Ataxia Digest

News from the Johns Hopkins Ataxia Center

JOHNS HOPKINS MEDICINE

Spring 2026

Letter from the Director

Liana S. Rosenthal, MD, PhD

Dear Ataxia community,

One of the most important parts of caring for people with ataxia is making sure there will be enough well-trained specialists in the years to come. Many of you know firsthand how difficult it can be to find a clinician who really understands these conditions. That's why training the next generation isn't separate from patient care—it's part of it.

This work happens in our clinic every month. At nearly every visit, we have movement disorders fellows seeing patients with us, learning how to recognize different types of ataxias, work through the diagnostic process, and help develop treatment plans. We also regularly have medical students, neurology residents, genetic counseling students, speech-language pathology trainees, and sometimes physical therapy trainees rotating through clinic. Each brings a different perspective, and I think ataxia care is better when the entire ataxia care team learn together from the start.

For many trainees, one of the biggest lessons is that ataxia care isn't about a single visit or a single test. It takes careful listening, a detailed exam, thoughtful use of imaging and genetic testing, and an understanding of how symptoms evolve over time. Just as importantly, it means learning how to talk with patients and families about uncertainty, about progression, about symptom management, and about the supports that can improve daily life. These are things best taught through direct patient care.

We've also been fortunate to support deeper training through fellowship opportunities. At Johns Hopkins, our current ataxia fellow, Dr. Dylan Del Papa, is generously supported by The Daniel B. & Florence E. Green Foundation. With that support, he's getting focused clinical training in ataxia beyond what is part of our usual movement disorder fellowship program, and he is also advancing research projects aimed at improving our understanding and management of these disorders. Programs like this matter because they help create physicians who won't just care for patients today—they'll help move the field forward tomorrow.

I've also had the chance to help with training efforts beyond our own center. Since 2022, the National Ataxia Foundation has run an annual Ataxia Clinical Training (ACT) program—an in-person course designed to help residents, fellows, and attending physicians build practical skills in diagnosing and managing ataxia. I've spoken at the course most years as part of my own effort to support this need. One of the best parts of ACT is that it combines lectures and workshops with direct interaction with patients affected by a wide range of ataxias. That kind of hands-on learning is unusual, and it gives participants a much deeper understanding of what ataxia looks like than any lecture could.

For families living with ataxia, advances in treatment and research are understandably the most visible signs of progress. But just as important is making sure more people are prepared to do this work well. Training future specialists takes time, patience, and the generosity of patients and families who let trainees learn from their experiences. I'm deeply grateful to all of you who make that possible.

Warmly,

Liana S. Rosenthal, MD, PhD

Swallowing and Ataxia

By Mackenzie Greulach, M.S., CF-SLP,
Johns Hopkins Hospital

What is Dysphagia?

Dysphagia, in short, is difficulty with swallowing. Some symptoms of Dysphagia may include:
Feeling that food goes down the "wrong way"

- Coughing on liquids, solids, and/or saliva
- Difficulty controlling food in the mouth
- Difficulties with tongue control
- Food or liquid left in the mouth or throat
- Difficulty with chewing food

How is Dysphagia assessed?

To assess swallowing, your doctor will refer you to a Speech-Language Pathologist, or SLP, for a swallowing evaluation. They will ask you questions about what you are currently eating and drinking and about any symptoms of Dysphagia that you may have. They will examine your head and neck, including your mouth, tongue, teeth, and lips. They may also have you eat and drink different consistencies of food and liquid during your appointment.

Based on your swallowing evaluation, you may be referred for a Videofluoroscopic Swallow Study, or VFSS, in order to see how you are eating and swallowing under an X-Ray machine.

How is Dysphagia managed?

Dysphagia in individuals with Ataxia is primarily managed through compensatory strategy training. An SLP can provide specific recommendations for home exercises, positioning changes, diet modifications, and, in some cases, alternative nutrition.

What can I do today to help with my swallowing?

If you are having any symptoms of Dysphagia, please consult your physician. A referral may be made to have an SLP evaluate your swallowing. Additionally, here are some strategies that you can implement today!

