Ataxia Digest

News from the Johns Hopkins Ataxia Center

JOHNS HOPKINS MEDICINE

Spring 2024

Letter from the Director

Dear Ataxia community,

As we all know, better treatments are needed to improve the lives of individuals with ataxia. Whether you are someone with ataxia or a family or friend of someone with ataxia, you can be part of developing those therapies through participation in ataxia-related research. Research is how we learn more about how and why the cerebellum changes in ataxia and, from there, what we can do to treat slow or stop those processes. To explain research, let me first explain a few key terms:

- **Clinical research:** Any research that involves or includes humans.
- **Observational research:** Research in which we collect information from participants. We may "observe" participants' clinical exam, blood, spinal fluid, skin biopsies, and imaging including MRI or CT scans.
- **Interventional research/clinical trials:** Research in which participants undergo an intervention. This can include medication, but it can also include medical devices, activity such as exercise, or other procedures.
- **Controls/healthy controls:** Individuals who participate in research who do not have the disease that is being investigated and usually, but not always, do not have other significant medical difficulties. Controls usually should be around the same age as individuals with the disease.
- **Symptomatic therapies:** Interventions that help treat specific symptoms. For example, individuals with ataxia who have significant stiffness can take muscle relaxant medications.
- **Disease modifying therapies:** Interventions that slow or stop the disease from getting worse over time.

Here at the JHU Ataxia Center we have several research studies that we are doing to try and understand the disease process of ataxia and help the larger ataxia community (see Pages 6-7 for an up-to-date list). There are also many valuable research studies going on at other institutions. For many research studies, we need both individuals with ataxia and controls to participate. In fact, it is only through the participation of those without ataxia that we can learn what is different in those with ataxia. Each research study has specific, and usually relatively strict, criteria regarding who is eligible to participate in the study. Importantly, most research does not directly benefit the research participants but instead future individuals with ataxia.

If you are interested in participating in research, please discuss with your physician the study or studies that you are looking at, and whether those would be good and safe options given your specific ataxia and medical conditions. It is only through your help that we can develop safe and effective drug treatments that can lead to better health outcomes and improved quality of life.

Sincerely,

Liana S. Rosenthal, MD, PhD

Liana S. Rosenthal, MD, PhD

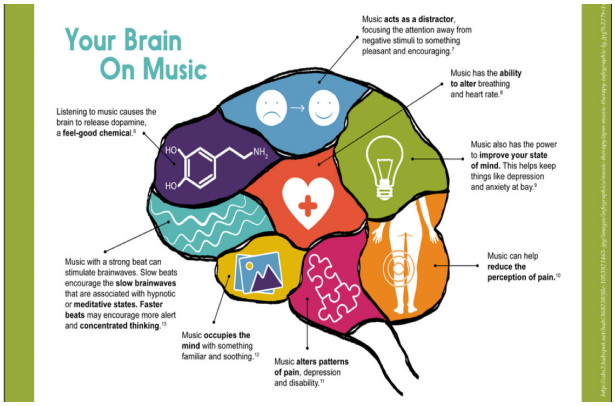
Upcoming in-person events!

* Adaptive sailing in the Baltimore Inner Harbor will take place on Saturday, June 8. Stay tuned for the email invite to RSVP.
* The Fall 2024 Ataxia picnic is scheduled for September 28th at Centennial Park in Columbia, MD. Stay tuned for the email invite to register.
* For more information contact the health educator, Melissa Egerton at megerto2@jhmi.edu

Reflections on a Year of Music Therapy

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

I have enjoyed the privilege of offering music therapy groups at the Johns Hopkins Ataxia Clinic over the past year. It has allowed me to meet so many wonderful people across the world who share the experience of living with ataxia, and allowed me to better understand the unique challenges that those with ataxia face.



Mental Health & Community Music Therapy

Most people enjoy music, and it's all around us. A catchy song comes on, and we start tapping our toes instinctively. Music has never been easier to listen to, with streaming platforms like Spotify, Youtube, and AI software that seems to know what we want to hear before we do. But when was the last time you sat down and really listened intently to a piece of music, noticing the melody, rhythm, and the lyrics? Or a time you chose a song that really resonated with how you were feeling? Or the last time you let yourself sing along, even if you're not a "singer"? These are some of the things you might find us doing in music therapy. Music can help us energize, relax, and process emotions by feeling them.

We, ourselves, are inherently musical; there is rhythm to our heartbeats, our breath, and our movements. Our introductions to the world as babies are filled with music to help us learn, music to lull us to sleep, music to remind us that we are loved. As we grow up, the highs and lows of our lives are marked by music; graduations, heartbreaks, weddings, funerals. The right song may trigger deep feelings of nostalgia, laughter, or tears. Through music, we can explore our feelings, and we can relate to others. Life throws so much at us, and music helps us to ride the waves of our highest joys and our deepest grief.

