing him in six months. He smiled, spun away, opened the door and disappeared.

I slipped on my coat and strode through the threshold, from the examination room into the hall and back into life.

A life born of thresholds, waiting patiently for us to simply brave up and cross.

**Jay Armstrong** is a writer, Ataxian, and motivational patient advocate. Jay uses the power of writing, speaking, and storytelling to motivate and inspire others. See what Jay is up to and read more of his writings at [writeonfighton.org](http://writeonfighton.org). You can contact Jay at [writeonfighton@gmail.com](mailto:writeonfighton@gmail.com).





## From Riding a Tricycle to a Harley Davidson to a Catrike

Mary Fuchs

Ataxia is not who I am, it is what I have. I was diagnosed with Hereditary Friedreich's Ataxia (FRDA) in August 1998. But that is not where my story begins.

I was born in a small Michigan German farming community in 1949, was the third of four children, had a normal childhood (but clumsy). After high school, I had a career as a Licensed Practical Nurse (LPN) and ultimately owned a home health care business "Home Sweet Home Services". Married three times (third time a charm), have 3 wonderful kids, 3 grandkids, & 3 step-grandkids. I am now married to Frank, my soulmate & caregiver - when I let him. We married in May 1998, before I knew I had Ataxia. We got married on a Harley Davidson, rode it out of the church side saddle, dress and all. BEST DAY EVER. We continued living in Michigan, but after the FA diagnosis, Frank and I moved to the Phoenix, Arizona area in April of 2002, to get away from cold and snow (2nd best thing I did).

If you cannot tell yet, I have lot of adventure or WILD in me. We try to do at least one out of the ordinary thing each year, we call them "Fat Moments". Examples: zip lining in a Costa Rica in 2004, a 14 day Australian cruise that included horseback riding in 2007, a 21 day white water rafting called "Colorado River Rampage Adventure" in 2008 (this was with a handicap adventure group), and Hawaii for 10 days in 2013, (I was able to get into the ocean, easy for most but not if you are in a wheel chair.) My next desire or item on my "bucket list" is to sky dive or parasail.

Back to my FA. I had strange neurological issues for a few years before being tested (e.g. gait or the airplane walk, slurred speech, balance, weak muscles). In 1997, a year before my FDRA diagnosis. I had a mild head injury from an auto accident. So, when my balance and gait got worse,

I thought it was from that auto accident. But no such luck. I had a very sharp neurologist at Michigan University Hospital in Ann Arbor, MI who tested me for Ataxia/FDRA, when genetic diagnosis was still in its infancy. The results indicated I had FDRA. My older sister, Charlotte also has late adult-onset FA. We have no clue where it came from but could blame it on one of our grandparents, who married as first cousins, which was common in the 1800's among ethnic groups. At least we have the adult late onset version and our repeat numbers are low, yeah! Diagnosis was FDRA, now what? No big deal at first, just a wiggle and wobble to my walk for the first 4-5 years. Then I began to fall and started using a walker, then I started to fall with the walker. I fractured my wrist 11-years ago and had to progress to a motorized wheel chair which was and still is safer for me. Safety is always first.

My story does not stop there! For me, it is now about the "Fight for a Cure" and "Ataxia Awareness". I found the AZ Ataxia Support group when we first moved here. I needed them, and they needed me. They were experiencing "burn out" and I had lots of energy, time, and new ideas

to give. I am now co-coordinator since August 2010. After these 20+ years of living with Ataxia. My priorities are diet, exercise, support, education, encouragement, safety, and faith which is huge for me. We cannot "stick our head in the sand" and hope it goes away. We all have gifts and talents. My message is: USE THEM. It's not about what

**"It's not about what you lost or cannot do; it is about doing things differently."**

you lost or cannot do; it is about doing things differently. so you have ataxia! You also have Muscular Dystrophy Association (MDA), National Ataxia Foundation (NAF), you have Facebook, chat rooms, you have support groups, and you have yearly conferences, YOU ARE NOT ALONE. In today's world of electronics and communication, it is so easy to connect. You can't and should not be alone, you need people.

I saved the best for last, I am not sure if you know about "RIDE ATAXIA" and Kyle Bryant. Through the Ataxian Athletic Incentive (AAI) grant

program, Ride Ataxia has facilitated the funding and purchase of adaptive cycling equipment for aspiring Ataxian Athletes, so they may start their personal Ride Ataxia adventure. I applied for an AAI grant last year, what a job, didn't get picked. I was so disappointed. Saw Kyle in Las Vegas, he said TRY again. So, I did, sent in ALL my 5 letters from last year, plus 5 more (only needed 2), with the application. I WAS PICKED, I am so excited!!!! Think when Kyle called, I may have hurt his ear drum with my scream for joy. Riding my Catrike is so cool, gives me New Energy (like I needed that), feel Normal (almost), I feel Free and Empowered.

I may or may not ride in a race. But I will ride it A LOT in my community, ATAXIA AWARENESS here I come. So, from a tricycle as a youth-to getting married and riding a Harley Davidson out of the church-back to riding an adaptive trike by Catrike. It's not about me or you it's about HELPING FIND A CURE.

---

**Mary Fuchs** is a wife, mother and grandmother who doesn't let Ataxia slow her down. If you are ever in the Phoenix area look out for her on her Catrike.

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## Using DIY to Feel in Control

*Julia Henrichs*

My three-year-old identical twin daughters have Ataxia and Nystagmus. One twin is slightly more impacted than the other and has "cerebellar vermian low volume" which shows up on her MRI. My husband and I are waiting on pins and needles for the genetic test results, which we should receive by the end of the month.

In the meantime, to help distract myself, I like to keep busy and work on projects with my girls or around the house. Of course, my DIY nature has come in handy when I have to sew Velcro extensions on my daughters' shoes, so she can fit in her Ankle Foot Orthotics while still being able to wear cute summer sandals to match her sister and her dress! I've also used DIY tricks to make a creative bench-like step all the way along the foot of the bed to help my daughter safety get in and out of her new big girl bed.

This last week, after one too many falls resulting in stitches, bumps, bruises, or fears of concussion, I decided to make foam toddler "concussion bands" for my girls out of repurposed items from around the house. I like my girls to take risks and push themselves physically but it's hard to see them get hurt. Making something myself was really my only options since I'm not going to have them wear a helmet all day and the concussion bands online are only made for youth size soccer players.

The girls had rented weighted vests that we had to return this summer when the girls turned three and graduated from Early Intervention. Since we are

temporarily without a PT until school starts in the fall and they were dropped from their PCP this past month when they qualified for a Medicaid Waiver, I didn't know who to call for a referral. I decided to go the DIY route and make



one myself. I sewed a series of pockets inside one of my daughter's vests and let my girls attempt to fill mini zip lock bags with black beans, which of course ended up scattered all over my carpet. Even if it did take an hour to clean up the mess, we had fun and stayed focused on our goal. With so much waiting, for doctors, equipment, test results, and other things outside our control, I've found that it helps

me to be able to take action towards small tasks that are within my control.

