National Ataxia Foundation

Cerebellar Ataxia with Neuropathy and Vestibular Areflexia Syndrome (CANVAS)

What is CANVAS?
CANVAS is a rare neurodegenerative disorder. CANVAS is an acronym meaning Cerebellar Ataxia (CA), Neuropathy (N) and Vestibular Areflexia (VA) Syndrome (S). As the cause of CANVAS was only discovered in 2019, there remains a lot we do not know about this condition. This includes how common CANVAS is. From initial research, CANVAS has been found across the globe, affecting people from all ethnic backgrounds.

As more people are diagnosed with CANVAS and more research is completed, we will have a better understanding of this disorder. This page will be updated with new information as it becomes available.

What are the symptoms of CANVAS?
CANVAS is an acronym for the three most common symptoms caused by this disorder. Most people with CANVAS have at least two of the three common symptoms:

- **CA (Cerebellar Ataxia):** Incoordination, marked by an unsteady gait, stumbling, and imbalance. There can also be problems coordinating muscles that control speech and swallowing.

- **N (Neuropathy):** Problems with the nerves that carry signals about what people feel through their skin, including touch, pressure, pain, and temperature. People with CANVAS might feel too much (feeling cold when it is room temperature) or not enough (not realizing you have touched something hot) sensation.

- **VA (Vestibular Areflexia):** Reflexes relating to balance aren't working properly. The Vestibulo-ocular reflex (VOR) is impacted the most. VOR helps keep your eyes steady when you move your head. People with CANVAS will have blurry or jumpy vision when they are walking, running, or moving their head.

It is also common for people with CANVAS to have an unexplained, chronic cough. Sometimes this is the first symptom noticed by patients. Less common symptoms include chronic constipation, urinary difficulties, or altered sweating (too much or too little).

What is the prognosis for CANVAS?
The age at onset of CANVAS symptoms is usually around 50 years old. However, there can be a lot of variation, with some people having symptoms early in their 20s or later in their 70s. Since the first signs of CANVAS can be subtle, such as a cough or small changes to your sense of touch, some people don't notice when symptoms first occur. It can be decades later, when balance symptoms begin, that people seek help from a neurologist. The severity and combination of CANVAS symptoms also vary considerably, even within families.

CANVAS usually progresses very slowly. Most people with CANVAS will eventually use upright walking aids such as a cane or rollator. However, most do not require the use of wheelchairs until much later in life. Lifespan generally is not shortened by the disease. Treatments such as physiotherapy, occupational therapy, and speech-language therapy can significantly improve the lives of people with CANVAS.

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Is CANVAS genetic?

CANVAS is a genetic disorder which means that it is an inherited disease. The abnormal gene responsible for this disease is passed along from generation to generation by family members who carry it. Genetic diseases like CANVAS occur when one of the body's 20,000 genes does not work properly. Genes are microscopic structures within the cells of our bodies that contain instructions for every feature a person inherits from his or her parents.

In the case of CANVAS, it is caused by a mutation called an AAGGG repeat expansion in the RFC1 gene. Healthy RFC1 genes typically have a repeat called AAAAG, but in CANVAS the ratios of A and G are messed up. Whether or not you develop CANVAS depends on how many AAGGG repeats you have.

Between 10-200 AAAAG Repeats: This is the typical number of AAAAG repeats in the RFC1 gene. People with this number of repeats are healthy.

Over 400 AAGGG Repeats: This is the current threshold number of AAGGG repeats to be diagnosed with CANVAS. If you have over 400 repeats, you will develop CANVAS symptoms. Some people with CANVAS can have over 2000 AAGGG repeats.

There are other types of mutations in the RFC1 gene that have been discovered to cause CANVAS. However, the research is so new on these mutation types that we don't fully understand why they cause CANVAS yet. More research is needed.

How is CANVAS diagnosed?

A neurologic examination can determine whether a person has symptoms typical of CANVAS. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia. Due to the variety of CANVAS symptoms, follow-up testing may look different from individual to individual. Some potential follow-up tests include MRI brain imaging, vestibular testing, or sensory neuropathy testing.

A definitive diagnosis of CANVAS is established following genetic testing. This confirms that someone has the number of AAGGG repeats needed to cause CANVAS. Genetic testing for CANVAS is currently available through the University of Chicago and other academic centers around the world.

What kind of support is available after the diagnosis?

As Ataxia research moves into the clinical phase, pharmaceutical companies will begin recruiting participants for clinical trials. Individuals with Ataxia or who are at-risk for Ataxia are encouraged to enroll in the CoRDS Ataxia Patient Registry. To access the Registry, go to NAF's website www.ataxia.org and click on the "Enroll in the Patient Registry" tab and follow the directions on the CoRDS website.

NAF provides accurate information for you, your family, and your physician about Ataxia. Please visit the NAF website at www.ataxia.org for additional information, including a listing of ataxia support groups, physicians who treat Ataxia, social networks, and more. For questions contact the NAF directly at (763) 553-0020 or naf@ataxia.org.