What is Ataxia?

Ataxia is typically defined as the presence of abnormal, uncoordinated movements. This term is most often, but not always, used to describe a neurological symptom caused by dysfunction of the cerebellum. The cerebellum is responsible for many motor functions, including the coordination of voluntary movements and the maintenance of balance and posture.

Those with cerebellar ataxia often have an “ataxic” gait, which is walking that appears unsteady, uncoordinated and staggered. Other activities that require fine motor control like writing, reading, picking up objects, speaking clearly and swallowing may be abnormal. Symptoms vary depending on the cause of the ataxia and are specific to each person.

Regardless of the type of ataxia a person may have, it is important for all individuals with ataxia to seek proper medical attention. For the vast majority of ataxias, a treatment or cure for the disease is not yet available, so the focus is on identifying symptoms related to or caused by the ataxia. By identifying the symptoms of ataxia it becomes possible to treat those symptoms through medication, physical therapy, exercise, other therapies and sometimes medications.

The Johns Hopkins Ataxia Center has a multidisciplinary clinical team that is dedicated to helping those affected by ataxia. The center has trained specialist ranging from neurologists, nurses, rehabilitation specialists, genetic counselors, and many others. This edition of the Ataxia Digest will provide you with information on living with ataxia and the multidisciplinary center at Johns Hopkins.

Letter from the Director

Welcome to the second edition of the Ataxia Digest. Since the inaugural edition of the Ataxia Digest, published in 2013, the Ataxia Center at Johns Hopkins has continued to make strides in its commitment to serve the ataxia community inside and outside the clinic. With the generous support of the Gordon and Marilyn Macklin Foundation and the National Ataxia Foundation, we have added new faculty and allied health professionals to our multidisciplinary clinical team, initiated new outreach programs for both our patients and their care partners and reconfirmed our commitment to cutting-edge ataxia research.

We look forward to introducing you to all of these new and continued initiatives in this newsletter. If you would like to learn more about the Ataxia Center at Johns Hopkins please visit our website at www.hopkinsmedicine.org/neuro/movement or contact our Health Educator, Bailey Vernon, at 410-616-2811 or bvernon1@jhmi.edu.

Sincerely,
Liana S. Rosenthal, MD
Meet the Neurologists

The Ataxia Center has a multidisciplinary team dedicated to comprehensive patient care. Patients have the opportunity to see multiple providers in addition to the neurologist during their visit with the Ataxia Center to help develop a comprehensive plan of care.

**Kristin Barañano, MD, PhD** - Dr. Kristin Barañano joined the Ataxia Center team in 2013. She received her MD and PhD degrees from Johns Hopkins, where she stayed for her residency training in pediatrics and neurology. She went on to the Kennedy Krieger Institute in Baltimore for her fellowship in neurogenetics. She has a particular interest in understanding the genetic control of cerebral cortex and cerebellar development. Dr. Barañano is a Clinical Associate in the Division of Pediatric Neurology and has particular expertise in the diagnosis and management of children with ataxia, adults with childhood onset ataxia and suspected genetic ataxias.

**Kelly Mills, MD** - Dr. Kelly Mills joined the Department of Neurology as an Assistant Professor in July of 2014. He attended medical school at the University of Maryland then completed his residency and movement disorders fellowship at the University of California, San Francisco. Dr. Mills sees patients with various movement disorders including dystonia, ataxia and Parkinson’s disease and helps with the care of some patients who need surgical treatments for their symptoms. He is particularly interested in describing and treating cognitive impairment in neurodegenerative movement disorders and exploring how various treatments for movement symptoms impact cognitive function.