**"With so much waiting, for doctors, equipment, test results, and other things outside our control, I've found that it helps me to be able to take action towards small tasks that are within my control."**

---

**Julia Henrichs** is a former counselor who now spends her days driving her 5-year old and 3-year old identical twin girls to and from school, play dates, and appointments. She has a gift for taking what others see as junk and transforming it into a beautiful treasure and blogs about it at [projectsandparenting.com](http://projectsandparenting.com). She also coordinates a local MOPS group to help encourage other moms.



## *A clinical study designed for patients with Friedreich's ataxia*

A clinical study is now enrolling individual with Friedreich's ataxia. MOXIe is a Phase 2 clinical study evaluating the safety and effectiveness of omaveloxolone (an oral investigational drug) for the treatment of Friedreich's ataxia (FA). The study has two different parts.

### **MOXIe (Part 1) completed enrollment in February 2017. Key observations from MOXIe (Part 1) are:**

- Omaveloxolone significantly improved mFARS (modified FA Rating Scale) from baseline across all doses
- In omaveloxolone-treated patients, mFARS was improved at Week 4 and further improved by Week 12
- Omaveloxolone at 160 mg dose showed large mFARS improvements as early as Week 4
- Omaveloxolone was well-tolerated and adverse events were generally mild in severity

### **MOXIe (Part 2) is now enrolling.**

#### **You may be eligible for this study if you:**

- **Are 16 to 40 years of age**
- **Have been genetically diagnosed with Friedreich's ataxia**
- **Are willing to maintain a consistent exercise routine and stable medication doses throughout the study**
- **Are willing to discontinue taking all antioxidant supplements and vitamins, or any other medication intended to treat Friedreich's ataxia, before beginning this study and throughout your participation in the study**

#### **Other eligibility criteria must also be met.**

The investigational drug, study-related procedures, and doctor visits will be provided at no cost. If you travel to the site for your study visits, travel expenses will be reimbursed, and compensation for study-related time may be provided.

For more information or to see if qualify, contact.

#### **Reata Pharmaceuticals**

Hanh Nguyen

hanh.nguyen@reatapharma.com

(469) 442-4754



This study is being sponsored by Reata Pharmaceuticals. [www.clinicaltrials.gov](http://www.clinicaltrials.gov) (NCT02255435)  
ENG V1.0, Protocol Version 9.0 – 17August2017

# 2018

# SUPPORT GROUP DIRECTORY AND EVENTS BY STATE

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

## Why Attend an Ataxia Support/Social Group?

Support groups can remind us that we are not alone and that while each individual may experience Ataxia in a different way, together we have many things in common. A benefit of attending a support group is simply to have a chance to talk with others and learn how different people deal with the same disease.

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 loved one 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 of the NAF staff at [lori@ataxia.org](mailto:lori@ataxia.org) or (763) 231-2743.

Attending a support group meeting can leave you with a sense of hope and inspiration.

**Come. Learn. Share. But most of all, know that you are NOT alone.**

## ALABAMA

### Alabama Support Group Leader

Becky Donnelly-Hoover, AL  
(205) 987-2883  
E-mail: [donnely6132b@aol.com](mailto:donnely6132b@aol.com)  
Facebook Page: <https://www.facebook.com/alAtaxia/>  
Facebook Group: <https://www.facebook.com/groups/154027955194806/>

### Ambassador

Dianne Blain Williamson-Huntsville-AL  
(256) 429-9092 or (256) 520-4858  
E-mail: [diannebw@aol.com](mailto:diannebw@aol.com)

### AL Meetings and Events: Alabama Ataxia Support Group Meeting

October 27, 2018 • 10:00am - 2:00pm  
Covenant Presbyterian Church  
65 Old Montgomery Hwy  
Homewood, AL 35209

### Alabama Ataxia Support Group Christmas Social

December 1, 2018 • 11:30am  
B&A Warehouse  
1531 First Avenue South  
Birmingham, AL 35233

## ARIZONA

### Phoenix Area Support Group Leaders

Angela Li-Peoria, AZ  
(847) 505-4325  
E-mail: [angelali1010@gmail.com](mailto:angelali1010@gmail.com)

Mary Fuchs- Casa Grande, AZ  
(480) 212-6425  
E-mail: [maryr10101@gmail.com](mailto:maryr10101@gmail.com)  
Facebook Group:  
[www.facebook.com/groups/arizonaAtaxia/](http://www.facebook.com/groups/arizonaAtaxia/)  
SG Email: [ArizonaAtaxia@gmail.com](mailto:ArizonaAtaxia@gmail.com)

### Ambassador

Bart Beck-Tucson, AZ  
(520) 885-8326  
E-mail: [bbeck15@cox.net](mailto:bbeck15@cox.net)

## AZ Meetings and Events:

### Arizona Support Group Meeting

November 3, 2018 • 1:00 - 3:00pm  
Ability 360  
5025 E Washington St, Phoenix, AZ 85034

## ARKANSAS

### Ambassadors

Judy and David King-Springdale, AR  
E-mail: [davidrkingpd@cox.net](mailto:davidrkingpd@cox.net)

## CALIFORNIA

### Los Angeles Area Support Group Leaders

Lora Morn-Santa Monica, CA  
(310) 664-8808  
E-mail: [loramorn@gmail.com](mailto:loramorn@gmail.com)

Harvey Kahn-Whittier, CA  
(562) 789-5776  
E-mail: [jkhk@aol.com](mailto:jkhk@aol.com)

### Northern California Area Support Group Leaders

Brian Wong - Santa Clara, CA  
Fernando and Rocio Wu - Danville, CA  
S.G. E-mail: [info@norcalataxia.org](mailto:info@norcalataxia.org)  
Facebook Group: <https://www.facebook.com/groups/592006361008986/>  
Support Group Website: <http://norcalataxia.org>

### Sacramento Area Support Group Leader

Teresa Bredberg-Sacramento, CA  
(916) 215-2686  
E-mail: [tbredberg@sbcglobal.net](mailto:tbredberg@sbcglobal.net)  
S.G. Website: [norcalAtaxia.org](http://norcalAtaxia.org)  
Facebook Group:  
<https://www.facebook.com/groups/592006361008986/>  
Secondary Contact: Darrell Owens - Davis, CA  
Email: [droopydog36@hotmail.com](mailto:droopydog36@hotmail.com)

## Orange County Area Support Group Leaders

Cindy De Mint-Yorba Linda, CA  
(714) 970-1191  
E-mail: [cindyocAtaxia@gmail.com](mailto:cindyocAtaxia@gmail.com)

S.G. Website: <http://orangecountyAtaxia.org/>  
Facebook Group: <https://www.facebook.com/groups/1980393985511432>

## Ambassadors

Deborah Levi-Morro Bay, CA  
(805) 407-0437  
E-mail: [Debbielevi213@yahoo.com](mailto:Debbielevi213@yahoo.com)  
Deborah Omictin-Hayward, CA  
(510) 783-3190  
E-mail: [rsisbig@aol.com](mailto:rsisbig@aol.com)

Martha Elliott-Camarillo, CA  
(805) 987-2490  
E-mail: [DOCElliott268@gmail.com](mailto:DOCElliott268@gmail.com)

## CA Meetings and Events: Los Angeles Ataxia Support Group Meeting

November 17, 2018 • 2:00 - 4:00pm  
Disability Community Resource Center  
12901 Venice Blvd, Los Angeles, CA 90066

## Orange County Ataxia Support Group Meeting

October 20, 2018 • 2:00 - 4:00pm  
Orange Coast Memorial Hospital Medical Center  
18035 Brookhurst St, Fountain Valley, Ca 92708

## Orange County Ataxia Support Group Holiday Party

December 1, 2018 • 12:00 - 3:00pm  
Marie Callender's  
18889 Brookhurst St, Fountain Valley, CA 92708  
\*\$20 to be paid to Cindy De Mint before  
November 30, 2018

## Sacramento Ataxia Support Group Meeting

October 20, November 17, & December 15, 2018  
1:00 - 3:00pm  
UC Davis Medical Center Campus  
The Lawrence J. Ellison Ambulatory Care Center Bldg.  
4860 Y St, 3rd Floor - Conference Room 3010A  
Sacramento, CA 95817  
\*We meet on the 3rd Saturday of each month. Location/  
Room subject to change please contact Teresa to confirm  
location.

