Ataxia Digest

News from the Johns Hopkins Ataxia Center

JOHNS HOPKINS MEDICINE

Fall 2025

Letter from the Director

Liana S. Rosenthal, MD, PhD

Dear Ataxia community,

In late October, clinicians, researchers, pharmaceutical partners, and representatives from the U.S. Food and Drug Administration (FDA) and the European Medicines Agency (EMA) met in Amsterdam to discuss the future of ataxia clinical trials. The meeting focused on a central question: how do we design studies that can clearly show whether a treatment is helping?

A major part of the discussion centered on choosing the right endpoints—the measurements we use to determine whether a therapy is working. Traditional endpoints, such as walking tests or clinician-rated scales, are important. At the same time, the field is placing increasing emphasis on objective measures, meaning tests that rely on concrete data rather than clinical judgment.

Biomarkers—specific, measurable changes in the body that reflect disease activity—are a key category of these objective endpoints. We reviewed several promising imaging and fluid biomarkers that may support future ataxia trials. The strongest long-term data come from certain genetic ataxias, including SCA1, SCA3, and Friedreich ataxia. These biomarkers are not yet ready for routine clinical use, but they are being evaluated as exploratory measures and, potentially, as primary endpoints in upcoming studies. Because biomarkers are specific to each type of ataxia, identifying and validating them requires careful, long-term research.

Natural history studies play a crucial role in selecting meaningful endpoints and designing successful trials. These studies follow patients over time to understand how symptoms evolve and how biological changes relate to those symptoms. Participants complete a variety of clinical assessments, so researchers can examine how changes in certain proteins or other components of the fluids correspond to changes in coordination, balance, and daily function.

Another theme highlighted throughout the meeting was the importance of the patient voice-what matters most to YOU. Your experiences—what symptoms affect you most, what changes feel meaningful, and what burdens are reasonable during a study—are central to shaping both trial design and the measures we use to judge success.

We also reviewed the broader clinical trial landscape. While no trials of new drugs or gene therapies are currently enrolling in the United States, several are underway in Europe as well as internationally. Within the U.S., there are numerous clinical trials and observational studies that help us better understand how ataxia changes over time. If you are interested in participating in this research, please refer to pages 6&7 in the newsletter for information about studies at Johns Hopkins.

Importantly, the momentum across the field continues to build. Each year brings new tools, deeper understanding, and more collaboration between scientists, clinicians, advocacy groups, and industry partners. While progress takes time, the pace of discovery is clearly accelerating, and the pipeline of upcoming therapeutic approaches is stronger than ever. There is genuine reason for hope, and many promising developments on the horizon for individuals and families living with ataxia.

Warmly,

Liana S. Rosenthal, MD, PhD

Finding Harmony Within: Music, Mindfulness & Self Compassion

By Emily Mahony, MMT, LPMT, MT-BC, LCPC

Life often pulls us in countless directions, leaving us overstimulated, distracted, or even harsh toward ourselves. In the midst of this noise, music offers something grounding: a way to return to the present moment. When paired with mindfulness and self-compassion, music can become a powerful tool for healing and inner balance.

Music as a Path to Presence

Mindfulness is the practice of intentionally bringing awareness to the here and now. Music naturally invites us into this state. Whether it's noticing the gentle rise and fall of a melody, the resonance of a single note, or the rhythmic pattern of percussion, music draws our attention to sensation. By listening deeply—without judgment—we can learn to find rest in the present, even if just for a moment.

The Soundtrack of Self-Compassion

Self-compassion asks us to turn toward ourselves with the same kindness we would offer a friend. Music can support this by softening our inner critic and giving voice to emotions we may struggle to articulate. A soothing playlist, a favorite childhood song, or even humming quietly to ourselves can become a form of emotional care. Music becomes a mirror, reminding us that all feelings are valid and that we are not alone in them.

Practices to Try

Mindful Listening: Choose a piece of music and set aside five minutes to listen with your full attention. Notice the layers of sound, the shapes of the melody, the pauses, and how your body responds.

Compassionate Playlist: Create a playlist filled with songs that comfort and uplift you. Return to it during stressful moments as an act of self-kindness.

Vocal Expression: Singing and humming can help release tension and bring a sense of calm. This isn't about performance—it's about connection.

