

Letter from the Director



Liana S. Rosenthal, MD, PhD

Dear Ataxia community,

In November 2024, research scientists and physicians from around the world gathered in London, England, for the International Congress on Ataxia Research. This meeting was a unique chance to learn about the newest and most exciting advances in understanding and treating ataxia.

One major focus of the conference was on the “cellular and molecular” causes of ataxia. This means studying what happens inside cells in the cerebellum to cause ataxia. Researchers tried to better understand why those cells stop working or are dysfunctional. These discoveries could lead to new treatments by targeting the root causes of the disease. But the research didn’t stop at the lab—there were also many studies aimed at improving care for those currently with the disease and potential treatments that are already being tested in patients.

Gene therapy was a big topic of discussion. For example, one study looked at how antisense oligonucleotides (ASOs) might help, though the final result of that study is still pending. ASOs are tiny pieces of genetic material that block the gene from making the dysfunctional protein. Researchers also shared early findings from a gene therapy study for Friedreich Ataxia (FA), which gave hope for decreasing the cardiac complications from FA.

While not gene therapy, another study compared the effects of troriluzole, a drug being tested for ataxia, to how the disease naturally progresses. These comparisons help researchers know if a treatment truly works. Based on the results of that study, the drug company that makes troriluzole is applying to the Food and Drug Administration to try and get the drug approved to be prescribed in the US.

For patients looking for ways to manage ataxia today, there was good news about home-based exercise programs. A new study showed that such programs could improve motor function and adds to the long list of studies showing that exercise can help patients. This means patients can take steps now to improve their quality of life while waiting for future therapies.

Improving clinical trials was another key theme. Scientists want to make sure that trials are as effective and patient-friendly as possible. Ideas included using advanced imaging, artificial intelligence (AI) methods, and patient-reported outcomes to measure progress in new and more meaningful ways. This will help get better treatments approved faster.

The energy at the conference was inspiring, and the research shared gave real hope for the future. Whether it’s breakthroughs in gene therapy or practical advice like exercise, the work being done brings us closer to a world where ataxia is better understood and treated.

Sincerely,

Liana S. Rosenthal, MD, PhD

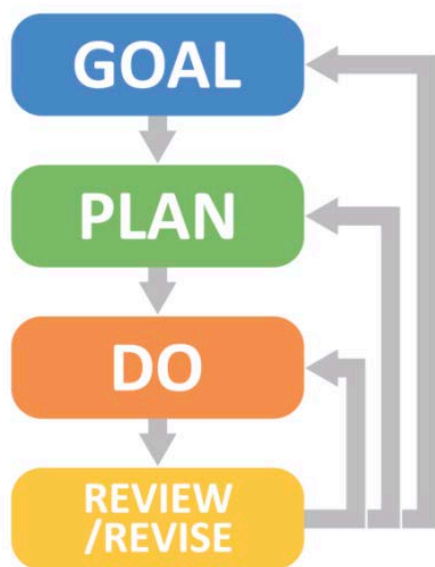
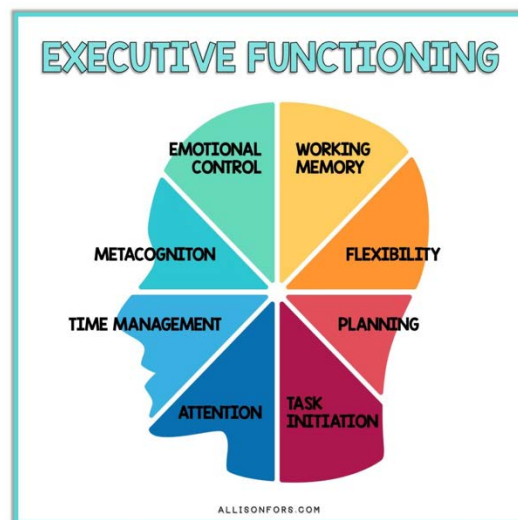
Executive Functioning

By Lindsey Kelly, M.A., CCC-SLP

Executive functioning is an umbrella term that includes critical skills such as planning, problem-solving, reasoning, initiation, persistence, organization, and awareness. These skills allow us to adjust our thoughts and actions during novel tasks. Inventing strategies, finding creative solutions, and persisting through roadblocks will help us achieve our goals.

It is important to emphasize that behaviors can change, but practice and training is required to do so. To invest in all versions of yourself, take some time to reflect on your current and future goals. Be realistic and understand that errors are a part of the process.

Goal, Plan, Do, Review and Revise is a framework that can support executive functioning skills to facilitate success in goal attainment if practiced regularly.



1

Set a realistic goal that you want to accomplish!

2

Create plan; identify problems and solutions.

3

Execute your plan of action.

4

Reflect on actions and progress; re-assess goal.

"Your future is created by what you do today, not tomorrow." - Robert Kiyosaki

(Kamath, 2023)

Hope Can Be Learned

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

Source: *Hope: Why it matters - Harvard Health, Hope Psychology: What Are The Benefits of Hope? | Psych Central*

There's no doubt that living with ataxia is hard! Little by little you are probably experiencing loss. Loss of physical strength and mobility, emotional and cognitive challenges and grieving the loss of your old self before your disability. It can be difficult to move forward with an optimistic attitude, especially when there is no cure for most ataxia.