## Abilities Expo - San Mateo

October 26 - 28, 2018  
San Mateo County Event Center  
1346 Saratoga Dr, San Mateo, CA 94403  
For nearly 40 years, Abilities Expo has been the go-to  
source for the Community of people with disabilities,  
their families, seniors, veterans and healthcare  
professionals. Every event opens your eyes to new  
technologies, new possibilities, new solutions and new  
opportunities to change your life. For more information  
visit: [www.abilitiesexpos.com](http://www.abilitiesexpos.com)

## COLORADO

### Greater Denver Area Support Group Leader

Charlotte DePew-Aurora, CO  
(720) 379-6887  
E-mail: [cldepew77@comcast.net](mailto:cldepew77@comcast.net)

Facebook Group: [www.facebook.com/groups/denverAtaxia2011/](http://www.facebook.com/groups/denverAtaxia2011/)

## CO Meetings and Events: Denver Ataxia Support Group Meeting

October 20, 2018 • 1:00 - 4:00pm  
Swedish Medical Center  
501 E Hampden Ave, Englewood, CO 801133  
\*We meet in the 2nd floor Conference Rooms.

## CONNECTICUT

### Connecticut Support Group Leader

Susan Masse - Broad Brook, CT  
(860) 528-1875  
E-mail: [smasse1875@gmail.com](mailto:smasse1875@gmail.com)

### Tri-State Support Group Leader

Kathy Gingerelli-Parsippany, NJ  
(201) 681-7639  
E-mail: [kgingerelli@msn.com](mailto:kgingerelli@msn.com)

## CT Meetings and Events: Tri-State Ataxia Support Group Meeting

November 8, 2018 • 6:30 - 8:30pm  
Mount Sinai Beth Israel Medical Center  
Phillips Ambulatory Care Center  
Conference Room 3  
10 Union Square East  
New York, NY 10003

## DELAWARE

### Delaware Support Group Leaders

Joe & Cathy DeCrescenzo-Bear, DE  
(302) 369-9287  
E-mail: [jdecr26@verizon.net](mailto:jdecr26@verizon.net)  
E-mail: [cdecres@verizon.net](mailto:cdecres@verizon.net)

## FLORIDA

### South Eastern FL Support Group Leader

Jose Fernandez de Castro - Plantation, FL  
(954) 864-1436  
E-mail: [jfcmv@yahoo.com](mailto:jfcmv@yahoo.com)

### Tampa Bay Support Group Leader

Darlene Harris -Tampa, FL  
(813) 431-2859  
E-mail: [Msdee004@gmail.com](mailto:Msdee004@gmail.com)  
Facebook Group: <https://www.facebook.com/groups/204933669913963/>

### Treasure Coast Support Group Leader

Lisa Cole- Port St. Lucie, FL  
(772) 370-3041  
E-mail: [lisacoleAtaxia@gmail.com](mailto:lisacoleAtaxia@gmail.com)  
<https://www.floridatreasurecoastAtaxiasupportgroup.club/>  
Facebook Group: <https://www.facebook.com/groups/572410932959895/>

## Ambassadors

Meghan McBrearty - Tallahassee, FL  
(850) -524-9060  
E-mail: [megra10@hotmail.com](mailto:megra10@hotmail.com)

Dennis Hill - Deland, FL  
(407) 399-8332  
E-mail: [dennisrevdh@gmail.com](mailto:dennisrevdh@gmail.com)

### **FL Meetings and Events:**

#### **Tampa Bay Ataxia Support Group Meeting**

November 17, 2018 • 12:30 - 3:00pm  
University of South Florida - Morsani Center  
13330 Laurel Dr, Tampa, FL 33612

#### **Treasure Coast Ataxia Support Group Christmas Party**

December 1, 2018  
PSL Community Center  
2195 SE Airoso Blvd, Port St. Lucie, FL 34984

## **GEORGIA**

### **Greater Atlanta Support Group Leaders**

Dave Zilles-Atlanta, GA  
(678) 596-6751  
E-mail: [dzilles@earthlink.net](mailto:dzilles@earthlink.net)

Greg Rooks-Atlanta, GA  
(404) 822-7451  
E-mail: [rooksgj@yahoo.com](mailto:rooksgj@yahoo.com)

Lealan Sims -Hilton Head Island, SC  
(678) 234-6600  
E-mail: [Lealan@mac.com](mailto:Lealan@mac.com)  
S.G. E-Mail: [atlantaAtaxia@yahoo.com](mailto:atlantaAtaxia@yahoo.com)  
Facebook Group:  
[www.facebook.com/groups/317380459539/](http://www.facebook.com/groups/317380459539/)

### **GA Meetings and Events:**

#### **Greater Atlanta Ataxia Support Group Christmas Party**

December 1, 2018 • 1:00 - 3:00pm  
Rush Center Annex  
328 Mell Ave, Suite B, Atlanta, GA 30316  
\*Parking is free in Lots A, B, and C. Do not park on the street along Mell Ave.

## **ILLINOIS**

### **Chi-Town Friendship Group Leader**

Jonas Cepkauskas- Matteson, IL  
(708) 381-5555  
E-mail: [jonas@fightAtaxia.org](mailto:jonas@fightAtaxia.org)  
[www.fightAtaxia.org/Chitown](http://www.fightAtaxia.org/Chitown)

### **Chi-Town Metro Friendship Group Leader**

Christopher (Topher) Marsh-Chicago, IL  
(312) 217-7737  
E-mail: [512crockett@gmail.com](mailto:512crockett@gmail.com)

### **Ambassador**

Elaine Darte-Coffeen, IL  
(618) 397-3259  
E-mail: [elainedarte@yahoo.com](mailto:elainedarte@yahoo.com)

### **IL Meetings and Events:**

#### **Chi-Town Ataxia Friendship Holiday Pot Luck**

November 18, 2018 • 8:00am - 5:00pm  
Good Samaritan Hospital  
3801 Highland Ave, Downers Grove, IL 60515

## **INDIANA**

### **Indiana Support Group Leader**

Cheryl (Cheri) Bearman-Hoagland, IN  
(260) 452-6231  
E-mail: [cheribearman@gmail.com](mailto:cheribearman@gmail.com)

### **IN Meetings and Events:**

#### **Indiana Ataxia Support Group Meeting**

November 10, 2018 • 11am - 2:00pm  
St. Vincent Fishers Hospital  
13861 Olio Rd, Fishers, IN 46037  
\*Join our support group for Ataxians and families. Bring something to share for lunch plus your own drink. Paper goods supplied. We will meet in Conference Room #A & B (Park & enter at entrance One - follow the hallway to the right). Please R.S.V.P to Amy Draves, (765)610-2866 or [amy4kids@msn.com](mailto:amy4kids@msn.com), Teresa Coccaro, (317)439-2515 or [tcoccaro12@gmail.com](mailto:tcoccaro12@gmail.com).

