

## Letter from the Director



Liana S. Rosenthal, MD, PhD

Dear Ataxia community,

I am excited to share with all of you that there is now an FDA approved medication to treat one type of genetic ataxia! The medication Skyclaris, whose generic name is omaveloxolone, was approved in late-February 2023 to treat a type of ataxia called Friedreich's Ataxia (FA). Individuals with FA typically develop ataxia symptoms in their teens and will often end up requiring a wheelchair to move around about 15 years after symptoms begin. They also develop speech difficulties, scoliosis, and heart problems. It is currently estimated that about 5,000 people in the US have FA. Research studies showed that individuals with FA who took Skyclaris had less decline in function over time than those who did not take the medication. This is an exciting finding for individuals with FA and we are hopeful that the medication will make a meaningful difference in the lives of individuals

with this disease. If you are reading this and have FA, please contact your physician about whether the medication would be appropriate for you.

Skyclaris specifically treats the way the neurons change in people with FA, so it cannot be used to treat any other forms of ataxia. Nevertheless, the approval of a drug for FA is a big step forward for the entire ataxia community. It demonstrates to pharmaceutical companies that they too could get a medication approved for use in those with ataxia. It also even further motivates scientists, researchers, and funding agencies to apply more effort to our understanding of how ataxia works and identifying better treatments.

Even with this new medication, we will continue with our current management of FA that tries to improve quality of life and alleviate symptoms as much as possible. We will continue to recommend exercise, physical therapy, speech therapy, and occupational therapy as well as referral to and management by other medical specialists as needed. In addition, we will also continue to search for even better medications that slow or stop the progression of FA. For the other forms of ataxia, we will continue to search for medications that improve symptoms and slow the disease while also working to optimize current function. The approval of Skyclaris is an exciting and groundbreaking step, but it is only the beginning.

With much hope,

Liana S. Rosenthal, MD, PhD

## Swallowing Problems and Ataxia

*By Rachel Glendenning, MS/CCC-SLP*

Difficulty with swallowing, also known as “dysphagia”, can be a common symptom of ataxia. Just like the coordination required for walking and talking, swallowing requires coordination and timing of multiple muscles to be safe and efficient. What does difficulty with swallowing look like? Below are some common signs and symptoms:

- Food, liquid or saliva escaping from the lips (includes drooling)
- Difficulty to chew solid foods completely
- Pocketing of food in the cheeks
- Runny nose or sneeze with food or drink
- Watery eyes with food or drink
- Coughing or clearing throat
- Choking
- Fever
- Respiratory illness
- Feeling of something ‘stuck’ in throat after swallowing
- Difficulty swallowing pills



If you experience any of the symptoms listed on a regular basis, you should contact your doctor, who can provide a referral to see a speech-language pathologist. With a speech-language pathologist, or “SLP”, you may participate in various evaluations to measure muscle strength, range of motion, and coordination. You may also be asked to try to swallow food and liquid of various consistencies for the clinician to look for the signs and symptoms listed above. The patient may be referred for further testing to evaluate swallowing. This may include a videofluoroscopy (VFSS; modified barium swallow study) or a fiberoptic endoscopic evaluation of swallowing (FEES). These objective tests are crucial for establishing treatment recommendations, diet recommendations, and compensatory strategies. In some scenarios, an SLP may prescribe exercises and/or postural modifications to facilitate safe swallowing, as well as diet modifications (i.e. thickened liquids, avoiding certain food items, etc.). Recommendations often include compensatory strategies that may help in reducing aspiration with meals. Below are common compensatory strategies that may be helpful:

- Sit fully upright for all oral intake (meals and while drinking)
- Remain upright for at least 30 minutes after meals (longer if you suffer from reflux)
- Alternate solids and liquids
- Take small, single bites and sips, swallowing completely before taking another bite or sip
- Minimize distractions with meals
- Take your time!

When it comes to aspiration, remember that aspiration alone does not necessarily lead to an infection such as pneumonia. Good oral health and a patient’s immune status are also important in minimizing infections. Therefore, being sure to complete thorough oral hygiene can help in minimizing risk of infection, even in the presence of aspiration. If you are experiencing increased difficulty with swallowing, consult with your physician, who can refer you for a swallowing evaluation with a speech-language pathologist, for personalized care and recommendations.