I invite you to consider how you use music in your life, and to consider ways that you might intentionally incorporate more music into your life. One of those ways might be through attending a music therapy session or series. Two virtual options are available through the Johns Hopkins Ataxia Clinic: music therapy sessions are offered as a 8-week series each quarter, as well as a monthly drop-in session. The next 8-week series will begin Wednesday April 3, at 1:00 pm, EDT, and requires pre-registration. The next monthly groups will be held April 8, May 13, and June 10 at 1:00 pm, EDT.

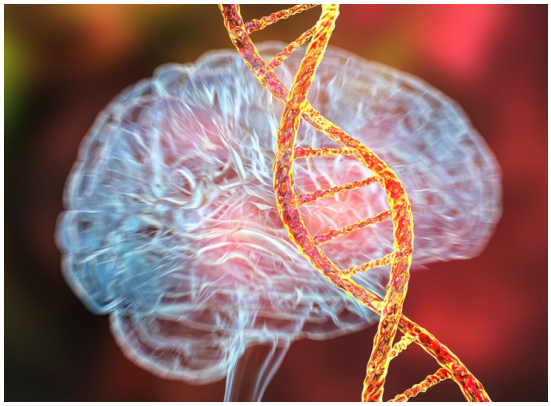
For more information about attending a group music therapy session, please email Emily Mahoney at emahone8@jh.edu

Learn More About Music Therapy:
https://www.musictherapy.org

Four Things to Know About SCA27B

By Weiyi Mu, Sc.M.
Genetic Counselor, Johns Hopkins University

Many people with undiagnosed ataxia are thought to have a yet undiscovered genetic cause. Early last year, an ataxia research breakthrough was announced – the discovery of a new type of hereditary ataxia linked to the FGF14 gene. This new genetic ataxia was named spinocerebellar ataxia type 27B, or SCA27B. The discovery involved a decades-long effort through the international collaboration of two different research teams with numerous ataxia patients and their families. Here are 4 things to know about this newest type of genetic ataxia.



Spinocerebellar ataxia variants show vast heterogeneity | BioWorld

1. **SCA27B may be one of the most common adult-onset hereditary ataxias, especially in people of European ancestry.** In some of these research studies, SCA27B was diagnosed in as many as 6 in 10 people who had undiagnosed ataxia starting in their 30s or later. Symptoms typically start in someone's 40s through 60s. SCA27B seems to be most common in people of French Canadian ancestry, but is found across the globe.

2. **The symptoms of SCA27B can start with episodic ataxia.** In about half of people who have SCA27B, the ataxia starts with episodes of imbalance, slurred speech, double vision, brain fog, lasting for a few minutes to a few days. Over time the symptoms become continuous. SCA27B is very slowly progressive – twice as slow as SCA6 (another slowly progressive genetic ataxia) – and does not affect lifespan.

3. **SCA27B is passed down directly from parent to child.** This is a pattern called autosomal dominant inheritance. This means that if a parent has SCA27B, each child has a 50% chance of inheriting the disease-causing gene. However, not every person who inherits the disease gene may end up developing symptoms.

4. **Genetic testing is available to see if you have SCA27B.** Only a few genetic testing labs currently offer this testing, and many commercial insurances will cover it. If you have Medicare, in most cases you may need to pay out of pocket (~$400-800) for this testing as Medicare usually does not cover it. We hope this will change over time.

It is important to keep in mind that despite the latest advancements, we still do not know every gene for ataxia. The majority of people with likely genetic forms of ataxia remain undiagnosed. If you are undiagnosed, check back with your Ataxia Center often to learn about updates. There are still more questions to be answered, and it is our privilege to be part of your journey.

Look for the latest information including webinars from experts about SCA27B from the National Ataxia Foundation: https://www.ataxia.org/sca27b/

It's Ok to Ask for Help

By Melissa Egerton, MS., Health Educator, Johns Hopkins Ataxia Center
Source: How to Get Better at Asking for Help: 5 Tips from Therapists (happify.com)

Whether you are a patient with ataxia or a care partner caring for someone with ataxia, asking others for help can be very difficult. Everyone likes having a certain level of independence and dignity, and it's often hard to have to surrender control to someone else. However, we need to learn how to be both independent and dependent, it isn't always one or the other. Asking for help is not a sign of weakness nor is it being a burden to others, it's a sign of being human. More times than not, people want to make a difference in people's lives and they feel good conducting acts of kindness.

Here are some tips for learning how to get better at asking for help:

1. **Practice:** the more you ask for help the easier it will become by building up your confidence. Begin with simple requests like asking someone to pick you up something.

