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National Ataxia Foundation

GENERATIONS

THE OFFICIAL PUBLICATION OF THE NATIONAL ATAXIA FOUNDATION

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Deadline to submit materials for the Summer issue of *Generations* is Monday, May 21.

Please direct correspondence to:

National Ataxia Foundation
Connecting Ataxia families, researchers, clinicians and the community

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Generations Schedule:

Issue	Spring	Summer	Fall	Winter
Mail Date	April	July	Octover	January

NAF Update

CHECK OUT PAGE 28 FOR MORE INFORMATION ABOUT THE GO THE EXTRA MILE CAMPAIGN

After more than 16 months in the role of Executive Director for the National Ataxia Foundation, Joel Sutherland is returning to his original position of Director of Development. Sutherland became E.D. shortly after long-time Executive Director, Mike Parent retired in July of 2016.

"To say the last 16 months have been a whirlwind is a tremendous understatement," Sutherland said. "As an office we implemented re-branding efforts, developed a new website, realized tremendous growth in our communication efforts, have taken on new fundraising campaigns and moved our national headquarters. Many of these initiatives have taken me away from what I think I do best and what I enjoy doing and that is generating revenue for our foundation," Sutherland concluded. NAF Board President, Bill Sweeney, will oversee the daily operations on an interim basis.

In addition to overseeing development efforts with Major Donors, Sutherland will continue to work on campaigns like the Walk n' Roll, the mid-year "Matching Gift" program, and the 2018 Go the Extra Mile campaign. Go the Extra Mile is the extension of the 2017 "60 for 60" effort. "It is our goal to get 100 people from around the country to "Go the Extra Mile" in 2018," Sutherland said. Getting the community energized to the point of participating is key to generating the additional revenue needed to fund important research.

THE DEADLINE FOR SUBMITTING MATERIALS

for the Summer issue of *Generations* is Monday, May 21st. Please send articles, your personal story, recaps of Ataxia-related events, photos and reports to *naf@ataxia.org*. Thank you.

"Reality"...Make the Most of It

Cathy DeCrescenzo

As the years pass, living with Spinocerebellar Ataxia can be daunting, not only for the person afflicted with this disorder, but for the spouse and children, as well. As many of you remember, when first diagnosed, it is surreal. For us, this was 13 years ago when my husband Joe was diagnosed with SCA2.

Speaking from the caregiver/spouse role, keeping a positive attitude is key, though some days are easier than others. As time marches on and the Ataxia progresses, you see changes in your loved one from year to year, and, at times, from day to day. It is heart wrenching to watch the person you married, in our case, 40 years ago, struggling daily to walk and using every muscle and fiber in their being to make it through the day. We continue with our normal life style, as much as is possible. Traveling is becoming more difficult but we adapted by purchasing a motorized scooter that I can handle alone, and Joe can drive to the door of the plane, which is then stored underneath in the luggage compartment. Making plans to travel, even if it's only a long weekend away, is something we look forward to. Wherever we travel, it is a much-needed and refreshing change. We are down to one car, as Joe no longer drives; therefore, I am the designated driver... no big deal whatsoever. Every Wednesday we enjoy going to dinner with our dear friends, and I country line dance at least once a week. Joe is retired two years now but I continue to work part-time. So, for us, life goes on, which is a blessing indeed.

It is not within either of us to EVER give up, there is always hope; though we are realistic in what lies ahead. We have taken steps over the last couple of years to prepare ourselves for the inevitable. Firstly, we met with an Eldercare attorney and had the appropriate legal/medical documents drawn up; copies of which were given to various parties, including our children. Secondly, we visited our local funeral director and made our final arrangements, which took a tremendous load off our minds. In fact, not to be morbid, Joe wrote his own obituary and he will update it occasionally. Knowing our children will not have to be subjected to making decisions under duress made it a much easier task. Joe has also arranged to have his brain tissue donated for research. The funeral home has all the contact information they will need; therefore, taking the burden off the family.

We are truly thankful for each day we are given. Joe and I continue to pray for a treatment and/or cure, and we rely heavily on our Faith. We believe that Faith is not a feeling, but it is a choice to trust God even when the road ahead seems uncertain.

In the meantime, we do not sit idly by. Instead, we organize fundraisers to help NAF to fund research towards a cure. It is amazing how our friends, family and church family have come out in support of NAF whole heartedly over the years. We make our "fun"draisers "FUN", which entices between 225-250 people to attend and make generous donations. Years ago, with guidance from NAF, we started a local support group here in Delaware, as the closest group was two hours away in Maryland. In addition, Joe is very active on the national level of NAF and we volunteer at the annual conferences.

"Speaking from the caregiver/ spouse role, keeping a positive attitude is key."

There are numerous avenues you can explore to help your own situation as well as others in the community who are struggling with Ataxia. Unfortunately, we cannot wave a magic wand and the Ataxia will disappear; that is in the hands of a higher power, and in the hands of the researchers who are working tirelessly to find a cure. We can; however, work in unison to make the lives of those afflicted with Ataxia, and the lives of the caregivers and family members less stressful and more enjoyable. Please reach out to the Ataxia community in your area, use your imagination and

plan an event such as a spaghetti dinner, a walk n' roll, or any activity or get-together you can arrange that will raise awareness as well as funds. Staying busy is important and is great therapy, so why not put that energy to good use and plan an event. Believe me, once you organize your first event, you will look forward to the next.

In closing, we only go around once in this life, so take what you have been given, stay positive BUT realistic, and live life to the fullest, to the best of your ability. Remember, there is always someone out there with no hope for the future. We are hopeful that, in our lifetime, a cure will be found and Ataxia will be nothing more than a distant memory.

Cathy was born and raised in Fairfield, CT and relocated to Delaware 15 years ago with her husband Joe to be near their girls and granddaughters. Two years later, Joe was diagnosed with SCA2. Being near family was and is a comfort. Cathy cared for their three granddaughters until they were of school age; she is now an administrative assistant at their church. Cathy and Joe feel truly blessed and fortunate for the gift of their loving family, church family, and their Pastor, Fr. Roger DiBuo, all from whom they draw strength and support as their Ataxia journey progresses.

Contact info: cdecres@verizon.net
302-388-2758 (c)

We want to hear your personal stories!
Send them to *naf@ataxia.org*

Don't Underestimate Yourself

Dana Creighton

I was diagnosed with SCA2 in 2006. In 2011, I was giving blood at a Red Cross blood drive at a local surgical center. There was going to be a raffle for prizes so I filled out a ticket. Two weeks later, they called me to tell me I won 8 sessions of personal training. I would have never paid $50/hr for a personal trainer, but I was thrilled to do this free of charge.

"I never would have done this had I stayed in my comfort zone."

I was amazed at the strength I was able to gain and how quickly it happened. I realized that initially I had way underestimated what I believed I could achieve. That if I had not been pushed to do more than I thought I could I would not have gained such strength both physically and especially mentally. My first 30 min session was so hard that when I met my friend that evening for dinner and I was utterly exhausted.

One reason for the dramatic increases in strength I experienced was that working at something that was a challenge physically was also a challenge mentally. That next year at 40, my friend talked me into completing my first (and last) sprint triathalon, "The Ramblin' Rose" with her. I'm glad I did this, and I never would have done this had I stayed in my comfort zone. I don't aspire to do another triathalon nor do I have a personal trainer. I am however, not afraid of pushing myself physically or limiting myself mentally to trying something new.

Dana grew up in northern Indiana and has lived in North Carolina for 18 years.

Travelling with Ataxia

Kory Macy

I have Ataxia. Specifically, AOA2. I also love to travel so I decided traveling is more important. I have been to Kenya, Mexico, a cruise to England, Ireland and Scotland, Amsterdam, a cruise to Alaska, canoeing down the St. Croix River, sea-kayaking in the Apostles Islands, camping in Yellowstone, Ecuador and soon I am going dogsledding. I have several more trips in mind. Sure, it is a little bit harder and you need to ask very specific questions but where there is a will there is a way and it helps that I am very stubborn. The more you say I can't, the more I will want to do it.

The first thing you need to do is decide what you want to do. The next thing you need to do is take a long, hard and honest look at what your abilities are and what you may need help with. Then do an internet search asking for accessible activities in that area or search for groups that work with people with disabilities. After you find that group or activity, call and ask questions. You can also work with a travel agent. The agent does not need to specialize in travel for people with disabilities but needs to be open to what you specifically need.

I use all three methods. I have made mistakes but have learned from them. I've learned more about what I can do and what I need help with. Not every country has American ADA standards.

"I have made mistakes but have learned from them. I've learned more about what I can do and what I need help with."

Mostly I have learned that in order to have an enjoyable trip, I must ask for help when I need it.

Travel with Ataxia is possible as long as you want it. To find out more contact me at kstab77@yahoo.com

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Kory works full-time in Madison, Wisconsin. She and her husband love to travel and she does not let her Ataxia stop her. She runs the Wisconsin Ataxia Support Group and was diagnosed with AOA2 in 1997.

The Last Laugh...Based on a True Story

Jason Wolfer

One year ago, I wrote a book. I titled the book, The Last Laugh...Based On A True Story...and I am going to include here an excerpt from one of the chapters from that book. The chapter is titled The Last Laugh, and I picked this section to share with you because it describes, in the last paragraph, why I picked that title for the book, and what it means to me. So for your consideration, I present to you the end of chapter fifteen....

> **"I feel that maintaining the ability to laugh at myself, even in the light of a neurological disease, helps me to stay in control."**

> But, I do try to maintain a good sense of humor and spend a lot of my day finding the humor in my situation. No, it's not always easy, but as they say, laughter is the best medicine. I just hope that some of the large pharmaceutical companies don't find out about this. If they hear that laughter is truly the best medicine, then I'm afraid that they might try to patent it, put it in pill form, and try to sell it to us at astronomically high prices. Most insurances, and for sure Medicare, wouldn't cover it either because laughter would most likely be viewed as a life choice and one that was only needed recreationally. Either it would be one of these...or they will refuse to pay for laughter because it will be deemed to be a pre-existing condition.
>
> In an effort made by these corporations to maximize their profits, I'm sure that the laughter available to us would not even be sold in its pure form. Most likely, it would be about fifteen-percent pure raw and uncut humor; that was mixed with various other fillers... some of which would cause horrendous side effects like uncontrollable bodily noises. I, for one, do not need any help in this department from a pill.
>
> I feel that maintaining the ability to laugh at myself, even in the light of a neurological disease, helps me to stay in control. My SCA might be able to dictate how a situation may go, or how I may react initially, but it cannot and will not influence my overall outlook, or attitude. I think, that in light of my Ataxia, humor's biggest encouragement to me is the knowledge that it will always be there to provide the last laugh. Not for SCA...but for me.

...If you would like to read more and would like to purchase the book, please contact me...either through Facebook Messenger or by emailing me at wolfer.jason@gmail.com. The book has not been distributed to any stores, such as Amazon, and can only be purchased through me. The cost is $15 and can be paid through a Paypal link which I will provide or by mailing me a check.

Jason is currently the leader of two support groups and among several other projects, began writing a blog in 2013, titled My Life...With Ataxia Along For The Ride. He has 3 kids, two of which are married and one in College. Jason lives in Oregon with his wife, two dogs, and his cat Pearl.

MOXIe

a study in Friedreich's ataxia

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
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REATA PHARMACEUTICALS

This study is being sponsored by Reata Pharmaceuticals. www.clincialtrials.gov (NCT02255435)
ENG V1.0, Protocol Version 9.0 – 17August2017

Shaky Finish

Susan Harris

Chapter 1: In the beginning

I wasn't always this way. Growing up the daughter of a military man I can honestly say red, white and blue run through my veins. I love America and my family for years has fought vehemently for this glorious country. Allow me to say here how conservative my financial values are and have always been and always will be. Socially, not so much. But that's a story for later.

I never regarded my family as special. But now I know that's not true. Yes, every cousin and aunt and uncle is successful. Not Microsoft successful but we are happy.

As a descendant of Abraham Lincoln what makes my family "special" indeed is that SCA5 is running through our DNA (thanks Bathsheba, his granny). My brother decided not to have kids because his symptoms started to emerge way too young – 19. I had no symptoms so I went along my merry way and had two children which are the biggest blessing to my family. The stress about did me in but I can't even imagine my sweet mother having no grandkids to spoil rotten

My background was that of a pre-Olympic swimmer, so teaching swim lessons and water aerobics was a no brainer. I did it for the income and so I could stay-at-home with my children but little did I know how therapeutic water really was.

Chapter 2: The benefits of water exercise

Do I really need to say a lot here? The benefits are well documented. Water helps with balance, strength and conditioning. Plus, there is a lot we can't do (gosh, I'd love to go for a run or a speeding bike ride to feel the warm air blow onto my face...). I love my adult tricycle. It really entertains the neighbors with my doggie in the basket as I try to pedal FAST! Whoa, watch the feet as they slip off the pedals and I bruise ankles constantly. Bruising is not possible in the pool unless you run into the sidewalls. Please don't do that. Trust me, it hurts. But do get in the pool. It doesn't matter, instructor or not, just MOVE. Jumping rope, cross-country skiing, or simply walking back and forth (which are movements you may or may not be able to do on land). Doing the exercises recommended by the Arthritis Foundation™ will benefit each and every person out there. It doesn't matter what issues. Unless you are dead and if you are reading this you most definitely are NOT dead yet. Get out there and MOVE!!

I am competing in master's meets although I am now unable to dive off the starting block. I just start off the side and swim as fast as I can. At least I'm out there.

"I never wanted to lament over the lost abilities. I am thankful for the abilities I do still have."

Chapter 3: Focus on what you can do

I never wanted to lament over the lost abilities. I am thankful for the abilities I do still have. So this swimming adventure is my own mental exercise. I also enjoy traveling and meeting new people. It is another gift from God. I will also be embarking on that adventure and cherishing each moment! Might be in a wheelchair, who cares. The finale is coming!

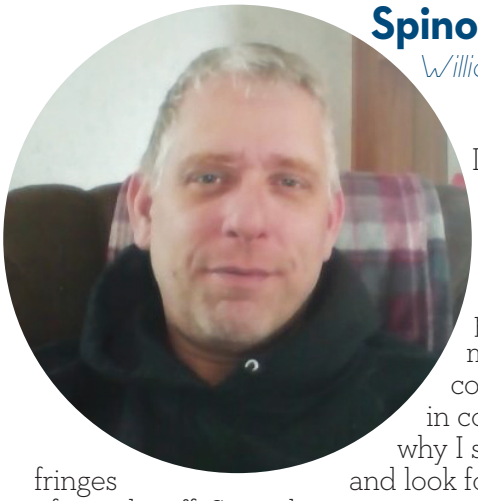
Chapter 4: Ataxia Sucks

We all have problems. Yes, that's true. But ours are something we must confront every single day. It's very personal to each of us. We are a gift from God and what we do with that is our gift back to God. Always look up. Get out there and move each and every day. Thanks for taking your valuable time to read this. You are kind and you are beautiful. Each in his or her own way PERIOD.