**Liana S. Rosenthal, MD** – Dr. Liana Rosenthal is an Assistant Professor of Neurology and the Director of the Ataxia Center. Dr. Rosenthal received her medical degree from Johns Hopkins, where she stayed for her residency training in Neurology and her fellowship training in movement disorders before joining the faculty. She specializes in the treatment of movement disorders, including Parkinson’s disease and ataxia. Dr. Rosenthal’s primary research interests include understanding the natural history of ataxia and identifying biomarkers in Parkinson’s disease. As the Director, she strives to continually expand the multidisciplinary team and provide care to underserved individuals through telemedicine.

**David S. Zee, MD** – Dr. David Zee is a Professor of Neurology with joint appointments in Ophthalmology, Otolaryngology - Head and Neck Surgery and Neuroscience. Dr. Zee received his medical degree in 1969 from Johns Hopkins, where he stayed for his residency training in Neurology and then joined the faculty. He specializes in ataxia as well as vertigo, dizziness and disorders of eye movements (including nystagmus and strabismus). One of Dr. Zee’s major research interests is to understand how the brain (and especially the cerebellum) compensates for disease and how compensation can be promoted. Dr. Zee has published over 450 scientific papers.
Partnering With Your Doctor

Liana Rosenthal, MD, Assistant Professor of Neurology

One of the most important tools in effectively managing a chronic condition, such as ataxia, is for patients to take an active role in the management of their condition by becoming a partner with their doctor. Maintaining a strong, open relationship with your physician is a powerful factor in optimizing your physical and emotional well-being. Therefore, it’s important to keep the following points in mind when choosing a physician, going to appointments and communicating with your physician in between visits.

When identifying a physician to manage an ataxia diagnosis it is important to find a physician that is:

- familiar with ataxia
- easily accessible
- willing to prioritize the patient’s wishes in the larger context of the disease

While it may be ideal for such a physician to be a trained movement disorders specialist, that may not be realistic for patients that do not live close to a movement disorders center. In such a situation, it may be more important to find a local general neurologist or family practitioner that has an interest in treating ataxia and is willing to collaborate with a movement disorder specialist for day to day management.

Beyond evaluating a physician’s experience with ataxia, it’s equally important to choose a physician who is willing to work with you to coordinate a treatment plan that works best for your needs. The same treatment plan will not work for every person with ataxia so it is important to identify a physician with whom you feel comfortable talking about your condition. Once you are able to identify a physician you feel comfortable with, it will become easier to build a relationship of openness, caring and mutual respect.

The next step in partnering with your doctor is to make the most of your clinic time. Before the appointment, create a current medication list and compile any questions you may have for the physician. Also, it is important that your physician has access to your past medical records and any genetic testing that has already been completed. During your visit, try to be an active participant. Make sure to speak up if you do not understand your physician’s advice and articulate your hesitation if you do not agree with the treatment plan. Additionally, take notes or have a companion take notes throughout the visit so that you can remember important directions after the appointment.

After your clinic visit, make all appointments immediately for any follow-up tests, therapy or other orders. Fill medication prescriptions as soon as possible and begin taking them as prescribed, reporting any troublesome side effects immediately. If there is anything about the treatment plan you are unclear about upon returning home, feel free to call your physician’s office for clarification.

Many physicians are also open to communicating with their patients in between appointments via electronic communication such as e-mail or the Johns Hopkins MyChart. When emailing your physician include your name, date of birth and pharmacy phone number. Also, include brief background information about your question, your medication list and dosing regimen. While email can be a great way to communicate, if it is an emergency you should call 911 or go to the emergency room.

One of the most important steps in treating your ataxia is building a strong relationship with your physician. Through a working relationship with your doctor, you will put yourself in a good position to effectively manage your ataxia.
A Look Back at Ataxia Programs

Sailing with Ataxia

On July 11, 2015 the Downtown Sailing Center, in conjunction with the Ataxia Center, hosted accessible sailing for people with ataxia and their families. Individuals were able to have a day full of fun and sail throughout the Baltimore Inner Harbor, regardless of their disability. Over 60 people attended including 17 people with ataxia and their families and friends and 5 team members from the Johns Hopkins Ataxia Center (including Dr. Liana Rosenthal, Dr. Ankur Butala, Dr. Laura Tochen, Carrie Berlett and Bailey Vernon). After sailing, the group enjoyed lunch at Little Havana and raised money for ataxia through raffles. The group is looking forward to sailing next year on June 11, 2016!

