

FREQUENTLY ASKED QUESTIONS ABOUT...

## 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, SCA10 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, wheelchair will become necessary. Within a few years of onset, slurred speech (dysarthria) is typical due to inability to coordinate 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 common 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 reflexly 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.

### 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.)

SCA10 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 has 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 and Brazilian heritage; so far, six apparently unrelated families have been found with this disease. It remains to be determined if SCA10 exists in other ethnic groups; it has not been found in Caucasian and Japanese persons with ataxia.

### **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 E46L, 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 issue with SCA10 because uncontrolled seizures may lead to status epilepticus (seizures occurring in rapid succession without consciousness being recovered between attacks), which may be fatal.

Supportive therapy is available to help manage other symptoms of SCA10, and there are resources to provide emotional support. *Living With Ataxia: An Information and Resource Guide*, published by the National Ataxia Foundation, includes a range of practical information and lists additional resources. NAF also provides and participates in many support and chat groups on the Internet. Visit our Web site (see address below) for a listing of these groups.

Contact the National Ataxia Foundation for a more complete listing of resources and of support groups affiliated with the NAF.

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