Susan is loving life and thankful for God's many blessings in her life. Proud American.

Spinocerebellar Ataxia: A Cultural Imperative

William R Vetter Jr.



Looking back 40 years, it all makes sense: why I avoid certain people, why my tongue gets confounded in confrontation, why I stay on the fringes and look for a way out of social stuff. Something so innocent as not being able to balance on one foot in gym class, and lacking appropriate common sense, and not having typical recollection of faces and information, and being clumsy and awkward in my movements... should pave such a chaotic existence? The bullying, the ridicule, the ambushes...I cry for that boy now that I know this genetic something in my brain made tough circumstances devastatingly self-convictive. I thank God often that I didn't follow through as Leelah Alcorn did.

I believe, deep down, I am always going to have a harder time adjusting socially. I believe much of that is due to lack of awareness and acceptance of SCA, and the lack of definitive biobehavioral red flags. I believe there is no cure and few treatments for SCA. I believe it's going to get way worse--degenerating further my balance, memory, coordination, speech, sight, and motor control. I believe I am going to have a difficult time making a living, especially now that I've left an established career in electrical construction. And I believe I am having a difficult time getting to the point because I feel like I have to justify what I believe in order to implore you to relate the things you hold true.

Looking forward is harder: Getting a clear sight of who I am versus who I am not means seeing this condition as it is and not taking it personal. But more than that, it means daring to look at my condition as a unique human perspective for adding value in at least biology, sociology, psychology, medicine, art, and education, with implications running a gamut of social landscapes and phenomena.

For instance, let's just say right this very instant I found enough courage to suspend my belief in my disability (in my particular ability-hobbeling condition). Suppose I suspend my disbelief in having something to offer that will change the lives of millions for the better. It's not such a far stretch. Suppose I opened the vault of my memory and rewired all the messages that convince me to believe "I am not enough" so that I understood "I once believed I was not enough."

So, here we are...maybe I offer little more than fanciful philosophy and maybe you got some inspiration. I hope. I also hope someone here gets the intention. These words are from the deepest realm of my heart, yet they seem frail and short-fallen for what I would say if I believed enough. Let's at least examine the dialog about and around Ataxia and disability in general, and how we might change our beliefs and talk to bring about better results.

For you and I, life may have reached a tolerable quell. But I suspect there are little ones developing with an unknowing about what is going on inside their skin. I suspect the number of unknowns could easily outnumber the knowns. I suspect many of them become statistics in avoidable circumstances. And I suspect this society could thrive beyond expectations should it discover better ways for individual expression. Let's start the dialog.

"I suspect this society could thrive beyond expectations should it discover better ways for individual expression. Let's start the dialog."

I am in.

William is a 48-year old servant of Jesus Christ, a father of 2 beautiful adult boys, and a brother to as many as I'm led. He attained his BA in Communications in 2016, and now aspires to be a freelance copywrite-artist and innovator. It's been rather easy for him to see his disability as obstacle and misfortune--It's much more difficult to see the gift and opportunity it naturally manifests.

Ramblings Of A Disabled Nature Lover

Brett Mitchell

Floors of malls
And big box stores
They are no fun for me
My legs like them no more
I like the outdoors
Sun overhead
Grass beneath my feet
Breezes flowing through the trees
You can have your concrete
I do better on a dirt path
Through the woods
Or next to a body of water
Parks- city, county or state
Are where are I like to be
I may need something
To keep myself upright
But I'm still walking
Getting exercise
For my muscles
Vitamin D for my bones
No screens to stare at
Nature all around
I may be disabled
But yet I still feel free.

Brett Mitchell was diagnosed with Spinocerebellar Ataxia in 2011. Nature walks are one of his favorite activities that he can still do.

NIH Grants $6 Million over 5 Years to Support Clinical Trial Readiness Efforts for SCA1 and SCA3

In a milestone grant, the National Institutes of Health will support a US-European collaboration that will seek to bridge the gap between the current state of clinical trial readiness and that needed to best support future Ataxia clinical trials. The recent rebooting and funding of the Clinical Research Consortium by NAF re-engaged CRC-SCA investigators in collaborative research, which led to the success of this international, multi-million-dollar trial readiness grant.

The research project will enroll patients with SCA1 and SCA3 at the early stages of symptoms, including pre-symptom onset as well as those who are at-risk for SCA1 and SCA3. Research participants will be needed at 17 sites across the United States and two sites in Europe.

Leading this important study is Tetsuo Ashizawa, MD at Houston Methodist Research Institute, who is a member of NAF's Medical Research Advisory Board and co-Principle Investigator of the NAF Clinical Research Consortium.

We applaud Dr. Ashizawa for successfully securing this important Ataxia research funding.

Information about enrollment and the study can be obtained from the HMRI team:
Email: U01SCA1&3@houstonmethodist.org
Phone: 346-238-9068

If you or family members are affected by any form of Ataxia or are at-risk for Ataxia, please enroll in the CoRDS patient registry. The registry helps identify patients who will be eligible to participate in future studies. You can enroll online in just a few minutes at http://www.sanfordresearch.org/SpecialPrograms/cords/

*Please note: Research reported in this publication was supported by the National Institute of Neurological Disorders and Stroke of the National Institutes of Health under Award Number U01NS104326. The content is solely the responsibility of the authors and does not necessarily represent the official views of the National Institutes of Health.

Fundraiser and Awareness Event Recaps

Chesapeake Chapter Represents NAF at DC Abilities Expo

Submitted by Carolyn Davis

The Chesapeake Chapter represented NAF at the DC Abilities Expo in December. Pictured Glenn and Carolyn Davis. They were able to help others learn about Ataxia and provide information about NAF.

Kid to Kid Contest Winner Gives Back!

I would like to give a big shout out to an amazing young lady, Ellioni Durrant!

Ellioni is an incredible young lady with a big heart and determination to make a difference in the lives of others. Elli is very kind and compassionate. She exemplifies this strength by giving back to the community. In particular, she has voluntarily taken on the responsibility to help raise awareness and funds for the Utah Ataxia Walk-n-Roll Fundraiser for the past 2 years. September 23, 2017, marked the 4th Annual Fundraiser. Elli chose to be more involved and take on more responsibility in the planning and execution of the fundraiser. She spent countless hours devoted to this worthy cause. She showed her commitment by showing up and following through with assignments in preparation for the event and during the event radiating positive energy and enthusiasm.

Ellioni's effort and time have contributed greatly in the success of the Utah Walk-n-Roll Fundraiser(s). Funds raised not only help those with Ataxia in Utah, but throughout the United States and other parts of the world. Her service and dedication in raising funds for specialized doctors and critical research to find a cure will impact many lives now and in the future.

This young ambassador serves many members of her family on a daily basis who live with Ataxia. She wears many hats with the varies responsibilities she carries. Ellioni is also an amazing, helpful, and responsible daughter, big sister, niece, cousin, granddaughter, student, and friend.

Elli has shown what a great role model she is by her desire to serve and help those in need by giving back to her community.

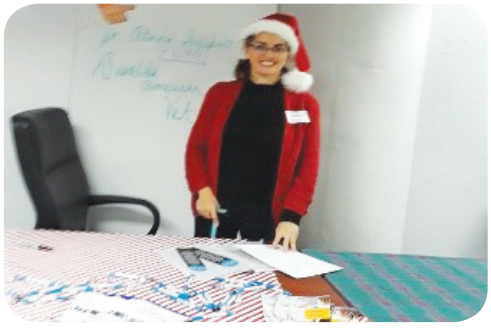
Because of all these reasons, and so many more, she was nominated for the Kid to Kid Contest in November 2017, sponsored by iheart Media, in Salt Lake City, Utah. Elli was one of the top four nominees chosen for the community to vote upon. Thanks to all the votes in her favor, she was the Kid to Kid Contest recipient! As part of her winnings, Elli was given the opportunity to choose an organization to donate $100.00 to their cause. Elli chose to support the National Ataxia Foundation. When asked what made her decide upon this foundation, Ellioni stated, "I chose the National Ataxia Foundation because it is very personal to me. Members of my family have Ataxia and I want to help them."

Thank you, Ellioni, for your dedication, service, and contribution! YOU make a difference!

St. Louis Ataxia and the CFC

Submitted by Shannon Dunphy

The Federal Government participates in a fundraiser called the Combined Federal Campaign (CFC). Every year, Federal employees can choose from a large list of nonprofits/charities to donate via payroll deduction. To give the employees a taste of the participating charities, agencies invite organizations to staff a table, and employees circulate and visit different charities. Shannon Dunphy hosted an exhibit for NAF at the CFC Charity Fair, offering NAF bookmarks, info cards, pens, and Children with Ataxia booklets. Overall, this CFC Charity Fair was worth the effort, and I'm glad I could represent the St Louis Area Ataxia Support Group and the NAF.



Tampa Bay Ataxia Group – 60 for 60 Walk for a Cure

Submitted by Jan Colon and Darlene Harris

The Tampa Bay Ataxia Group 60 for 60 Walk for a Cure to Ataxia was held on January 13 at USF Marshall Student Center in Tampa, Fl. Even though it was cold we did it!!! Nothing can stop us. My daughter and I had the opportunity to Raise Ataxia Awareness with #AtaxiaRocks during the walk.

Thanks to Dr. Z and her wonderful staff for all of their assistance with our Ataxia 60 for 60 walk. And a special Thank You to Dr. Z for her treating the volunteers with breakfast from Chick-Fil-A.

Thanks to Dr. Stephenson and her great students for all of their assistance with our Ataxia 60 for 60 walk. Their help in pushing our participants as well as providing the Wheelchairs was really appreciated. We could not have done it without their help and for that we are grateful.

I would like to list & thank all of the vendors and contributors that supported us:
Rare Patient Voice, Dress for Success, SISTUHS, Ask a Pharmacist - Christina Wilson-Smith, Tampa Metropolitan Chapter of Delta Sigma Theta Sorority, Inc, My Ride or Die Crew from Miami, Total Nutrition, Perfume Express at University Square Mall, The Hair Cuttery off of Bears, Sears at University Square Mall, Famous Footworks at University Square Mall, Chili's on Fowler, Walmart on Fletcher Ave, The Dollar Tree on Fowler and The members of Tampa Bay Ataxia Group.

We raised $312.00 with donations & from the silent auction to benefit the National Ataxia Foundation. We were happy to support the NAF.

The Ataxia Community is always looking for great ideas to share in *Generations*.

If you have Pearls of Wisdom or a personal story you would like to share in a future issue of *Generations*, please submit it to *naf@ataxia.org*. Please keep your "pearls" short and personal stories to 1000 words or less.
Those submitting a personal story are asked to please include a photo or two and a brief author bio (1-2 sentences).

YOUR DOLLARS AT WORK:
A Look at NAF Funded Research

The following are lay summaries from research projects that NAF was able to fund because of generous contributions from our donors. All of these research summaries are of grants funded by NAF for fiscal year 2018. Thank you to each of you who made a donation to last year's Research Drive "Proud Past... Focused Future."

Unless you are a scientist, these research summaries can seem like "Greek" to you, however, it does demonstrate the complexity of science, particularly neuroscience. These summaries were submitted directly from the researchers. While they may be difficult to read, we at NAF think it is important to keep you up-to-date on the science that your membership and donations support.

SEED MONEY GRANTS

Deciphering the pathophysiology of SCA35

Manuela Basso, PhD
University of Trento
Trento, Italy

Spinocerebellar ataxia type 35 (SCA35) is a rare, autosomal dominant neurodegenerative disorder associated with mutations in TGM6 gene. Recently, we provided evidence of seven new mutations identified in three different Centres in Europe and USA. By analyzing the protein behaviour of all the TG6 mutations known to date, we divided the mutations in two groups, a less toxic and a more aggressive group, according to different acquired properties of the mutant proteins. While we studied the newly identified mutations in cells lines, neurons reproduced in dish and in the fruit flies, we deem necessary to develop a mouse model where to reproduce the clinical ataxic phenotype and understand how different mutations impact on the cerebellum physiology with the final goal to propose a successful therapeutic treatment. Of note, the preliminary data of this work has been presented in a poster at the Investigator meeting in Orlando, FL in 2015 thanks to a travel grant by the NAF.

Investigating the connection between DNA damage repair and nervous system maintenance in a Drosophila model of SCAN1 disease

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The main goal of our project is the development of a powerful genetic model to investigate pathogenesis of spinocerebellar ataxia with axonal neuropathy-1 (SCAN-1) disease. Patients affected by SCAN-1 develop an adult onset devastating pathology characterized by peripheral axonal motor, sensory neuropathy, distal muscular atrophy, pes cavus and steppage gait. The genetic alteration causative of SCAN-1 is a mutation altering the function of a gene called *tdp1* (tyrosyl-DNA phosphodiesterase 1). The product of this gene is an enzyme that plays a key role in DNA repair processes whose defective activity causes failure to reseal broken DNA strands causing

neurodegeneration. The tiny fruit fly, *Drosophila*, is an organism extremely useful for studies on human biology, health and a wide range of pathologies including neurodegenerative diseases. This is because *Drosophila* genes controlling fundamental cellular functions, such as cell growth and death, are quite identical to those found in human cells. We plan to obtain a *Drosophila* model of SCAN-1 disease by applying a well-known genetic approach. This focuses on the expression of the human *tdp1* mutant gene in brain tissue of flies lacking the endogenous Gkt protein. This approach would create a fruit fly model that mimics the human pathological condition.

Genome-wide transcriptome analysis of this SCAN-1 fly model will get insight into the mechanism of the disease. The identification of the steps of the SCAN-1 pathological cascade in turn will help the development of therapies targeting key molecules acting in these steps.

