National Ataxia Foundation

Spinocerebellar Ataxia Type 27B (SCA27B)

What is SCA27B?

SCA27B is a rare neurodegenerative disorder. As the cause of SCA27B was only discovered in 2022, there remains a lot we do not know about this condition, including how common it is. From initial research, SCA27B is found across the globe, with high prevalence rates in French Canadians.

As more people are diagnosed with SCA27B 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 SCA27B?

Like many other forms of Ataxia, SCA27B is marked by incoordination. In fact, the word Ataxia means incoordination. In most cases, the first symptoms of SCA27B are unsteady gait, stumbling, and imbalance. Other common symptoms include vision problems (double vision, bouncy vision, or blurry vision), poor hand coordination, vertigo or dizziness, and difficulty speaking. Less common symptoms include swallowing difficulties, tremors, urinary urgency, stiff or rigid muscles, and numbness or pain in your limbs.

In about 50% of cases, SCA27B symptoms begin as episodes – where people experience bouts of ataxia lasting from minutes to days, but then regain their sense of balance and other symptoms go away. Over time, episodes increase to a more permanent form of ataxia, where symptoms are constant. SCA27B ataxia episodes can be triggered by alcohol, physical activity, and caffeine.

What is the prognosis for SCA27B?

Age at onset of SCA27B symptoms usually ranges from 50-80 years old. However, symptoms can occur as early as 30 years old and as late as 90 years old. The severity of symptoms also varies considerably, even within families.

SCA27B usually progresses very slowly. Most people with SCA27B will eventually use upright walking aids such as a cane or rollator, however, most do not require the use of wheelchairs. 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 SCA27B.

How is SCA27B diagnosed?

A neurologic examination can determine whether a person has symptoms typical of SCA27B. This suspected diagnosis is then confirmed through brain imaging, such as MRI, and genetic testing to detect the presence of the abnormal gene that causes SCA27B. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia.

Genetics of SCA27B

SCA27B 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 SCA27B 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.

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Genetics of SCA27B (continued)

SCA27B is an autosomal dominant disease which means that individuals of either sex are equally likely to inherit the gene and develop the disease. Each child of a person with SCA27B has a 50 percent chance of inheriting the gene that causes SCA27B.

In the case of SCA27B, it is caused by a mutation called a GAA repeat expansion in the FGF14 gene. Whether or not you develop SCA27B depends on how many repeats you have.

- Under 200 GAA Repeats: This is the normal number of repeats in the FGF14 gene. People with this number of repeats are healthy.
- 200-249 GAA Repeats: Due to limited data, it is not clear what this number of repeats means. Some people may develop symptoms, others may not. There is active research to understand what this number of repeats does to the body, but right now it results in an uncertain diagnosis.
- 250-300 GAA Repeats: This number of repeats causes 'incomplete penetrance' of SCA27B, meaning that individuals may or may not develop symptoms.
- Over 300 GAA Repeats: This is the current threshold number of repeats to be diagnosed with SCA27B. If you have over 300 repeats, you will develop ataxia symptoms.

GAA repeat numbers can grow or shrink between generations. This can make it seem like ataxia 'skips' a generation, if the GAA repeat number shrinks below the number needed to cause SCA27B. If in the next generation, the GAA repeat number grows to over 300 again, then people will develop symptoms.

GAA repeat numbers are more likely to grow when passed to children by their mothers. GAA repeat numbers are more likely to shrink when passed to children by their fathers.

Gene tests can be performed for diagnostic purposes to determine what kind of Ataxia is within a person or family. Genetic testing also can be done, in some circumstances, even before there are symptoms to determine whether a person carries the abnormal gene or genes that cause Ataxia. This is called predictive or presymptomatic testing. A gene test also can be used to determine whether a fetus has an abnormal Ataxia gene. This is called prenatal testing. Anyone who is considering a predictive or prenatal test should consult with a genetic counselor to discuss the reasons for the test, the possible outcomes, and how those outcomes might affect the person emotionally, medically, or socially.

What kind of support is available after the diagnosis?

NAF is committed to providing information and education about Ataxia, support groups for those affected by Ataxia, and promoting and funding research to find the cause for the various forms of Ataxia, better treatments, and, hopefully someday, a cure. NAF has been at the forefront funding promising worldwide research to find answers.

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.