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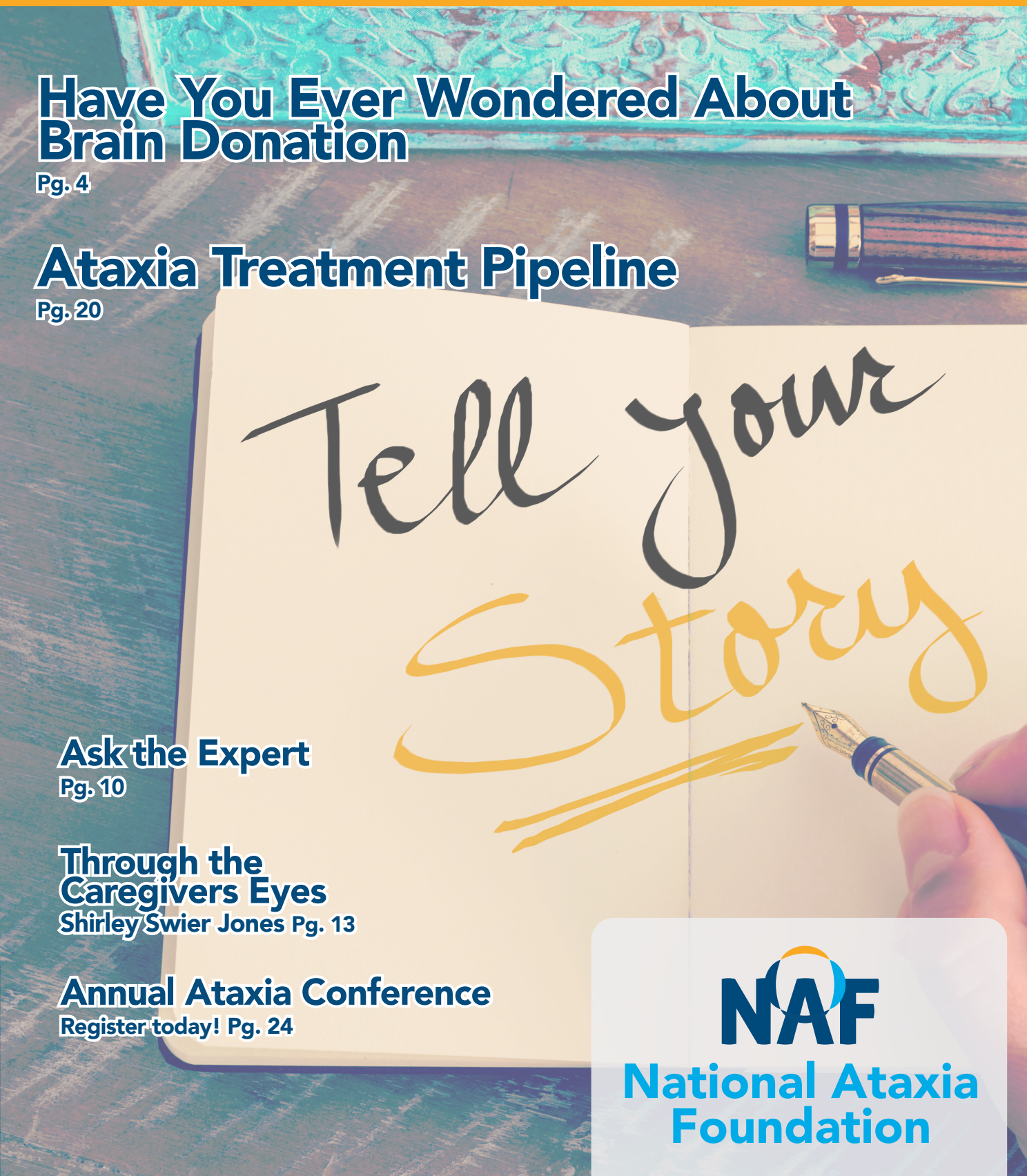

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NAF

National Ataxia Foundation

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NAF Update:
Letter from the Executive Director

Andrew Rosen,
Executive Director

The power of simply being together. I have been struck recently by how wonderful it has been to be with other people in the Ataxia community in 2022. While the pandemic forced AAC 2022 to be held online, the rest of the year has allowed us to feel the energy, compassion, and resolve of this remarkable group of people. From live Walk N' Rolls around the country to the first in person meeting of our CRC-SCA natural history study principal investigators since 2019; from NAF staff collaboration days where we're not staring at each other in little Zoom boxes to the first International Congress for Ataxia Research with more than 450 scientists and pharma executives sharing ideas to advance Ataxia research, I will remember this year for the simple thrill of being together. On that note, I can't wait to see so many of you at our Annual Ataxia Conference in Las Vegas next March! If you're able, please join us to share in the power of this community.

While we still wait for the first approved treatment for any type of Ataxia, you should know that there is so much good happening at NAF. Our small but mighty staff is now 15, and there is not a harder working or more caring group out there. You will see the impact of their work in the following pages. I wish you all a peaceful holiday season and here's to seeing you, in person, in 2023.

Andrew

VISION

A world without Ataxia.

MISSION

To accelerate the development of treatments and a cure while working to improve the lives of those living with Ataxia.

Have you Ever Wondered about Brain Donation?

Many feel powerless when they are first diagnosed with Ataxia. Brain donation gives the donor and family the power to provide researchers knowledge to fight back against the disease. The examination of diseased brain tissue by a trained researcher remains the gold standard to gain a better understanding of the disease.

Have you ever wondered what the benefits are in brain donation? Brain research benefits all those who are diagnosed with Ataxia making brain donation an ideal way to achieve a positive outcome despite a difficult situation. Brain donation has the capacity to help find treatments and a cure!

Have you ever wondered what happens to the donated brains through NAF's Brain Donation program? The majority of the donations are housed at the University of Florida (UF) and are used strictly for research on Ataxia. Tissue is sent to various Ataxia investigators across the country who are studying the disease. UF is establishing agreements with partners in the pharmaceutical industry who are in various stages of drug development for Ataxia research. Dr. Ranum heads the NAF Brain Donation Program at UF; she writes, "Autopsy donations are critical for understanding how Ataxia affects the brain and for guiding research efforts. Investigators use donated tissue to explore why neurons in the brains of Ataxia patients die and to develop therapies to improve the lives of patients living with Ataxia."

About Brain Donation - People with a Plan in Place

You might also wonder about the thoughts and experiences of those who currently have a brain donation plan in place with NAF and those whose loved one has passed and has donated tissue for research.

Comments from those with a plan in place:

Gigi

I was finally diagnosed in 2006 with Spinal Cerebellum Ataxia. I found NAF on my first internet search right after my diagnosis and I learned valuable and current information about Ataxia from the NAF website. I signed up to receive the NAF newsletter and that is where I read about brain donations.

I understood the importance of research from my career in Medical Laboratory and Pharmaceutical Research, so I did not hesitate in signing up. The application was easy to complete. I appreciate receiving periodic emails to make sure all my information is up to date.

I have informed my husband and daughter about my wishes to donate and what to do when my time comes. I would encourage anyone thinking about brain donation to learn if this is the right decision for you.

Cheri

I decided to become a donor to help further the research and the efforts being made to develop a cure for Ataxia. I believe our bodies are just "dust in the wind". It gives me great pleasure to know my brain may be of use in developing a cure for Ataxia.

The process to become a donor was quite simple (filling out a brief form, submitting it to the NAF, and updating it each year if there are changes in one's contact information or health status). You or your loved ones may call NAF at any time to ask questions. It is best to enroll in the program as soon as possible (call Mary Ann Peterson at NAF TODAY @ 763.231.2750) as one never knows when this life will end. I feel completely at peace knowing I am enrolled in the pre-plan program, and the necessary steps can be taken by my surviving loved ones when I die. Taking care of enrolling in the program early allows plenty of time for loved ones to ask any and all questions, so they will be prepared to proceed at the time of death.

If this fits with your views, I encourage you to GO FOR IT! Your donation may perhaps find a cure for Ataxia and prevent this disease from affecting future generations!!

Lisa

I made the decision to become a brain donor because I want to help researchers discover further knowledge about the disease. The process to enroll in the program was extremely easy – it was one-page form, and I am glad to be reminded every year to update any changes or additions. Remember – you are helping researchers, you are helping future generations, and you are continuing to help even after you are gone.

Pat Berry

I am donating my brain for research to, hopefully, help researchers better understand and provide answers for future generations, to get a more knowledgeable understanding of the disease, and to help.

Jesse Diehl – My Ataxia Story

I started showing signs of coordination problems in my mid 30's. My older brother had been diagnosed earlier with SCA8. He has since died as a result of his condition. I was diagnosed with SCA8 in the year 2000. I worked as a mechanical engineer until Sept. 2015. I stopped working and went on disability in Feb. 2016.