Apoptosis-Related Genes BCL2, BAX And TP53 As Biomarkers Of Machado-Joseph Disease (MJD/SCA3)

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Portugal

Despite undeniable progresses in the knowledge concerning the molecular pathology of Machado-Joseph disease (MJD)/Spinocerebellar ataxia type 3 (SCA3), therapeutic compounds remain to be discovered and validated. Interventional trials face several obstacles, namely those related with the clinical outcome measures used, which lack sensitivity for slow-progressing diseases such as MJD, and are devoid of utility in the preclinical stage, a time where molecular alterations are known to be already present. Identification of molecular biomarkers, accessible in a peripheral tissue such as the blood, is therefore of particular importance to allow the fine tracking of disease progression, starting at the preclinical stage, thus facilitating the detection of subtle therapeutic benefits during interventional therapeutic trials. Furthermore, once ameliorating drugs are available, molecular biomarkers could help identifying the molecular onset of disease and aid in the therapeutic strategy, since such drugs are expected to be more efficient if administrated to mutation carriers before overt disease. Building on promising preliminary results, the goal of this project is to validate the apoptosis-related genes as peripheral molecular biomarkers of MJD and disease progression. Expression of these genes will also be studied in brains of MJD patients, where we expect a similar expression pattern to that seen in blood samples. Drug discovery relies on the translation from animal model studies to patient trials - confirming the similarity of molecular alterations occurring in animal models of disease with those observed in patients is therefore of high relevance. To ensure translatability of pre-clinical trials using YACMJD84.2 transgenic mice we will assess the expression of the apoptosis-related mouse homologue genes in blood and brain tissues from these commonly used mice in such trials. As YACMJD84.2 express the human disease target and replicate the disease we expect to observe similar expression alterations to the ones found in MJD patients. Here, our goal is to hopefully contribute to include the assessment of expression levels of apoptosis-related genes in a set of outcome measures for MJD and to a better definition of the molecular onset of this disease. Furthermore, understanding alterations in the intrinsic apoptotic pathway, particularly in the brain, could be valuable to pinpoint novel therapeutic targets for MJD.

HAX-1 is a molecular biomarker for cardiomyopathies in Friedreich's Ataxia

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Frataxin deficiency, responsible for the hereditary disease Friedreich's Ataxia (FRDA), is crucial for cell survival as it critically affects viability of neurons, pancreatic beta cells and cardiomyocytes. The heart is affected in approximatively two thirds of FRDA patients with typical manifestation of hypertrophic cardiomyopathy, which can progress to heart failure and death. Except for frataxin, very few genes have been correlated with ataxia and cardiac disease in FRDA. To investigate FRDA pathogenesis, we conducted a wide expression analyses on cells derived from a FRDA patient and we observed HAX-1 as the highest over-expressed gene. HAX-1 is highly expressed in the heart and belongs to a family of proteins involved in the protection of cardiomyocytes from cell death. Frataxin and HAX-1 are therefore both involved in cell death regulation, a mechanism underlying the progression of cardiomyopathies. We further analyzed frataxin and HAX-1 expression level in blood cells from FRDA patients and controls and we found a positive correlation, i.e. in FRDA patients, the levels of frataxin and HAX-1 were lower than in controls. This correlation was stronger when we compared frataxin and HAX-1 levels in FRDA patients showing cardiomyopathy. This project is aimed to confirm HAX-1 expression as new molecular biomarkers to enhance a personalized medicine for cardiomyopathy and to render wise decisions to families being evaluated for the presence or absence of this potentially lethal yet treatable cardiac disorder. The role of biomarkers to predict the onset of future cardiomyopathy, to identify its presence when fully developed, to risk stratify affected patients, and possibly to serve as a biological tool to guide therapy for cardiomyopathy is indeed fundamental. Another aim of this project is to analyze the expression level of circulating molecules such as microRNAs in FRDA and to investigate their link to HAX-1 expression and regulation. This information will be useful in terms of prognosis and therapeutic impact.

Disclosure of genetic information within families affected by hereditary ataxia

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People with hereditary ataxia face the complex and difficult challenge of communicating their diagnosis and genetic risk information to family members. Communication about an inherited condition implicates unaffected family members as well as future children – information which may have an impact on important life decisions. Previous studies have found that early disclosure and an open family communication style can help in family members' development of effective coping strategies, adaptation to the genetic condition, and emotional well-being. While studies have found that many people want to disclose genetic risk information to their family members, some people encounter barriers that can cause them to delay or avoid disclosure. While a number of qualitative studies have explored disclosure behavior in families with other neurological conditions, this process has not been described in families with ataxia.

The purpose of this study is to explore disclosure behavior and to identify factors that affect the communication of diagnosis and genetic risk information within families affected by hereditary ataxia. This study also aims to identify barriers, if any, to the disclosure process for this population, and to explore people's attitudes about their disclosure decisions. The results will be critical to understanding the experience of family members about genetic risk, and in predicting how health professional and disease advocacy groups can help facilitate better genetic risk communication for families.

Phenotypic Characterization of Mouse Models of Infant- and Adult-Onset SCA13

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The goal of this project is to characterize the phenotypes of three unique, newly-generated knock-in mouse models of spinocerebellar ataxia type 13 (SCA13). SCA13 is caused by mutations in the KCNC3 gene, which encodes the Kv3.3 voltage-gated potassium channel. Voltage-gated channels such as Kv3.3 control the passage of ions across cell membranes. In the nervous system, voltage-gated channels are responsible for generating action potentials, which are rapidly moving electrical signals that underlie brain function. Kv3.3 is highly expressed in cerebellar neurons, where it regulates action potential firing. Different Kv3.3 mutations cause two clinical forms of SCA13. In one form, SCA13 emerges in infancy or early childhood, characterized by cerebellar atrophy early in life, motor delay, persistent motor deficits, and intellectual disability. In the other form, SCA13 is an adult-onset disease characterized by progressive ataxia and progressive degeneration of the cerebellum. Our lines of knock-in mice carry distinct Kcnc3 point mutations, R421H, R424H, and F449L, which correspond to the human mutations R420H, R423H, and F448L, respectively. In humans, R420H causes late-onset SCA13, whereas R423H and F448L cause the early-onset form of the disease. The new mouse models provide a novel opportunity to investigate mechanisms that determine the age of onset and trigger cerebellar degeneration and ataxia in SCA13. Such studies are important for identifying new therapies for the disease. Our preliminary data, obtained in zebrafish, a lower vertebrate, suggest that early- and late-onset mutations have differential effects on action potential firing that trigger the age-dependent degeneration of cerebellar Purkinje cells. The knock-in mice now make it feasible to test this hypothesis in a mammalian model system. With support from the National Ataxia Foundation, I propose to begin characterizing the phenotypes of the three lines of SCA13 mice by accomplishing the following Specific Aims: 1) to investigate motor performance at different ages using standard behavioral assays; 2) to investigate age-dependent changes in cerebellar anatomy and the viability of Purkinje cells; and 3) to characterize the effects of SCA13 mutations on action potential firing in Purkinje cells.

National Ataxia Database

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The Natural History Study of and Genetic Modifiers in Spinocerebellar Ataxias (ClinicalTrials.gov

Identifier: NCT01060371), under the direction of Dr. Tetsuo Ashizawa, continues to recruit subjects and monitor changes in their neurologic examinations. The participating 15 sites see and examine subjects and enter data into the National Ataxia Database (housed at UCLA under the direction of Dr. Jeanette Papp in the Department of Genetics). The Database has served as a valuable repository for data collected in this important collaborative project that has enrolled over 500 individuals with Spinocerebellar Ataxia types 1, 2, 3, and 6. It will now be adding individuals with types 7, 8, and 10. There are over 15,000 patient forms entered in the National Ataxia Database.

Nuclear-mitochondrial pathways of neurodegeneration in cerebellar ataxia

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Autosomal recessive cerebellar ataxias are heterogeneous neurodegenerative diseases, characterized by incoordination of movement and unsteadiness, due to cerebellar dysfunction. Cerebellar ataxia has emerged as the most common clinical presentation of deficiency of Coenzyme Q10 (CoQ10), a vital molecule required for cells to generate energy and to prevent damage from toxic oxygen radicals. The proposed studies will define the causes underlying CoQ10 deficiency in cerebellar ataxia, and its role in neurodegeneration, and may lead to the identification of novel therapeutic targets. The results of our studies may provide important information relevant also to other neurodegenerative diseases.

Characterization of the disease phenotypes of Ataxia Telangiectasia patient Purkinje cells in vitro

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Ataxia-Telangiectasia (A-T) is a rare genetic disorder that is caused by mutations in ATM gene. There are currently no treatments for the cerebellar deficits that cause motor dysfunction and balance problems in the A-T patients. Main output neuron type of the cerebellum are the Purkinje cells, which are GABAergic inhibitory neurons that are innervated by cerebellar granule cells and to project to deep cerebellar nuclei that regulate motor coordination and balance. Previously the restricted availability of human brain tissue for medical research has limited the detailed characterization of the causes of cerebellar degeneration and neurological deficits in the A-T patients. To overcome this limitation, researchers have recently developed new cellular model called human induced pluripotent stem cells (hiPSCs), which can be derived from human somatic cells and converted in a dish into different cellular types, like different neuronal cell populations. These hiPSC derived neurons facilitate the studies of patient specific neurons in laboratories, and allow identification of molecular mechanisms behind the development of various neurological diseases, including A-T.

Previously the Sahin lab has developed a novel protocol to differentiate the hiPSC into cerebellar precursors and Purkinje cells. In addition, the Lerou lab has discovered genetic abnormalities in control of NRXN1 expression in the human neuronal precursor cells. To discover the molecular causes for the development

of cerebellar dysfunction in A-T patients, we propose to study functional properties of the A-T patient derived hiPSC-derived Purkinje cells compared to healthy control Purkinje cells, and study differentially regulated genes in these cells. Our study will provide valuable insights into the molecular mechanism and potential targets that can be utilized in the future for development of new therapies for the neurological deficits of the A-T patients.

Suppression of polyglutamine aggregation by a new class of chaperone

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We all have proteins that contain repeats of the amino acid glutamine. However, some individuals have an expansion of these glutamines resulting in very long glutamine repeats. These long glutamine repeats cause the polyglutamine diseases including Spinocerebellar ataxia types 1, 3, 6, 7, and 17. ln these diseases the long strings of glutamine cause proteins to clump up and this results in neuronal death. We have recently shown that the ameoba, Dictyostelium discoideum, normally expresses very long glutamine tracts and is naturally resistant to clumping up of proteins with long polyglutamine repeats. More recently we have identified a single protein that is only found in Dictyostelium that is responsible for suppressing the clumping of these polyglutamine proteins. Here we propose to figure out how this protein prevents the clumping of proteins with long polyglutamine repeats, and produce a mouse that expresses this Dictyostelium protein to determine if it can suppress the underlying cause of polyglutamine diseases including SCAI ,3,6,7, and 17.

Modulation of neuronal proteostasis by serotonin: impact on SCA3 pathogenesis

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Spinocerebellar ataxia type 3 (SCA3), also known as Machado-Joseph disease (MJD), is a neurological disorder affecting the specific areas of the brain and the spinal cord of patients. The first symptoms usually start around middle age and include difficulty in walking, due to lack of coordination and balance. Other complications, such as problems with vision, speech, and swallowing may also occur. This is a progressive disease and many patients ultimately need a wheelchair or become bedridden, as there is no treatment currently available. We have previously found that chronic treatment with an antidepressant very strongly improves SCA3-like symptoms in model organisms of the disease. Worms (C. elegans) and mice expressing the mutant human gene and protein display abnormal movement and lack of balance and coordination. Antidepressant treatment reverted these phenotypes and decreased the presence of aggregated ataxin-3. Our next step will be to understand what is the drug doing in the brain that protects against disease. We have recently found that antidepressant treated cells show increased expression of protective genes that

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enhance protein homeostasis capacity and reduce the risk of formation of aggregated mutant protein. Here, we propose to study in which neurons this protective mechanisms are activated and to study this augmentation in protein homeostasis aptitude as a therapy for SCA3. This study will help us to decipher a series of events that occur in the brain of SCA3 animals and are prevented when the animals are treated with the drug. A better understanding of those events may help the future development of clinical trials in patients and define novel therapies. Besides being useful for SCA3 patients, this antidepressant therapy may also be important for patients suffering from other neurodegenerative disorders, hence it is essential to clarify its mode of action, before advancing to clinical trials.

Function of ATM in Neuronal Survival in Response to Growth Factor Signaling

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Ataxia-telangiectasia (A-T) is a neuronal degeneration disorder characterized by progressive cerebellar ataxia, oculocutaneous telangiectasias, and variable immunodeficiencies. The gene mutated in this disease, ATM (A-T, mutated), encodes a protein kinase. The main characteristic of A-T disease is the progressive neuronal degeneration of cerebellar Purkinje and granular cells. Patients with A-T also exhibit various symptoms ranging from insulin resistance, glucose intolerance, to growth retardation. Many of them also develop type 2 diabetes at an early age. However, the reason for progressive neuronal degeneration in A-T patients remains unclear. ATM is traditionally considered a nuclear protein and a common hypothesis is that defective nuclear function of ATM in response to DNA damage is responsible for neuronal degeneration of A-T. However, it is known that ATM is predominantly in the cytoplasm in human Purkinje cells and mouse cerebellum neuronal cells. Our previous findings have shown that cytoplasmic ATM is an insulin responsive protein that stimulates Akt phosphorylation. We also discovered that in response to insulin, ATM protects differentiated human neuron-like SH-SY5Y cells from serum starvation-induced apoptosis. Based on these results, we hypothesize that ATM, through activation of Akt, promotes neuronal survival in response to insulin and other neural growth factors. We have recently compared the functions of ATM in proliferating and differentiated human neuron-like SH-SY5Y cells. Our results clearly show that ATM switches function from a sensor of DNA damage in proliferating cells to a mediator of growth factor signaling in differentiated human neuron-like cells. Our results further demonstrate that ATM mediates Akt signaling and promotes cell survival in differentiated SH-SY5Y neuron-like cells, which suggests that impaired activation of Akt, rather than defective response to DNA damage, is the reason for neuronal degeneration in human A-T. The goal of this proposal is to further examine the functional link between the ATM protein kinase and neuronal survival in response to insulin and other neural growth factors. Findings from this project may provide novel insights into the role of ATM in the progressive cerebellar ataxia observed in patients with A-T. Furthermore, as many of the ataxia or neuronal degeneration disorders are closely related to insulin resistance and type 2 diabetes, our research may provide an overall better understanding of the ataxia and neuronal degeneration process and may lead to

the discovery of novel therapeutic strategies and agents not only for A-T but also for other ataxia or neuronal degeneration disorders.