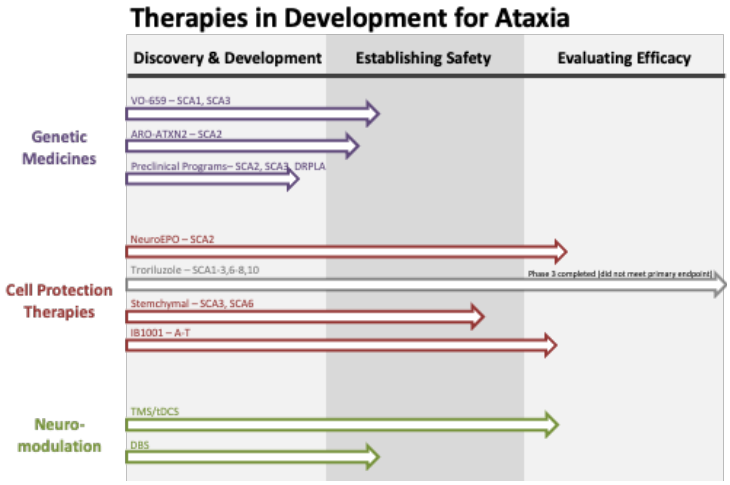
- Slow your pace of eating
- Take small bites of food and small sips of liquids
- Sit upright during and after eating and drinking
- Avoid distractions while eating and drinking
- Avoid talking while eating or drinking

New Frontiers in Therapeutic Development for Ataxia

By Dylan Del Papa, MD – Movement Disorders Fellow, Johns Hopkins University

Over the past decade, our understanding of ataxia has grown significantly. Researchers are developing therapies that go beyond treating symptoms and now working on approaches that target the underlying causes of disease.

Many genetic ataxias—including SCA1, SCA2, SCA3 (Machado–Joseph Disease), SCA6, SCA7, and SCA17—result from abnormal proteins that disrupt how normal brain cells work. These proteins all have one thing in common: abnormally long repetitive sequences called polyglutamine (polyQ) tracts. Because these ataxias all result from proteins with this common problem, this offers a potential target for treatment. Several strategies are being developed to reduce production of the harmful protein.



Genetic Medicines

Anti-sense oligonucleotides (ASOs) and silencing RNA (siRNA) are the leading approaches for stopping this abnormal protein production. These genetic medicines block the instructions that tell cells how to make the harmful protein. The ASO that is the most advanced in its development, called VO-659, is delivered by spinal tap. It is in early safety trials for SCA1 and SCA3. Early data show it is well-tolerated. ARO-ATXN2, an siRNA for SCA2, is being tested in a small safety study outside the U.S. Additional ASO, siRNA, and gene-editing programs for SCA2, SCA3, and DRPLA are in development but not yet tested in people.

Cell Protection Therapies

Other therapies aim to protect or stabilize vulnerable brain cells. These may have more modest effects on slowing disease progression. NeuroEPO is a nasal spray that has shown safety and early signs of benefit in SCA2. A larger trial is underway in Cuba. Stemchymal is a stem-cell therapy given through an IV. It has shown safety with early signs of benefit for SCA3 and SCA6. Troriluzole is an oral medication that affects glutamate, a chemical messenger in the brain. It did not meet its primary goal in Phase III trials, though combined data suggested a small slowing of progression. The FDA denied approval last year based on the available evidence. This creates uncertainty about its future. N-acetyl-L-leucine/IB1001 is in a trial for ataxia-telangiectasia, with results expected later this decade.

Neuromodulation

Neuromodulation uses electrical or magnetic stimulation to change brain activity. It is an emerging therapy for managing symptoms. Non-invasive techniques include transcranial magnetic stimulation (TMS), which uses magnetic pulses, and transcranial direct current stimulation (tDCS), which uses mild electrical currents. Both have shown modest improvements in coordination and balance.

Deep brain stimulation (DBS) is a more invasive approach. It involves surgery to place electrodes in the brain. This offers more precise control over brain circuits. A small 2025 study showed improved coordination six months after DBS in patients with SCA. Several U.S. centers are planning clinical trials. At Johns Hopkins, an FDA Investigational Device Exemption application is in early development. Although still experimental, DBS provides a highly targeted approach that non-invasive methods cannot achieve.

A Legacy Tomorrow, an Impact Today-Johns Hopkins Legacy Match

Thank you for being a partner in our mission. Supporters like you improve outcomes and maximize quality of life for ataxia patients by helping us provide a comprehensive approach to care that incorporates the latest findings of our researcher-clinicians.

We know that treatments will continue to improve and that the most important discovery, a cure, is yet to be made. That's why support for our future is so important. Right now, when you make a legacy gift benefiting the Johns Hopkins Ataxia Center, **10% will be matched with an outright gift of up to $25,000** — amplifying your impact at no additional cost to you.

