

SPINOCEREBELLAR ATAXIA TYPE 7 (SCA7)

Spinocerebellar ataxia type 7

Spinocerebellar ataxia type 7 (SCA7) is a type of inherited cerebellar ataxia. It is caused by a defect in a gene. This results in damage to certain parts of the brain, particularly the cerebellum (ie the co-ordination centre) and to the eyes.

What are the symptoms?

SCA7 is a progressive cerebellar ataxia.

All people with SCA7 experience balance problems, inco-ordination of limbs and slurred speech (dysarthria). Progressive macular degeneration, which results in progressive visual failure eventually leading to blindness is very common. This is in fact generally considered one of the main distinguishing features from other spinocerebellar ataxias. People who have a later age of onset may not experience visual problems. [*The Lancet Neurology* 2004; 3:291-304]

Other possible symptoms include problems with swallowing, spasticity and problems with eye movements.

Symptoms and their severity vary between individuals.

What causes SCA7?

SCA7 is an inherited condition caused by a defect in the so-called SCA7 gene, located on the 3rd chromosome. (Each person has 23 pairs of chromosomes). This gene is more extended than normal in people with SCA7.

All genes are made up of nucleotides that are held together in a chain. Each nucleotide is identified by a letter (A, T, C or G). The SCA7 gene is extended because of extra copies of a series of nucleotides identified by the letters C-A-G. In general, the more additional copies there are, the more severe the condition.

The SCA7 gene has instructions for the production of a protein called ataxin 7. The abnormally extended SCA7 gene results in the production of an extended ataxin 7 protein. Research is underway to understand what the function of the ataxin 7 protein is and what happens when it is extended. This information is important in order to develop treatments.

How is SCA7 inherited?

SCA7 is inherited in an autosomal dominant way. For more information on inheritance see Ataxia UK's *Genetics and ataxia*' leaflet.

When do symptoms start?

The age of onset varies greatly, from 3 months or less to over 70 years. The average age of onset is 30. In general, the later the onset of symptoms, the slower the progression. Generally people will need to use a wheelchair after a number of years.

Often, SCA7 results in earlier age of onset and more severe symptoms as the gene is passed down the generations. This phenomenon is known as anticipation. The explanation for this phenomenon is the instability of the extended SCA7 gene, which gets longer as it passes from parent to child. [Handbook of Ataxia Disorders. Ed. Klockgether, 2000]

Some studies have shown that when the father has SCA7 it generally results in a larger extension in the SCA7 gene as it is passed to a child than when the mother has SCA7. Anticipation is more marked in SCA7 than in other spinocerebellar ataxias.

There have been cases where a person without symptoms of SCA7 has a child who develops the symptoms, because the expansion in the parent was small enough not to manifest itself and it increased as it was passed down to the child. [*The Lancet Neurology* 2004; 3:291-304]

Rarely the opposite happens ie later age of onset and less severe symptoms as the gene is passed down the generations. [Handbook of Ataxia Disorders. Ed. Klockgether, 2000]

How is SCA7 diagnosed?

A genetic test will confirm a diagnosis of SCA7. This involves taking a blood sample to detect the abnormal gene.

How common is SCA7?

There is no recent information on the prevalence of SCA7 in the UK. Ataxia UK is currently funding research to address this lack of knowledge. The first study published from this research estimates that the prevalence of all the dominant ataxias (of which SCA7 is one) in North East England is 1 in 12,500. [Ann Neurol. 2004 May; 55 (5):752-5].

Management of SCA7

As with other cerebellar ataxias, physiotherapy and speech therapy can be helpful. A visit by an occupational therapist will be useful in order to assess the need for items such as walking aids, or for adaptations to the home.

It is important to see a neurologist, who will monitor the condition, on a regular basis.

For more support or information please contact:

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We have a number of other publications on the ataxias available free of charge. In addition we publish a quarterly magazine called *The Ataxian* containing articles on research, living with ataxia and other relevant information. Our website also contains news of research projects.

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