YOUNG INVESTIGATOR AWARDS

Delineating the PUM1 functional network in mice and humans

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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 the disease-driving protein can cause the same disease as if the protein were mutated. What if we could find a way to lower the levels of these proteins in the brain? Could we slow disease progression? These questions led me to search for the factors that regulate the levels of ataxin1, the protein that is mutated in SCA1. I discovered that ataxin1 levels are controlled by an RNA-binding protein called Pumilio1 (PUM1). Take away PUM1 in a mouse model of SCA1, and ataxin1 returns to normal levels, and the SCA1 mice no longer have ataxia.

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. We reached out to medical geneticists to search for such patients and so far have identified 20 patients with PUM1 deficiencies: those with deletions or severe missense mutations that eradicate protein function suffer from a neurodevelopmental disorder that causes physical, cognitive, and/or psychosocial delay with seizures and ataxia, whereas patients with a hypomorphic mutation develop a late-onset disorder that causes a mild, slowly progressive ataxia. PUM1 is thus a newly identified genetic cause of two ataxia syndromes.

This proposal aims to (1) generate mice carrying the PUM1 mutations identified in humans to better understand the molecular pathogenesis of the human disease and (2) characterize the Pum1 protein-protein interaction network in the mouse brain in order to identify potential druggable targets. Not only is this necessary to understand how PUM1 deficiency causes early-onset ataxia and neurological dysfunction, but understanding the roles PUM1plays in neurons will help us understand whether manipulating PUM1 levels in the brain could help SCA1 patients (or, indeed, patients with other neurodegenerative diseases). As my preliminary data indicate that Pum1 regulates a number of proteins involved in other neurodevelopmental and neurodegenerative conditions, the proposed studies should reap a considerable return on the NAF's investment.

Uncovering pathogenic mechanisms of cerebellar atrophy in SNX14 deficiency

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Cerebellar ataxias are a heterogeneous group of disorders characterized by imbalance and poor coordination due to impaired function or damage to the cerebellum, which regulates balance and movement. The condition is often genetically inherited and affects ~9 per 100,000 individuals. Although more than 100 genes have been implicated so far, disease mechanisms for most of the cerebellar ataxias are still unknown and treatment options unavailable. This highlights the need for further studies that will provide better understanding of disease mechanisms and advance in treatment options.

Spinocerebellar ataxia autosomal recessive 20 (SCAR20), is a particular form of cerebellar ataxia diagnosed early after birth and presenting with progressive shrinking of the cerebellum, intellectual disability and dysmorphic facies. Our recent work identified that mutations in the gene Sorting Nexin 14 (SNX14) are the cause of SCAR20. SNX14 is involved in regulating the function of lysosomes, which are the cellular compartments responsible for the degradation of unwanted materials. Lysosome dysfunction has previously been associated with cerebellar degenerative diseases. However, it is not well understood how lysosome dysfunction leads to cerebellar damage.

In order to uncover how SNX14 mutations lead to lysosome dysfunction and cerebellar ataxia, we have generated a mouse model that shows impaired movement reminiscent of patients with SCAR20. Our current work aims to characterize the type of cerebellar damage present in these mice and to look for sign of lysosome dysfunction that may be present in their cerebellum. Future work will be focused on seeking for cellular materials that may toxically accumulate in SCAR20 cerebellum due to lysosome dysfunction, and on identifying routes to prevent cerebellar damage.

With this study, we expect to uncover disease mechanisms of SCAR20. Moreover, our research will help us better understand the relevance of lysosomes for the cerebellum and may reveal targets for the development of treatment options for a broader spectrum of spinocerebellar ataxias.

Edgotype: a platform to stratify variants of unknown significance ataxias

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Since the release of the first reference sequences of the human genome, incredible advances have occurred in the identification of gene mutations that cause ataxia. Next-generation genetic sequencing is now inexpensive enough to bring this diagnostic capability to clinical practice. This is a critical first step toward therapeutics because it allows patients to be stratified, and for treatments to be matched to specific genetic lesions. And, yet, daunting challenges remain. Our ability to sequence genomes is greatly outpacing our ability to usefully interpret the data.

Genes code for proteins, the building blocks of our cells. They are involved in chemical reactions, signaling, interactions with DNA and other proteins, as well as a host of other functions. Some gene mutations will lead to a clear loss of function

because no active protein is made, or the protein is made with a slight alteration. The alteration may profoundly affect protein function, or not affect it at all, and we are actually very poor at predicting which of these will occur. When these changes are present and there are no previous reports of this genetic change in association with disease, these are called variants of unknown significance (VUS).

Here, we propose a new method developed to look at how gene mutations affect the protein interactions within a living cell. We plan to apply our methods to a large number of ataxia patients. We expect that the data generated in this study will have direct impact on clinical care. First, it will shed light on determining if the variants are disease-causing and stratifying patients for clinical trials and potential therapies. Second, it may have the potential to increase knowledge about disturbances in protein-interactions in ataxias, leading to novel therapeutic targets. Finally, it may lay the groundwork for a new test that can be used in many other types of genetic diseases.

Elucidating the mechanism of novel spinocerebellar ataxia gene TMEM16K in endolysosomal maturation

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Mutations in the evolutionarily conserved endoplasmic reticulum (ER) protein TMEM 16K are causative for autosomal recessive spinocerebellar ataxia (SCAR10), a debilitating progressive neurodegenerative disease. I have discovered that TMEM16K acts as a critical mediator of endolysosomal maturation. Abnormal protein degradation due to endolysosomal dysfunction has been implicated as the primary contributor in multiple neurodegenerative diseases.

This proposal seeks to uncover the molecular mechanics of how TMEM16K and its interactome mediate endolysosomal maturation in aging neurons to provide insight into the pathophysiology of neurodegeneration and could open new avenue for therapeutics.

POST-DOC FELLOWSHIP AWARD

Identification of novel pathogenic tandem repeat expansions in spinocerebellar ataxia

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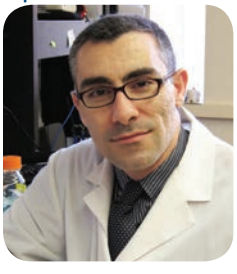
Many types of spinocerebellar ataxia (SCA) are caused by expansions of tandemly repeated stretches of DNA (or "TRs"). These are segments of DNA where multiple copies of a sequence are serially repeated dozens, hundreds or even thousands of times, e.g. CAG-CAG-CAG-CAG-CAG. The large size of many TR expansions in SCA patients makes them difficult to detect by conventional sequencing methods, such as PCR, or standard genome sequencing technologies that only look at short segments of DNA. As a result, the identification of novel expanded TRs that cause SCA is very difficult. To overcome this limitation, in this project, we seek to identify novel pathogenic TR expansions in patients with unexplained SCA using two novel approaches. First, we will study four candidate TRs we have identified in the genome that each show strong signatures of instability using an optimized Repeat-

Primed PCR method that can amplify expanded TRs. Secondly, we will perform whole genome sequencing to look for expanded TRs across the entire genome of individuals with inherited SCA using Pacific Biosciences long-read technology. This approach produces long sequencing reads (read length 10,000-15,000 bp) that, unlike short reads, are able to fully span even the very longest expanded TRs found in patients with SCA. In addition to providing an accurate molecular diagnosis to patients with unexplained SCA, we expected that the results arising from this proposal will contribute to expand our understanding about the mechanisms involved in the development of SCAs as well as to develop novel approaches for diagnosis, genetic counseling and follow-up.

YOUNG INVESTIGATOR-SCA AWARDS

Cellular Mechanisms Underlying Spinocerebellar Ataxia Type 13



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Spinocerebellar ataxia type 13 (SCA13) is an autosomal dominant genetic disease caused by mutations in the KCNC3 gene. This gene encodes the Kv3.3 voltagegated potassium channel, which plays an essential role in facilitating proper electrical activity in cerebellar Purkinje neurons. There are two identified SCA13 mutations and depending on the mutation, SCA is characterized by ataxia and cerebellar neurodegeneration during aging or persistent motor deficits and cerebellar mal-development starting early in life. We plan to test the hypothesis that SCA13 mutations have adverse effects on the electrical activity of developing Purkinje neurons that lead to their abnormal development. Changes in neuronal function have been reported in several neurodegenerative diseases, including those caused by toxic, misfolded proteins, but whether changes in neuronal electrical activity are involved in pathogenesis is unknown. SCA13 provides a novel opportunity to investigate the relationship between neuronal activity, development and motor control in the absence of other complicating factors such as misfolded proteins. We are using zebrafish to investigate these questions because zebrafish provides numerous technical advantages making it an attractive system to address our research interests. One important advantage is that neuronal function can be assessed using genetically-encoded calcium indicators to monitor changes in electrical activity that are due to the SCA13 mutations, and we can follow the development of the cerebellum in living animals.

Polyserine protein toxicity in spinocerebellar ataxia type 12

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Spinocerebellar ataxia type 12 (SCA12) is a rare neurodegenerative disease caused by a repetitive CAG sequence in exon 7 of PPP2R2B, a gene encoding regulatory units of the protein phosphatase 2A. Two ATG start codons upstream of the repeat may translate the repeat into a protein containing a polyserine tract, giving rise to short polySer tracts from the normal allele and long polySer tracts from the mutant allele. We therefore hypothesize that proteins containing long polySer tract may contribute to the pathogenesis of SCA12. PolySer neurotoxicity has never been studied in SCA12, or in any other expansion disease, and our hypothesis therefore represents a novel approach to neurodegeneration. To test this hypothesis, we

have successfully generated 8 SCA12 induced pluripotent stem cell (iPSC) lines from three different human SCA12 fibroblast lines. We have generated polyclonal antibodies that could recognize the predicted proteins containing the polySer tracts. Our preliminary evidence suggests that long polySer tracts are toxic to cells and its production may be triggered by repeat assisted non-ATG (RAN) translation. Based on these findings, and the tools that we now have available, we have developed two aims to begin systematically testing the polySer hypothesis of SCA12 pathogenesis.

In Aim 1, we will examine the expression of the predicted polySer-containing proteins in 8 SCA12 patient iPSC lines and three control lines, both in undifferentiated state and after differentiation into cortical neurons, using western blots and immunocytochemistry. We will also perform immunohistochemistry to detect polySer-containing protein expression in the one formalin fixed SCA12 patient brain that is available for our study, along with control, Huntington's disease (HD), and Alzheimer's disease (AD) brains. We will also determine if RAN translation contributes to the expression of long polySer containing proteins, by modifying an SCA12 iPSC line to prevent ATG translation of PPP2R2B exon 7.

In Aim 2, we will explore the hypothesis that expression of long polySer containing protein in SCA12 is neurotoxic, and we will test this using SCA12 iPSC models. We will use vulnerability to BDNF-withdrawal as the readout for toxicity. First, we will examine whether SCA12 cortical neurons differentiated from iPSCs are more vulnerable to BDNF-withdrawal induced toxicity, compared with the control lines. Second, we will determine whether interruptions in long polySer region modify toxicity, by comparing the vulnerability of SCA12 cells with that of an isogenic "interrupted" SCA12 line edited to mimic single amino acid replacement in long polySer region. Third, we will ascertain whether long polySer toxicity can be reproduced by polySer encoded by non-repeat RNA, by replacing the hairpin-forming AGC repeat in an SCA12 iPSC line with a TCC/TCT repeat.

This project will allow us to test for the first time the novel hypothesis that proteins containing long polySer tract may contribute to SCA12 pathogenesis, with potential relevance for other neurodegenerative diseases caused by repeat expansions.

Regulation of cerebellar development and connectivity by βspectrins: implications for the pathophysiology of spinocerebellar ataxias

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Spinocerebellar ataxia type 5 (SCA5) and spinocerebellar ataxia autosomal recessive 14 (SCAR14) are progressive neurodegenerative disorders characterized by cerebellar atrophy and profound Purkinje cell loss. In addition, patients with SCAR14 also present with cognitive impairment. Genetically, these disorders are caused by mutations in SPTBN2, the gene encoding the protein βI-spectrin, which is highly present in the cerebellum. Since the discovery of the genetic cause of SCA5 over ten years ago the number of βI-spectrin mutations found in patients affected with these ataxias has grown, but our understanding of the underlying cellular mechanisms has lagged and promising treatments for the disease continue to evade us.

We have new evidence that suggests that βI-spectrin's deficits affect the normal function of

β-spectrin, another member of this protein family. β- spectrin along with βI-spectrin, actin, and other partnering proteins appear to cooperate to build a periodically organized, structurally robust tubular membrane skeleton in neurons. Simultaneously, these spectrins seem to modulate intracellular transport of multiple synaptic components. In turn, we believe that the combined structural and transport defects caused by βI-spectrin lead to severe neuronal dysfunction in the cerebellum.

In this project, we will characterize mouse models of β- and βI-spectrin-deficient cerebella, and conduct cellular studies of βI-spectrin with selected SCA5 human mutations to understand the functional duality of βspectrins as scaffolding proteins and transport effectors. We hope to integrate this knowledge with previous findings into a comprehensive mechanism that explains the pathology of SCA5 and SCAR14, and serves as roadmap for judicious design of therapies.

Longitudinal Profiling of Spinocerebellar Ataxia Type 3 Mouse Model's Molecular Signatures and Specific Biomarkers

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University of Michigan
Ann Arbor, MI

Spinocerebellar ataxia type 3 (SCA3), the most common dominantly inherited ataxia in the world, is a relentlessly progressive and fatal disease for which currently there is no disease-modifying therapy. With the goal of preventive therapy, several ongoing studies seek to reduce levels of the SCA3 disease protein. Leveraging tissue collected from our recently published gene silencing preclinical study in SCA3, I will longitudinally assess the molecular events in disease-relevant brain regions from two complementary SCA3 mouse models. This global gene profiling over time, the first to be completed in these models, will help identify the molecular events underlying disease and may elucidate protective pathways and biomarkers for SCA3.