Ataxia Wellness Program & Ataxia Wellness Day

For six weeks starting in March of 2015, seven individuals with ataxia and their care partners attended the Ataxia Wellness Program. The program provided exercise, education and social support. Each participant was given a specialized home exercise program and had the opportunity to try yoga, Qi gong, Pilates and dance. The Ataxia Center is currently expanding the program and seeking new participants.

The center sponsors an Annual Ataxia Wellness Day every fall that includes a variety of exercise demonstrations, group discussions, massages and more. If you are interested in learning more about these wellness programs and live within commuting distance to Baltimore, please contact Bailey Vernon at bvernon1@jhmi.edu or 410-616-2811.
In the Community

Resources

**National Ataxia Foundation (NAF)**  |  www.ataxia.org  |  763-553-0020
A non-profit organization dedicated to helping those affected by ataxia through education, support and research.

**Chesapeake Chapter of the NAF (CC-NAF)**  |  ccnafpres@gmail.com  |  703-759-2008
The local NAF chapter provides support to families, raises funds for research and holds educational meetings.

**Friedreich’s Ataxia Research Alliance (FARA)**  |  www.curefa.org  |  1-484-879-6160
FARA is a national non-profit organization that is dedicated to curing Friedreich’s ataxia through research.

**National Institutes of Health, National Institute of Neurological Disorders and Stroke (NINDS)**
www.ninds.nih.gov/disorders/ataxia  |  1-800-352-9424
The NINDS provides information on ataxia and conducts research on cerebellar and spinocerebellar degeneration.

**International Network of Ataxia Friends (Internaf)**  |  www.internaf.org
Internaf is an online resource for people with ataxia and is volunteer led.

“The League” - A Community Service for Maximizing Independence
Becky Dunlop, RN, MS

Maintaining your independence while living with a neurodegenerative disease such as ataxia can be extremely difficult without the help of others. In Baltimore, there are many resources available that are designed to help individuals maintain their independence as the disease progresses. One such organization dedicated to helping individuals with neurodegenerative diseases is The League for People with Disabilities.

Founded in 1927, The League in Baltimore is an organization committed to offering opportunities for individuals with multiple, physical, cognitive and neurological disabilities to gain independence through a comprehensive continuum of vocational, rehabilitative, educational, medical, wellness and social services.

The Adult Medical Day Program is designed to address the challenges of adults with a variety of disabilities, including neurodegenerative diseases. Individuals in this program are assessed and given an individual plan of care with the goal of allowing participants to continue to live within their communities. The Adult Medical Day Program is also certified by the Maryland State Department of Health and Mental Hygiene to provide adult medical day services to participants eligible under multiple Maryland State Waiver Programs.

There are a variety of other services available at The League as well as a wellness center that specializes in multifaceted exercise and rehabilitation. Although there are fees associated with these services, they do accept medical assistance, veterans benefits, private pay or any other form of insurance that pays for medical day services.

For more information on The League, please visit their website at www.leagueforpeople.org.

Simple Technology Offers Alternative Ways to Exercise
Becky Dunlop, RN, MS

For people with ataxia, research suggests that exercise can improve gait, flexibility and motor coordination. If you are having significant trouble with your mobility or you are simply looking for a way to incorporate a low intensity workout into your daily routine, it may be beneficial to invest in an inexpensive folding exercise peddling machine. Stationary cycles are an excellent option for those with balance difficulties or a fear of falling. These peddling machines can be placed on the floor to exercise the legs in a manner that is similar to riding a bike or can be placed on a table top to exercise the arms. Most models require no assembly. Online retailers such as Amazon, Walmart and Target offer many different models that range in price from $25 - $50.
Cognitive Neuropsychiatric Research Lab