## Why In-Person Events Help Build Community

By Melissa Egerton, M.S., Johns Hopkins Ataxia Center Health Educator

Source: [How to Build Community and Why It Matters So Much](#)

During my time as the Johns Hopkins Ataxia Center's health educator, I have experienced both the pros and cons of performing my job during the Covid-19 pandemic. Thanks to the Covid-19 vaccine and boosters we are now returning to our pre-covid lives. My main role as a health educator is to provide educational outreach to promote overall health and well-being and help people living with ataxia manage their condition and improve their quality of life. As the Ataxia Center has started to transition to having more in-person events, I have noticed the importance of what it means to be a part of a community. It was so evident at our holiday party in December how excited everyone was to reconnect in person with their friends and feel socially connected to each other.



Community is integral to our overall well-being. According to a study by Queen's University, 27% of people said they were suffering from loneliness. Having a strong community provides a deeper sense of purpose and each individual in the group adds to the value of the whole. It's wonderful to experience people sharing their perspectives, life experiences and challenges. Each person's experience matters and people are eager to exchange their life stories and resources with one another and demonstrate compassion.

By attending in-person events, we thrive as an ataxia community and feel a strong sense of belonging. People feel valued for who they are and invested in each other's success. It's a lot easier to really get to know each other in-person versus online and when you know each other, you are more likely to feel supported.

While it's not always easy to get out of the house for various reasons, it's worth the effort and time to try and attend as many in-person events as possible. We all know how socially isolating living with ataxia can be and attending in-person events can help people feel less alone in their journey.

We look forward to seeing you at our upcoming in-person events!

## Upcoming In-Person Events!

- \* We are excited to announce that our **Annual Adaptive Ataxia Sailing Day** is happening on **Saturday, June 10, 2023**. This event is through the Downtown Sailing Center in Baltimore. They provide the community with accessible quality sailing programs. This event allows individuals with ataxia and their family members the opportunity to sail around the Baltimore Inner Harbor in accessible boats. After the event, there will be a lunch social gathering at Little Havana restaurant. This event is very popular, and registration is limited.
- \* On **Saturday, May 13, 2023** the Johns Hopkins Ataxia Center along with the Johns Hopkins Parkinson's Disease and Movement Disorders Center is hosting an in-person **Holistic Wellness Event** in Pikesville, MD to promote and foster overall health and well-being. This event will feature a formal presentation on Cognition and Emotion, break-out sessions from health clinicians and a resource fair.
- \* Our **Annual Fall Picnic** will be held on **Saturday, October 7, 2023** at Oregon Ridge Park.



For more information and to register for these events, contact the health educator, Melissa Egerton at [megerto2@jhmi.edu](mailto:megerto2@jhmi.edu)

## Nutrition Tips for a Healthy Body and Happy Brain

*By Leah Fitchett, RD, LDN*

A healthy diet can be a powerful tool for recovery from illness and management of chronic conditions. Good nutrition provides the body with the building blocks for muscles, nerves, body cells, and much more. Studies have shown that good nutrition is associated with improved functional outcomes like strength and stamina, as well as overall longevity. A healthy diet can also help to reduce your risk of developing other chronic conditions like diabetes, heart disease, and cancer. One of the most well studied diets associated with longevity and reduced risk of diseases is the Mediterranean Diet.

### Components of a Mediterranean Diet:

- Include lots of fruits and vegetables
- Choose whole grains like oats, whole wheat, brown rice, and quinoa
- Choose fish and chicken, limit red meat and pork
- Include plant-based proteins like nuts, seeds, and beans
- Choose healthy fats like olive oil, seed oils, and avocado
- Choose low fat dairy products
- Limit excess salt and sugar

Cooking food at home has many advantages, like being healthier and less expensive than takeout or frozen premade meals. Homemade meals tend to be lower in salt, fat, sugar, and preservatives. They also tend to be higher in vitamins, minerals, and fiber. However, cooking food at home can be time consuming and challenging for anyone, but especially for those with ataxia who may have fine motor, coordination, and balance difficulties.



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### Here are some tips to make home cooking easier:

- Choose frozen, pre-cut vegetables: frozen vegetables are just as healthy and nutritious as fresh vegetables. This is because they are picked and flash frozen at the height of freshness to preserve their flavor and nutrition.
- Choose fruits and vegetables that do not require additional chopping: such as baby carrots, snap peas, berries, applesauce, bananas, mini sweet peppers, spinach, and cherry tomatoes.
- Buy pre-portioned and/or pre-seasoned meats like chicken breast and fish fillets to save prep time and effort.
- Cook food in bulk and freeze for later: foods like soups, stews, pasta, and casseroles freeze well for later use.
- Cooking in bulk can seem daunting at first but will save you time and effort later on. Make sure to label and date foods when freezing for later use.
- Look for “One Pot” or “One Pan” recipes. These tend to be quick, easy, and require fewer dishes.
- Gather all of your materials into one place before you start cooking to reduce the need for moving around during preparation. Materials include ingredients, spices, cooking oils, bowls, prep tools, and cookware.
- Sit down at a table while doing prep work to reduce fatigue and limit balance difficulties.
- Invest in adaptive equipment: identify aspects of food preparation that may be difficult for you. Talk to your occupational therapist, doctor, or dietitian about what adaptive cooking equipment may be right for you. Examples of adaptive equipment include grippy pads for opening jars, utensils with weighted handles, and hands-free can openers.

