SPINOCEREBELLAR ATAXIA TYPE 6 (SCA6)

Spinocerebellar ataxia type 6

Spinocerebellar ataxia type 6 (SCA6) 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 (i.e. the co-ordination centre).

What are the symptoms?

SCA6 is a slowly progressive cerebellar ataxia. Most people experience the following symptoms; balance problems (gait ataxia), slurred speech (dysarthria), swallowing problems (dysphagia), involuntary eye movements (nystagmus), and vertigo. [Handbook of Ataxia Disorders. Ed. Klockgether, 2000]

Symptoms and their severity vary between individuals. It is sometimes episodic, i.e.: people may experience fluctuations in the severity of the ataxia. This is sometimes related to stress and fatigue. Some people may have episodes of vertigo or migraine. [Ann Neurol. 2004 May; 55 (5):752-5].

The progression is generally quite slow. In some cases people can still walk 20 years after onset of symptoms. [Handbook of Ataxia Disorders. Ed. Klockgether, 2000]

What causes SCA6?

SCA6 is an inherited condition caused by a defect in the so-called SCA6 gene, located on the 19th chromosome. (Each person has 23 pairs of chromosomes). This gene is more extended than normal in people with SCA6.

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 SCA6 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 earlier onset of the symptoms. [J Hum Genet. 2004;49(5):256-64.]

The SCA6 gene has instructions for the production of a protein that is the main component of calcium channels that are found mainly in cells of the cerebellum. These calcium channels are normally important for maintaining the correct amount of calcium in cells, which is necessary for the cells to perform their normal functions. In people with SCA6 the expansion in the SCA6 gene results in a change in the calcium channel prevent them from controlling the levels of calcium in cells.

How is SCA6 inherited?

SCA6 is inherited in an autosomal dominant way. For more information on inheritance see Ataxia UK's *'Ataxia: what's that'?'* leaflet.

When do symptoms start?

Symptoms typically start in the late 40s to early 50s, although there is variation in the age of onset. The range is from mid-20s to mid-70s. Unlike some of the other spinocerebellar ataxias, SCA6 is not normally associated with anticipation (i.e. earlier age of onset and more severe symptoms as the gene is passed down the generations). [Handbook of Ataxia Disorders. Ed. Klockgether, 2000]

A genetic test will confirm a diagnosis of SCA6. This involves taking a blood sample to detect the abnormal gene.

How common is SCA6?

There is currently little information on the prevalence of the cerebellar ataxias in the UK. Ataxia UK is funding research to rectify this lack of knowledge. The first study published from this research estimates that the prevalence of SCA6 in the North East England is 1.59 in 100,000 (for people over 16 years of age). In this same study researchers estimated that the number of people who actually had SCA6 or were at risk of developing it was 1 in 19,210. [Ann Neurol. 2004 May; 55 (5):752-5].

Management of SCA6

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.

People with SCA6 who have episodic features may respond to the drug acetozolamide. Please note that the use of any medicines would of course always need to be discussed with a doctor or neurologist who would know the specific circumstances of the patient. Not all people respond well to azetozolamide. As stress may triggers attacks, stress management techniques (e.g. meditation) may be helpful in controlling symptoms.

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

This information leaflet was written by Ataxia UK in collaboration with Dr Giunti, Neurologist at National Hospital for Neurology and Neurosurgery, London.

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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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