National Ataxia Foundation

Spinocerebellar Ataxia Type 12 (SCA12)

What is Spinocerebellar Ataxia type 12?

Spinocerebellar Ataxia type 12 (SCA12) is a rare neurodegenerative disorder. It is caused by a CAG triplet repeat expansion in the PPP2R2B gene. SCA12 is very rare worldwide. However, SCA12 is very prevalent in Northern India. Approximately 16% of autosomal dominant ataxia patients in India have SCA12.

What are the symptoms of SCA12?

Like many other forms of Ataxia, SCA12 is marked by poor balance and coordination. In fact, the word Ataxia means incoordination. There can also be problems coordinating muscles that control speech and swallowing.

The first symptom for many people with SCA12 is tremors in the head and arms. These tremors are called action tremors, they occur when muscles are being voluntarily used. Many people with SCA12 have neuropsychiatric symptoms, such as anxiety, depression, cognitive difficulties, and dementia.

Less common symptoms of SCA12 include vision problems, eye spams, muscle contractions, increased reflexes, Parkinson-like symptoms, and neuropathy.

How is SCA12 acquired?

SCA12 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 SCA12 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.

SCA12 is an autosomal dominant disease, meaning individuals of either sex are equally likely to inherit the gene and develop the disease. Each child of a person with SCA12 has a 50% chance of inheriting the gene that causes SCA12.

In the case of SCA12, it is caused by a mutation called a CAG repeat expansion in the PPP2R2B gene. Whether or not you develop SCA12 depends on how many repeats you have.

- Between 4-31 CAG Repeats: This is the typical number of repeats in the FGF14 gene. People with this number of repeats are healthy.
- Between 32 and 49 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.
- Over 50 repeats: People with more than 50 repeats usually develop SCA12.

More repeats are associated with an earlier age of onset of symptoms. Unlike some other forms of ataxia caused by repeat expansions, SCA12 repeats do not tend to expand or increase in number between generations.

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.

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How is a diagnosis made?

A neurologic examination can determine whether a person has symptoms typical of SCA12. 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 SCA12. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia.

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.