Spinocerebellar ataxia type 3 (SCA3)

Spinocerebellar ataxia type 3 (SCA3) 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). SCA3 is also known as Machado Joseph Disease (MJD).

What are the symptoms?

SCA3 is a progressive cerebellar ataxia. Most people experience the following symptoms; balance problems (ataxic gait), incoordination of hands, slurred speech (dysarthria), and some problems with blurred vision (due to problems with eye movements). Other possible symptoms include problems with swallowing, stiffness, muscle spasms or a reduced ability to discriminate temperature.

There are sometimes similarities between SCA3 and Parkinson's disease, such as rigidity and slowness in movements and resting tremors. Dystonia can also be experienced by people who have these Parkinson-like symptoms. These symptoms may respond to treatments given to people with Parkinson's disease.


What causes SCA3?

SCA3 is an inherited condition caused by a defect in the so-called SCA3 gene, located on the 14th chromosome. (Each person has 23 pairs of chromosomes). This gene is more extended than normal in people with SCA3.

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 SCA3 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 SCA3 gene has instructions for the production of a protein called ataxin 3. The abnormally extended SCA3 gene results in the production of an extended ataxin 3 protein. Research is underway to understand what the function of the ataxin 3 protein is and what happens when it is extended. This information is important in order to develop treatments. Research using cells models of SCA3 has recently given promising results for a form of gene therapy (known as RNAi). [PNAS 2003; 100 (12): 7195-200.] More information in Ataxia UK’s magazine.

How is SCA3 inherited?

SCA3 is inherited in an autosomal dominant way.

When do symptoms start?

The age of onset varies greatly, from 10-70 years of age. In general, the later the onset of symptoms, the slower the progression. There are people in their seventies who have very mild symptoms. Generally people will need to use a wheelchair after a number of years. Sometimes, SCA3 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 SCA3 gene, which gets longer as it passes from parent to child. The opposite can happen in rare cases 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 SCA3 diagnosed?

A genetic test will confirm a diagnosis of SCA3. This involves taking a blood sample to detect the abnormal gene.

How common is SCA3?

There is no recent information on the prevalence of SCA3 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 ataxies (of which SCA3 is one) in North East England is 1 in 12,500. [Ann Neurol. 2004 May; 55 (5):752-5].

Management of SCA3
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. A referral to a neuro-ophthalmologist may help if blurred vision is a problem. There are medications that can be taken for problems with sleeping, muscle spasms or Parkinsons-disease like 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, Consultant Neurologist at National Hospital for Neurology and Neurosurgery, London.

Ataxia UK, 12 Broadbent Close, London N6 5JW
www.ataxia.org.uk  helpline@ataxia.org.uk  Tel: 020 7582 1444  Helpline: 0845 644 0606
Ataxia UK works across the whole of the UK and is a charity registered in Scotland (no SC040607) and in England and Wales (no 1102391) and a company limited by guarantee (4974832).

Disclaimer
This leaflet is for guidance purposes only and, while every care is taken to ensure its accuracy, no guarantee of accuracy can be given. Individual professional advice should be sought before taking or refraining from taking any action based on the information contained in this leaflet and nothing should be construed as professional advice given by Ataxia UK or any of its officers, trustees or employees. No person shall have any claim of any nature whatsoever arising out of or in connection with the contents of this leaflet against Ataxia UK or any of its officers, Trustees or employees.