Legacy gifts that cost you nothing during your lifetime:

Will or Trust — Retain control and use of your assets and modify your plans if your needs change.
Beneficiary Designation — Use a simple form to make Johns Hopkins the beneficiary of all or any part of the remainder of a financial account..

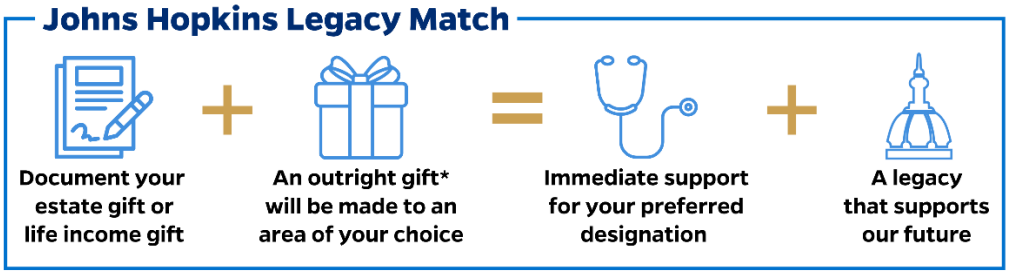
Legacy gifts that pay you back:

Charitable Gift Annuity — Receive guaranteed lifetime income and potential tax benefits; fund with cash, stock, or retirement assets.

Charitable Remainder Unitrust — Receive potential tax benefits and variable income; fund with cash, stock, real estate, or business interests.

If you have included the Johns Hopkins Ataxia Center in your plans — or are considering a gift through your will or trust, a gift by beneficiary designation, a charitable gift annuity, or a charitable remainder trust — please let us know; your gift could be matched today!

To ensure your gift qualifies for the Johns Hopkins Legacy Match, please contact the Office of Gift Planning at *410-516-7954* or *giftplanning@jhu.edu*. Learn more at giving.jhu.edu/legacy-match.



For more information about the Johns Hopkins Ataxia Center and ways to support our work, please contact:

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Upcoming In-Person Johns Hopkins Ataxia Center Events

We have had an exciting Fall 2025 and Spring 2026 at the Johns Hopkins Ataxia Center, marked by meaningful connections and exciting new and returning programs. Our community came together for annual traditions including our fall picnic and holiday party.

Thanks to the generous grant funding from the National Ataxia Foundation, our highly popular and well attended Let's Move Exercise and Wellness Program is going strong. This past January in partnership with a licensed mental health therapist, we launched a new 6 week Mindfulness Program on cultivating self-compassion. Participants learned valuable mindfulness strategies such as deep breathing and self-soothing techniques to manage their anxiety as well as learning how to improve their emotional regulation. To date two cohorts have completed the program with positive feedback. We look forward to welcoming the next cohort in the fall.

Mark Your Calendars — Upcoming Events:

- **Ataxia Adaptive Sailing Day** — Saturday, June 20, followed by a lunch gathering at Little Havana
- **Annual Fall Picnic** — Saturday, September 26, at Oregon Ridge Park in Hunt Valley, Maryland

Please be sure to check your email inboxes for details on these and other educational and social events. Whether in person or virtual, these gatherings are designed to foster an inclusive, supportive community — helping individuals reduce isolation, learn ataxia self-management strategies, and improve overall quality of life.

A strong support network truly makes all the difference. We hope to see you soon!

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MEDICINE

Johns Hopkins Ataxia Research Studies (Current as of 5/01/2026)

IRB approved					
Condition	**Study Name**	**Eligibility/Information**	**Enrollment (Current or Closed)**	**Principle Investigator**	**Contact**
Ataxia	Natural History Study of Genetic Modifiers in SCA **NA_00034854**	Positive **genetic testing either in participant or family** for **SCA 1,2,3,6,7,8,10** Blood sample, neurological exam, and other tests; study visit every 12 months Ages: over 6 years old Reimbursement: $50/session	Open enrollment	Chiadi Oniyike, MD Liana Rosenthal, MD, PhD	Vanessa Nesspor vjohns23@jhmi.edu 410-616-2815
Ataxia	Multimodal Bio-Signal Repository for Parkinson Disease and Movement Disorder **IRB00234370**	Eligibility: Established diagnosis of ataxia or other movement/neuro degenerative disorder English native speaker 1 required visit, lasting ~60-75 minutes total Reimbursement: parking compensation	Open enrollment	Ankur Butala, M.D.	Seneca Motley Cmotley1@jh.edu 667-776-1908
Ataxia	Cerebellar Ataxia Clinical and Genetic Investigation **IRB00414884**	All types of ataxia Yearly blood draw This study aims to identify genetic and blood-based markers of cerebellar ataxia and to better understand the progression of functional and physical changes that occur during the disease process.	Open enrollment	Liana Rosenthal, MD, PhD	Ashley Kucharski akuchar4@jh.edu