For healthy recipe inspiration created by dietitians, visit <https://www.eatright.org/recipes>

## The Impact of Cerebellar Ataxia on Quality of Life

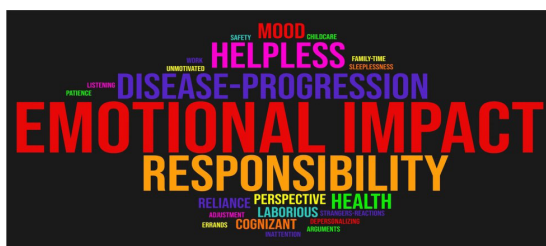
By Cherie Marvel, PhD and Prianca Nadkarni, BS

Having a deep understanding of how cerebellar ataxia impacts quality of life can be helpful to patients, their families, and clinicians. A recent study conducted by researchers at the Johns Hopkins Ataxia Center studied the impact of cerebellar ataxia on the quality of life (QoL) of both patients and their “informants” (e.g., a family member). Interviews were given to patients and informants separately to obtain their perspectives regarding the motor, cognitive, and psychosocial variables associated with the disease. Open-ended questions were used to understand how people were doing in terms of their activities of daily living and real-world tasks. Responses were then categorized into 15 QoL themes that represented the changes experienced by the patients following the onset of ataxia symptoms.

The patients indicated that their biggest challenge was in fitness and physical activities. Informants, however, responded that, from their perspective, independence was the biggest challenge to patients. The patients and informants generally agreed on the severity of symptoms, including posture and gait, fine motor tasks (e.g., using utensils, buttoning a shirt, tying shoelaces), speech, feeding, swallowing, and vision impairments. Interestingly, the patients’ verbal report of their symptom severity of motor-related functions strongly correlated with scores from the International Cooperative Ataxia Rating Scale (ICARS), a commonly used neurological examination tool used by clinicians and researchers to assess symptom severity.

The study also identified associations between motor impairments and specific psychosocial difficulties, which could have important prognostic implications. For example, the study found that impairments of posture/gait and for daily activities/fine motor skills were related to an inability to complete light household duties or responsibilities, such as sending emails or organizing files. These impairments were also related to difficulties with multitasking. Additionally, impairments of posture/gait and daily activities/fine motor skills were associated with impulsive behaviors. In contrast, impairments in posture/gait and daily activities/fine motor skills were not related to memory changes.

Anxiety and depression, along with increased agitation and mood lability, is a common feature of cerebellar ataxia, reported in over 95% of the patients who were interviewed. It is important to recognize that a diagnosis of cerebellar ataxia impacts the patient and family and should be approached in a holistic manner. Informants were asked an open-ended question, “What are the biggest challenges while supporting your loved one?” Responses are shown below in a word cloud, where bigger words represent a higher instance of reporting.



These results provide a useful resource for patients, caregivers, clinicians, and researchers by characterizing the symptoms and challenges associated with ataxia. The findings suggest that clinicians should be mindful of the interplay between motor and psychosocial variables in the assessment and management of patients with ataxia. Moreover, the study highlights the importance of integrating the perspectives of both patients and caregivers to gain a more comprehensive understanding of the impact of the disease.

Joyce, M.R., Nadkarni, P.A., Kronemer, S.I. et al. Quality of Life Changes Following the Onset of Cerebellar Ataxia: Symptoms and Concerns Self-reported by Ataxia Patients and Informants. *Cerebellum* 21, 592–605 (2022). <https://doi.org/10.1007/s12311-022-01393-5>

Kronemer, S.I., Slapik, M.B., Pietrowski, J.R. et al. Neuropsychiatric Symptoms as a Reliable Phenomenology of Cerebellar Ataxia. *Cerebellum* 20, 141–150 (2021). <https://doi.org/10.1007/s12311-020-01195-7>