Since 2016, I have been reinventing myself. I have taken over most of the domestic duties of our household and volunteer when I can. It is an ongoing learning process for me. I have been involved with Ataxia research studies and try to learn information about this progressive rare disease. In 2018, I started a support group in the Western New York area so I can learn what has worked for others and share my successes.

The decision to join the brain donor program was easy for me because I am an advocate for myself. I have two grown sons who are not showing signs of this disease, but I know that they have a 50% chance of having inherited the affected gene. I think it is important to help where I can. More needs to be learned about this disease and I think that I can help.

Family Members Whose Loved Ones Were Donors

Nelta

My husband, David, had Spinocerebellar Ataxia. Before he passed away, he decided to donate his brain to help with the research of his disease. My daughter contacted NAF and requested a brain donation form, which was sent with much needed information. After filling out the form and sending it back to NAF, we received an email from Mary Ann Peterson verifying she received it. Next, we contacted our chosen funeral home and explained our wishes and took copies of all conversations and forms from NAF. We also sent the name of the funeral home to NAF.

Within a short time, David went to Hospice and passed away within 24 hours. Hospice was informed of David's wishes and was in contact with the funeral home, so retrieving the tissue would be done within a certain time frame. Everything went smoothly with no problems. David was a selfless man and wanted to help others in life, and even in death. From my end, the whole process was simple and uplifting. My family and I miss David everyday—thank you for your help and kind words.

Betty

My husband's long, slow physical and mental decline was noticeable in the mid to late 1980's. He saw a neurologist who diagnosed olivo-ponto cerebellar atrophy.

In the mid 2000's, I received a phone call from my youngest stepson as he was starting to exhibit similar symptoms. I have spoken several times with him and his mother over the years since then about my husband's illness. They became active with the National Ataxia Foundation. As my husband's health declined, they requested that when the time came that I donate my husband's brain for Ataxia research. Having worked for many years at a medical

school, I understood that such a donation would be invaluable to researching such a terrible disease. I was glad to do such a donation to further the understanding of Ataxia.

Pre-planning for brain donation was very helpful. I was out of town when my husband passed away in hospice care. Having pre-arrangements with the National Ataxia Foundation, a local funeral home, and medical school was a source of comfort especially since it fulfilled his request that his body be donated to science/medical research in addition to brain donation for Ataxia research.

Having made prior arrangements, everything went smoothly as I could do much over the telephone since I was out of town. I found the preplan forms from NAF to be easy to understand and fill out. Thank you.

Carrie Edge

The disease that my mother had crippled my family for generations. Knowing that my siblings and I are possibly next in line to get this rare disease, we are all very focused on finding a cure and participating in research.

When we were approached with this opportunity to help in finding a cure for this disease, we were eager to assist. However, brain donation came with a set of concerns that had to be addressed and naturally we were hesitant. Not only did I have to discuss this delicate subject with my mother who rarely wanted to focus on the aftermath of her passing, but I was concerned with how it would affect her physical condition for purposes of a viewing at her funeral. After careful consideration we decided that this would be a great opportunity to further support the advancement of SCA3 research. To my relief, not only did my mother agree to donate her brain tissue, but the entire experience was handled delicately and efficiently by all parties involved.

After my mother's passing, she was transported immediately to the Health System via the funeral home. The procedure was then completed, and she was brought back to the funeral home in time for the viewing without any obvious physical signs of a procedure taking place. Since her passing, my family and I are grateful and find comfort in the fact that our mother had an opportunity to give one last final attempt at finding a cure for this rare disease and that even in spirit, she is able to continue fighting for a cure.

Allen

(Editor's Note: Allen's son, Ken, was a brain donor. Ken's aunt has a plan in place with NAF for brain donation. Ken's grandfather's brain is at Massachusetts General Hospital. There is added value to have related brains available for research.)

Why donate? I think that it just felt like the right thing to do. NAF has a process for donating brain tissue and researchers can make use of brain tissue that will help future Ataxia research.

The staff at NAF and the University of Florida were wonderful to work with. They were very

Allen cont.

responsive and very human in helping families through this. They made sure that the nursing home, hospice, and I all knew what would be needed at the time of death.

Everyone made it clear that they were happy to talk about this while giving no pressure to make the donation. They understand the decision is very personal. They also want everyone who is wondering if this might be right for them to be comfortable, and if it isn't right for them to be able to say, "No". They are also very grateful for the families and the donors who say, "Yes".

Richard

My wife, Judith's, mother, grandmother, and an aunt all had or have SCA6. Judith saw what the disease does and thus took a DNA test to find out if she had SCA6. The test was positive.

About five years after taking the test, the Ataxia symptoms started and continued until her demise. The progression was as expected, but not enjoyable. She and I had discussed what her wishes were for her remains and she included a desire to donate her brain tissue for research.

After contacting the National Ataxia Foundation (NAF) and informing them of our desires, the process continued by NAF and went very easily. After her death, her brain was sent to the University of Florida (UF). The donations that are housed at UF through NAF's Brain Donation program are used strictly for research on Ataxia. I was glad to hear that good news. Bottom line: I would encourage anyone considering a donation to not hesitate to contact the National Ataxia Foundation for information about all aspects of the process of brain donation. I sincerely hope such research eventually finds a remedy.

Kathy

Kathy and Harry went to NAF's Annual Ataxia Conference in Orlando in 2016. At a Birds of a Feather session, one doctor mentioned needing brain tissue for research. After the session, Harry made the decision to become a donor to provide researchers the necessary tools (brain tissue) to do their research. The majority of the tissue available was for the more well-known Ataxias and they needed tissue from the lesser known Ataxias. Harry felt strongly that his donating may help to find a cure for Ataxia.

If you have ever wondered about brain donation, NAF understands how difficult yet important this decision is and we want to provide you with as much assistance as we can in helping you and your family members with any questions you may have. We are so grateful for the courage of each family member who helped their loved ones' wishes of making brain donation a reality. We also thank those who shared their story with the Ataxia community.

If you are interested to learn more about NAF's Brain Donation program, please do not hesitate to contact NAF at mary@ataxia.org.

My SCA1 Story, I Share it With You!

By Rusty LeJeune

Basically, I had a completely normal life growing up. I ran track and played football in high school, I went to college and majored in music, and played in multiple marching bands during my college career. I continued running after college, competing in 5K and 10K races and running twelve miles a day just to keep in shape. I taught at LTC for 14 years, I am currently working in the IT department with the Pointe Coupee Parish school district. I started having trouble with my balance, walking, and falling frequently. I did not know what was causing this. I had been a runner for many years, and it was difficult for me to do anything around the house.

Since I turned forty, I have had open heart surgery, prostate cancer, my urologist had to remove my prostate, three neck fusion surgeries, and one lower back surgery. I have a full-time pain management doctor who noticed that my walking was getting worse, my speech was getting slurred, and recommended I see a neurologist. I made an appointment, after intensive examinations and several visits, she told me that it could be one of two things that was causing my issues. But the tests were expensive and there was no cure for either disease. So, I didn't rush out to get a diagnosis, because what's the point if there isn't a cure. After talking with my insurance, I discovered they paid most of the expenses for genetic testing. I did the test and was diagnosed with Spinocerebellar Ataxia Type 1. That was three years ago.

I am presently working full time, but it's getting harder and harder for me to get ready in the morning. What used to take me 15 minutes takes over an hour now. Walking one hundred yards feels like I have just run a mile. I get out of breath doing things. I have difficulty Just walking. I must use a cane or walker to aid my walking now. My neurologist has given me orders for an upright rollator to assist me.

My symptoms are frustrating, tiring, depressing, and annoying, but I have a decent life. Running and walking are terms of the past. To my friends and family, it looks like nothing is wrong with me. I try to explain this rare disease, but some cannot grasp my whole situation. I stay in the house a lot, because it is hard for me to do anything now. I want to share my story so that others know they are not alone and to not give up. Some diagnostic journeys are short, and some are long but you have to keep fighting for yourself.

This is My Ataxia Journey

By Vickie C. Weaver

I am retired, I no longer work, and I have Sporadic Ataxia.

I'm 64 now, and my life has totally changed from how it once was. I was a very active person, an outdoorsy type. From elementary age up until I had my 1st child, I played softball, catcher and first base, and I loved it. I was a cheerleader in Jr. High and in High School I marched with the band. I used to love to work in the yard, i would mow the front and back yard-with a push mower, in the same day and think nothing of it.

