

Spinocerebellar Ataxia (SCA)

Last updated November 2010

Spinocerebellar ataxia (SCA) is one of a group of genetic disorders characterized by slowly progressive in-coordination of gait and often associated with poor coordination of hands, speech, and eye movements. Frequently, atrophy of the cerebellum occurs. As with other forms of ataxia, SCA results in unsteady and clumsy motion of the body due to a failure of the fine coordination of muscle movements, along with other symptoms.

The symptoms of the condition vary with the specific type (there are several), and with the individual patient. Generally, a person with ataxia retains full mental capacity but may progressively lose physical control.

Treatment and prognosis

There is no known cure for spinocerebellar ataxia, which is a progressive disease (it gets worse with time), although not all types cause equally severe disability.

Treatments are generally limited to softening symptoms, not the disease itself. The condition can be irreversible. A person with this disease will usually end up needing to use a wheelchair, and eventually they may need assistance to perform daily tasks.

The treatment of in-coordination or ataxia mostly involves the use of adaptive devices to allow the ataxia individual to maintain as much independence as possible. Such devices may include a cane, crutches, walker, or wheelchair for those with impaired gait; devices to assist with writing, feeding, and self cares if hand and arm coordination is impaired; and communication devices for those with impaired speech.

Many patients with hereditary or idiopathic forms of ataxia have other symptoms in addition to the ataxia. Medications or other therapies might be appropriate for some of these symptoms, which could include tremor, stiffness, depression, spasticity, and sleep disorders, among others.

Both onset of initial symptoms and duration of disease can be subject to variation. If the disease is caused by a polyglutamine trinucleotide repeat CAG expansion, a longer expansion may lead to

an earlier onset and a more radical progression of clinical symptoms.

Diagnosis

It can be easily misdiagnosed as another neurological condition, such as multiple sclerosis (MS). One means of identifying the disease is with an MRI to view the brain. Once the disease has progressed sufficiently, the cerebellum (a part of the brain) can be seen to have visibly shrunk. The most precise means of identifying SCA, including the specific type, is through DNA analysis. Some, but far from all, types of SCA may be inherited, so a DNA test may be done on the children of a person with the condition, to see if they are at risk of developing the condition.

SCA is related to olivopontocerebellar atrophy (OPCA); SCA types 1, 2, and 7 are also types of OPCA. However, not all types of OPCA are types of SCA, and vice versa. This overlapping classification system is both confusing and controversial to some in this field.

Types

The following is a list of some, not all, types of spinocerebellar ataxia. The first ataxia gene was identified in 1993 for a dominantly inherited type. It was called "Spinocerebellar ataxia type 1" (SCA1). Subsequently, as additional dominant genes were found they were called SCA2, SCA3, etc. Usually, the "type" number of "SCA" refers to the order in which the gene was found. At this time, there are at least 29 different gene mutations which have been found (not all listed).

Identifying the different types of SCA now requires knowledge of the normal genetic code, and faults in this code, which is contained in a person's DNA (Deoxyribonucleic acid). The "CAG" mentioned below is one of many three-letter sequences that makes up the genetic code. Thus, those ataxias with poly CAG expansions, along with several other neurodegenerative diseases resulting from a poly CAG expansion, are referred to as polyglutamine diseases.

Others include SCA18, SCA20, SCA21, SCA23, SCA26, SCA28, and SCA29. Four X-linked types have been described (302500, 302600, 301790, 301840), but only the first of these has so far been tied to a gene (SCAX1).

Inheritance

The hereditary ataxias are categorised by mode of inheritance and causative gene or chromosomal locus. The hereditary ataxias can be inherited in an autosomal dominant, autosomal recessive or X-linked manner.

Numerous types of autosomal dominant cerebellar ataxias are now known for which specific genetic information is available. Synonyms for autosomal dominant cerebellar ataxias (ADCA) used prior to the current understanding of the molecular genetics were Marie's ataxia, inherited olivopontocerebellar atrophy, cerebello-olivary atrophy, or the more generic term "spinocerebellar degeneration." (Spinocerebellar degeneration is a rare inherited neurological disorder of the central nervous system characterized by the slow degeneration of certain areas of the brain. There are three forms of spinocerebellar degeneration: Types 1, 2, 3. Symptoms begin during adulthood.)

There are five typical autosomal recessive disorders in which ataxia are a prominent feature: Friedreich ataxia, ataxia-telangiectasia, ataxia with vitamin E deficiency, ataxia with oculomotor apraxia (AOA) and spastic ataxia.

Information for this fact sheet has been sourced from the Living with Cerebral Palsy website <http://www.livingwithcerebralpalsy.com>

Another website with more information about specific Ataxias can be found at: <http://www.ataxia.org/>

Spinocerebellar Type 1 (SCA1) -

<http://www.ataxia.org/pdf/NAF%20Web%20Content%20Publication%20SCA1.pdf>

Spinocerebellar Type 2 (SCA2) -

<http://www.ataxia.org/pdf/NAF%20Web%20Content%20Publication%20SCA2.pdf>

Spinocerebellar Type 3 (SCA3) -

<http://www.ataxia.org/pdf/NAF%20Web%20Content%20Publication%20SCA3.pdf>