Movement with Music: Let your body respond naturally to the rhythm. Even gentle swaying can anchor you in your body and ease self-criticism.

Music, mindfulness, and self-compassion are not separate practices but interconnected ways of tuning into ourselves and our needs. By listening—truly listening—we cultivate presence. By offering ourselves gentleness in that presence, we nurture resilience. And through music, we are invited back to our own inner harmony.

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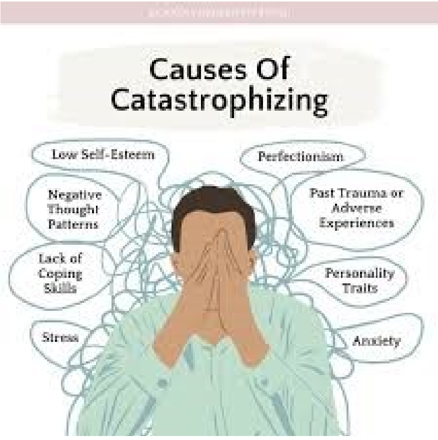
Tips to Cope with Catastrophic Thinking

By Melissa Egerton, M.S., Health Educator, Johns Hopkins Ataxia Center
Source: https://www.choosingtherapy.com/catastrophizing/



Catastrophizing refers to assuming the worst possible outcome will happen. It's something that we all do from time to time, especially when you're feeling anxious, depressed and stressed. Having a chronic progressive condition like ataxia can cause a person to worry about things in the future like injuries from falling, and disease progression. However, when the worrying becomes so constant that it impacts your daily life, that is a sign of a serious mental health condition. There have been several published research studies that show a connection between chronic pain and catastrophizing that is related to pain intensity, severity and distress.
Some common signs of catastrophizing include:

- Persistent anxiety and racing thoughts
- Ignoring positive aspects of a situation
- Making assumptions about the future
- Talking negatively about yourself

Helpful coping strategies:
1. Mindfulness-based stress reduction (MBSR). This teaches how to engage in practices like meditation and yoga to cope with life challenges. It can be an effective tool to keep yourself focused on the present moment and reduce worry about the past and future.

2. Take care of yourself by making sure you get adequate sleep, eat nutritious foods and engage in physical activity. If you are feeling sleep-deprived or stressed, you are more likely to experience catastrophic thinking.

3. Reframe your thinking and challenge your thoughts. Instead of thinking about the worst case scenario, start thinking about the best or better case scenario.

4. Get comfortable with the discomfort. It's perfectly normal to have some anxiety. It's important to sit with your thoughts and know that they will pass if you don't constantly think about them. This takes practice!

5. Take a social media break. The internet is full of people sharing worst-case scenarios to get attention from others, even when those scenarios are not very common. By taking a social media break you can refrain from doomscrolling.

Example: "Bob" has cerebellar ataxia and experienced a fall when going for a walk. He begins to excessively worry about falling and not being able to get back up. His thoughts begin to spiral about worst case scenario of needing to use a wheelchair and becoming a burden to his family. To challenge this thought, you could say to yourself "what if I can handle whatever comes my way. I know I have support available from my family and healthcare team and assistive devices to keep me safe". "Needing help doesn't make me a burden, I am loved by my family and we support each other".
Remember that your fears are understandable, especially when you have a progressive condition that causes uncertainty. Your thoughts are not facts, worst case scenarios are possibilities not absolute certainties. Focus on things that you can control today to build confidence. Be sure to connect with others for support and know you are not alone in your journey.

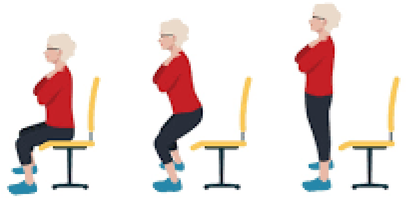
How Much Should I Be Exercising?

By Nicole Miller, PT, DPT & Jennifer Millar, PT
Physical Medicine and Rehabilitation, Johns Hopkins

If exercise came in the form of a pill, it would be the most popular drug on the market as there are so many benefits to exercise. Exercise can help you feel better and function better. You may ask, "How much should I be exercising?"

The ACSM (American College of Sport Medicine) recommends for the general population:

o 150-300 minutes of moderate intensity exercise (this breaks down to 5 days per week of 30-60 minutes of moderate intensity) **OR** 75-150 minutes of vigorous intensity exercise per week (this breaks down to 3 days per week of 25-50 minutes or 5 days per week of 15-30 minutes of vigorous exercise.)



www.steptohealth.com

You may then ask, "What type of exercise should I be doing?"