Around 2005-2006, I was around 49 years old and noticed the my balance was off and some of my words were not clear. After several falls and an unsteady balance (I could not perform the heel to toe walk) my physician referred me to a neurologist. I had an MRI of the head and was diagnosed with Spinocerebellar Ataxia. I had genetic testing, and my results showed no evidence of hereditary. My neurologist, Dr. George Wilmot at Emory in Atlanta, Georgia, classified mine as Sporadic Ataxia. I do believe that my father had symptoms of Ataxia and my older brother has since been diagnosed with gait Ataxia. He used to be an avid runner and ran Peachtree Road Race in Atlanta every year, now he uses a cane to steady himself.

My journey continues. My Ataxia has progressed with age. I'm thankful that I can still walk even if I look like I've been drinking. I'm so compassionate to everyone who has this hidden and not talked about disease. Let's make people aware and understand Ataxia.

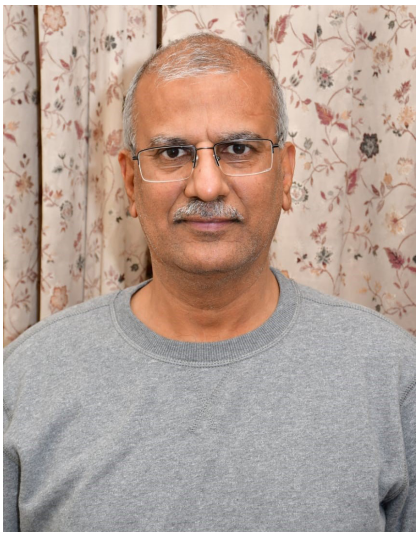
ASK THE ATAXIA EXPERT

A bimonthly series that empowers you with the knowledge you need to live better with Ataxia. Dr. Susan Perlman joined us for 2022. You can find recordings of all her discussions at

Visit **www.ataxia.org/asktheexpert/**

Past Experts include:

- Susan Perlman, MD
- Theresa A. Zesiewicz, MD, FAAN
- Pedro Gonzalez-Alegre, MD, PhD
- Liana S. Rosenthal, MD, PhD
- Christopher Gomez, MD, PhD
- Jeremy Schmahmann, MD, FAAN, FANA, FANPA



My Family and SCA12

By Salil Gupta

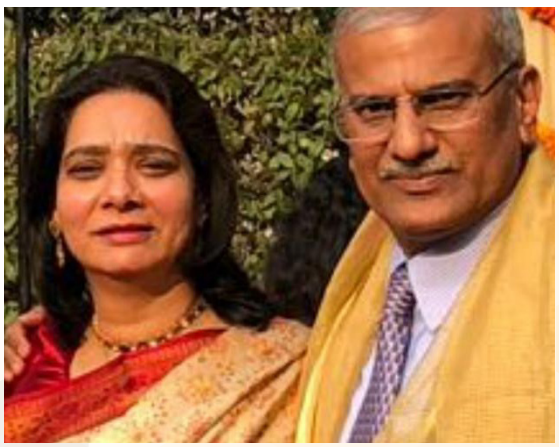
Hello, I am Salil Gupta, age 58 years and a resident of New Delhi, India. I am an Accountant (CPA) and have worked in the higher Indian bureaucracy (Internal Revenue Service) for many years. Now I hold a leadership position at a corporate employment while living a healthy lifestyle. I am vegetarian, I don't drink or smoke, I exercise regularly with Yoga and walks.

I started showing symptoms of mild tremors in 2014 at age 50. I tested positive for SCA12 in 2018. In a recent gene test, I tested positive for biallelic for SCA12 i.e., from both parents, a rarest of rare occurrence. The tremors started primarily in one hand with gait unsteadiness, reduction in walking pace/duration with rotational imbalance etc. I was prescribed Beta Blockers in varying dosage along with Trihexyphenidyl hydrochloride but those did not help the symptoms much. This medication caused several side effects like my legs below knee remaining cold and numb, throat and mouth dryness, urinary issues etc.

After I tested positive and became symptomatic, I set up family groups on both sides of the family. The gene has transmitted from my great grandfather. Several others in the extended family were known and treated for Parkinson's as SCA12 was unknown at that time. Now however, others in the extended family have similarly tested positive for SCA12. We are now attempting to gain knowledge and share our experiences within our family and amongst our community. Believe that this it is my duty to help the community better understand Ataxia and those affected by it.

I recently experimented my medication under my neurologist advice. I wish to share this with you, I am off all medications for past few months. My SCA12 related symptoms haven't deteriorated but all the side effects of the medications have vanished completely. So, my next plan is to remain off my medication and try some alternate medication such as Tibetan medication of Dalai Lama doctors and traditional Indian medicine of Ayurveda. I practice aggressive Yoga exercises and Pranayama meditation including inverted postures like head stand etc. which I believe have helped me in delaying the onset and progression of the disease. Remember to always check with your healthcare provider before starting or stopping medication and physical activity.

I have come to terms with SCA12 by making necessary lifestyle changes and living with the insidious progression of the symptoms. Most important part of our learning and experience is to stop further genetic transmission.



NAF offers FREE Genetic Counseling and Testing for SCA1, SCA2, and SCA3

NAF is excited to offer a program to remove barriers to genetic counseling and testing. Eligible participants will receive genetic counseling at no-cost to them. After counseling, if the participant and genetic counselor determine that testing is the right decision, genetic testing will be offered at no-cost to the participant.

Who is eligible?
At this time, the program is being offered to those who are at-risk for SCA1, SCA2, and SCA3 because they have a family member with a confirmed genetic diagnosis of SCA1, SCA2, or SCA3. Participants must be a member of NAF, live in the United States, and be 18 years of age or older.

Not a member yet? Sign up for free membership: **www.ataxia.org/JoinNAF**

Program Description: A diagnosis confirmed with genetic testing can be helpful to a person who is at-risk for a rare inherited disease. The confirmation of a clinical diagnosis takes away uncertainty and becomes a tool for a patient and their doctor. It helps identify clinical trials and therapies that may be a fit for the patient. Some individuals might not have access to genetic testing. NAF, in partnership with our Drug Development Collaborative, offers this program to remove barriers to genetic counseling and testing. Eligible participants will receive genetic counseling at no-cost to them. After counseling, if the participant and genetic counselor determine that testing is the right decision, genetic testing for SCA3 will be offered at no-cost to the participant.

Why aren't other types of Ataxia eligible for the Genetic Counseling Initiative?
While we hope to expand this program in the future, NAF had to develop limited eligibility criteria for the pilot program to meet the immediate needs of NAF's pharmaceutical partners. Focusing on SCA1, SCA2 and SCA3 at the launch of the program allows NAF to develop a system that is simple and seamless for the participant, their physician and the genetic counselors who have been contracted for this program.

How it works:
1. Contact NAF to determine if you are eligible for no-cost genetic counseling.
2. Enroll in the program using a secure online portal.
3. Schedule your appointment with a genetic counselor.
4. Attend a pre-test genetic counseling session.
5. Schedule your lab appointment if you decide to pursue genetic testing for SCA1, SCA2 or SCA3.
6. Attend a post-test genetic counseling session. You may opt to have your results shared with your neurologist.
7. Optional – Share your results with the National Ataxia Foundation. If you were diagnosed with SCA1, SCA2 or SCA3, you will be informed of future studies or clinical trials for which you may qualify. If you tested negative, we will update your record.

How to get started: If you or a family member are eligible because you have a direct relative who has genetically tested positive for SCA3, please contact NAF's Research Manager, Kelsey Trace at **kelsey@ataxia.org.**

Through the Caregivers Eyes

By Shirley Swier Jones

In May 2020, the book I wrote, "*Remembering the Ride, A Memoir, One Family's Journey Through A Hereditary Illness*", was published. The illness I write about is SCA 1. I do not have SCA, but I have lived it through the illnesses of my husband, two sons and a granddaughter. Due to this, myself and my other children have experienced tremendous grief.

My husband, Vernon and I met in 1956. He was twenty-one, handsome, kind, fun loving and Christian. He stole my heart quickly. This was the beginning of our love story that would evolve over the next twenty-three years through the blessing of five children and the sadness of living with Ataxia. That illness would impact our family for more than fifty years.

Vernon was never told the disease his mother had was hereditary. He heard it from talk in the community he grew up in and from putting together bits and pieces he gathered. When his older brother showed symptoms that progressed with time, it all made sense. When I met him, he told me on our first date that his mother had died from a disease he could one day get, but, he said, he didn't think he would—he felt fine, walked well, and showed no other evidence that he might have inherited it. I believed that he would not ever have Ataxia.

Vernon was diagnosed with SCA in 1963. We had been married six years and had three children. Even after that diagnosis we went on to have two more children. Our children were a joy to us, so it's certainly, difficult to say I know we should have been more responsible in family planning. I cannot imagine life without them, and for the most part I believe their lives have been good and full. Like my son who is affected said, "mom, my life has been so good, except for one thing—Ataxia. It has been difficult to watch my two younger sons and granddaughters live and struggle with SCA. This has been difficult for my three children who were not affected as well. They have all expressed, in different ways, living with survivors' guilt.

