

Spinocerebellar Ataxia Type 10 (SCA10)

What is spinocerebellar Ataxia type 10?

Spinocerebellar Ataxia type 10 (SCA10) is one specific type of ataxia among a group of inherited diseases of the central nervous system. As in other inherited ataxias, SCA 10 is caused by genetic defects that lead to impairment of specific nerve fibers carrying messages to and from the brain, resulting in degeneration of the cerebellum (the coordination center of the brain).

What are the symptoms of SCA10?

Typically, problems with balance and coordination (ataxia) are noticed first. (In fact, “ataxia” means incoordination.) The affected person has trouble walking and this difficulty gradually worsens. Falls occur with increasing frequency. As time goes on, the use of a cane, walker, and eventually, a wheelchair will become necessary. Within a few years of onset, slurred speech (dysarthria) is typical due to inability to coordinated movements of the lips, cheeks, tongue, vocal cords, diaphragm, etc. Also, within a few years of onset, coordination of hands and arms becomes impaired. Fine motor skills such as handwriting and fastening buttons are affected first; eventually basic daily tasks such as feeding and dressing become difficult. Abnormal tracking movements of the eyes are common. In the later stages of the disease, difficulty swallowing (dysphasia) results from inability to control muscles in the mouth and throat and aspiration pneumonia may become a life-threatening issue.

Recurrent seizures affect 20 percent to 100 percent of those afflicted with SCA10. The seizures are usually seen after the ataxia symptoms first appear. Most commonly recognized are generalized motor seizures, but complex partial seizures also have been seen.

Some individuals with SCA10 have additional symptoms such as mood disorders; weakness or loss of feeling in the feet, legs, or hands (peripheral neuropathy); cognitive dysfunction; and mild pyramidal signs such as overly active reflexes (hyperreflexia) and/or Babinski sign (when the sole of the foot is tickled, the big toe reflex turns up instead of down).

What is the prognosis for SCA10?

Age of affected persons at onset of SCA10 symptoms ranges from 10 to 50 years. SCA10 is a slowly progressive disease, which means symptoms develop gradually over many years. Life span may be shortened by the progressive nature of the disease. Severe seizures may become fatal if not adequately controlled.

How is SCA10 acquired?

SCA10 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 occur when one of the body's 30,000 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.)

SCA 10 is an autosomal dominant disorder. This means that individuals of either sex are equally likely to inherit the gene and develop the disease and the gene passes directly from one generation to the next without skipping generations. Each child of a person with SCA10 had a 50 percent chance of inheriting the gene that causes it.

How common is SCA10?

The exact prevalence of SCA10 is not known. SCA10 has been identified only in families of Mexican, Brazilian and several other Latin American heritages. Whenever traceable, the affected ancestor has been of a Native American origin.

How is the diagnosis made?

A neurologist often is the most helpful specialist in recognizing symptoms and diagnosing the diseases that cause Ataxia. Initially, a neurologic examination can determine whether a person has symptoms typical of one of the SCAs. DNA-based testing can accurately detect the presence or absence of the abnormal gene that causes SCA10.

DNA tests for SCA10 involve analysis of a gene located on chromosome 22q13. (Each person has 23 pairs of chromosomes.) Genes are made up of substances known as nucleotides linked together in chains. Each nucleotide is identified by a letter. In SCA10, a gene mutation in a gene called ATXN10, which is located on the 22nd chromosome, results in extra copies of a series of five nucleotides (pentanucleotide) identified by the letters ATTCT.

What kind of support is available after the diagnosis?

Although there is no specific treatment to delay or halt the progression of SCA10, medication may help to control the seizures. Conventional anticonvulsants such as carbamazepine, phenytoin, and valproic acid may be effective. Control of seizures is a very important management issues with SCA10 because uncontrolled seizures may lead to statue epilepticus (seizures occurring in rapid succession without consciousness being recovered between attacks) which may be fatal.

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. NAF has been at the forefront funding promising worldwide research to find answers.

Ataxia research has moved into the clinical phase, and pharmaceutical companies have begun 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.

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