

Spinocerebellar ataxia type 2: SCA2



Spinocerebellar ataxia type 2 (SCA2) 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).

What are the symptoms?

SCA2 is a slowly progressive cerebellar ataxia.

All people with SCA2 experience the following symptoms; balance problems, incoordination of limbs and slurred speech (dysarthria). Another common problem is slower eye movements, meaning people tend to move their heads to compensate. In SCA2 the nerves in the limbs are generally affected resulting in reduced feelings in hands and feet (peripheral neuropathy).

Other possible symptoms include problems with swallowing, painful muscle cramps, tremors, other involuntary movements. Urinary incontinence can occur in the later stages. Some people with SCA2 develop cognitive problems, such as a decline in verbal memory or dementia in the later stages of the condition. [Handbook of ataxia disorders. Ed. Klockgether, 2000; Storey et al. Arch Neurol. 1999;56(1):43-50].

Sometimes SCA2 presents with similar symptoms with Parkinson's disease, and may respond to medications used for that condition. There have been cases where people with SCA2 have been misdiagnosed as having Parkinson's disease. [Wilkins et al. Mov Disord. 2004;19(5):593-5; Furtado et al. Mov Disord. 2004;19(6):622-9.] Symptoms and their severity vary between individuals.

What causes SCA2?

SCA2 is an inherited condition caused by a defect in the so-called SCA2 gene, located on the 12th chromosome. (Each person has 23 pairs of chromosomes). This gene is more extended than normal in people with SCA2.

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 SCA2 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 the age of onset of symptoms and the more severe the condition.

The SCA2 gene has instructions for the production of a protein called ataxin 2. The abnormally extended SCA2 gene results in the production of an altered ataxin 2 protein. Research is underway to understand what the function of the ataxin 2 protein is and what happens when it is extended. This information is important in order to develop treatments.

How is SCA2 inherited?

SCA2 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?

The average age of onset is 30 years of age. However, the age of onset varies greatly, ranging from 6 months to 78 years of age. 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 SCA2 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 partly the instability of the extended SCA2 gene which gets longer as it passes from parent to child. [Handbook of ataxia disorders. Ed. Klockgether, 2000]

How is SCA2 diagnosed?

A genetic test will confirm a diagnosis of SCA2. This involves taking a blood sample to detect the abnormal gene.

How common is SCA2?

SCA2 was first described in a Cuban family. There is a particularly high prevalence of SCA2 in a particular part of Cuba (one study quotes a prevalence of 1 in 200). [Rev Neurol. 2001;16;32(7):606-11].

There is no recent information on the prevalence of SCA2 in the UK. Ataxia UK is currently funding research to rectify this lack of knowledge. The first study published from this research estimates that the prevalence of all the dominant ataxias (of which SCA3 is one) in North East England is 1 in 12,500. [Ann Neurol. 2004 May; 55 (5):752-5].

Management of SCA2

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. There are also medications that can be taken for muscle cramps and for urinary incontinence.

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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