I am delighted with the progress that has been made by the NAF since 1963 to today. It was that year that my husband was diagnosed. At the time, we believed we were the only family living with Ataxia. Progress has been tremendous, including, discovering the gene in 1993, identifying over 50 types of SCA over the years, learning of Ataxia families worldwide, and an increasing interest in SCA as evidenced by the number of doctors and researchers who have made Ataxia their focus. Patients can take part in clinical trials and there is progress being made toward finding a drug that, hopefully, will arrest SCA. This gives hope to those living with the disease today, and it gives hope to their families. Today, through social media, Ataxia patients and their families can connect with other patients and families. They can know, unlike in 1963, that they are not in this alone.

"*Remembering The Ride*" elaborates on my story. It is available online at Amazon and Barnes and Nobel and can be ordered at bookstores worldwide through Ingram Spark.

How Am I? Not Ashamed Anymore

By Richard Brown, MBE

Part of my coping mechanism has always been to remember the great number of people who are experiencing much worse than I – 'It could be worse' 'keep a stiff upper lip' or keep calm and carry on'. Now I realize that minimizing the physical realities of living with a disability in this way is actually a form of denial in itself. Focusing only on the social side of disability may come at the expense of recognizing our physical health and the very real pain and fatigue we all feel.

Friends and family say they are impressed that I don't complain much. This is a back-handed compliment. I don't talk about my disability because that would be seen as complaining and I would feel embarrassed and ashamed. After all, nobody wants to hear how you really are. To avoid judgement, disabled people should keep quiet about their health. An ableist society puts us under great pressure to fight, to show grit, steel and courage instead.

This dilemma catches many disabled people out when applying for benefits they are entitled to. Rightly, disabled people want to focus on what they can do. To not be defined by their disability. Cruelly, the usually underqualified DWP assessors only award points for what claimants can't do. Since the Government's transfer of people from Disability Living Allowance (DLA) to Personal Independence Payment (PIP), over 100,000 disabled people have lost their mobility vehicles and a further 200,000 who used their mobility allowance to cover taxis or general care needs to live independently have lost that too. This is an absolute disgrace and shows how badly our society needs to reconsider disability.

My own approach was challenged in 2020, when a friend presented a video about her life with Ataxia at our virtual conference. As she spoke openly about her care needs, I began to think "Whoa, this is way too much information" and "I don't need to hear all this." But it made me consider the extent of my own efforts to cover up my own disability, and why I was doing it. I think it is right that people know the truth. I don't want sympathy, just recognition that it is ok to be disabled, that my life, though different and difficult, is as valuable as any other. So, thanks to Emma B, for the very first time, here is a full and frank description of how my disability affects my life. Prepare to be surprised!

I was diagnosed with Friedreich's Ataxia, a progressive and incurable neurological condition as a teenager in 1991. Today, my energy is limited. I feel tired all the time. My wrists, elbows, and shoulders ache from the strain of twenty years of propelling myself in a wheelchair. My transfers are generally poor and require a lot of concentration. I fall a few times a year. Although I have my feet attended to by a podiatrist and need some help getting dressed, I can manage all my own personal care. My scoliosis (curvature of the spine) makes sitting or lying down uncomfortable. My poor circulation means my hands and feet are cold through Winter. In the

summer, my feet swell up and sweat soaks my back and seat from sitting in a wheelchair all day. I have no reactions to speak of but jump out of my skin and have an adrenaline-fueled fight-or-flight reaction to both loud and sudden noises.

I have a hearing loss known as Auditory Neuropathy. I can still hear sound clearly, I just cannot separate the frequencies of different sounds. For example, in a noisy restaurant, I cannot hear a person next to me talking, all I can hear is the background noise. This kind of hearing loss is linked cognitively impaired perception of speech. I have always been frustrated at my inability to follow what people who have difficulties with speech are saying and have always found it difficult to focus on details and remember names. I never understood what depression was, but now I feel its pull when I am at my lowest, perhaps short of sleep or fighting off illness.

My eyesight is stable, but is now at the very limits of what corrective lenses can do. Difficulty focusing (Nystagmus) makes watching quick movement difficult. My speech is slower and slightly slurred and I can manage to write a signature, or single words which soon become unreadable, but nothing more.

As I eat one-handed, I use my other arm to keep myself upright and find cutting food difficult. I need to concentrate on swallowing or I'll choke. My involuntary leg-spasms make for the most interesting dinner table disruptions. I have difficulties with urgency when toileting and need to be careful what I eat and make sure I drink enough water. Fatigue and poor mobility coupled with being middle-aged and having two children make sexual fulfilment much more of a challenge.

My middle-aged body just cannot do all the things it used to, and certainly not as quickly. I have streaks of grey in my hair and a lot of white in my beard. Instead of being bitter about the things I can't do, I need to recognize them and appreciate the things I can do. I use a speech-to-text program to help me write for sustained periods. I listen to music and audiobooks, watch TV and film, play on my PlayStation and can use computers and phone apps adequately. I get out and about on my trike and I exercise with my Thera-trainer for ninety minutes every day. This helps with my digestion and cardio. I sleep fitfully enough. I am grateful to my Wife, Helen, for enabling much of my independence.

You might think my life sounds terrible. But it is my life, and I'm used to it. And that's the point, we all have challenges, but we should recognize them, not ignore them. We are set apart by how we face them. I live a full and rewarding life with mine, and that is alright.

About the Author

Richard Brown MBE is 46 and diagnosed with Friedreich's Ataxia (FA) at 15. He worked full-time in local Government until he retired in 2012. He was a Trustee of Ataxia UK from 2012 to 2022 and was awarded an MBE for services to disability and my community in 2018. He is an Ambassador for Ataxia UK and recently featured by FARA. A busy volunteer with several charities, he lives in Oxfordshire, England with his Wife and 2 children. He has been sharing his FA story and life as a disabled person on his blog, World According to Me: Award Winning Observations of a Disabled Person. This was an excerpt from Richard's blog. You can find more at worldaccordingtome.blog.

Snapshot: What is a Gene

A gene is the basic physical unit of **heredity**. Every living cell contains genetic information that determines an organism's development, form, and function. This genetic information is encoded by two macromolecules: DNA and RNA.

DNA consists of two strands of phosphate and sugar molecules connected by pairs of nitrogenous bases to form a double helix structure. The four nitrogenous bases in DNA are adenine, thymine, cytosine, and guanine (abbreviated A, T, C, and G). Genes are sequences of nucleotides (composed of a sugar, a phosphate group, and a base) provide the instructions that cells need to make molecules that give rise to an organism's characteristics. Within the nucleus of each cell, DNA is tightly coiled around specialized proteins called **histones**, forming compact structures called **chromosomes**. Each gene occupies a particular position, or **locus**, on a chromosome.

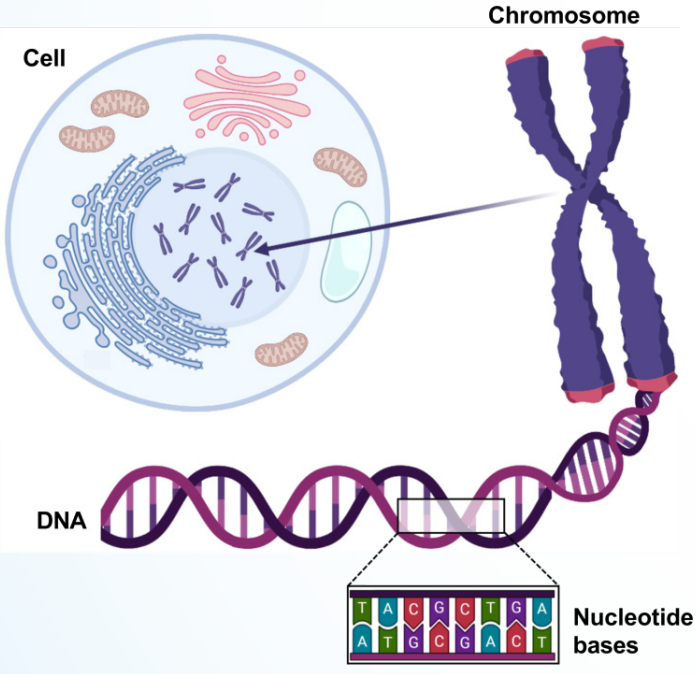


Figure1 created by Chloe Soutar using Biorender.com.

Genes are sequences of nucleotides that give rise to an organism's traits. DNA is tightly coiled around structural proteins and compressed to form chromosomes. Chromosomes are housed in the cell's nucleus and replicated prior to cell division.

