National Ataxia Foundation

Spinocerebellar Ataxia Type 50 (SCA50)

What is SCA50?

Spinocerebellar Ataxia type 50 (SCA50) is a rare neurodegenerative disorder. It is caused by a missense mutation in the *NPTX1* gene. SCA50 is very rare worldwide, with only a handful for families known to have this condition. However, as *NPTX1* genetic testing becomes more widely available, more individuals are being diagnosed with SCA50.

SCA50 Symptoms

Like many other forms of Ataxia, SCA50 is marked by poor balance and coordination. In fact, the word Ataxia means incoordination. There can also be problems coordinating muscles that control speech, swallowing, and vision. Other potential symptoms include tremor and cognitive impairment. The severity of SCA50 symptoms may vary considerably between patients, even within families. More research is needed to better understand the symptoms of SCA50.

Prognosis

SCA50 symptoms usually begin in adulthood. However, a few cases of symptoms beginning in childhood have been reported.

SCA50 usually progresses very slowly, with patients eventually needing the use of a wheelchair. Treatments such as physiotherapy, occupational therapy, and speech-language therapy can significantly improve the lives of people with SCA50.

Genetics

SCA50 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 SCA50 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.

SCA50 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 SCA50 has a 50% chance of inheriting the gene that causes SCA50.

Gene tests can be performed for diagnostic purposes to determine what kind of Ataxia is within a person or family. Genetic testing can also 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 can also 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.

Diagnosis

A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia. A neurologic examination can determine whether a person has symptoms typical of SCA50. MRI brain imaging may be used to confirm cerebellar atrophy. A definitive diagnosis of SCA50 is established following genetic testing. This confirms that someone has a mutation that causes SCA50 in their *NPTX1* gene.

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What kind of support is available after the diagnosis?

The National Ataxia Foundation (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. 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.