PIONEER TRANSLATIONAL SCA AWARDS

Exploring the therapeutic capacity of Aripiprazole and related compounds for Machado-Joseph disease

Maria do Carmo Costa, PhD

University of Michigan
Ann Arbor MI

Disease-modifying therapies are lacking for fatal neurodegenerative disorders including Machado-Joseph disease (MJD), also known as Spinocerebellar ataxia type 3 (SCA3). Our long-term goal is to identify small molecules that are effective to reduce the abundance of toxic ATXN3 protein in brains of MJD patients/carriers and hopefully alleviate disease progression. This project aims 1) to evaluate the potential of aripiprazole to be repurposed for MJD by carrying out a chronic pre-clinical trial of this drug in MJD transgenic mice, and 2) to develop novel therapeutic compounds structurally related with aripiprazole for this disease that are effective to decrease levels of mutant ATXN3 in neurons. Because aripiprazole is already commonly used in humans, a positive trial outcome shown by reduced ATXN3 levels in brains of MJD transgenic mice chronically treated with this drug, and/or by decreased progression of motor dysfunction in these mice, would accelerate the path to clinical testing of aripiprazole in MJD patients. To develop novel therapeutic compounds for MJD, we will carry out structure activity

relationship studies building on chemical structures of aripiprazole and two related molecules newly identified by us as efficacious to reduce mutant ATXN3 abundance in a MJD cell line. By tackling two different stages of drug discovery for MJD, we expect to identify a fast route for therapy for MJD patients and/or to develop novel compounds of increased therapeutic potential for this disease.

Combinatorial Gene Therapy for Spinocerebellar Ataxia Type 1

Beverly Davidson, PhD

Children's Hospital of Philadelphia
Philadelphia, PA

There are currently no therapies that delay onset or progression of spinocerebellar ataxia. In earlier work, we showed that gene silencing approaches or gene over-expression approaches delivered individually had a profound positive impact on disease readouts in two animal models of spinocerebellar ataxia type 1 (SCA1). Additionally, our gene silencing therapies in SCA1 mice reversed behavioral deficits and neuropathology, even when delivered after onset. These data are the foundation for future clinical application of gene silencing studies in SCA1 patients. Here, we propose to expand and improve on this work by testing a combinatorial approach. The goal of this newer approach is to reduce the overall dose of material needed to achieve therapeutic benefit. If our tests in mice are successful, we will seek additional funding to move this forward to SCA1 patients.

Launching a blood and spinal fluid based biomarker program for SCAs

Puneet Opal, MD, PhD
Northwestern University
Chicago, IL

There is a critical need for establishing objective, reliable and sensitive biomarkers will enhance our understanding of the natural history of spinocerebellar ataxias (SCAs), provide insights into pathophysiology, and most importantly improve our ability to perform well-powered interventional clinical trials using fewer patients and shorter time-lines. Current biomarker strategies in neurodegeneration fall into two broad categories (1) those based on brain imaging and (2) those based on detecting biochemical differences from bodily fluids such as CSF or blood.

In this proposal we will take the lead in establishing a comprehensive biochemical biomarker program for the National Ataxia Foundation. This will involve establishing the operational infrastructure that will include regulatory materials (IRB, consent forms and standard operating procedures) and facilities for the collection and storage of blood and CSF. We will align the biomarker program with the NAF funded natural history studies that are being conducted at the CRC-SCA sites. Patient materials will be linked to the existing web-based CRC-SCA database to serve as a resource to the CRC-SCA consortium of investigators.

The study is thus designed as operationalizing a biorepository linked to observational, prospective, multi-center natural history study linked to the CRC-SCA. Although, the request is for one year, we are launching this initiative with the intention that it will become an open-ended, prospective study and will be self-supporting in the future by serving as a platform for clinical trials funded by disease foundations and philanthropy (including NAF), government funding agencies (such as the NIH), and pharmaceutical companies.)

Ataxia Tips

Hints and Tricks for Ataxians...from Ataxians

For many with Ataxia, swallowing can become increasingly difficult. One person's shared tip about swallowing in our enewsletter prompted a flood of different strategies from others. See what everyone had to say about ways to help with swallowing...

1. I have ILOCA with spasticity. I use coconut yoghurt to swallow my pills. It works better than apple sauce for me. (Submitted by Vera Olcott)

2. Tilt your head and chin forward if you feel like you might start choking. (Submitted by Mary Oshaughnessy)

3. I often have trouble swallowing liquids and find it easier to drink when I use a straw. (Submitted by Mary Mattison)

4. I have trouble swallowing medication and my doctor recommended a throat specialist to me. He suggested that I swallow my pills using yogurt instead of water, because water can possibly go down the wrong tube and that's bad for you (Submitted by Dave Roy)

5. Put your head down to drink water. Put your pills in pudding. Use thickener for drinks. Cut food into small pieces and eat a little at a time. Don't use straws. My husband has a severe swallowing problem and this is what he has been told to do by therapist. (Submitted by Cathy Schall)

6. On taking pills: those that are gel type can be frozen. Even big ones go down the hatch easily as my three-year olds demonstrated a year ago. (Submitted by Pam Wetzels)

7. Take pills with Ensure, or at least, tip your head forward if taking pills with water. (Submitted by Virginia)

8. Taking pills while eating a banana will make them go down without the pills lodging in your throat. (Submitted by Frank Muellersman)

For the next issue, submit your tips on Falls and Fall Prevention.

1. Ataxians usually fall backward. Tuck your chin to avoid hitting your head. Better yet, don't fall. (Submitted by Mary Donahue)

***Ataxia Tips must be submitted by 5/21/18 to be eligible for inclusion in the next issue of *Generations*.**

Submit them via email to naf@ataxia.org.

WALK N'ROLL TO CURE ATAXIA

National Ataxia Foundation

What is Walk N' Roll?

It is NAF's largest grassroots fundraising event! Walk N' Roll to Cure Ataxia currently takes place in cities across the U.S. Since its inception in 2007, Walk N' Roll has raised more than $2,680,000 thanks to support and tireless commitment from walkers, rollers, runners, volunteers, donors, and sponsors.

Why Walk N' Roll?

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

How Can I Participate?

For more inforamtion, or to start a Walk N' Roll in your community, please contact Lori Shogren, NAF Community Programs and Services Director at 763-553-0020 or **lori@ataxia.org**.

For more info and to find Ataxia events visit **ataxia.org/events**

2018 GO THE EXTRA MILE 2 Cure Ataxia

The FLEXIBLE way to help raise funds that benefit the Ataxia community!

NAF National Ataxia Foundation

Let's Go The Extra Mile!

Last year, in celebration of 60 years of NAF operations, dozens of people walked 60 miles in a fundraising effort for NAF. For 2018, we are going to "Go the Extra Mile."

While some people walked 60 miles over a three day weekend, others completed their 60 miles over several weekends; others usded a week's vacation to camp and hike their 60 miles at their own pace. This program is flexible! How would you like to do it? It's up to you!

ABOUT THE PROGRAM

Who?

Anyone! Have friends and/or family join you in this effort...more people equals more memories.

When?

Pick a time of year, commit to it, announce it, and begin fundraising as soon as you can. This year we are asking participants to make a $5,000 commitments.

How?

Pick a team name and NAF will create your fundraising page for you! The most efficient way to raise funds is through an online letter writing campaign asking for support from your friends and family.

NAF Can Help!

Given ample time, the NAF office will distribute a press release in your area seeking local publicity for your efforts. The NAF Development Office will work with you on the other ideas to supplement your fundraising efforts. Just give us a call...

"I proved to myself that I am stronger than I thought I was, and that I am capable of doing so much more."
- Joni Lahr-Moore 2017 60 for 60 Participant (pictured left)

NAF Rated 4-Stars by Charity Navigator For 5th Time in a Row

We're excited to announce that NAF has received the top rating by Charity Navigator – the largest independent evaluator of charities – for the 5th time in a row!

Why is that a big deal? Because only 9% of charities evaluated have received at least five consecutive 4-star ratings!

The coveted 4-star rating means that Charity Navigator has recognized NAF's commitment to accountability and transparency. It means that we provide our donors with the information that they need to be confident that their money is spent responsibly; contributing to the research, support, and education that our mission proclaims. We are proud to continue to earn the trust and support of our donors – and will continue to do so in the years to come. Thank you to our supporters! You are the reason we do what we do. Together, we will find the cure.

Check out the full report at
https://www.charitynavigator.org/index.cfm?bay=search.summary&orgid=8543

A Cherished Life: Tribute to Denise Drake

There are some people who seem to have a contagious demeanor — they spread joy to every life they touch. Denise Drake was one of those people, as evidenced by the loving declarations from those who knew her. Despite facing a horrendous disease and bleak outlook from her doctors, Denise approached life with a fierceness and positivity that inspired all around her. Denise Drake passed away in January of 2018, surrounded by friends and family, and ushered on with a flood of well wishes on social media. It is with great sadness that we bid her farewell and look over her life's contributions.

At the tender age of 12, Denise's doctors diagnosed her with Friedreich's Ataxia, bluntly informing her that she may never have children – and worse, could die from the disease. Yet, faced with this devastating news, Denise's response was to find the silver lining. She wrote, "I feel lucky, I guess, for not having to endure years of misdiagnoses and ridicule, as many have." She decided that she would not allow something that happened to her, that she could not control, dictate her life. She fought back.

Denise Drake quickly became a pillar of the Ataxia community. First, becoming involved in a local NAF support group – then going on to become a longtime President of NAF's LA Chapter and respected member of the Board of Directors to the national office. Denise was an integral part of building the National Ataxia Foundation into the respected institution that it is today. Denise, speaking about her involvement with NAF, once wrote, **"I wanted to share experiences, truth, and hope, and learn about my disease at the same time."** She served tirelessly, remaining an active member of the Board of Directors until she took her last breath. Her contributions will not be forgotten.

Read Denise Drake's published article, "Finding balance" from The Lancet at http://bit.ly/DeniseDrake

Fellow Board Members were quick to remember her vigorous efforts throughout the years with an outpouring of thanks and memories. NAF Board Member Harold Crawford noted that she endured through the early days of the Foundation "when all we had to work with was dedication." NAF's Research Director reached out with sentiments shared by the Medical Research Advisory Board, pointing out that Denise's long battle with Friedreich's Ataxia and devotion to NAF reminds them that their dedication as researchers and clinicians to aid those who confront Ataxia is important every day of the year – for Denise and those like her.

Denise's impact was not limited to NAF. She served as an avid advocate in her community as well, one time even suing (successfully) to obtain wheelchair accessibility at precinct voting sites. Now, thanks to Denise, more than two dozen polling sites in her community comply with ADA requirements. There are countless other examples of the positive changes that Denise created. While Denise's fight may have concluded – her legacy lives on.

Support Group News

Arizona Ataxia Support Group

Submitted by Mary Fuchs

The Arizona Support Group held a meeting on Saturday, February 3rd. It was held at Ability 360, Classroom B, 5025 E. Washington Street, Phoenix, AZ 85034. The meeting opened welcoming everyone, plus 2 new members. We had a short information meeting.

Our GUEST SPEAKER was Jerry Ketelhut from "Daring Adventures". He showed a short 4 minute video and also a Power Point presentation. The presentation covered their mission, history and the outdoor recreation and services they provide to the community. All activities and trips are ADA accessible. Also he told us how people can get involved with them and volunteer. We had time for questions and answers at the end. We closed the meeting with a SPECIAL tribute to Jolanta Chyla, who passed away January 21st, 2018.

Central PA Ataxia Support Group

Submitted by Charlotte Depew

The Central PA Ataxia Support Group, a member of the National Ataxia Foundation, is excited to be chosen as a participating organization in Pennsylvania College of Art & Design's 10th annual

Central PA Support Group

Designathon, which will be held on February 23 and 24, 2018. During the Designathon's 24 hours, teams of students and faculty mentors create all-new pieces for eight non-profits. Learn more at PCAD.edu/news.

Greater Atlanta Ataxia Support Group

Submitted by Greg Rooks

The Greater Atlanta Ataxia Support Group meet on Saturday February 17, 2018 at the Rush Center Annex Building. There were approximately 30 people in attendance with several first timers. A special welcome to those attending their first meeting. Our guest speaker was Denae Morris, representative with Tobii Dynavox and she demonstrated the I-12+. The I-12+ is a tablet device that enables effective augmentative and alternative communication in all forms. The device can be controlled by touch, switch, or gaze interaction. Other topics discussed were the Annual Ataxia Conference and meeting dates for 2018. Our next meeting is scheduled for April 21, 2018 at the Dunwoody Library.

Greater Denver Area Ataxia Support Group

Submitted by Michael Cammer

The Greater Denver Ataxia Support Group met at Swedish Medical Center on Saturday, January 20, 2018. Of the thirty or more who attended, nine individuals came for the first time. Active visiting happened during our potluck meal and after. Charlotte welcomed everyone and gave each an opportunity to introduce themselves, state their ataxia type,

and anything else they wished to talk about very briefly.

Dr. Trevor Hawkins from The University of Colorado (UC) Neurology Department was our guest speaker and gave a brief overview of current research foci and findings. Excitement is currently in genetic modification. In addition, UC's Ataxia Clinic's research is expanding to include historical data collection in several SCA's. After his talk, many attendees spoke with Dr. Hawkins regarding research and data collection involvement. These are the beginning of exciting times for ataxia patients and ataxia research particularly for those who have a Denver access. The next meeting is at Swedish Medical Center, April 21, 2018 from 1:00 to 4:00 PM. The speaker will be Ellen Belle, a physical therapist at Colorado Neurological Institute. She will discuss the weighted balance vest and demonstrate it on anyone who volunteers.

India Ataxia Support Group (SAMAG)

Submitted by Chandu George

Hello all. I want to share that I received yet another prestigious award recognizing my NGO leadership efforts by bringing in positive changes to society in my country, India.

The event was organized by World CSR Day. I was presented with a lovely award. My acceptance speech spoke on three topics which are always close to my heart – Ataxia Awareness, Rare Diseases, and Accessibility for wheelchair users in our society.

The event took place at the wonderful hotel Taj Deccan, Hyderabad which had a great atmosphere. I was able to meet and get to know many people from different walks of life: HR Managers, Entrepreneurs'-Start-Ups, NGO Leaders, Academic Teachers, Doctors, Researchers, etc. The setting reminded me of wonderful memories when I was lucky to attend NAF AAC meeting in Denver, 2015.
Would not say this is another feather in my cap, but the Real support I get is from my family. Thanking all.

India Support Group

Indiana Ataxia Support Group

Submitted by Cheri Bearman

The Indiana Ataxia Support group held their first official meeting on Saturday, November 11, at the St. Vincent Fishers Hospital in Fishers IN. The meeting was attended by nine members of the group.

Amy Draves coordinated the meeting and provided a folder filled with information on Ataxia for each person in attendance. Cheri Bearman, group leader, gave a short welcome. Then, each person shared a little bit about themselves, their Ataxia, and what they are doing to cope with their Ataxia symptoms. After the meeting, a carry-in lunch and social time were enjoyed by all.