Figure 1 created by Chloe Soutar using Biorender.com. Most genes contain instructions for creating proteins, amino acid-based macromolecules with a wide range of structures and functions. Among their numerous essential functions, proteins contribute to cell structure and repair, signal transmission between cells, and biochemical reactions within cells. Genes are used to create proteins through a two-step process. Double-stranded DNA is first transcribed into single-stranded messenger RNA (mRNA) that serves as a template for protein synthesis. mRNA exits the nucleus and interacts with cellular machinery called ribosomes. Ribosomes then read mRNA and translate its nucleotide sequence into long chains of amino acids, which then fold to form proteins.

Genes encode proteins through a two-step process. During transcription, enzymes within the nucleus build single-stranded mRNA molecules that are complementary to one strand of DNA. At this stage, the base thymine (T) is substituted for uracil (U). During translation, cellular structures called ribosomes "read" the mRNA within the cytoplasm and translate the nucleotide sequence into a sequence of amino acids. Linear chains of

amino acids then undergo patterns of folding to yield intricate protein structures. Figure created by Chloe Soutar using Biorender.com.

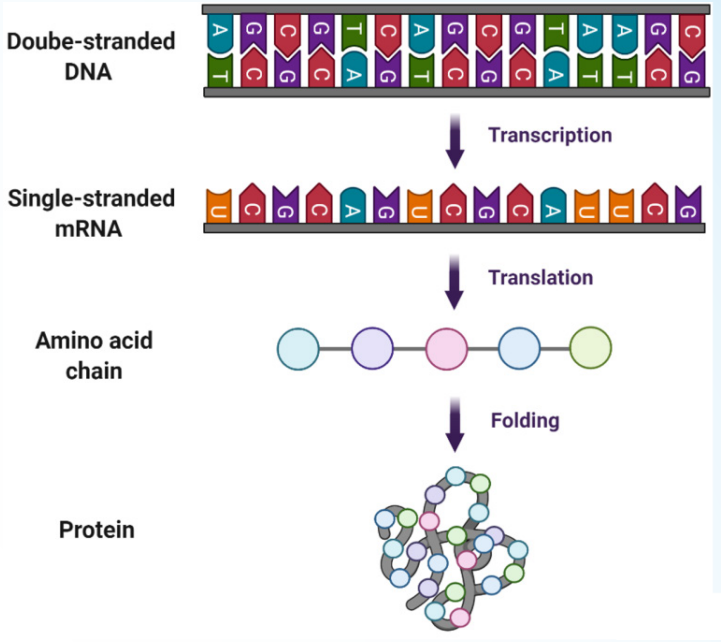


Figure 2 created by Chloe Soutar using Biorender.com.

An organism's complete set of genetic material is called a **genotype**. The human genome is estimated to contain between 20,000 and 25,000 protein-coding genes, varying in size from thousands of nucleotides to over 2 million nucleotides. The complete set of observable traits that results from gene expression is called a **phenotype**. An organism's phenotype includes all of its outward characteristics, including height and eye colour, as well as less apparent characteristics such as blood group and intelligence. For example, the genes that determine the amount of pigment in my skin are part of my genotype, but my skin colour is part of my phenotype. Whereas one's genotype is determined solely by biological factors, one's phenotype is determined by complex interactions between biological and environmental factors. This distinction between genotype and phenotype is evident in the case of identical twins – even though they have the same genotype, they often look and behave differently due to environmental and lifestyle factors.

GENES AND HEREDITY

Heredity is the transmission of genes and traits from one generation to the next. Nearly all human cells have 23 pairs of chromosomes. Egg and sperm cells are unique in that they only have one set of 23 chromosomes. We inherit one set of chromosomes from our mother's egg and the other set from our father's sperm, yielding two copies of each gene. The exception to this is a small set of genes that are unique to the X and Y chromosomes, which determine an organism's sex.

Variants of genes that differ slightly in nucleotide sequence are called **alleles**. An individual with two copies of the same allele is said to be **homozygous** for the corresponding gene. Conversely, an individual with two different alleles for a given gene is said to be **heterozygous** for that gene. **Dominant** alleles are alleles for which a single copy is sufficient to result in a particular trait; the trait will be expressed in a heterozygote. **Recessive** alleles are alleles for which two copies are required for the corresponding trait to manifest; recessive alleles are masked, or hidden, by their dominant counterparts in a heterozygote. Most traits are **polygenic**, meaning that they are determined by the expression of multiple genes.

GENETIC DISEASES AND ATAXIA

Permanent changes in DNA sequence called **mutations** can be inherited or acquired and can affect one or multiple genes. A genetic disease is a disease caused by an inherited change in an organism's DNA sequence. Scientists now believe that most disorders have a genetic component.

Many forms of ataxia are hereditary, resulting from gene mutations and the creation of faulty proteins. In hereditary ataxias, these proteins impair the function of neurons in the cerebellum and spinal cord, resulting in clinical symptoms such as impaired balance and coordination. Ataxias resulting from dominant genes include the **spinocerebellar ataxias** (SCA1 through SCA37). Ataxias resulting from recessive genes include **Friedreich's ataxia and ataxia-telangiectasia.**

As we advance our understanding of genetics, we can better understand and treat complex genetic conditions such as hereditary ataxias. We can also apply an understanding of genetics to improve our general health. Given the intimate interaction between genes and the environment, we can modify our lifestyle factors and activities based on our genetics to improve our health and quality of life. For example, someone with a genetic predisposition to heart disease may choose to adopt a low cholesterol diet. Similarly, someone who has inherited a genetic ataxia may choose to engage in exercises that improve coordination and balance.

Written by Dr. Chloe Soutar
Edited by Celeste Suart

This article was written by Ataxia scientists and originally published by SCAsource. SCAsource works to make research more readily accessible and understandable for Ataxia patients. They partnered with NAF to host their content and publish their articles. They have written more than 100 articles and you'll find new articles each month.

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ataxia.org/support-groups/

ACAREF Foundation – Who We Are

By Silvio Sivieri

My name is Silvio Sivieri, I live in Italy in a town called Ferrara. I am 63 years, and I don't have the disease of Ataxia, but my first wife Alessandra had SCA1. My son Matteo who is 41, is affected due to inheritance from his mother. 10 years ago, together with a group of friends I founded ACAREF Foundation. This reality was born with the aim to support scientific research on Spinocerebellar Ataxia type 1 and 2. For the last 6 years we have been funding a research project on SCA1. Last year, I met professor Dr. Harry Orr, who attended one of our conferences on Ataxia. We believe in alliances and wanted to let you know, we are in Italy fighting with you to find a treatment for Ataxia.

My Personal Note to Ataxia

by Shundra Wooten

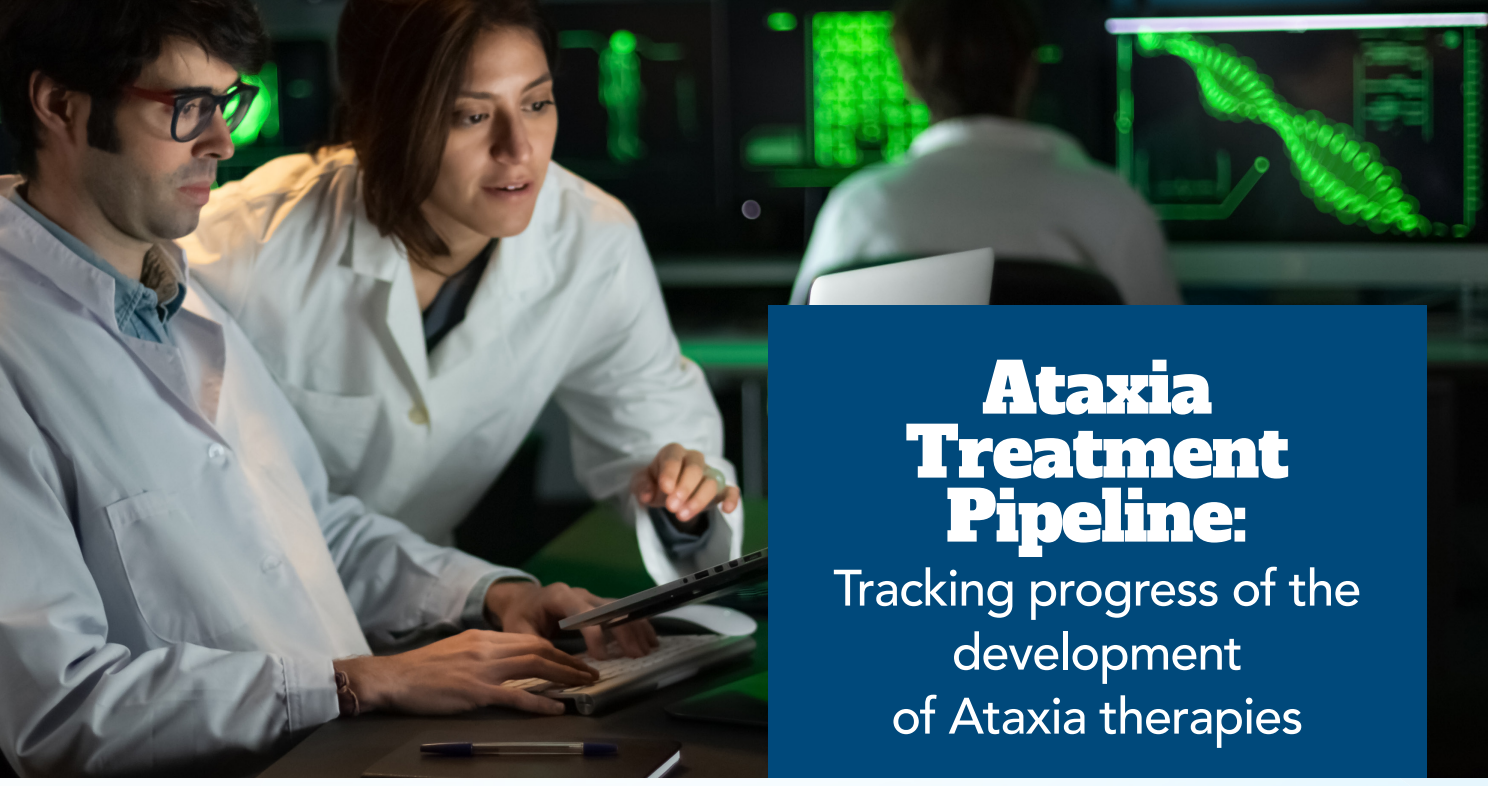
You caught me by surprise. Even though you weren't invited in my life, and I had no idea that you were coming, you took it upon yourself to start showing up when I was 43 years old. Causing me to retire from my world of educating young children because I didn't want to risk my safety or theirs. But however, I do thank you for allowing me the chance to let me go through childhood, be in marching band in high school without any issues, raise my daughter and put her through college as a single mom and for giving me the opportunity to baby sit Ryan and Tyler and to take them to events whenever I had the chance.