To quote Michael J. Fox, "Gratitude makes optimism sustainable". Having hope requires optimism and willpower, despite experiencing adversity and difficult emotions. Hope is a belief that some of your future will be better than the present and you have the ability to make it happen. This definition is derived from "Hope Theory" which is a positive psychology concept developed by American psychologist, Charles Synder. It involves both optimism and a can-do-attitude.



<https://hope-action.com/>

If hope is a state of being or way of thinking, then hope can be learned. According to Hope Theory there are three main parts:

Goals - Have a goal is most important, they can be big or small

Willpower - You need to stay motivated to meet your goal. You need to believe good things will come from your actions

Pathways - Specific routes to meet your goal. Understanding that setbacks happen so you need to problem solve to find a new pathway. It may take several tries to reach your goals. Develop a plan A, B and C

Here is an example of how this may look for someone with ataxia:

"Sue" is feeling scared about being diagnosed with ataxia, but she is determined to not let her diagnosis define who she is and what she is capable of accomplishing.

Goal - "Sue" made a goal to become as healthy as possible despite her mobility challenges

Willpower - "Sue" is motivated to stay healthy so she can be as active as possible and be around for her children and grandchildren.

Pathways - "Sue" develops several strategies to meet her goal of being more healthy. She attends physical therapy and completes her daily exercise routine at home. She becomes more mindful about developing healthier eating habits. She tries to minimize her stress levels and does everything she can to educate herself about ataxia. She attends support groups, meets regularly with her healthcare providers and accesses the resources available on the National Ataxia Foundation Website.

Those that have hope have improved coping skills, well-being and tend to engage in healthy behaviors. Hope helps to protect from depression, loneliness and suicide and is essential for developing resilience.

Tips for becoming more hopeful:

- Think of your goals as exciting challenges and how good you'll feel when you reach them
- Expect roadblocks, things don't come easily
- Take one step at a time, think of one small thing you can do each day to reach your goal
- Set realistic goals
- Gain strength from others

Games as Research Tools

*By Jennifer Keller, PT, M.S.
Assistant Professor PM&R, JHMI, Kennedy Krieger Institute*

Games are a means to increase engagement in a range of research activities. Games are used not only to encourage greater participation in an activity but also to study various aspects of our behavior. You may be familiar with some games that have been used to study and improve memory (e.g. Lumosity), attention (e.g. Constant Therapy) and fitness (e.g. Wii Fit). As technology and game development improves, the simulated activities allow researchers to answer questions about what motivates us to change our behavior and how we might fine tune our treatments. In the Center for Movement Studies at Kennedy Krieger Institute, we are using interactive gaming and virtual reality in our studies of motor learning, memory formation, and decision-making.



<https://gamequitters.com/video-games-make-you-smarter/>

Humans can learn to perform incredibly complex and precise sequences of movements—think about the skill of an Olympic level gymnast performing on the balance beam. Some gymnastic coaches have adopted rapid auditory feedback (using a clicker) to reinforce each correct step in a movement sequence as it is happening. This is a replacement for feedback only at the end, i.e. explaining what went right and what went wrong in the movement sequence after it is completed.

The most recent experiments from Dr. Amy Bastian's lab show that stepwise feedback is more effective than feedback just at the end for learning complex sequences of movements. Participants who received stepwise feedback were twice as accurate with a reaching task in a 3D virtual reality task than those who received only feedback at the end. This result could be important not only for expert learning in sports but also for relearning of everyday tasks that require complex movement sequences by patients with motor impairments (e.g., drinking water from a glass). We have also found that people with cerebellar ataxia learn similarly to age-matched controls when they are given stepwise feedback via virtual reality. Overall, this bodes well for developing new training methods for people with cerebellar ataxia.

Our movements are often guided by things we see or experience around us. Imagine a gymnast who needs to perfectly time their routine or a musician hitting the right notes while reading sheet music. Our brain doesn't just rely on muscles to make these movements—it also draws from memories. In Dr. Vikram Chib's lab, we are exploring how the brain's movement regions work together with brain regions responsible for storing memories. In one of our studies, participants learned to link different arm movements with simple symbols. We found that these memory-based tasks helped people fine-tune their movements. Moreover, by using brain scans, we saw that the brain's memory regions created a mental map, connecting each symbol with a specific movement. By strengthening the connection between memory and movement, memory-based motor training could potentially help people with cerebellar ataxia improve their ability to fine-tune their movements and better interact with the environment.

These studies show that innovative methods can enhance motor learning, from learning motor sequences with precise timing to building memory-based movement associations. Whether helping athletes achieve peak performance or developing new therapies for those with motor disorders like cerebellar ataxia, these findings underscore the power of combining neuroscience, gaming, and technology. Through this work, we hope to unlock new approaches for rehabilitation and performance improvement.

Johns Hopkins Ataxia Research Studies (Current as of 10/01/2024)

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 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 1 st 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	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 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.

Friedreich'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 Green Family 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.