Ataxia and vestibular	Identification of relationships of abnormal eye movements and activity in individuals with balance disorders including ataxia and vestibular dysfunction **IRB00246479**	This study aims to understand the relationships of oscillopsia symptoms (bouncy vision and/or dizziness), eye/head coordination, balance and gait in people living with ataxia. Eligibility: Diagnosis of ataxia Ambulatory, without a device Age 18-80 English native speaker 1 session, 2-3 hours No reimbursement, parking pass and test results provided.	Open enrollment	Jennifer Millar, PT	Jennifer Millar jmillar1@jhmi.edu
Ataxia	Mechanisms and Rehabilitation of Cerebellar Ataxia **IRB 00182673**	4-85 years with cerebellar ataxia; this research is being done to learn about how we control movement and how movement is altered when parts of the brain are damaged	Open enrollment	Amy Bastian	Jennifer Keller, PT, MS keller@kennedykrieger.org
Ataxia	Motivated Decision-Making and Performance Cerebellar Ataxia **IRB 00283000**	18-75 years with cerebellar ataxia; this research is being done to understand physical and mental fatigue in individuals with cerebellar ataxia	Open enrollment	Vikram Chib	Jennifer Keller, PT, MS keller@kennedykrieger.org

Other Research Resources

Clinicaltrials.gov ClinicalTrials.gov is a registry and results database of publicly and privately supported clinical studies of human participants conducted around the world.

Connecting Organizations for Regional Disease Surveillance (CORDS) http://www.cordsnetwork.org

Non-Governmental Organization comprised of six international networks, working to reduce and prevent the spread of infectious diseases by exchanging information between surveillance systems globally.

National Ataxia Foundation http://www.ataxia.org/ Dedicated to improving the lives of person affected by ataxia through support, education and research.

Fredreich's Ataxia Research Alliance (FARA) http://www.curefa.org/index.php The Friedreich's Ataxia Research Alliance (FARA) is a national, public, 501(c)(3), non-profit, tax-exempt organization dedicated to the pursuit of scientific research leading to treatments and a cure for Friedreich's ataxia.

The Johns Hopkins Ataxia Center: How to Become a Patient in Our Clinic

Welcome to our Ataxia Center at Johns Hopkins! The first step in the process to becoming one of our patients is to have neurology records sent to us. Please include demographic information (so we know who to contact when we get the records), neurology clinic notes within the past year, reports of your most recent MRI, lab results, and any genetic testing results. These notes can be faxed to 410-367-3212; Attn: Ataxia Center for review by one of our physicians. The decisions to accept a patient into our clinic is based on our neurologist's assessment of whether the patient would benefit from being seen by physicians and therapists with an expertise in neurodegenerative cerebellar ataxia. Based on review of the clinic records, patients may also be scheduled with a physical therapist, occupational therapist, speech therapist, genetic counselor and for vestibular testing, neurocognitive testing, and/or neuro-ophthalmology. Our center believes in a multidisciplinary approach to recognize and treat cerebellar ataxia. All of these appointments are geared towards diagnosing and providing treatment recommendations. Each appointment provides a thorough work up and concentrated care to our patients.

When coming to your appointment day, please make sure to have a copy of the most recent MRI on a CD, and questions to ask your physician. Before you leave the appointment please make sure you have all referrals, orders, prescriptions or refills placed for you. Right after the visit please make sure you call to get a follow up appointment right away, since we tend to book up quickly. We always look forward to assisting in your care!

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Please consider supporting our center!

The work of the Johns Hopkins Ataxia Center would not be possible without the generous support of the Gordon and Marilyn Macklin Foundation, the National Ataxia Foundation, our patients and the community.

For more information about supporting the center, please contact Kimberly Willis, Executive Director of Development at 410-440-3984 or kwillis@jhmi.edu