## **IOWA**

### **Ambassador**

Emily Medina-West Des Moines, IA  
(515) 727-8713  
E-mail: [emily061578@yahoo.com](mailto:emily061578@yahoo.com)  
Facebook Group:  
[www.facebook.com/groups/107944351294/](http://www.facebook.com/groups/107944351294/)

## **KANSAS**

### **Ambassador**

Jalean Retzlaff-Park City, KS  
(316) 303-2351  
E-Mail: [jlrtrolls@yahoo.com](mailto:jlrtrolls@yahoo.com)

### **KS Meetings and Events:**

#### **Kansas City Ataxia Support Group Meeting**

December 8, 2018 • 12:00 - 2:00pm  
KC Public Library - Trails West Branch  
11401 E 23rd St, Independence, MO 64052

## **KENTUCKY**

### **Ambassador**

Janice Johnson-Brownsville, KY  
(270) 597-3854

## **MAINE**

### **Maine Support Group Leaders**

Alan and Paula Nadeau-Belgrade, ME  
E-mail: [psn92871@roadrunner.com](mailto:psn92871@roadrunner.com)

## **MARYLAND**

### **Chesapeake Chapter President**

Carolyn Davis-Vienna, VA  
(703) 759-2008  
E-mail: [ccnafpres@gmail.com](mailto:ccnafpres@gmail.com)

### **Johns Hopkins Ataxia Center Support Group Leader**

Donna DeLeno Neuworth, Ataxia Clinic Health Educator  
(410) 616-2811  
Lutherville, MD  
E-Mail: [ddeleno1@jhmi.edu](mailto:ddeleno1@jhmi.edu)

## Ambassador

Karen DeVito-Frederick, MD  
(301) 682-5386  
E-mail: [karen.devito@yahoo.com](mailto:karen.devito@yahoo.com)

## MASSACHUSETTS

### Boston Area Support Group Leaders

Denise Mindle-South Dartmouth, MA  
(508) 369-7925

John Mauro-Auburn, MA  
(508) 736-6084  
Email: [john@ataxia.org](mailto:john@ataxia.org)  
S.G. E-mail: [ngataxia@outlook.com](mailto:ngataxia@outlook.com)

### Central MA Support Group Leaders

John and Dana Mauro-Auburn, MA  
(508) 736-6084  
Email: [john@ataxia.org](mailto:john@ataxia.org)  
Email: [danamauro63@msn.com](mailto:danamauro63@msn.com)  
Facebook Group:  
[www.facebook.com/ataxiadidyouknow?ref=hl](http://www.facebook.com/ataxiadidyouknow?ref=hl)

## MICHIGAN

### Detroit Area Support Group Leader

Tanya Marshall-Detroit, MI  
(313) 736-2827  
E-mail: [tinyt48221@gmail.com](mailto:tinyt48221@gmail.com)

### Western Michigan Support Group Leader

Lynn K. Ball-Grand Rapids, MI  
(269) 330-1975  
E-mail: [lynnkball@aol.com](mailto:lynnkball@aol.com)

## MINNESOTA

### Central MN Support Group Leader

Marsha Binnebose-St. Cloud, MN  
(320) 248-9851  
E-mail: [mbinnebose@hotmail.com](mailto:mbinnebose@hotmail.com)

### Twin Cities Ataxia Social Group Leaders

Lenore Healey Schultz-Minneapolis, MN  
(612) 724-3784  
E-mail: [schultz.lenore@yahoo.com](mailto:schultz.lenore@yahoo.com)

### Maryann Sweeney-Minneapolis, MN

(612) 924-4947  
E-mail: [maryann.sween@gmail.com](mailto:maryann.sween@gmail.com)

## Ambassadors

Julie Schuur-Luverne, MN  
(507) 283-2555  
E-mail: [jschuur@vastbb.net](mailto:jschuur@vastbb.net)

### Lori Goetzman-Rochester, MN

(507) 990-4506  
E-mail: [logoetz@gmail.com](mailto:logoetz@gmail.com)

## MN Meetings and Events: Central Minnesota Ataxia Support Group Meeting

November 10 & December 8, 2018 • 9:45 - 11:45am

## Harvest Bank

34952 Country Rd 7, St. Cloud, MN 56301

## Twin Cities Ataxia Social Group Meeting

October 20 & December 15, 2018 • 10:00am - 2:00pm  
Langton Place

1910 W. CTY RD D, Roseville, MN 55112

\*The Twin Cities Ataxia Support Group meets once a month. Family and friends of an afflicted individual are always welcome! We meet on the third Saturday of the month in a meeting room at Langton Place which is located on the south side of the road on County Road D roughly four tenths of a mile east of I35W in Roseville. We wanted to provide a central location that it easy to access which is why we picked this place. Please join us and make new connections!

## MISSISSIPPI

### Mississippi Chapter President

Camille Daglio-Hattiesburg, MS  
E-mail: [dagliot@bellsouth.net](mailto:dagliot@bellsouth.net)

## MISSOURI

### Kansas City Support Group Leaders

Stephanie Wilkins  
Lee's Summit, MO  
(816) 623-3318  
Cell: (949) 463-6102  
E-Mail: [swilkins@yahoo.com](mailto:swilkins@yahoo.com)

Laurie Colby  
Kansas City, MO  
(816) 429-6456  
Cell: (816) 745-4549  
[Lcolby61@gmail.com](mailto:Lcolby61@gmail.com)

### St. Louis Support Group Leader

Shannon Dunphy-Lazo - St. Louis, MO  
(202) 306-2738  
[Shan\\_d@hotmail.com](mailto:Shan_d@hotmail.com)

## Ambassador

Roger Cooley-Columbia, MO  
(573) 474-7232 before noon  
E-mail: [rogercooley@mediacombb.net](mailto:rogercooley@mediacombb.net)

## MO Meetings and Events: St. Louis Ataxia Support Group Meeting

December 8, 2018 • 11:00am - 1:00pm  
The Center for Advanced Medicine  
4921 Parkview Place, St. Louis, MO 63110  
\*We meet the second Saturday of every month on the 3rd Floor in Conference Room 1.