Cognition, the Cerebellum and Cerebellar Ataxia

Cherie Marvel, PhD, Assistant Professor of Neurology and Psychiatry

The aim of the Cognitive Neuropsychiatric Research Laboratory (CNR) is to examine the impact of motor impairment on cognition. The lab is currently investigating working memory in people with cerebellar ataxia. Working memory, or short-term memory, is the cognitive system that holds a limited amount of information (≈ 5-9 units) for a short period of time (≈ 30 seconds). For example, working memory will be engaged while trying to remember a phone number that was just spoken, or solving a complex math problem.

Recent data shows a trending decline in working memory capacity among people with cerebellar ataxia, especially for information that is verbal in nature (e.g., words, numbers, shapes, written instructions, etc.). Working memory problems may be significant enough to affect professional productivity and home-life management.

The overarching goal of this research is to characterize cognitive abilities in cerebellar ataxia in order to develop rehabilitation tools. Such therapies may improve cognition as well as a patient’s personal and professional life.

If you are interested in participating in this type of cerebellar ataxia research, please contact the CNR Lab for more information at 410-502-4664 or email Dr. Cherie Marvel at cmarvell@jhmi.edu.
Research

Lab for Medical Image Computing

Natural History Study of Genetic Modifiers in Spinocerebellar Ataxia

Chiadi Onyike, MD, MHS, Assistant Professor of Psychiatry and Behavioral Sciences

The Johns Hopkins Laboratory for Medical Image Computing is participating in the Natural History Study of Genetic Modifiers in Spinocerebellar Ataxia, a longitudinal study of the progression of spinocerebellar ataxias (SCAs). This multi-center research effort is being conducted by allied researchers of the Clinical Research Consortium for the Study of Spinocerebellar Ataxia (CRC-SCA), part of NIH’s Rare Diseases Clinical Research Network. Dr. Chiadi Onyike is currently recruiting patients with SCA 1, 2, 3 and 6 for participation in the Natural History Study. Patients with other forms of ataxia and unaffected family members are also encouraged to participate in research on the cerebellum, cognition and ataxia. Testing may include an MRI, neurological exam, testing of thinking and memory and a blood sample. To learn more, please contact Ann Fishman at ataxiaresearch@jhu.edu or 410-502-5816.

Motion Analysis Lab

Rehabilitation Strategies for Cerebellar Ataxia

Amy Bastian, PhD, PT, Professor of Neuroscience

The Motion Analysis Lab investigates both reaching and walking to try to develop rehabilitation strategies for people with movement disorders, including cerebellar ataxia. A robotic arm is used to study arm movements, allowing us to move people’s arms from point to point very precisely. Recently the lab found that people with cerebellar ataxia have a harder time telling how fast and for how long their arm is moving. Sometimes this affects their ability to determine the position of their arm, but this is not always the case. Currently the lab is studying how movement sense and position sense are related, and how much these rely on the cerebellum.

The lab is also studying how people learn new walking patterns by using a special split-belt treadmill that can drive the legs at different speeds. It has been found that healthy adults can adapt to these new environmental demands and learn a new walking pattern – going from asymmetric to symmetric walking. Some data also shows that not only are there changes to the motor system but there are also sensory changes in how leg speed is perceived. Previous data has shown that patients with cerebellar ataxia, compared to age-matched adults, have difficulties in learning a new walking pattern when being perturbed by a treadmill. Preliminary data shows that there are sensory deficits that accompany the motor deficits. This provides insight on the role the cerebellum is playing during walking adaptation and the relationship between motor and sensory systems. Please contact the Motion Analysis Lab at 443-923-2716 to inquire about cerebellar studies.
Johns Hopkins University Ataxia Center

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www.hopkinsmedicine.org/neuro/movement

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Please consider supporting our center!
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