Now you are here to stay with no intentions of leaving. Even though you are progressively taking over my physical and neurological health, I refuse to let you take over my mental health.

You have affected and destroyed so many families including mine, but I will continue to be a study participant to assist researchers in any way possible to put a stop to you.

With God's help and the knowledge of the researcher's there will be an end to you one day.

Sincerely,
Shundra Wooten



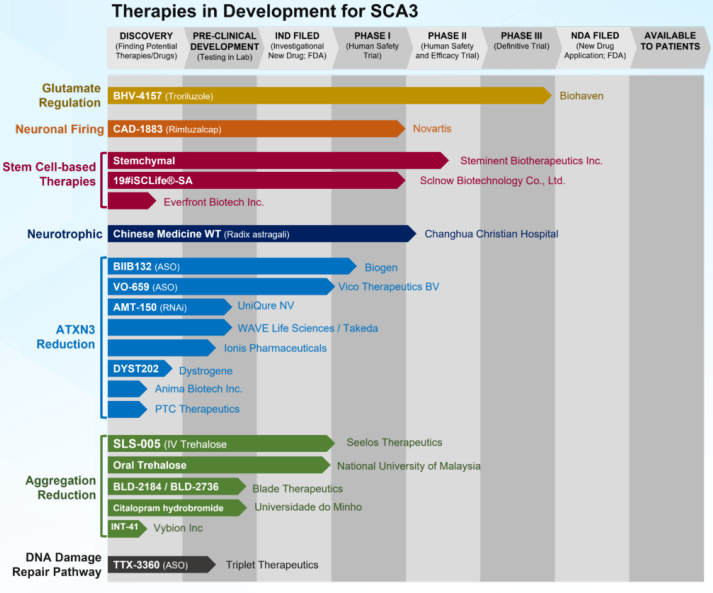
Ataxia Treatment Pipeline:

Tracking progress of the development of Ataxia therapies

We know that treatment development for Ataxias has been changing rapidly. It can be hard to keep track of the different clinical trials and research studies. We developed the Ataxia Treatment Pipeline to make it easier for you to find all the information you need. Treatment development is a long process that involves multiple phases of a clinical trial and oversight by the Food and Drug Administration (FDA). NAF is committed to empowering you with up-to-date information about Ataxia treatment development. The pipelines are visual tools that were created to show the progress of all Ataxia therapies that are currently being developed. There is a pipeline for each type of Ataxia that currently has therapies in development.

These are

- SCA1
- SCA2
- SCA3
- SCA6
- SCA7
- SCA8
- SCA10
- SCA17
- DRPLA
- Friedreich's Ataxia



Sample Pipeline

To view the pipelines **www.ataxia.org/pipeline**

My Fight Against SCA7

By Derrian Hollingsworth

My name is Derrian Hollingsworth. Due to complications caused by having SCA-7, in 2021 (at only 40 years old), I was forced to retire and go on disability permanently. My sister and mother both have SCA-7 and are fully disabled. I have several other family members struggling in different ways with SCA-7. Most heartbreaking of all, I have 2 teenage daughters with SCA-7. My 19-year-old is wheelchair-bound, is legally blind (rapidly approaching complete blindness), and has difficulty speaking, swallowing, and completing any coordinated movement. Due to genetic testing results, we know that my 15-year-old will be there in about 2 years. They will never go to college, live on their own, have careers or families, or any of the options and experiences life affords most people. I started the Hollingsworth SCA-7 Fund (now known as The Hollingsworth Foundation) because, even though there is no treatment or cure, I could not sit back and do nothing…and just watch their health deteriorate and lives fall apart. I want to help them, and anyone affected by SCA-7, in any way that I can for as long as I can. If you have (or know someone who has) SCA-7, you know the situation is humbling, heartbreaking, and hopeless…but we will fight back against it.

It All Started with a Fall

By: Margaret "Peggy" Dawson-Goldberg

I am 86 years old and thank God or as one doctor told me "thank you for grandparents," I'm in wonderful health.

However in 2015, I had a horrific fall. I felt dizzy and nauseous but it quickly went away. I didn't do anything because it didn't seem necessary. Three months later I started to feel "strange". I was driving but turned over the car to my employee. She commented that to her she thought I was sleepy. Several hours later I found that I couldn't walk down some stairs.They took an MRI. It showed that I had a Sub-Duro-Hematoma. They operated immediately. Gradually the dizziness and imbalance started to affect me even more. I don't know if it was caused by the surgery or the fall. I was told that I have Ataxia and there is no cure at this time but walking and exercising were strongly encouraged. Daily, I walk with my service dog and ride my bike. I am able to continue playing tennis and golf. I don't understand how I can play tennis. Maybe it's because I play doubles and dominate the net. While golfing, I try to walk more than ride to help the legs and feet be more active.

I have always been a fan of the healing powers of Acupuncture. An MD specializing in acupuncture comes to my house twice weekly to give me acupuncture. She also adds current to the leg and feet needles at the end of the session. Wow, that really helps activate those nerves!

That's my story of being affected by Ataxia. I'm so grateful to NAF for the informative support. It takes courage to do the activities that are supported by NAF. The results are nothing but marvelous in my opinion. Thank you all! Keep moving my people.

How do I Know if I Qualify for a Research Study?

If you've been involved in research or read about research studies involving human participants, you've likely heard the terms "inclusion and exclusion criteria." While the definitions of inclusion criteria and exclusion criteria seem fairly self-explanatory, it is important to understand some background on why these criteria are utilized.

Let's start by defining a couple of terms and then we'll dive into some of the reasoning behind utilizing inclusion and exclusion criteria.

- **Eligibility Criteria:** A list of factors used to determine whether a person is eligible (inclusion criteria) or not eligible (exclusion criteria) to participate in a study. Eligibility criteria may include disease type and stage, other medical conditions, previous treatment history, age, and/or gender.
- **Inclusion Criteria:** Characteristics of a participant defined by the study team that specifies specify who can participate in the research study.
- **Exclusion Criteria:** Characteristics of a potential participant that disallows them from participating in a given study. The list is produced by the study team to keep all interested participants safe.
- **Remember:** Inclusion and Exclusion criteria are never meant to personally reject potential participants but rather to keep them safe.

While eligibility criteria may feel restrictive, they are an essential element for research involving human subjects because they help ensure that treatments are safe and effective for a range of patients.

Inclusion and Exclusion criteria vary from study to study, especially because a new treatment may utilize a very specific approach and a very specific group of participants is needed. Inclusion and Exclusion criteria are never meant to reject potential participants personally, but rather to keep them safe. Often, the initial criteria are very specific, and if the treatment is successful, the criteria may broaden during a future phase of the study.

What are some common inclusion criteria for Ataxia related studies?