Indiana Support Group

Indiana Ataxia Support Group

Submitted by Cheri Bearman

The Indiana Ataxia Support group held their second meeting on Saturday, January 20, at the St. Vincent Fishers Hospital in Fishers, Indiana. The meeting was attended by eight members of the group (Kevin Draves is not pictured).

Amy Draves and Teresa Coccaro coordinated the meeting. Cheri Bearman, group leader, gave a short welcome. Since two new people attended this meeting, the first hour members enjoyed getting to know one another, sharing their Ataxia stories, and explaining what they are doing to cope with Ataxia. The group then took a break for a delicious carry-in lunch and a social time.

The last hour of the meeting, Cheri shared some important information about joining the NAF, signing up for the CORDS registry, and getting involved in research opportunities. She also asked for volunteers to represent the NAF at the second annual Humanifest event to be held on March 24. This Indianapolis event promotes health and well-being and raises Ataxia awareness and funds for research.

Cheri also gave a short report on the first ever Walk n' Roll event in Indiana which took place October 7, 2017 in Fort Wayne. The event raised a total of $8,634.25!! Cheri gave a special "thank you" to Jessica Lebrato who organized the Walk n' Roll. Jessica announced that Fort Wayne's second annual Walk n' Roll will take place on Saturday, October 13, 2018.

As the meeting came to a close, group members volunteered to schedule the following professionals to share at upcoming meetings: a genetic counselor, a speech pathologist, an adaptive yoga teacher, and a service dog representative.

Seated L. to R. Tom Dobey and Jessica Lebrato, Standing L. to R. Mick and Chris Cozmanoff, Amy Draves, Teresa Coccaro, and Cheri Bearman

Los Angeles Ataxia Support Group

Submitted by Lora Morn

Dr. Fogel joined us and presented information for one of our support group meetings. The meeting was a huge success! We even had a family on vacation from London – a brother and sister that have Ataxia.

Los Angeles Support Group

Northern California Ataxia Support Group (NCASG)

Submitted by Shirley Hanks

Our October meeting featured "Transitions with Care," presented by Steve Villa. Steve helps families with loved ones that can no longer live safely at home to find a new living situation. Steve reviewed different types of living community options.

Types of communities:

- Independent living community –No assistance with activities of daily living (dressing, bathing, grooming) is included. Many communities provide meals in a communal setting. Housekeeping and laundry may be offered.
- Assisted living – Residents get their own apartments (studio, 1 bdrm or 2 bdrm) within a larger building. Caregivers are on staff, 24 hours a day. Assistance with activities of daily living (ADL)

is available. Residents tend to be more active and can manage some ADL on their own.

- Residential care homes (RCFE) – These are homes in residential communities. Usually they are licensed for 6 residents and have 2 caregivers on staff. The residents tend to be older and less active.
- Memory care – This is usually a section within a larger assisted living community. There is a higher staff to resident ratio than the other section.

Northern California Support Group

Northern California Ataxia Support Group (NCASG)

Submitted by Brian Wong

Alan Acacia, a vibrant member of our Northern California Ataxia Support Group, passed away unexpectedly from lung cancer on December 26th, 2017. He was 69 years old. Alan was often seen smiling, laughing, and socializing with people at our meetings. Yet, there was so much more to Alan than the man we knew from the quarterly meetings. Writing was very important to him. He was most proud of having written poetry for over 50 years. He taught philosophy and creative writing at a Franciscan seminary college. He worked for 4 years in an agency that promoted positive roles for men in family planning. He was a senior salesman for 10 years at a Japanese import company. He also worked as a concierge at the Oakland Marriott. Most recently, he volunteered to teach English as a Second Language to Chinese students around his Berkeley neighborhood.

Alan is survived by his brother and his sister. As you can see, Alan lived a full life and involved himself in a variety of experiences, however he rarely shared the details of his life with many people. I feel honored to have been able to get to know him. He was a great friend and will be dearly missed.

Alan Acacia

Sioux Empire Ataxia Support Group

Submitted by Mary Beth Farley

At the last meeting, Mary Beth Farley opened the meeting by introducing participants. Mary Beth said she would like to have a social type get together every other month and suggested Saturday, February 10th to meet over lunch. Discussion was held about having a Walk & Roll fundraiser in the local area.

Matt Leedom, a Physical Therapist with the Avera Orthopedic Institute and Board Certified in working with neurological conditions gave a presentation on "Exercising with Ataxia." He provided a handout that covered why it is important to keep exercising and the various types of physical activities. The participants with Ataxia agreed that exercising in the pool was great therapy. Fatigue is a problem. It is a challenge to find a physical therapist to help you with your specific needs.

Discussion was held about parental guilt and the hereditary aspects of Ataxia and the only way to stop it is to not have children. Matt ended his presentation by saying that safety is the key. It is

Sioux Empire Support Group

important to find an environment where you feel safe and confident. He also suggested finding a buddy to work out with.

Tampa Bay Ataxia Support Group

Submitted by Jan Colon and Sharon Farley

On January 20, 2018, Tampa Bay held their first support meeting of the year at the USF Morsani building. This is arranged by a USF Neurologist, Theresa Zesiewicz, MD, who specializes in Ataxia and is most compassionate to our cause.

The speaker that Darlene had scheduled to speak with the caregivers was unable to attend due to illness. Therefore, we had ample time to meet & greet and to discuss various subjects of interest. Some of the discussions included pool exercises and their benefits. One of the auxiliary aides (provided by Dr. "Z"), whose name is Dan, informed us that hotel pools will often allow the use of their pools and that it is law for public facilities to have disability accessibility. We mused over whether to have this activity as a fellowship time. Another attendee, Frank Fountas who accompanied his Ataxian wife Sue, suggested a bi- weekly meeting where healing would take place because of relationship and laughter. Everyone seemed very excited by that!

We also discussed the process of obtaining a motorized scooter thru insurance and opinionated about different fellowship activities while stressing the importance of everyone contributing their time, talents & abilities to any and all NAF events & fundraisers as research availability was for us

Tampa Bay Support Group

ALL! All things considered, it was a very fruitful meeting and we learned much about each other and the opportunities available.

Treasure Coast Ataxia Support Group

Submitted by Lisa Cole

The support group meeting was held on Saturday December 9th at the Port St Lucie Community Center. We had nine people and started out with introducing ourselves. Same as last year's meeting, in December we had everyone who came and who wanted to participate bring a wrapped gift. When they left, they took one. Happiness was shared by all!

Our guest speaker was Jan Field-Byrne. She spoke about Pilates and Micro-Movements. The presentation was very informative. We would greatly love to have her back again next year.

Lastly, a list of the 2018 tentative dates for our Ataxia Support Group Meetings are all on a Saturday, they are April 21st, June 2nd, August 4th, October 6th and December 1st. We will have one of the above dates in Palm Beach Gardens in addition to there will be a joint Support Group Meeting in Orlando, either August 18th or 24th.

I do want to say "Thank you!" to Helen and Carly Magnuson for my lovely orchid.

I want to thank everyone, Lisa Cole

Tampa Bay Support Group

Treasure Coast Ataxia Support Group

Submitted by Jackie Hernandez and Lisa Cole

The Treasure Coast Ataxia Support Group met on Saturday, February 3 from 1-4:00p.m. in Port St. Lucie, Fl. There were 7 people who attended the lunch meeting in the Port St. Lucie Community Center. Our special guest speaker

was Jake McCandless who is a personal trainer. Jake demonstrated various exercises to help with balance and strengthening. He recommended various inexpensive equipment and household items such as suspension trainers, resistance bands & grippers. We discussed The Treasure Coast 1st Annual Walk n' Roll to Cure Ataxia that will be held on Saturday, May 19, 2018 in Tradition-Square and Lake, 1087 S.W. Tradition Square Port St Lucie, Fl. Registration for the event is at 7 a.m. and the Walk N' Roll will commence at 8 a.m. We also discussed how important it is to spread the word about the event with friends, family and social media. Our goal is to raise awareness and fundraise in support of NAF. Lisa shared a basket of hand painted "Ataxia Rocks" for us to take and place in special

Treasure Coast Support Group

places to help raise awareness. Our next meeting will be held in North Palm Beach from 12:30 p.m.-3:30 p.m.

Treasure Coast and Tampa Bay Ataxia Support Joint Meeting

Submitted by Jannete Colon

The Tampa Bay and The Treasure Coast Ataxia Support Group had a Joint meeting in Orlando at The Southwest Library Community Room, December 2, 2017. We met new members from Orlando.


Tampa Bay Support Group

Our guest speaker, Dr. Subramony, talked about the research he's been working on with other colleagues and also answered our questions. It was a very informative meeting.

We will do another joint meeting next year.

Willamette Valley Ataxia Support Group

Submitted by Jason Wolfer

On January 7, 2018, ten of us met (3 were new members and it was great to have them)! Thanks to Tyler and Casey, who arranged and hooked up the group for a chat via Skype with Dr. Perlman who works at UCLA and is the Neurologist for the National Ataxia Foundation. I felt the time was well spent and I hope that those who were there did as well.

Willamette Valley Ataxia Support Group

Submitted by Jason Wolfer

Hey Everyone! We had a good meeting on February 25, 2018, as ten of us met together to hear from a PY student named Michelle. The emphasis was on movement no matter our situation, and to challenge ourselves. It was a good reminder to me to not take shortcuts or compromise in areas of normal daily tasks because they have become more difficult. As we struggle with our challenges, it's good for us to keep these kinds of issues and encouragements before us...and I appreciated the reminder about our well-being yesterday...and I hope those of you who attended did as well.

Have a great story to share from the 2018 Annual Ataxia Conference?

Submit it by May 21st to be included in the Summer issue of *Generations*. Email your photos and stories to *naf@ataxia.org* today!

NAF Staff Directory, Directory of Chapters, Support Groups, Social Networks and Ambassadors

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

If you or a 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 or (763) 553-0020.

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.

NAF Staff Directory:

Jerry Frick Director of Finance and Operations Manager	(763) 231-2745 *jerry@ataxia.org*
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Stephanie Lucas, Communications Manager	(763) 231-2744 *stephanie@ataxia.org*
Lori Shogren, Community Program and Service Director	(763) 231-2743 *lori@ataxia.org*
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Joel Sutherland, Director of Development	(763) 231-2748 *joel@ataxia.org*
Mollie Utting Administrative Coordinator	(763) 231-2741 *mollie@ataxia.org*

Social Networks

NAF Facebook Page
www.facebook.com/ataxiafoundation/

NAF Facebook Group
www.facebook.com/groups/NAFmail

NAF YouTube Channel
www.youtube.com/user/NatlAtaxiaFound

NAF Twitter
www.twitter.com/NAF_Ataxia

NAF LinkedIn
www.linkedin.com/company/nationalataxiafoundation

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*** **or (763) 231-2742**.

Chapters, Support Groups and Ambassadors

Please note: Hometown of each Support Group Leader or Ambassador is noted below. For group meeting locations please refer to the Support Group Calendar of Events

Alabama

Alabama Support Group Leader
Becky Donnelly-Hover, AL
(205) 987-2883
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Facebook Page: *https://www.facebook.com/alataxia/*
Facebook Group: *https://www.facebook.com/groups/154027955194806/*

Ambassador
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Arizona

Phoenix Area Support Group Leaders
Angela Li-Peoria, AZ
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Facebook Group:
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Ambassador
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Arkansas

Ambassadors
Judy and David King-Springdale, AR
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S.G. Website: *http://orangecountyataxia.org/*
Facebook Group: *https://www.facebook.com/groups/1980393985511432*

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Tampa Bay Support Group Leader
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Treasure Coast Support Group Leader
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https://www.floridastreasurecoastataxiasupportgroup.club/
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Mississippi Chapter President
Camille Daglio-Hattiesburg, MS
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Kansas City Support Group Leaders
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INTERNATIONAL SUPPORT GROUPS AND AMBASSADORS

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Facebook Group:
www.facebook.com/groups/1468963499991380/

India Support Group Leader (Samag)
"Seek a Miracle Ataxia Group"
Chandu Prasad George
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Mobile: 0091-9989899919, 0091-9885199918
E-mail: *sam_ataxiaindia@yahoo.com*
Facebook Group: *https://www.facebook.com/groups/135999194491/*
SG E-mail: *india.ataxiagroup@gmail.com*

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Participants Needed for Research in **Spinocerebellar Ataxia**

CLINICAL TRIAL READINESS FOR SCA1 AND SCA3

-READISCA-

DO YOU MEET THE FOLLOWING STUDY CRITERIA?

- You have the clinical or genetic diagnosis of spinocerebellar ataxia type 1 or type 3 (SCA1 or SCA3) **OR**
- Diagnosis of SCA1 or SCA3 in one of your first degree relatives

For example

- You have early ataxia and your parent has SCA1 or SCA3
- You have no symptoms but you were tested positive for SCA1 or SCA3
- You have no symptoms and have not taken DNA testing, but your sibling tested positive for SCA1 or SCA3

If so, then you may qualify for participating in this international multi-institutional study. Please note that various restrictions may apply for the eligibility.

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 clinical trials for spinocerebellar ataxias.

IRB# Pro00022607

If you are interested and would like to have further information, please contact:

National Ataxia Foundation
Sue Hagen, Patient and Research Services Director

Phone: 763-231-2742

Email: susan@ataxia.org

Please note that there are 18 study sites across the US where you can participate in this project

- This is not a treatment trial. Rather, this study is to get ready for treatment trials we anticipate within 5 years.
- This research visit will likely take about half a day. You will be asked to return annually for the next five years.
- We will draw blood and perform DNA testing to confirm your genetic diagnosis, and if you wish to know your gene status, we will release the DNA results to your doctor or genetic counselor at no cost to you.
- You will be asked about optional spinal fluid collection by spinal tap (you can say "no" but the spinal fluid is extremely important for developing new drugs for SCAs).
- If qualified, you will be asked to participate in an imaging study using an MRI machine in Boston, Baltimore, Minneapolis or Gainesville (FL).
- There will be no cost for participation, and all expenses will be paid.

HOUSTON Methodist® NEUROLOGICAL INSTITUTE

2018 ATAXIA SUPPORT GROUP CALENDAR OF EVENTS

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

Why Attend an Ataxia Support/Social Group?

Many of you may ask, "Why should I attend a support group meeting?"
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.

Attending a support group meeting may give you a glimpse into the many different stages and types of the disease. This can help by using some of the strategies that have been beneficial to others in order to avoid and/or plan for some of the same challenges that others have faced in the progression of their Ataxia.

