Spinocerebellar Ataxia Type 17 (SCA17)

Spinocerebellar Ataxia type 17 (SCA17) is a rare neurodegenerative disorder. It is caused by mutations in the TBP gene. SCA17 is very rare worldwide. The estimated prevalence of SCA17 is less than 1 in 1,000,000 people, with approximately 100 families with SCA17 reported in the scientific literature.

There is some evidence that the prevalence of SCA17 might be underestimated, as the symptoms of SCA17 appear very similar to those of Huntington Disease. In fact, an older name for SCA17 was “Huntington Disease-Like 4”.

Symptoms
Like many other forms of Ataxia, SCA17 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. People with SCA17 also develop dementia and involuntary movement symptoms, such as chorea or dystonia.

Other common SCA17 symptoms include pyramidal signs, psychiatric symptoms, rigid muscles, and Parkinsonism. These symptoms may be mistaken for other neurodegenerative disorders, such as Huntington Disease, Parkinson Disease, ALS, or Frontotemporal Dementia.

Prognosis
SCA17 symptoms usually begin around age 30, with most cases occurring in people between the ages of 19 and 48. However, there have been cases of SCA17 occurring in people as young as 3 years old and as old as 75 years old.

The severity of symptoms also varies considerably between people, even within families. SCA17 symptoms are progressive. People with SCA17 may need to use a wheelchair after 5-15 years of experiencing symptoms. SCA17 is a life-shortening condition. Treatments such as mental health support, physiotherapy, occupational therapy, and speech-language therapy can significantly improve the lives of people with SCA17. Some medications may be helpful for symptom management

Genetics
SCA17 is an inherited genetic disorder. It is caused by an abnormality of a single gene called the TBP gene. The abnormality can be passed from generation to generation by family members who carry it. Males and females are equally likely to inherit the genes that cause SCA17.

Genetic diseases like SCA17 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 SCA17 a person inherits from his or her parents. Two copies of each gene are inherited; one copy from the mother and one from the father. SCA17 is an autosomal dominant disease, meaning that someone will develop symptoms if they inherit at least one mutated copy of the TBP gene. Each child of someone with SCA17 has a 50% chance of inheriting the gene that causes SCA17.

In the case of SCA17, it is caused by a mutation called a CAG repeat expansion in TBP gene. Whether or not you develop SCA17 symptoms depends on how many repeats you have.

- **Less than 40 repeats:** This is the typical number of repeats in the TBP gene. People with this number of repeats are generally healthy.

- **Between 41-48 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.

- **More than 48 repeats:** People with over 48 repeats usually develop SCA17. If someone has over 62 repeats, they tend to develop symptoms as a child.

More CAG repeats are associated with an earlier age of onset of SCA17 symptoms. The number of repeats can sometimes grow between generations, a phenomenon we call “genetic anticipation”. Due to genetic anticipation, each generation of a family with SCA17 may experience their first symptoms earlier than the previous generation.

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 neurologic examination can determine whether a person has symptoms typical of SCA17. A neurologist is often the most helpful specialist in recognizing symptoms and diagnosing the disease that causes Ataxia. There are several potential follow-up tests. MRI brain imaging is often used to confirm cerebellar atrophy or degeneration. Clinical exams are performed to measure various movement, behavioral, and speech-related symptoms. However, a specific diagnosis of SCA17 ataxia can only be made by a genetic test.