## Kansas City Ataxia Support Group Meeting

December 8, 2018 • 12:00 - 2:00pm  
KC Public Library - Trails West Branch  
11401 E 23rd St, Independence, MO 64052

## NEBRASKA

### Nebraska Ataxia Support Group Leader

Linda Snider-Omaha, NE  
(402) 212-3060  
E-mail: [lindasnider@cox.net](mailto:lindasnider@cox.net)

## NEVADA

### Ambassador

Debbie Grant-Las Vegas, NV  
(770) 885-3092  
E-mail: [grantgirl2004@aol.com](mailto:grantgirl2004@aol.com)  
Facebook Group: <https://www.facebook.com/groups/2090650814550409/>

## NEW HAMPSHIRE

### New Hampshire Support Group Leaders

Doug Place - Melrose, MA  
(781) 307-0800  
E-mail: [douglas.place@comcast.net](mailto:douglas.place@comcast.net)

Donna Gorzela - Billerica, MA  
(978) 490-9552  
E-mail: [donna.gorzela@gmail.com](mailto:donna.gorzela@gmail.com)

## NEW JERSEY

### Tri-State Support Group Leader

Kathy Gingerelli-Parsippany, NJ  
(201) 681-7639  
E-mail: [kgingerelli@msn.com](mailto:kgingerelli@msn.com)

### New Jersey Support Group Leader

Priya Mansukhani - Manville, NJ  
(908) 342-5675  
E-mail: [priyamans@gmail.com](mailto:priyamans@gmail.com)

### **NJ Meetings and Events:** **Tri-State Ataxia Support Group Meeting**

November 8, 2018 • 6:30-8:30  
Mount Sinai Beth Israel Medical Center  
Phillips Ambulatory Care Center  
Conference Room 3  
10 Union Square East, New York, NY 10003

## NEW YORK

### Tri-State Support Group Leader

Kathy Gingerelli-Parsippany, NJ  
(201) 681-7639  
E-mail: [kgingerelli@msn.com](mailto:kgingerelli@msn.com)

### Western New York Support Group Leader

Jesse Diehl - Spencerport, NY  
(585) 315-1578  
E-Mail: [jesse.diehl61@gmail.com](mailto:jesse.diehl61@gmail.com)

### **NY Meetings and Events:** **Tri-State Ataxia Support Group Meeting**

November 8, 2018 • 6:30-8:30  
Mount Sinai Beth Israel Medical Center  
Phillips Ambulatory Care Center  
Conference Room 3  
10 Union Square East, New York, NY 10003

## NORTH CAROLINA

### Tarheel Support Group Leaders

Ron and Donna Smith- Garner, NC  
(919) 779-0414  
E-mail: [rsmith@sacherokee.com](mailto:rsmith@sacherokee.com)

E-mail: [dsmith@sa-pr.com](mailto:dsmith@sa-pr.com)  
Facebook Group: <https://www.facebook.com/groups/539411309575761/>

### Western NC Support Group Leader

Jodie Kawa - Brevard, NC  
(828) 384-8414  
E-mail: [jodiekawa@comporium.net](mailto:jodiekawa@comporium.net)  
Facebook Group: <https://www.facebook.com/groups/919905244740316/>

### **NC Meetings and Events:** **Western North Carolina Ataxia Support Group Meeting**

October 26, November 30 & December 28, 2018  
12:30 - 3:30pm  
Foster SDA Church  
375 Hendersonville Rd, Asheville, NC 28803  
\*Foster 7th Day Adventist Church is right off Highway 40, so it is easy to get to. Please bring a covered dish and we will have lunch, listen to a speaker, and have confidential group time.

## OHIO

### Cincinnati Support Group Leaders

Julia Soriano-Cincinnati, OH  
(513) 899-1195  
E-mail: [julia@epivision.com](mailto:julia@epivision.com)  
Group Blog: [Ataxiafoundationcleveland.blogspot.com/](http://ataxiafoundationcleveland.blogspot.com/)

Willian Vetter Cincinnati, OH  
(513) 310-7530  
E-mail: [wjuniorvet@gmail.com](mailto:wjuniorvet@gmail.com)

### Cleveland Area Support Group Leader

Carmen Pieragastani - Willowick, OH  
(216) 272-5588  
E-mail: [willowpier@roadrunner.com](mailto:willowpier@roadrunner.com)  
Blog: <http://ataxiafoundationcleveland.blogspot.com/>

### **OH Meetings and Events:** **Cincinnati Area Ataxia Support Group Meeting**

October 21, November 18 & December 16, 2018  
2:00 - 4:00pm  
Sharp Turn Institute  
201 5th St, Suite 1901, Cincinnati, OH 45202  
\*We meet the third Sunday of each month at 2:00 p.m. Even numbered months will be support only meetings and odd numbered months will be programmed. Each meeting will include introductions and a show and tell time. All meetings are confidential, except for agendas and presenter materials.

The Sharp Turn Institute is an accessible building in downtown Cincinnati across the street from Government Square, the end point for many Cincinnati public bus routes and a transfer point for the TANK system serving Northern Kentucky. Parking is available on the street or in a parking structure attached to the building (fee).

## OKLAHOMA

### Central Oklahoma Ataxia Support Leader

Carrie Stanley  
(405) 387-9227  
E-mail: [cdstanley1977@gmail.com](mailto:cdstanley1977@gmail.com)

Facebook Group: <https://www.facebook.com/groups/421890291503549/permalink/428669084159003>

## OREGON

### Albany Support Group Leader

Jason Wolfer - Gervais, OR

Phone #: 503-502-2633

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

Facebook Group:

[www.facebook.com/groups/388993597939205/](http://www.facebook.com/groups/388993597939205/)

### Portland Support Group Leader

Tyler Kalina - Portland, OR

(541) 892-3519

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

Facebook Group:

[www.facebook.com/groups/388993597939205/](http://www.facebook.com/groups/388993597939205/)

## OR Meetings and Events:

### Portland Ataxia Support Group Meeting

October 21, 2018 • 3:00 - 4:30pm

Capitol Hill Library

10723 SW Capitol Hwy, Portland, OR 97219

## PENNSYLVANIA

### Central PA Support Group Leader

Michael Cammer- Downingtown, PA

(610) 873-1852

E-mail: [michael.cammer62@hotmail.com](mailto:michael.cammer62@hotmail.com)

Facebook Group: [www.facebook.com/groups/1475283086068548/](http://www.facebook.com/groups/1475283086068548/)

### Western PA Support Group Leaders

Ed Schwartz- McMurray, PA

(724) 986-8088

E-mail: [eds@Ataxia.org](mailto:eds@Ataxia.org)

Facebook Group: [www.facebook.com/wpaAtaxia.nafwesternpasupportchapter.weebly.com/](http://www.facebook.com/wpaAtaxia.nafwesternpasupportchapter.weebly.com/)

Donna Eiben - South Park, PA

(412) 655-4091

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

## RHODE ISLAND

### Rhode Island Support Group Leader

Anabela Azevedo - Bristol, RI

(401) 297-8627

E-mail: [azevedo70anabela@gmail.com](mailto:azevedo70anabela@gmail.com)

## SOUTH CAROLINA

### Ambassador

Brad Forth - Greenville, SC

(864) 415-8147

E-mail: [bradtf@photoforth.com](mailto:bradtf@photoforth.com)

## SOUTH DAKOTA

### Sioux Empire Support Group Leader

Mary Beth Farley - Sioux Falls, SD

(605) 941-2913

E-mail: [englishteacher1116@gmail.com](mailto:englishteacher1116@gmail.com)