- **Age ranges:** Each study varies, and if a study allows participants under 18, the participant will need consent from a parent or guardian. Because Ataxia typically affects middle aged people, the age range is often targeted to reflect middle age.
- **Diagnosis:** A confirmed diagnosis by a neurologist for the specific type of ataxia or neurodegenerative disease that the study is evaluating.
- **Willingness** to participate and give informed consent

What are some common exclusion criteria for Ataxia related studies?

- **Comorbidities:** Other conditions a person may have that could put them at risk if they participated. Some conditions are excluded if they may affect the results of the study. Some examples may include dementia, cancer, liver disease, and/or severe psychiatric disease.
- **Medications:** Some medications may not be compatible with the study's treatment protocol.
- **Inability to undergo MRI scan:** Some studies require an MRI scan as part of the study protocol. If a participant is unable to participate in an MRI related to weight, cognitive function, or other reasons, they may be excluded.
- **SARA score outside targeted range:** The Scale for Assessment and Rating of Ataxia (SARA) is often used to rate a person's severity of ataxia. Some studies require a specific performance on this scale to be included in the study. This is because this scale is often used to show progress and change in disease over time as a measurement during the study.

Eligibility criteria are a crucial part of research studies and often are determined strategically by the investigators and approved or modified by the Institutional Review Board (IRB) or Ethics Committee. The primary use of eligibility criteria is to ensure participants' safety and help researchers ensure that they can answer the questions they are studying to find future treatments for ataxia.

For a list of current Ataxia related studies, visit
www.ataxia.org/help-develop-new-treatments/

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Ataxia and My Journey with IVF

By Lucy Lin

My name is Lucy, I was diagnosed with SCA3 in 2020, when I was 26. Back when I was 21, my friends, and later, my now husband pointed out to me that I walked weird on hikes. Up until that point, I was a very active person. I got into boxing in university, ran 3-5 miles daily, and pre-pandemic, rock climbed weekly. I used to dream of being like Nikita (the female rogue kickass agent show), using my body as strength that carries through my life.

To be honest, the first year or two was rough, coming to terms with it. And I still have my moments now, but my driving philosophy has become: ataxia is not my fault, but it's my responsibility. To that end, I exhausted all the possible treatment options available to me, attended clinical trials, built a daily vitamins list, got on daily walks, and weekly yoga. I am not exactly thriving, but I am taking responsibility to improve my quality of life. That's important to me. And I have many things to be thankful for, like the clinical advances, the job I have, my husband, and my friends.

I am now in a life stage where I am planning for my future family. I learned of IVF soon after my diagnosis, it was the first thing that my neurologist referred me to, genetic counseling. This year, my husband and I finally made the decision to pursue it. I used fertilityiq.com to narrow down the IVF provider I am interested in.

The first barrier to IVF for many people is the finances. We knew that we wanted to have two kids, that meant we needed multiple cycles statistically speaking to make sure we at least have 4-6 frozen embryos (healthy and Ataxia free). This is because transfer success rate of embryos is 1/3 times roughly. So, for two successful pregnancies, we are looking at 6 healthy embryos. It's not cheap. For ours, it was roughly 80k everything added for 3 cycles.

So far, I have gone through 1 cycle, and now have 3 embryos ready to go under Ataxia testing.

We are waiting for results and hopeful. The fact that this is available to my generation but wasn't when I was born is enough of a perspective for me to feel lucky and excited for the future of clinical advances.

Fingers crossed on my side.

I am open to answer any questions about my IVF journey - **xinyu.lin.lucy@gmail.com**

Visit **www.ataxia.org/webinars** to view past educational webinars about Ataxia that NAF has offered. From yoga to research, you can find them all!

Helping Others While I Still Can

By Lisa Cole

I was diagnosed in December of 2013, first I was misdiagnosed for several years prior. Unfortunately, this misdiagnosis is still going on for so many people.

Earlier that year, in 2013, I was doing a stress test on a treadmill, and I almost fell. That cardiologist said I should get that checked out. I blamed it on my knees and made an appointment with the orthopedic.

The Orthopedic said my walking is not due to my knees but said I should see a neurologist; he referred me to one and I finally went in October 2013.

The neurologist listened to me and sent me up to Gainesville for a second opinion. Both Drs. agreed and said Spinocerebellar Ataxia, SCA for short. I got my family history and my MRI's. My dad, my brother and my aunt all have it. My father passed away over 8 years ago, he was in a nursing home for about 8 months before he died. He was not walking/standing due to Ataxia, he also had non-Hodgkin's cancer and COPD. My family suspected my grandfather (his father) had it too because he showed signs of it. My family just passed it off as the Antonelli thing (my maiden name). Well, back then, no one was aware of what this was or seemed to care. Back then is long ago, I'm 53 today

I didn't go to doctors for about 4 years prior to my diagnoses. I was concerned about my walking. I was still working and going to several clients a week. However, as the time has passed, I would only do my accounting/bookkeeping work in my office. I complained for many years about being dizzy (starting in 2005-2007), not feeling balanced, no coordination and when I close my eyes, I would feel like I was going to fall over or felt my body swaying. I went to a local ENT (ear, nose, and throat). I was treated for vertigo; another long word and I was also sent to physical therapy. I stopped complaining when I saw "CHRONIC ANXIETY" in the office visit notes as one of the illnesses, even though the dizziness and unbalance feeling was still there, I stopped going to doctors and just dealt with it. I didn't have anxiety; I knew something was wrong.

When I told the ENT doctor what I have (the diagnosis), he said: "that's not his field". In a way he's right. But I do feel that all doctors should know what Ataxia is or possibly some other form of rare disease and steer people in the right direction, if possible. Not to just not know of it and pawn it off to anxiety and for us, the patient, to continue to suffer because some doctors do not listen or really don't care what you say. I know my body. I knew something was wrong. It is like you drive your car every day and when it doesn't feel right, you try to explain. Bad analogy, nothing gets fixed until it's broken. Usually then things are too late.

After a long journey with doctors, I was finally able to find the right one to help me, a neurologist that listened to me. I got the diagnoses of Ataxia, which is not a good one, but now I have a reason why I have no coordination, balance, dizziness, etc. At least I know and when it progresses, I can figure out how to deal with it.

Cont. Helping Others While I Still Can

I started a support group here on the East Coast of Florida (the Treasure Coast). Now, I share my journey and want to help others through theirs. I am happy to help as many as I can, while I can. Due to the pandemic, our 3rd and 4th Walk n' Roll to Cure Ataxia fundraisers were virtual. We still raised funds and awareness to help with research. I am so fortunate to have so many great friends that help me have a wonderful and successful event. Awareness is always spread and that is the main goal.

I enjoy bringing awareness to Ataxia. It not only sheds a light on this rare disease but it's helping so many others that might be stuck in their journey somewhere feeling alone. No one should have to go about this alone. We hope you will join us at one of our Treasure Coast Support Group meetings. We meet virtually two Saturdays a month from 12:30pm-2:30pm.

Find the dates at www.ataxia.org/events.

REMEMBERING NAF IN YOUR WILL

Throughout the years, individuals have named the National Ataxia Foundation as a beneficiary in their wills. Their thoughtfulness and foresight have enabled NAF to provide more research studies, more services to patients and families, and more education and Ataxia awareness to the public. We are grateful for the impact that has been made by these compassionate acts. If this is something you would like to consider, please contact Joel Sutherland at **joel@ataxia.org** or call (763) 231-2748.

Memorials and In Your Honor

NAF 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 January - November 2022.

IN HONOR

Alistair Thirkettle
Amy Messigian-Legault
Amy Nakai
Andrea Kiess
Angela Burnett
Anthony Alayo's Wife
Anabela Azeved
Anthony & Elise Matchett
Art Pinkston
Austin Brown
Ayres Family
Ben Lee
Benjamin Cox
Gabriel Cox
Benny Frei
Beverly Lima
Bill Moore
Brad Williams
Brett Masserant
Bryan Scott Palmer
Burl Turner
Cameryn Cobb
Carmen Pieragastini
Carol Biby
Carol, Matt & Nate Stabenow
Carol Tate
Caroline Heilweil
Carolyn Allen
Charlie and Mary Ruehl
Charlie Wong
Cheri Bearman
Chester White
Chicago Ataxia Support Group
Cindy & Jim Bean
Cindy Crook
Clyde Fano
Cynthia Brown
Dana Haven Crisp
Darlene Harris
Deborah Taylor-Omictin
Debra Charlesworth
Debra Covington
Debra Hazlin
Delaine Emmert
Dirk Desserault
Dr. Fogel
Dr. Michael Gorum
Dr. Sow-Hsin Chen
DuCharme Family
Ed Collard
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Emily Rose
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Erin Peterson
Erin Wenger and Sean Greenhalge
Evelyn Chu
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Frank Orlowski
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George Williams
Ginny Haley Gokenbach Gislers
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Kelli Fritz
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Kenneth Randall Harbison
Kennon and Page Davis
Kent Hardel
Keri Naccarato
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Maryellen Tseng
Matthew Lafleur
Michelle Baumgart
Michelle Pinard
Mike Anderson
Mike Clementz
Mike Puckett
Moomaw Family
Morgan Talevich
Mort Family
Nan Feldman
Nan Vail
Nancy Kochevar
Nancy Van Twuyver
Nebraska Ataxia
Ornella Liesenfeld
Patricia Browning
Paul Bice
Paul Richter
Paula Ureta
Penney Soboski
Penni Sutherland
Pennie Haydon
PJ & Richard Byrd
Randy and Beverly Whetstone
Raylan Hardigree
Renuka Kalaria
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Rita Garcia
Rob Razmus
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Robert Hrupek
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Robert St. George
Robert Tucci
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Roger Richard
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Sheng-Han Kuo, MD
Sherri Hubbard
Steven Legare
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Sweeney Family
Tanya Tunstull Marshall
The Demint Family
The Frei Family
Tim, Peter, & JT De Mint
Victor Masserant Family
Wayne Walters
Xander Barnes
Xander Merk