## Johns Hopkins Ataxia Research Studies (Current as of 4/1/2023)

Condition	Study Name	Eligibility/Information	Principle Investigator
Ataxia	Transcranial Direct Current Stimulation (tDCS) to augment dysarthria treatment in neurodegenerative ataxias <b>IRB00239380</b>	10 sessions of free speech therapy; 5 sessions combined with sham Age 18–80 years old  Right handed  Fluent speakers of English	Rajani Sebastian, PhD  <b>Contact</b>  Sarah Cust, SLP <a href="mailto:scust1@jhmi.edu">scust1@jhmi.edu</a>  410-502-2445
Ataxia	Natural History Study of Genetic Modifiers in SCA <b>NA_00034854</b>	Positive <b>genetic testing either in participant or family for SCA 1,2,3,6,7,8,10</b>  Blood sample, neurological exam, and other tests; study visit every 12 months  Ages: over 6 years old	Chiadi Oniyike, MD Liana Rosenthal, MD, PhD  <b>Contact</b>  Vanessa Nesspor <a href="mailto:vjohns23@jhmi.edu">vjohns23@jhmi.edu</a>  410-616-2815
Ataxia and MSA	Biomarkers for ataxia and Multiple System Atrophy <b>IRB00205116</b>	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	Liana Rosenthal, MD, PhD  <b>Contact</b>  Vanessa Nesspor <a href="mailto:vjohns23@jhmi.edu">vjohns23@jhmi.edu</a>  410-616-2815
Ataxia	Multimodal Bio-Signal Repository for Parkinson Disease and Movement Disorder <b>IRB00234370</b>	Established diagnosis of ataxia or other movement/neurodegenerative disorder  English native speaker  1 required visit, lasting ~60-75 minutes total	Ankur Butala, MD  <b>Contact</b>  Seneca Motley <a href="mailto:cmotley1@jh.edu">cmotley1@jh.edu</a>  667-776-1908
Ataxia	Understanding Fatigue in Cerebellar Ataxia <b>IRB00283000</b>	The study will take one session (2.5 hours)  You will perform a series of hand-gripping tasks and choices  Age 18-80 years  Fluent speakers of English	Vikram S. Chib, PhD  <b>Contact</b>  Agostina Casamento-Moran <a href="mailto:jacasame1@jhu.edu">jacasame1@jhu.edu</a>

Ataxia and vestibular	Identification of relationships of abnormal eye movements and activity in individuals with balance disorders including ataxia and vestibular dysfunction <b>IRB00246479</b>	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 years old English native speaker  1 session, 2-3 hours	Jennifer Millar, PT  <b>Contact</b>  Jennifer Millar <a href="mailto:jmillar1@jhmi.edu">jmillar1@jhmi.edu</a>
Ataxia	Ataxia Clinical Research Registry <b>IRB00191999</b>	Anyone who is seen at the Ataxia Clinic  Will serve as a recruitment database and a clinical data database  No additional visits are required	Liana Rosenthal, MD, PhD  <b>Contact</b>  Melissa Egerton <a href="mailto:Megerto2@jhmi.edu">Megerto2@jhmi.edu</a>
Ataxia	Using Motor Imagery and Machine Learning-Based Real-Time fMRI Neurofeedback to Improve Motor Function in Cerebellar Ataxia <b>IRB00281329 and IRB00300264 and NCT05436249</b>	To use MRI and motor imagery to improve motor function in cerebellar ataxia  Ataxia and health controls: 18–100 years old  Diagnosis of SCA or cerebellar ataxia	Cherie Marvel, PhD  <b>Contact</b>  Cherie Marvel, PhD <a href="mailto:Cmarvel1@jhmi.edu">Cmarvel1@jhmi.edu</a>
Ataxia	Cerebellar Involvement in Cognitive Sequencing	To use fMRI to determine how the cerebellum is involved in cognition  Age 18–75 years old  Fluent English speaker  Confirmed spinocerebellar ataxia diagnosis	John Desmond, PhD  <b>Contact</b>  Rida Saeed <a href="mailto:rsaeed5@jhu.edu">rsaeed5@jhu.edu</a> 410-502-2150
Ataxia	Monetary decision-making task to study reward perception in Cerebellar Ataxia <b>IRB00283000</b>	The study will take one session (2.5 hours)  Age 18–80 years old  Fluent speakers of English	Vikram S. Chib, PhD  <b>Contact</b>  Joonhee Lee <a href="mailto:jlee552@jhu.edu">jlee552@jhu.edu</a>

#### OTHER RESEARCH RESOURCES

**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 persons affected by ataxia through support, education and research.

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

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

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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 genetic testing results. These notes can be faxed to 410-367-3318; 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!

*- 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, Director of Principal Gifts at **443-287-7877** or **kaylin.kopcho@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](mailto:FJHMOptOut@jhmi.edu). Please include your name and address so that we may honor and acknowledge your request.