Facebook Group: <https://m.facebook.com/groups/1606527972792690>

## SD Meetings and Events:

### Sioux Empire Ataxia Support Group Meeting

November 10 & December 8, 2018 • 3:00 - 5:00pm

Community Room at River Tower Apartments

111 E 7th St, Sioux Falls, SD 57104

## TENNESSEE

### Middle TN Area Support Group Leader

Alex Cohn - Nashville, TN

(256) 504-0240

E-mail: [alex.j.cohn@us.pwc.com](mailto:alex.j.cohn@us.pwc.com)

## TEXAS

### Greater Houston Area Support Group Leader

Dave Cantrell - Montgomery, TX

936-588-5179; Cell: 936-206-1504

E-mail: [dcant7358@aol.com](mailto:dcant7358@aol.com)

Facebook Group:

<https://www.facebook.com/groups/Ataxia.houston/>

### North Texas Area Support Group Leader

David Henry Jr. - Trophy Club, TX

(817) 73 9-2886 (contact by email preferred)

E-mail: [chevelle@sbcglobal.net](mailto:chevelle@sbcglobal.net)

Facebook Group: [www.facebook.com/Ataxiasupport](http://www.facebook.com/Ataxiasupport)

### Ambassadors

Dana LeBlanc - Orange, TX

(409) 883-5570

E-mail: [tilessal@yahoo.com](mailto:tilessal@yahoo.com)

Debra Whitcomb - El Paso, TX

(915) 329-0721

E-mail: [debrawhitcomb@hotmail.com](mailto:debrawhitcomb@hotmail.com)

## TX Meetings and Events:

### Greater Houston Ataxia Support Group Meeting

November 17, 2018 • 1:00 - 3:00pm

Methodist Hospital - Woodlands

17201 Interstate 45, The Woodlands, TX 77385

### North Texas Ataxia Support Group Meeting

November 10 & December 8, 2018

10:00am - 12:00pm

Ben Washington Baptist Church

Rev JR Sheppard Educational Center

615 Davis St, Irving, TX 75061

\* There is lots of parking and it is handicap accessible.

The meeting room is in a separate bldg. from the church.

## UTAH

### Utah Support Group Leaders

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### Ambassador

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## VIRTUAL SUPPORT GROUP

### UNDER 30 WITH ATAXIA

This support Group is exclusively for those who are ages 16-30 who have Ataxia. Parents/Spouses/Friends of people with Ataxia are not permitted to join this group.

### Support Group Leader

Lauren Sormani  
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[lasormani@gmail.com](mailto:lasormani@gmail.com)

Facebook Group: <https://www.facebook.com/groups/Under30withAtaxia/>

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## Memorials and In Your Honor

The National Ataxia Foundation is grateful to those who have made contributions in memory of or in honor of their friends and families whose names are listed below. This list reflects contributions made in July 2018 - August 2018.

Alma L. Vane	Danny Gunabe	Krista Humes	Neal Meakin
Angela Brown	Darin Dobson	Linda Anderson	Rita Dean
Ann Duke Smith	Denise van Voorhis	Linda Meier	Robert J. Schriefer
Aymee G. Torres, Dr. Aymee	Donnette Pretzer	Lynn E. Neuendorf	Santa Croce Family
A. Torres -Michels, Ricardo	Ellen Moetsch	Lynne Dutton	Schmidt 25th Wedding
Luis Guerrero & Teresita	Gay Coakley	Marguerite (Rita) Skerchak	Anniversary
Guerrero	George Ratica	Michael Hensley and the	Sharon Baggett
Beverly Mendoca	Gualberto “Ben” Honoridez	Kansas City Ataxia Support	Stephen Scott Griswold
Buddy W. Madden Jr.	Joanne L. Huebner	Group (KC Wobblers)	Susan Hochberg
Carlos Garzia	John Louis Weir	Michael Kampfer	Teri Rudin
Cheryl Martin	Joseph King Cox	Mrs. Barbara Engler	Virginia McAdams
Colleen Thomas	Josephine Merlino	Nathan J. Stackle	Thompson

# Natural History Study needs SCA Research Participants

The Clinical Research Consortium for the Study of Cerebellar Ataxia (CRC-SCA) continues to recruit research participants who have a confirmed diagnosis of SCA 1, 2, 3, 6, 7, 8 or 10. This is an opportunity for anyone in the United States with those forms of SCA at any stage of the disease to participate. Contact the research coordinator at a site near you to learn more about how you might be able to help in Ataxia research efforts to discover a treatment.

**The National Ataxia Foundation encourages anyone with SCAs 1, 2, 3, 4, 6, 7, 8 and 10 to participate.**

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## **Johns Hopkins University**

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This research is generously supported by the Gordon and Marilyn Macklin Foundation and the National Ataxia Foundation.

Thank you to Dr. Henry Paulson, University of Michigan, who has provided hours of counsel and leadership to make this a successful research endeavor. And to each of the sites clinical researchers and research coordinators who perform the research necessary to move the field closer to treatments and a cure.

**For more information on the study, you may contact Sue Hagen at [susan@ataxia.org](mailto:susan@ataxia.org) or 763-231-2742**

## **Disappointed that you don't qualify for this research study?**

There is an important step you can take so that in future studies for which you might qualify, you will be notified. And that step is to enroll in the CoRDS Ataxia Patient Registry.

If you are affected with any type of SCA or any other form of ataxia, enroll in the registry by going to the website: <https://cordsconnect.sanfordresearch.org/BayaPES/sf/screeningForm?id=SF5FL>. If you have questions about enrollment in the registry, contact the CoRDS staff at 877-658-9192.

# Support Group and Community Events News



## Alabama Ataxia Support Group

*Submitted by Becky Donnelly*

The Alabama Ataxia Support Group held its summer social in Cullman, Al, on Saturday, July 21st, with 23 in attendance. Elaine Brooks and Stephanie Culbreth kept the group entertained with games and door prizes. We welcomed two new members, Louise Joe of Vestavia Hills, Al, and Susie O'Brien of Florence, Al., and guest Joel Sutherland, Director of Development at NAF.

The group enjoyed delicious meals at Allsteak Restaurant capped off with their famous orange rolls as dessert. Afterwards, Becky reminded members of future events: the Christmas social at B@A Warehouse on December 8th.

Joel Sutherland then addressed the group with a few updates on research, encouraged members to be registered with the CoRDS Ataxia Patient Registry, Walk n' Roll, "Go the Extra Mile" in 2018, and emphasized the importance of revenue needed to fund important research. He answered many questions posed by the group and especially expressed his impression of our group... that we are a social, fun-loving people in spite of the difficulties of living with Ataxia. Joel, we hope to see you in Alabama again in the future...hope you enjoyed your visit.



## Arizona Ataxia Support Group

*Submitted by Angela Li*

We had a wonderful support group meeting this August. We chatted about the upcoming convention, CORDs database, Amazon smile, finding someone to help as a 3rd co-leader, and the possibility of organizing a "The Ataxian" viewing IAAD event in September. Dr. Paarth Shah joined us from Gilbert Neurology to answer our questions about Ataxia, medications, treatments, etc. Thank you, Dr. Shah, for sharing all your knowledge and expertise with us. After our meeting we went to a nearby restaurant for a social meal. Join us for our next meeting on November 3rd.