There are a lot of different types of exercises and having a combination of these components will help us to optimize our movement and mobility. Let's break it down a bit further into categories:

Aerobic Activity is physical activity that uses your body's large muscle groups, is rhythmic and repetitive. Examples: Walking, swimming, using an elliptical, or riding a stationary bike. A general goal of 3-5 days/week for 15-30 minutes is recommended; yet start where you are and work on increasing your duration and level of difficulty.

Strengthening can incorporate everyday tasks (such as squats), use of your own body weight, resistance bands, or added weight. A general goal of 2-3 days/week with rest days in between is recommended

Balance – to be able to safely react to events challenging our balance in our everyday routine, and to help prevent falls. Research has shown that balance training may even have a direct impact on quality and speed of walking.

Flexibility involves stretching your muscles to help with the feeling of tightness, a few days per week if needed Combination of categories: Performing exercises that include more than one of the above category is a great way to incorporate a good mix of different types of activity. Example: A lunge exercise works on strength, balance, and flexibility all in one exercise

Key Considerations when starting an exercise program:

- When first beginning an exercise routine, utilize guidance from your doctor or medical professional, especially if you have underlying health conditions.
- Set up your environment for success – always keep your phone with you, practicing challenging tasks in a safe environment where you have supports or can sit down when needed.
- Start slow – gradually build up how much exercise you are doing. Increasing your activity too fast can lead to overfatigue, imbalance, and falls. It might be helpful to touch base with a physical therapist to help create an individualized plan for you.
- A good rule to follow is to let your symptoms be your guide and don't exercise to total exhaustion.
- Start where you are and gradually build on your abilities. The above guidelines are for the general population. Yet do what you can do. It's okay to modify your goals and expectations.

Mood Fuel

By Ashli Greenwald, MS, RDN, LDN
Johns Hopkins Medicine

“Whenever I get home from work, I just want to go to sleep. The dark evenings and cold weather are zapping my energy.” This is a common thing I hear from many patients in the winter. They might start noticing a dip in their mood and energy during November-February with a resurgence of energy in the spring. Another common theme among my Seasonal Affective Disorder (SAD) patients, is a change in appetite, specifically a craving for refined carbohydrates. Comfort foods may offer a temporary mood boost, but in the long run, they fuel the symptoms of SAD.

Although there’s no cure for SAD, there’s a lot of research on the gut-brain connection, theorizing that diet can play a critical role in managing SAD symptoms. It’s important to focus on nutritional choices that can positively impact mood and energy levels which may improve symptoms.

Here are some of the key nutrients to focus on:

1. Omega-3 fatty acids (FA)

Found in fatty fish, like salmon, mackerel, and sardines, as well as in flaxseeds and walnuts, omega-3 FA are known for their anti-inflammatory properties and their role in brain health. Research suggests they can help alleviate symptoms of depression.

2. Vitamin D

Often called the “sunshine vitamin,” vitamin D is crucial for mood regulation. During the darker months, getting enough sunlight can be challenging, making dietary sources like fatty fish, fortified dairy products, and egg yolks important.

3. Complex carbohydrates

While it’s common to crave simple carbohydrates like sweets and pastries, opting for complex carbohydrates such as whole grains, legumes, and vegetables can help stabilize blood sugar levels and provide sustained energy.

4. B vitamins

These vitamins, particularly B6, B12, and folate, are vital for brain function and mood regulation. Sources include leafy greens, beans, eggs, and lean meats.

5. Magnesium

Known for its calming effects, magnesium is found in leafy greens, nuts and seeds, whole grains, and legumes.

Johns Hopkins Ataxia Research Studies (Current as of 12/01/2025)

IRB approved					
Condition	**Study Name**	**Eligibility/Information**	**Enrollment (Current or Closed)**	**Principle Investigator**	**Contact**
Ataxia	Natural History Study of Genetic Modifiers **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

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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 jmilllar1@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

If you prefer not to receive fundraising communications from Johns Hopkins Medicine, please contact us at 1-877-600-7783 or FJHMOptOut@jhmi.edu. Please include your name and address so that we may honor and acknowledge your request.