IN MEMORY

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Addy Ancona
Albert Frei
Alice, Christopher & John Tatti
Amy Donnelly Hamrick
Amy Draves
Amy Lynn Bac
Ann and Givens Goodspeed
Appa V. Ramachandran
Arnold Berg
Aymee G. Torres, Dr. Aymee A. Torres -Michels, Ricardo Luis Guerrero & Teresita Guerrero
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Bob Schlickbernd
Bonnie Dunkelberg
Brent Masserant
Brittany Mumford
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Carol Haukos
Carol Heon
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Catherine O'Brien Edge
Cecil Bergman
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Charles Murphy
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Chris Buechel
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Curt Alan Wingerter
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Hope Eggleston
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Tyler John Wells
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Vernon Niebuhr
Victor Masserant
Virginia Horel
Vithal Kardani
Walter Herbert Jones
William Ellis
William Holland
William Oberndorf

Support Groups

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

Support groups can remind us that we are not alone. Each person experiences Ataxia in a different way, but we still have many things in common. NAF coordinates Support Groups as a way for the Ataxia to connect and learn from others. There are more than 65 support groups and hundreds of meetings each year. Our volunteer Support Group Leaders provide this invaluable service to the community. There are in-person and virtual meeting options that vary by group.

If you or a loved one has been newly diagnosed with Ataxia, please contact the support group 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 Sarah Pilato of the NAF staff at sarah@ataxia.org or (763) 553-0093.

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

Join one of our Facebook Support Groups!

NAF Support Group	*facebook.com/groups/NAFmail*
Under 30 with Ataxia	*facebook.com/groups/under30withataxia*
Parents of Kids with Ataxia	*facebook.com/groups/ParentsOfKidsWithAtaxia*
African Americans with Ataxia	*facebook.com/groups/501238100417901*
Spouses and Partners of Loved Ones with Ataxia	*facebook.com/groups/AtaxiaSpousesAndPartners*
U-MATTER, Unbroken - Military Members with Ataxia	*facebook.com/groups/922015755298690*

Learning How to Cope in My New World

By Ed Forshaw

I have hereditary Cerebellar Ataxia - specifically, SYNE1-related Spinocerebellar Ataxia (ARCA8).

Symptoms have slowly crept up during my 20's. It wasn't until I turned 30 in 2021 when I received my diagnosis.

How did it start?

I worked in the IT department for a university. During a meeting in 2015, I noticed it was difficult to speak with my colleagues. This was unusual for me; I usually have a lot to talk about when I'm engaged in something! Slowly, over the years, I noticed it more especially towards the end of the day and when I was tired. Eventually my colleagues, friends, and family noticed me struggling to get words out and occasionally slurring.

Referral to Neurology...?!

Mid-2019, I went to my GP about my speech issues, I was quickly told I had tongue-tie and I was referred to a local speech therapist to confirm and arrange some simple surgery to correct it. After two months of thinking "yes, I can finally look forward to speaking properly again!", I saw the speech therapist. She spent 5 minutes with me and said she needed to refer me to the local neurology department. I was shocked, neurology, they deal with issues affecting the brain, right? Having learnt this, and waiting another 3 months, I saw the neurologist and had an MRI scan of the head and spine arranged.

We're getting somewhere...

Mid-February 2020 I had the MRI. The 10th of March I learned my cerebellum was of diffused volume (i.e. it was scrunched up), and that the neurologist wanted to book me in for some further tests, including a Lumbar Puncture. Towards the end of March 2020, the UK went into a strict lockdown because of the novel coronavirus, and as the hospitals reacted to Covid-19 I expected more delays. It wasn't nice knowing my brain had this issue and I was unable to find out why.

After a few months of worry, eventually I heard I had been referred to the National Hospital for Neurology and Neurosurgery in London. Upon chasing the referral in September 2020, I found out I had been triaged to the "Ataxia Centre" based on my symptoms. This was the first time I had ever heard the word "Ataxia"!

By this time, I was noticing more issues, such as a struggle to maintain a grip (not good whilst playing the guitar!), fatigue, and balance issues.

Seeing the Ataxia Specialist

October 2020, I attended the National Hospital for Neurology and Neurosurgery Ataxia Centre for my initial consultation with a neurologist who specializes in Ataxia. I was told they highly suspected it'll be genetic, and a neurodegenerative disease. This was based on my age, symptom progression, and "clinical picture" (that's a term I've heard a lot!). It was here I found

out my double-vision (caused by weak muscles behind the eyes) was also related to the damaged cerebellum. I never thought my speech issues and double vision were linked!

Diagnosis

Fast forward to May 2021, found out I had a SYNE1-related Spinocerebellar Ataxia (ARCA 8) which is inherited recessively from both of my parents. This was a shock to my family, as I was the first to present symptoms - I have two brothers who are not presenting any symptoms.

Upon asking what my outcome looks like, I was told "we're not sure". My jaw dropped and I was reassured it's expected to be a slow progression and there's some brilliant research into the genetic Ataxias to learn more and work towards a cure.

In April 2022 my parents attended the National Hospital for Neurology and Neurosurgery because the Ataxia Centre is keen to confirm their findings from my genetic testing.

My Thoughts? And What's Next...

It's worrying. Finding out you have this genetic disease which does not have a cure and there's very little known about it. Suddenly a lot of future plans, like financial, relationships, and careers are thrown into the air. It's been almost a year since my diagnosis, and I'm not sure if I have fully accepted it yet. Mostly, I think, because there's a lot of uncertainty. Given I have been living with the symptoms that have very slowly crept up on me, it's difficult to accept such a life-changing diagnosis.

I now work with my dad running a small IT company, we work from our respective homes 99% of the time. Fortunately, this gives me a lot of flexibility, working from home and with a family member, managing our own workload. I also have several friendship groups offering lots of support and understanding of the condition I have.

After my diagnosis, I quickly got involved with Ataxia UK - the leading ataxia charity in the UK. I have joined a local support group, attended their 'All About Ataxia" sessions (for recently diagnosed patients to learn about the condition), and done some fundraising. The fundraising challenge was to walk to your GP's surgery and post the Ataxia UK Medical Guidelines, to raise awareness and improve Ataxia diagnosis and referrals - I had several friends and family members join too!

Unfortunately, one of my hobbies since I was a teenager, playing guitar, is too difficult nowadays. Though, I still have a couple of guitars on display in my flat - I love looking at them! Another hobby, kayaking, is also too difficult on my own however, my father and I bought a tandem sit on top kayak last year, so we can go out together and I don't get too tired (plus I do not have to worry about getting out of a sit-in kayak!)

In the UK, we're lucky to have the National Health Service (NHS). There are tons of health professionals covering many types of healthcare. Using my local GP, or the specialists at the Ataxia Centre, I can access the healthcare I need.

Like many Ataxia patients, I have found it useful talking to others in the Ataxia community and learning how to cope with the condition.

About the Author

Ed Forshaw is 31 years old, works as a Telecom/IT Company Director and lives in Guildford, UK.

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NAF Facebook Page	*www.facebook.com/ataxiafoundation/*
NAF YouTube Channel	*www.youtube.com/user/NatlAtaxiaFound*
NAF Twitter	*www.twitter.com/NAF_Ataxia*
NAF LinkedIn	*www.linkedin.com/company/nationalataxiafoundation*
NAF Instagram	*www.instagram.com/ataxiafoundation/*

Join the Conversation!

More than 10,000 people have joined our Facebook group as a place to connect and learn from one another.

Join at www.facebook.com/groups/NAFmail


Happy Holidays
from all of us at NAF, We wish you and your famlies all the best in the New Year!

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