Arizona Ataxia Support Group



## Denver Ataxia Support Group

*Submitted by Charlotte DePew*

The Denver Support Group met July 21st. With a smaller group it promoted conversation and exchange of experiences, suggestions, etc. One new couple came, and they left with many helpful suggestions from the group. There was a report from the Run, Walk 'n Roll Committee that met at City Park in the morning. Sign-up sheets for volunteers, drawing donations (baskets) and Silent Auction donations were circulated as well as event flyers

Our speaker was Ashley Elsasser, a yoga instructor in Golden, CO. Ashley demonstrated moves and positions that can help improve balance and movement. Of course, she also had us try most activities, which we could do in our chairs. Ashley was an engaging speaker and earned many positive comments.



## South Eastern Florida Ataxia Support Group

*Submitted by Ellie Rockwell and Lisa Cole*

Three Florida Ataxia support groups were well represented in Orlando: the Tampa Bay Ataxia Support Group, (Miami) SE Florida Ataxia Support Group, and Treasure Coast Ataxia Support Group affiliates, 30 people were in attendance which included caregivers, mothers, fathers, husbands and wives. Our guest speaker was Nantesha Chen, she is a Movement Therapist from Dunedin, FL, she demonstrated simple exercises that she had

prepared for us. Her advice was to move, wake up your muscles, water is very good and listen to your body.



L to R: Darlene Harris, Jose Fernandez de Castro, Lisa Cole

people who want to make living with Ataxia easier for all of us. We are open to suggestions and ideas, and these can be funneled to Lisa Cole at [lisacoleataxia@gmail.com](mailto:lisacoleataxia@gmail.com).



Treasure Coast Support Group



3 Florida Support Groups meet together

## Tampa Bay Ataxia Support Group

*Submitted by Jannete Colon*

The Tampa Bay Ataxia Support group met on July 21, 2018 at the Morsani Center at USF.

Our guest speaker was Nantesha Chen, a Physical Therapist from Dunedin, Fl. She showed us how to connect our brain with the body with simple exercises and everybody participated.



Tampa Bay Support Group

## Treasure Coast Ataxia Support Group

*Submitted by Ellie Rockwell and Lisa Cole*

The meeting was held on August 18, at Glen and Myra Gardiner's house in Tradition, Port St. Lucie, Florida. Attending were 12 group members, including six new people introduced to the group. The reason for expanding our scope is that we are a wonderful set of caring and positive-thinking

## Ataxia Awareness Table at LA Fitness Port St. Lucie

*Submitted by Lisa Cole*

For the past 3 months Lisa Cole has been raising awareness for the Ataxia community and the National Ataxia Foundation at LA Fitness in Tradition, Port St Lucie. Every month LA Fitness has a member appreciation where they have vendors come in with information for their members to learn about new equipment, nutrition, and health. Lisa is happy to have the Ataxia awareness table, so she can share awareness with so many people, people who have never heard of the word or the disease, sharing the awareness is what she loves to do and why she does it.



Lisa Cole working hard raising awareness about Ataxia



## Greater Atlanta Ataxia Support Group

*Submitted by Greg Rooks*

The Greater Atlanta Ataxia Support Group held a meeting on August 18, 2018 at the Emory Brain Health Center in Atlanta. The meeting was well attended by 36 individuals. This was our first meeting at the Emory Brain Health Center and everyone loved the location. Attendees found the meeting

informative and a great time was had by all.

Our speaker was Dr. George (Chip) Wilmot. Chip updated the group on current research and clinical trials. We also discussed plans for the upcoming Atlanta Walk n' Roll to Cure Ataxia on September 22nd and International Ataxia Awareness Day on September 25th. Lastly, we discussed the importance of the CoRDS Registry and encouraged all to be registered.

After the meeting everyone socialized and enjoyed having refreshments. Fifteen individuals continued to enjoy each other's companionship by going out to eat at Grub Burger Bar.



Greater Atlanta Support Group



## Indiana Ataxia Support Group

*Submitted by Cheri Bearman*

The Indiana Ataxia Support Group held their bi-monthly meeting on July 14, 2018, at St. Vincent Hospital in Fishers, Indiana at 11:00 am. The meeting was attended by eleven members of the group. Two special guest speakers were featured at the meeting. Anne Komafel, Certified Tai Chi Instructor, presented a brief introduction to Tai Chi. She then performed a seated Tai Chi demonstration with group members participating. Tamara McCord, Licensed Mental Health Counselor from Imagine Hope Counseling, Indianapolis, gave a presentation about the grief people experience with "ambiguous loss", such as the various losses by those affected by Ataxia. Tamara discussed various coping mechanisms. After the guest speakers, a carry-in tossed salad lunch and socializing was enjoyed by all. The meeting adjourned at 1:30 pm.



Indiana Ataxia Support Group

## Hotdog Festival Fundraiser for Ataxia

*Submitted by Suzanne Spicer*

Frankfort Indian held their annual Frankfort Hotdog Festival on July 28th and this year NAF supporter and volunteer Suzanne Spicer staffed a table to bring Ataxia awareness to the event. There were raffle prizes, games and information about Ataxia. The event also held a 5k run which Suzanne participated in to raise funds for the National Ataxia Foundation through her Go the Extra Mile campaign. Thanks to Suzanne for raising awareness and funds for the Ataxia Community.



Suzann Spicer gearing up for the Bun Run



Volunteers working at the Frankfort Hot Dog Festival

## Hu-manifest

*Submitted by Amy Draves*

The third Humanifest was held on August 11, 2018. This event was put on by Playground Productions

in Indianapolis and have chosen NAF as the recipient of half of the proceeds. This event is an avenue for the Holistic community to share their products and services that are available. During the evening there were several bands who entertained the crowd. NAF hosted a table with lots of information given out. The majority of the people who visited our table were totally unaware of Ataxia. It was great to be able to shed some light to these people.



Attendees enjoying Crab Feast



Indiana Ataxia Support Group at Hu-manifest



### Crab Feast to Cure Ataxia in Maryland

*Submitted by Letitia Diggs*

A great time was had by all at the Crab Feast to Cure Ataxia, the event which was in memory of Vivessia Horton "Tootsie" Jones took place at the Boulevard Heights Volunteer Fire and Rescue Department. Attendees for this ticketed event were able to enjoy all you can eat crabs, fried chicken, pulled BBQ pork, corn on the cob, watermelon, Chesapeake chips much more. The evening also had a silent auction to raise funds and awareness for the National Ataxia Foundation.



Attendees enjoying their feast



### Twin Cities Ataxia Support Group

*Submitted by Lenore Healey Schultz*

Our group meets every third Saturday of the month in a centrally located, accessible and free meeting space for 2 hours. Our group has generally mapped out for the year what we will be doing at each meeting. Over the last few years we are having less guest speakers at our meetings, and more meetings that we now call "Living with Ataxia".

Living with Ataxia meeting starts with the entire group together. We do introductions & announcements. Our meeting room has a room divider that we have pulled out before the meeting begins. Once intros & announcements have concluded, the caregivers, family members, and/or friends associated with someone who has Ataxia take their chairs to the other side of the split room while those of us with Ataxia form a circle in the current room. A person to facilitate each group has been determined beforehand. After an hour, the 2 groups reunite. This type of meeting has become so popular, that it was decided that this year we do 6 "Living with Ataxia" meetings. Members always point out that they learn the most about coping with Ataxia from talking to one another.