Hopefully attending a support group meeting will leave you with a sense of hope and inspiration, knowing that if others can cope, so can you. **Come. Learn. Share. But most of all, know that you are NOT alone.**

SUPPORT GROUP MEETINGS

Saturday, April 14, 2018

Mid-Atlantic Social Group Luncheon

Time: 8am-5pm
Location: Columbia, Maryland
Details: Ask the Lawyer, Ask the Doctor. A program with lunch provided that includes presentations and a large Q & A session segment on various topics on cerebellar Ataxia, estate planning, advance directives, living wills, trusts, and disability benefits including applications and appeals. Presenters include Liana Rosenthal, MD, Director, Ataxia Center; Director, Clinical Core of the Morris K. Udall Centers of Excellence for Parkinson's Disease Research and Asisstant Professor of Neurology; Mary E. O'Byrne, Esq., O'Bryne Law, LLC, Lutherville, MD, and Richard Neuworth, Attorney at Lebau & Neuworth, Towson, MD.
More Info: To register, contact Donna DeLano Neuworth at *ddeleno1@jhmi.edu* or 410-616-2811

Central Minnesota Ataxia Support Group Meeting

Time: 9:45am-11:45am
Location: Harvest Bank, 24952 County Rd 7, St. Cloud, MN 56301
More Info: Marsha Binnebose at 320-248-9851 or *mbinnebose@hotmail.com*

North Texas Ataxia Support Group Meeting

Time: 10am-12pm
Location: Ben Washington Baptist Church –Rev Jr Sheppard Educational Center, 615 Davis St., Irving, TX 75061
Details: There is lots of parking and it is handicap accessible. The meeting room is in a separate bldg from the church.
More Info: David Henry at *cheve11e@sbcglobal.net*

Kansas City Ataxia Support Group Meeting

Time: 12pm-2pm
Location: KC Public Library – Trails West Branch, 11401 E 23rd St., Independence, MO
More Info: Stephanie Wilkins at 816-623-3318 or *sfwilkins@yahoo.com*

Sioux Empire Area Ataxia Support Group

Time: 3pm-5pm
Location: Bennington Hills Apartments – Community Room, 5100 S Nevada Ave., Sioux Falls, SD 57108

More Info: Mary Beth Farley at (605) 941-2913 or *englishteacher1116@gmail.com*

Saturday, April 21, 2018

Twin Cities Ataxia Social Group Meeting

Time: 10am-12pm
Location: Langton Place, 1910 W. CTY. RD. D., Roseville, MN 55112
Details: 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 every month at 10:00am for 2 hours 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 at 1910 W. CTY. RD. D., ROSEVILLE, MN 55112. 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!
More info: Lenore Healey Schultz at 612- 724-3784 or *schultz.lenore@yahoo.com*

Treasure Coast Ataxia Support Group Meeting

Time: 12:30pm-3:30pm
Location: Florida Movement Therapy Center, 8645 N Military Trl #401, Palm Beach Gardens, FL 33410
More Info: Lisa Cole at *lisacoleataxia@gmail.com* or 772-370-3041

Sacramento Ataxia Support Group Meeting

Time: 1pm-3pm
Location: UC Davis Medical Center, The Lawrence J. Ellison Ambulator Care Center Building, 4860 Y Street, 3rd Floor, Conference Room 3030C, Sacramento, CA 95817
Details: We meet on the third Saturday of each month.
More Info: Teresa Bredberg at 916-215-2686 or *tbredberg@sbcglobal.net*

Denver Ataxia Support Group Meeting

Time: 1pm-4pm
Location: Swedish Medical Center, 501 E Hampden Ave, Englewood, CO 80113
Details: We meet in the 2nd floor Conference Rooms.
More Info: Charlotte DePew at 720-379-6887 or *cldepew77@comcast.net*

Greater Atlanta Ataxia Support Group Meeting

Time: 1pm-3pm
Location: Dunwoody Library, 5339 Chamblee-Dunwoody Rd., Dunwoody, GA 30338
More Info: Greg Rooks at 404-822-7451 or *rooksgj@yahoo.com*

Orange County Ataxia Support Group Meeting

Time: 2pm-4pm
Location: Orange Coast Memorial Hospital Medical Center, 18035 Brookhurst St, Fountain Valley, CA 92708. Pacific Coast Ballroom.
More Info: Cindy DeMint at *cindyocataxia@gmail.com*

Sunday, May 6, 2018

Willamette Valley Ataxia Support Group Meeting – Portland Location

Time: 3pm-4:30pm
Location: Capitol Hill Library, 10723 SW Capitol Hwy., Portland, OR 97219
More info: Jason Wolfer at *wolferjason@gmail.com* or 503-502-2633

Thursday, May 10, 2018

Tri-State Ataxia Support Group Meeting

Time: 6:30pm-8:30pm
Location: Mount Sinai Beth Israel Medical Center – Phillips Ambulatory Care Center – Conference Room 3, 10 Union Square East, New York, NY
More info: Kathleen Gingerelli at *dkgingerelli@msn.com* or 201-681-7639

Saturday, May 12, 2018

Central Minnesota Ataxia Support Group Meeting

Time: 9:45am-11:45am

Location: Harvest Bank, 24952 County Rd 7, St. Cloud, MN 56301
More info: Marsha Binnebose at 320-248-9851 or *mbinnebose@hotmail.com*

North Texas Ataxia Support Group Meeting

Time: 10am-12pm
Location: Ben Washington Baptist Church –Rev Jr Sheppard Educational Center, 615 Davis St., Irving, TX 75061
Details: There is lots of parking and it is handicap accessible. The meeting room is in a separate bldg from the church.
More info: David Henry at *cheve11e@sbcglobal.net*

South Eastern Florida Ataxia Support Group Meeting

Time: 2pm-4pm
Location: Baptist Hospital, 8900 N Kendall Dr, Miami, FL 33176
More info: Jose Fernandez de Castro at *jfcmv@yahoo.com* or 954-864-1436

St. Louis Ataxia Support Group Meeting

Time: 11am-1pm
Location: The Center for Advanced Medicine, 4921 Parkview Place, St Louis, MO
Details: We meet the second Saturday of every month at The Center for Advanced Medicine on the 3rd Floor in Conference Room 1.
More info: Shannon Dunphy Lazo at 202-306-2738 or *shan_d@hotmail.com*

Tampa Bay Ataxia Support Annual Picnic

Time: 12:30pm-3pm
Location: University of South Florida – Morsani Center, 13330 Laurel Dr., Tampa, FL 33612
More info: Darlene Harris at 813-431-2859 or *Msdee004@gmail.com*

Saturday, May 19, 2018

Twin Cities Ataxia Social Group Meeting

Time: 10am-12pm
Location: Langton Place, 1910 W. CTY. RD. D., Roseville, MN 55112
Details: 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 every month at 10:00am for 2 hours 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 at 1910 W. CTY. RD. D., ROSEVILLE, MN 55112. 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!
More info: Lenore Healey Schultz at 612- 724-3784 or *schultz.lenore@yahoo.com*

Sacramento Ataxia Support Group Meeting

Time: 1pm-3pm
Location: UC Davis Medical Center, The Lawrence J. Ellison Ambulator Care Center Building, 4860 Y Street, 3rd Floor, Conference Room 3030C, Sacramento, CA 95817
Details: We meet on the third Saturday of each month.
More Info: Teresa Bredberg at 916-215-2686 or *tbredberg@sbcglobal.net*

Los Angeles Ataxia Support Group Meeting

Time: 2pm-4pm
Location: Disability Community Resource Center, 12901 Venice Blvd., Los Angeles, CA 90066
More info: Lora Morn at *loramorn@gmail.com* or Harvey Kahn at 562-686-9720

Sunday, May 20, 2018

Greater Houston Ataxia Support Group Meeting

Time: 8am-5pm
Location: Methodist Hospital – Sugar Land, 16655 Southwest Freeway, Sugar Land, TX
More info: Dave Cantrell at Home: 936-588-5179 Cell: 936-206-1504 E-mail: *dcantr7358@aol.com*

Chi-Town Ataxia Friendship Group Meeting

Time: 1pm-3pm
Location: Good Samaritan Hospital, 3801 Highland Ave., Downers Grove, IL
More info: Jonas Cepkauskas at 708-381-5555 or *jonas@chitownataxia.org*

Saturday, June 2, 2018

Treasure Coast Ataxia Support Group Meeting

Time: 11am-1pm
Location: Buffalo Chophouse, 918 SW Gatlin Blvd., Port Staint Lucie, FL 34953
Details: The meeting will be in a private dining room. Our Guest Speaker will be Massage Therapist, Mark Bozzo. Friends and family are always welcome.
More info: Lisa Cole at *lisacoleataxia@gmail.com* or 772-370-3041

Greater Atlanta Ataxia Support Group Picnic

Time: 1pm-3pm
Location: Lanier Park, 1360 Buford Dam Rd., Buford, GA 30518
More info: Greg Rooks at 404-822-7451 or *rooksgj@yahoo.com*

Saturday, June 9, 2018

Mid-Atlantic Social Group Sailing Event

Location: Baltimore, Maryland – Downtown Sailing Association
Details: The fourth annual Ataxia Sailing Day at the Downtown Sailing Association (DSC). No sailing experience necessary. The DSC provides a safe and fun experience for children and adults with Ataxia. See their website for more information. After sailing, enjoy appetizers and a raffle with great prizes at a nearby restaurant. This is a popular event, register soon!
More info: Donna DeLano Neuworth at *ddelano1@jhmi.edu* or 410-616-2811

North Texas Ataxia Support Group Meeting

Time: 8am-10am
Location: Ben Washington Baptist Church –Rev Jr Sheppard Educational Center, 615 Davis St., Irving, TX 75061
Details: There is lots of parking and it is handicap accessible. The meeting room is in a separate bldg from the church.
More info: David Henry at *lcheve11e@sbcglobal.net*

Central Minnesota Ataxia Support Group Meeting

Time: 9:45am-11:45am
Location: Harvest Bank, 24952 County Rd 7, St. Cloud, MN 56301
More info: Marsha Binnebose at 320-248-9851 or *mbinnebose@hotmail.com*

St. Louis Ataxia Support Group Meeting

Time: 11am-1pm
Location: The Center for Advanced Medicine, 4921 Parkview Place, St Louis, MO

REMEMBERING THE 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 the 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* or call (763) 231-2748.

Details: We meet the second Saturday of every month at The Center for Advanced Medicine on the 3rd Floor in Conference Room 1.
More info: Shannon Dunphy Lazo at 202-306-2738 or *shan_d@hotmail.com*

Kansas City Ataxia Support Group Meeting

Time: 12pm-2pm
Location: KC Public Library – Trails West Branch, 11401 E 23rd St., Independence, MO
More info: Stephanie Wilkins at 816-623-3318 or *sfwilkins@yahoo.com*

Saturday, June 16, 2018

Twin Cities Ataxia Social Group Meeting

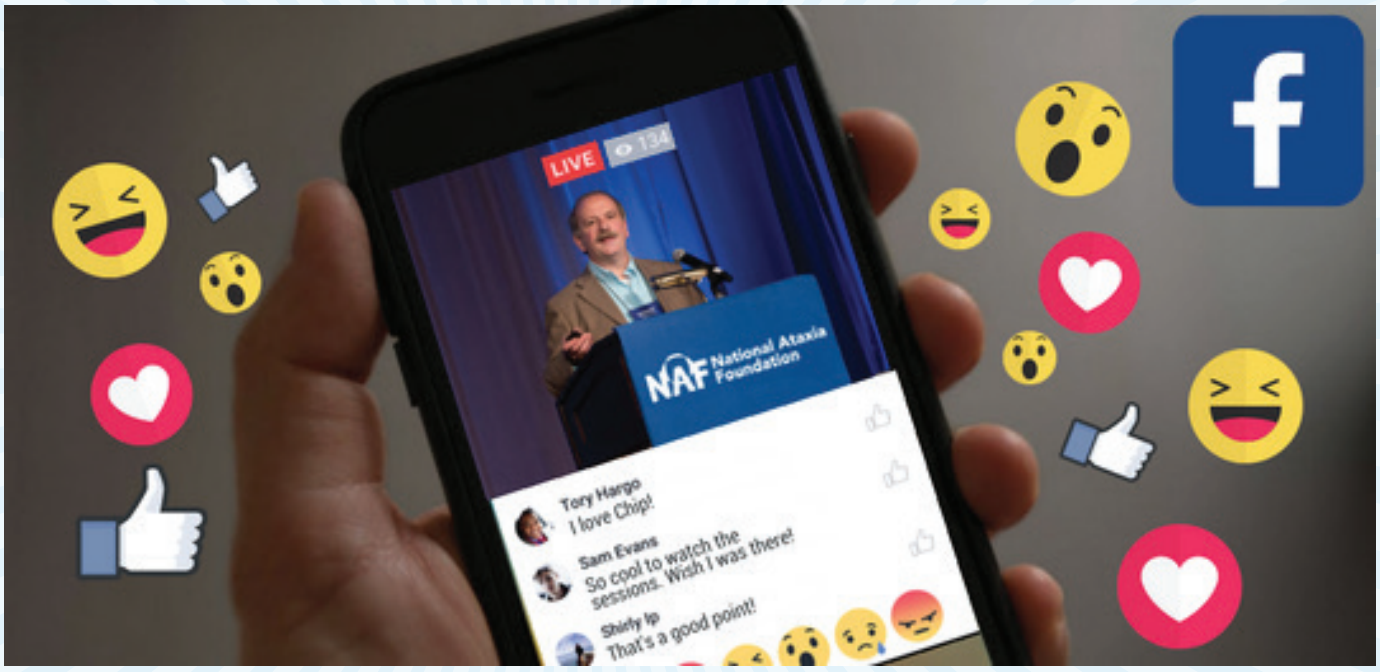
Time: 10am-12pm
Location: Langton Place, 1910 W. CTY. RD. D., Roseville, MN 55112
Details: The Twin Cities Ataxia Support Group meets once a month. Family and friends of an afflicted individual are always welcome! We meet on the third Saturday of every month at 10:00am for 2 hours in a meeting room at 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 at 1910 W. CTY. RD. D., ROSEVILLE, MN 55112. 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!
More info: Lenore Healey Schultz at 612- 724-3784 or *schultz.lenore@yahoo.com*

Sacramento Ataxia Support Group Meeting

Time: 1pm-3pm
Location: UC Davis Medical Center, The Lawrence J. Ellison Ambulator Care Center Building, 4860 Y Street, 3rd Floor, Conference Room 3030C, Sacramento, CA 95817
Details: We meet on the third Saturday of each month.
More info: Teresa Bredberg at 916-215-2686 or *tbredberg@sbcglobal.net*

The Annual Ataxia Conference was broadcasted FB Live!