2. **Skip sugar coating:** You may feel like you need to offer the other person some kind of return form of compensation. Asking for help should not feel transactional. You should be direct and to the point. If the person you are looking to for help insists that they want to be compensated, you should try asking someone else.

3. **Be clear on what your needs are:** before you ask for help, it's helpful to reflect on what you need and want. Make sure you clearly communicate your needs to increase the chance of receiving the help you requested.

4. **Lean into gratitude:** we all enjoy the feeling we get knowing we are appreciated by someone else. Showing your appreciation shows you value the other person and in return, that person will be more likely to help in the future.

It's Okay- Self-Care Poster

Use of Weighting

By Jennifer Millar, PT, MS, Johns Hopkins School of Medicine

While exercise is one of the most impactful ways to improve your movement while living with ataxia, you may be hoping for additional solutions to help you gain more control of your motor function. Some of you reading this article may either have heard of or have tried the use of weights – either use of limb or torso weights.

Use of upper limb weights in ataxia may help slow down a movement to facilitate using utensils when eating, yet the use of limb weights unfortunately does not translate to learning a new movement pattern that is retained when the limb weights are removed. Interestingly, the use of external limb weights to control acceleration and inertial forces has been proven ineffective in people with degenerative cerebellar ataxias (Zimmet et al, 2019). Use of weights to strengthen one's legs is of course fine!

Torso weighting on the other hand has been shown to have promise in improving postural stability and function in individuals with ataxia on a case by case basis in the short and/or long term. The type of weighted vests that I'm referring to is where light weights are strategically placed on the trunk to counter postural instability and change center of mass control (as opposed to wearing a heavy backpack or fishing vest with weights in the pockets).

Balance Based Torso-Weighting (BBTW), Balance Wear®, is commonly used in clinical practice and was developed based on previous evidence literature by Cindy Horn, a neurological specialized PT in Oakland, CA. For more information on Balance Wear®, including how to find a Balance Wear® clinician refer to the website, www.motiontherapeutics.com.

To pursue a formal assessment for determining if torso weighting is effective in your case, it's recommended that you schedule an in-person physical therapy assessment by a neurological PT with specialized training in torso weighting. Clinical and functional measures would be completed to compare your functional performance with and without the torso weighting.

In many cases we find that individuals with sensory ataxia and/or neuropathy are individuals who are ambulatory with or without a device achieve the most benefit from torso weighting. The nice features of the Balance Wear® vest is the garmet is customized for you, using lightweight material that may be worn under your clothes.

Implementing routine balance training and aerobic exercise may have the same or better efficacy as the torso weighting. So you are encouraged to keep up with your regular exercise routine first and foremost to help manage your motor control symptoms.

Zimmet AM, Cowan NJ, Bastian AJ. Patients with Cerebellar Ataxia Do Not Benefit from Limb Weights Cerebellum. 2019;18(1):128-136. doi:10.1007/s12311-018-0962-1

Johns Hopkins Ataxia Research Studies (Current as of 3/01/2024)

IRB approved					
Condition	**Study Name**	**Eligibility/Information**	**Enrollment (Current or Closed)**	**Principle Investigator**	**Contact**
Ataxia	Transcranial Direct Current Stimulation (tDCS) to augment dysarthria treatment in neurodegenerative ataxias **IRB00239380**	10 sessions of free speech therapy; 5 sessions combined with sham. Age 18-80 years Right handed Fluent speakers of English No reimbursement, free parking, and free speech therapy	Open enrollment	Rajani Sebastian, PhD	Sarah Cust, SLP scust1@jhmi.edu 410-502-2445
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 and MSA	Biomarkers for ataxia and Multiple System Atrophy **IRB00205116**	Cerebellar ataxia (of unknown etiology) with symptoms for at least 8 years or MSA diagnosis Blood draw, lumbar puncture, cognitive testing 1 visit with possible yearly follow ups $100 for 1st visit, $25 for subsequent visits	Open enrollment	Liana Rosenthal, MD, PhD	Vanessa Nesspor vjohns23@jhmi.edu 410-616-2815
Ataxia	Multimodal Bio-Signal Repository for Parkinson Disease and Movement Disorder **IRB00234370**	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 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	Ataxia Clinical Research Registry **IRB00191999**	Anyone who is seen at the Ataxia Clinic Will serve as a recruitment database and a clinical data database No reimbursement No additional visits are required	Open enrollment	Liana Rosenthal, MD, PhD	Melissa Egerton Megerto2@jhmi.edu

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 decision 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 upon 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!

~ *Teshome Wubishet, Ataxia Clinic Coordinator*

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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*, the *Green Family Foundation,* our patients and the community.

For more information about supporting the center, please contact Kaylin Kopcho, Senior Associate Director of Development at **443-287-7877** or **kaylin.kopcho@jhmi.edu**.