We will have a meeting in October and November. We then end the year with a holiday party in December.



## Tri-State Ataxia Support Group

*Submitted by Kathleen Gingerelli*

July 12, 2018, Tonight's meeting was our Walk 'N' Roll "kickoff"

with a large turnout of everyone wanting details about our upcoming event. The night started off with everyone enjoying some food brought for the evening while reviewing the website for our walk and learning how to register/form a team and make donations. After answering all questions dealing with this year's walk, which helps with fundraising, building awareness & energizing our community, the topic then moved onto Living with Ataxia. Try to find the humor in everyday life. Don't take everything so seriously and try very hard to have a low embarrassment level. Ataxia is not your life.....your diagnosis does not mean you can't do things anymore, you just need to learn to do it differently. As hard as it is giving up things, move on and do it a new way. Exercise, mental activities and relationships are important and we should try to do different things. Don't get stuck in the same routine every day. Remember, we all handle things differently and it's important to share your story, get involved and encourage/learn from others. Before ending for the night, Dr. Kuo spoke and answered questions about Ataxia updates.



## Dart Tournament Ataxia Fundraiser

*Submitted by Christopher DeHaven*

The 501/Cricket tournament took place at Booger Reds in Midwest City, Oklahoma. \$1,000 was raised for the National Ataxia Foundation for half of the event entry fee was donated to NAF and then matched by Booger Reds. It was a fun and friendly tournament that not only raised funds but also awareness for the Ataxia community.



Dart Tournament competitors



## Greater Houston Area Ataxia Support Group

*Submitted by Dave Cantrell*

On July 21st we had our 4th meeting of the year. Met at Methodist Woodlands Hospital

and had a nice turnout of 16 people. Participation in the meeting was high with everyone engaging in various topics. We had a visitor from a Wellness Center. She talked about how her center is taking a Holistic approach with an Ataxian at her clinic and while it is extremely rigorous they are seeing some positive results. She promised to keep coming back with updates and wants to bring her client with her (shyness among Ataxians seems to be a trend). The issue of leg cramps came up along with Restless Leg Syndrome (RLS). A couple of home remedies came up as providing relief, mixed reactions from the group as one was a tablespoon of mustard (regular old French's) and pickle juice approximately a shot glass. This got some noses turned up but who cares if it works.

## Houston Abilities Expo

*Submitted by Dave Cantrell*

Houston Abilities Expo August 3-5 was a huge success for the Texas support groups. Huge thanks goes out to the David Henry family for coming down from Dallas (North Texas Support Group) and working the NAF booth on Saturday, Amy Cantrell and Rob Fruth manned the booth on Friday along with some interloper that wanted his picture taken with them, said his name was Joel Sutherland but I remember Joel being taller. On Sunday Eddie and Andrea Shannon manned the booth with Amy and we saw lots of action all three days. Twenty-two people signed up to receive e-mail from Amy regarding our Support Group Meetings in Houston and any information we have to pass along so that was awesome. Jim and Cynthia Barton stopped by on Friday and offered to lend a hand and as usual we just wound up talking and giving each other heck. Ellie Cammock stopped by on Sunday and visited about her work with persons with neurological disorders and how painting and drawing helps them to cope and relax. Great exhibits from all over the country were in attendance with many of the better ones being simple everyday items to help everyone along, from wrist straps that attach to eating utensils to

help steady the hand to carry bags that attach to a walker or wheelchair. Service animals were also big in attendance and who doesn't like dogs, they will be speaking at a Houston support group meeting next year. A local contractor, Whately & Whately, that specializes in "accessible" homes stopped by and what a great conversation on building and remodeling homes for the disabled we saw. New technology related to wheel chairs to make them easier to maneuver and safer to operate. A lady stopped by the booth at Eddie's request with a wheelchair that she moved with levers attached to the hubs that she pushed with her arms to make go, great for exercise and getting around. Met many people just wondering what Ataxia was and we took them all on to try and explain, after hearing their stories it was surprising how many people felt they had some of the symptoms or knew of someone that did. We immediately directed them to the NAF site and encouraged them to learn all they could. There are many different forms of Ataxia out there and we need to be cognizant of them all and recognize the struggle they all have. Looking forward to next year for newer and better equipment to help everyone.



NAF Volunteers at the Houston Abilities Expo getting the word out there about Ataxia!

# Join CoRDS

You hold a piece of the puzzle.



Enroll now at [sanfordresearch.org/CoRDS](https://sanfordresearch.org/CoRDS) by selecting **Enroll Now**.



## Stay up-to-date – Get on our email list

Email blasts from the National Ataxia Foundation are sent out periodically on Ataxia research, events and other timely issues of interest.

Please email your contact information to [naf@ataxia.org](mailto:naf@ataxia.org) so you don't miss out on important news.



# NAF Staff Directory and Social Networks

The use of these names and contact information for any purpose other than requesting information regarding the NAF or joining a chapter support group without NAF's written permission is strictly prohibited.

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## Social Networks



NAF Facebook Page

[www.facebook.com/ataxiafoundation/](http://www.facebook.com/ataxiafoundation/)

NAF Facebook Support Group

[www.facebook.com/groups/NAFmail](http://www.facebook.com/groups/NAFmail)

Under 30 with Ataxia

[www.facebook.com/groups/under30withataxia](http://www.facebook.com/groups/under30withataxia)



NAF YouTube Channel

[www.youtube.com/user/NatlAtaxiaFound](http://www.youtube.com/user/NatlAtaxiaFound)



NAF Twitter

[www.twitter.com/NAF\\_Ataxia](http://www.twitter.com/NAF_Ataxia)



NAF LinkedIn

[www.linkedin.com/company/nationalataxiafoundation](http://www.linkedin.com/company/nationalataxiafoundation)

## LOOK FOR FULL RECAPS OF THE 2018 WALK N' ROLL AND IAAD EVENTS IN THE WINTER ISSUE OF *GENERATIONS*!

Please send your IAAD and Walk n' Roll event recaps and photos to [mollie@ataxia.org](mailto:mollie@ataxia.org) by November 5, 2018



## THE DEADLINE FOR SUBMITTING MATERIALS

for the Winter issue of *Generations* is November 5. Please send articles, your personal story, recaps of Ataxia-related events, photos and reports to [naf@ataxia.org](mailto:naf@ataxia.org). Thank you.

## Gift – Honor – Memorial

A contribution given in memory of a friend or relative is a thoughtful and lasting tribute, as are gifts to honor your friends or family. A Gift Membership is a wonderful gift to a friend or relative for special occasions like birthdays, graduations, anniversaries, and holidays. NAF will acknowledge your gift without reference to the amount. Simply fill out this form and mail with your check or credit card information to the National Ataxia Foundation. Honor/Memorial envelopes are available free of charge by writing or calling NAF.

My contribution is:     In Memory  In Honor  Gift Membership

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## 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 - \$65

Individual - \$40 Household - \$60     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).

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Address \_\_\_\_\_

City/State/Zip \_\_\_\_\_

Phone \_\_\_\_\_

E-Mail \_\_\_\_\_

Yes, sign me up for NAF e-mails

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## PAYMENT INFORMATION

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

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**2018  
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**November 1 through  
December 31...  
read more  
on page 26**