View the videos now on the NAF Facebook page at **www.facebook.com/ataxiafoundation.**



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 information to *stephanie@ataxia.org* so you don't miss out on important information

Upcoming Informational, Awareness Events, and Fundraisers

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

Friday, May 4 - Sunday, May 6, 2018
Abilities Expo – New York
Time: 8am-4pm
Location: New Jersey Convention and Exposition Center, 97 Sunfield Avenue, Edison, NJ 08837
Details: 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.
More info: *www.abilitiesexpo.com*

Saturday, May 19, 2018
Treasure Coast Walk N' Roll to Cure Ataxia
Time: 7am-11am
Location: Tradition Square and Lake, 1087 SW Tradition Square, Port St. Lucie, FL 34987
Details: : Come and learn about Ataxia while having fun raising funds for NAF. The event is up to a 1.75 mile Walk or Roll around the Lake in Tradition. The event is followed by refreshments, ataxia information, and recognition of funds raised. Registration at 7am. Walk N' Roll at 8am.
More info: To volunteer or for more information please contact Lisa Cole at *lisacoleataxia@gmail.com* or (772) 370-3041

Saturday, May 26, 2018
WPA Golf Outing to Cure Ataxia
Time: 10am-6pm
Location: LakeVue North Golf Course, 591 Pittsburgh Rd., Butler, PA 16002
Details: $80 entry fee per golfer. Includes:18 holes of golf, green fees and cart, lunch at the turn, steak dinner (chicken or fish upon request), beverages (beer, water, Gatorade, Iced Tea).
More info: *ataxia.donorpages.com/2018WPAGolfOuting/*

Sunday, June 3, 2018
Balloon Ride Contest
Time: 8am-5pm
Location: Online
Details: An online fundraising contest. The contest winner will receive a ride in a hot air balloon flown by Captain Crystal Stout. Wanna be a contestant to win a hot air balloon ride? Start a fundraiser *https://www.facebook.com/pg/ataxiafoundation/fundraisers/?ref=page_internal* and then post a link to your fundraiser on the event page *https://www.facebook.com/events/723485921373797/*. Every dollar you raise = A vote for you. All proceeds benefits the National Ataxia Foundation.
More info: Sherry McLaughlin at 360-344-2445 or *ccherilynmc@yahoo.com*

Saturday, June 23, 2018
Western North Carolina Walk N' Roll to Cure Ataxia
Time: 9am-12pm
Location: Transylvania County Recreation Center, 1078 Ecusta Rd., Brevard, NC
Details: Come and learn about ataxia while having fun raising funds for NAF. The event is up to a 1 mile Walk or Roll around the track. No Registration Fee – Donations Gladly Accepted. Registration: 9am, Walk n' Roll: 10am.
More info: Jodie Kawa (828) 384-8414 or *jodiekawa@comporium.net*

Friday, June 29 - Sunday, July 1, 2018
Abilities Expo – Chicago
Time: 8am-4pm
Location: Renaissance Schaumburg Convention Center, 1551 North Thoreau Drive, Schaumburg, IL 60173
Details: 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.
More info: *www.abilitiesexpo.com*

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 December 2017 through February 2018. We are sorry that we cannot separate the memorial contributions from those made in honor of someone, as sometimes the person making the contribution does not always let us know if the contribution is a memorial or in honor of their friend or family member.

Abbie Spellman
Alan Acacia
Alana & Alyssa Wolfson
Alice Battung
Alice Tapper
Alisa McFarland
Allan Yousten
Amy Cantrell
Amy Messigian-Legault
Ann Foster
Anna Lau
Antonio Pimentel
Ardella Tate
Arthur G. Brennan
Betty Worfel
Bettylou McIntosh
Blanche Buckley
Bob Sturdevant
Brandon Barker
Bruce Ayres, Barbra Jones & Cece Boyne
Carol Greenblatt
Carol Paige
Charles Adams
Chicago Ataxia Support Group
Claire J. (Mackey) Conway
Claire Mackey
Clete and Peggy Brunnert
Connie Fiasco
Connie Holden
Constance Holden
Dan Freedland
Danny Cook
Darren Martin
David D. Brown
Debi Adair
Debi Griffin-Cutten
Deborah Taylor-Omictin
Debra Covington
Denise Claire Drake
Derek Semler
Donna Reiser
Doris Campbell
Douglas Fickle
Dr & Mrs. Lawrence Schut
Dr Gerald Tadej
Dr Ken Kato
Dr. Gerald Tadej
Dr. Michelle Zhang
Earl McLaughlin
Ed Schwartz
Edith Chille
Ellioni Durrant
Emily Coble
Emily Penn (Baudin)
Eric Gisler
Eric Peterson
Everybody with FA
Florence Rinaldi
Frank Murphy
George Vine
Gerard Reidy
Gerry Neugebauer
Gian Parkash Jain
Gisela Kittel
Gordan Ball
Gordie Hoffmann
Greg Kreuter
Hans Peter Meyerhoff
Howard Hunnius
Jacqueline A. Devito
James Carr
Jane G. Shaw
Janet Hundelt
Janice Cohen
Jay Greenblatt
Jeffrey B. Gibson
Jeffrey Oram
Jennifer Leader
Jenny Spiller
Jerry and Cindi Tadej
Jessica Budreau
Jim Horne Hankins, Evelyn S. Hankins, Jimmy Hankins
Jim, Edward & Bob Burman
Joe Decrescenzo
Joe Machado
Joel Leventhol
John Barkasi
John Ebron, Jr.
John Frey
John G. Barkasi
John Zeller
Johna Leidholt
Joseph Fisher
Joshua Kirschbaum
Kai M. Chau
Katherine Gorman
Kathryn D Smithers
Kelly Oistad
Kelly Tambourino and Kim Michael
Kevin Whitaker
Kimberly Fisher Michael
Kjell Holmes
Leona Johnson
Lisa Jaffe
Louise Estabrook
Lucille Ellson
Lydia Ramsden
Maddie Ford
Marcella Schifrin
Marcia Saltmarsh
Marge Zaharis
Maria Alioto
Marie Schirling
Mark Dehebreard
Marvin Chell
Mary Hartmann
Mary Rotolo
Mary Schlickbernd
Maury McDonald
Merle
Michael Leader
Michael Lundquist
Mickey Mackinaw / Papa
Moira Greenway
Mr. Claude Decker
Mrs. Doris Pair Campbell
Nathan Bell
Nathaniel J Kowatch
Pansy Gooch
Patrica C. Rymut
Patricia Flynn O'Brien
Patricia Rymut
Phil Turnbull
Phyllis Hoekstra Meima
Ralph William Aiello
Jason Johnson
Refiye Miller
Rich Korosa
Richard Chin
Richard J. May Jr.
Robert James Carr
Robert Lawson
Roger Cooley
Rose Marsh
Russel Crystal
Ruth Magoto
Samuel Wilson
Sandra Burke, My Hero
Sandra Busalacchi
Seth Klausen
Sophie Barrera
Stephanie Peterman
Steve Havel
Summer B. Little
The Alexander Family
The Leader Family
The Silva Family
Thomas Cromer
Thon Phhrom
Tom Sander
Twila Lester
Tyrane Venessa Gates
Verlie Wilkins House
Vincent Ferranti
Virgie Corbin Wince
Virginia Marsh
Walter Robert Lowry
Wanda States
Wesley Tischoff
Western Washington Support Group
William Hachmann
William Kingery
Wyatt Brink

So you have a genetically confirmed form of Spinocerebellar Ataxia?

Invitae is looking for individuals with specific genetically confirmed forms of SCA: SCA7, SCA10, SCA12 or DRPLA who are willing to provide saliva sample and a copy of their genetic test results.

Invitae is a diagnostic testing company whose mission is to make genetic testing more widely accessible and affordable, especially to individuals with a rare disease who often face difficulty obtaining a diagnosis.

Next-generation sequencing panels have benefitted many rare disease communities, but due to the inherent technical difficulties, a reliable, low-cost, comprehensive pane has yet to be developed for Spinocerebellar Ataxia.

Invitae is offering $200 for your participation.

If you have a genetically confirmed diagnosis of one of these Ataxias and are interested in participating, please contact Invitae Genetic Counselor Hannah White at *Hannah.white@invitae.com* or (415) 231-5648 for more information.

INVITAE

Tissue donations for research in Friedreich Ataxia

If you have been diagnosed with Friedreich Ataxia and wish to contribute to its eradication by helping research, please consider donating your tissues after death. To do so, contact Dr. Arnulf H. Koeppen for detailed information. Tissues affected by Friedreich Ataxia are brain, eyes, spinal cord, dorsal root ganglia, sensory peripheral nerves, heart, and the insulin-producing beta-cells of the pancreas.

Arnulf H. Koeppen, MD • Professor of Neurology and Pathology
Research Service (151) • VA Medical Center
113 Holland Ave, Albany, NY 12208
Tel. 518-626-6377 • FAX 518-626-5628
E-mail: *arnulf.koeppen@va.gov* or *koeppea@mail.amc.edu*

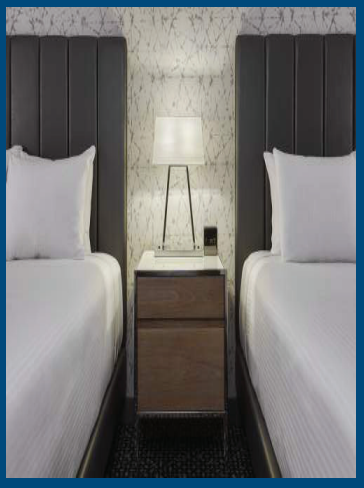
National Ataxia Foundation

62nd Annual Ataxia Conference

March 29-30, 2019

Flamingo Hotel and Casino

Las Vegas, Nevada



Join us in Las Vegas! • www.visitlasvegas.com

For the latest information on conference registration, program schedule, and area information, keep checking NAF's website - www.ataxia.org

Wanted: Those with SCA1 and SCA3

In early 2018, 20 study sites in the United States and 2 in Europe will be asking those with SCA1 or SCA3 whether they or any of their family members are willing to participate in an important study.

To be eligible for the study:

1. You must be a member of a family with SCA1 or SCA3.
2. You have a parent, a sibling(s) or a child(ren) who is (are) affected with SCA1 or SCA3 and you have no noticeable gait imbalance, hand clumsiness or slurred speech.
3. You are between the ages of 18 to 65 years old.
4. And you can walk independently without any support (that means no touching a wall, furniture, person, cane, walker or other assisting device).

It does not matter if you have had gene testing or not for SCA1 or SCA3. The research sites will provide free DNA testing. In addition, multiple members from the same family are welcome to participate.

For more information, enroll in the CoRDS Ataxia Patient Registry. By enrolling in the registry, when the details are available about the upcoming study for SCA 1 and SCA3, you will be notified and can make your own decision about whether to participate or not. It will be totally up to you. But if you don't enroll, you may not learn about this study and other studies for which you may qualify.

Go to Sanfordresearch.org, click on the Special Programs tab and select CoRDS from the drop-down menu. Follow the directions to enroll and if you have any questions or problems about enrollment, call Sanford directly at 1-877-658-9192.

If you want to learn more about the SCA 1 and SCA 3 study, please contact the National Ataxia Foundation. Sue Hagen, NAF's Patient and Research Services Director, will be happy to answer your questions. Send an email to **susan@ataxia.org or call 763-231-2742**.

RESEARCHER RECEIVES $1 MILLION FOR ATAXIA RESEARCH FROM NIH

THANKS NAF DONORS FOR HELPING MAKE IT POSSIBLE

In 2015, Miao Zhang, Ph.D. was early in his career on the Ataxia research scene, but he had a strong desire to look for answers. He applied for a Young Investigator Grant from the National Ataxia Foundation to help get his project started. Dr. Zhang said the grant "helped me establish my new lab, led me to new findings that resulted in new publications."

From there, Dr. Zhang was able to learn about the structural makeup of Ataxia that may lead to new Ataxia treatments. Fast forward to 2017 – Dr. Zhang, a junior faculty member of Chapman University School of Pharmacy, was awarded a $1,059,867 grant by the National Institutes of Health (NIH) for his Ataxia research. This grant will allow him to continue looking for the underlying causes of Ataxia. He says that SK ion channels have been associated with Ataxia in past studies, but their exact role is not yet known. Finding that role could be the key to treatments and drug development in the future – a future that NIH just invested in supporting with their IGNITE (R21/R33) grant to Dr. Zhang.

At a pivotal moment in his research, Dr. Miao Zhang took the time to write a letter of thanks to YOU – supporters and donors of NAF. Why? Because you helped get the ball rolling. An important part of the process that is not lost on Dr. Zhang. He says he would not have been able to establish his lab or perform the research project that led to the IGNITE grant without NAF. He writes, "I would like to thank the donors of the National Ataxia Foundation who provide the research grant funds. I will continue my research in the hope of finding a cure for ataxia."

Giant leaps in Ataxia research come from the small steps made in our Ataxia community. We come together to fight this disease – we will celebrate together when it is defeated! Thanks for your help – which gives NAF the ability to support researchers.

NAF National Ataxia Foundation

FIGHTING *for* FREEDOM

ANNUAL ATAXIA CONFERENCE 2018

APRIL 5-6, 2018 • PHILADELPHIA, PA

WE WANT TO SEE YOUR PHOTOS! DID YOU ATTEND 2018 ANNUAL ATAXIA CONFERENCE IN PHILADELPHIA? EMAIL YOUR STORIES/PHOTOS TO NAF@ATAXIA.ORG.

By submitting a photo, you give NAF the right to use it in current and future communications.

We hope to hear from you!

Gift – Honor – Memorial

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

My contribution is: ☐ In Memory ☐ In Honor ☐ Gift Membership
Name _____
Occasion _____
Send Acknowledgment Card to:
Name _____
Address _____
City/State/Zip _____
From:
Name _____
Address _____
City/State/Zip _____

Membership

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

Name _____
Address _____
City/State/Zip _____
Phone _____
E-Mail _____
☐ Yes, sign me up for NAF e-mails

PAYMENT INFORMATION

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

☐ Check. Please make payable to the NAF.
Total Amount Enclosed $ _____
Card: ☐ Visa ☐ MasterCard
☐ Discover ☐ AMEX

Name on Card _____
Card # _____
Exp. Date _____ CVV # _____
Signature _____
Phone Number _____

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