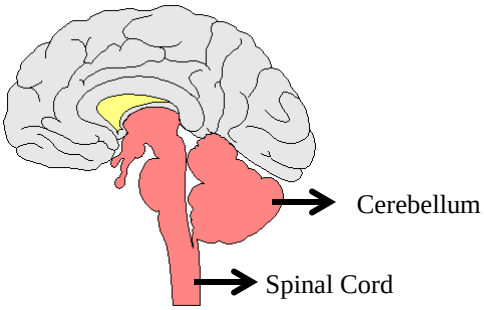
Division of Human Genetics
University of Cape Town

Spinocerebellar Ataxia (SCA)

What is Spinocerebellar Ataxia?

Spinocerebellar ataxia (SCA) is a genetically inherited disorder. Ataxia refers to problems in the control of body posture, motor (movement) coordination, speech control, and eye movements. Nerve tissues in the spinal cord and the cerebellum that control these functions become damaged (Fig. 1).



(http://home1.pacific.net.sg)

Fig. 1. **The Human Brain**. Position of the cerebellum and spinal cord. The cerebellum (meaning 'little brain') coordinates most muscles, movement and timing including eye movement, as well as learned skills.

Presently, over 25 types of SCA have been identified, each caused by a different genetic mutation (fault). The diagnosis of spinocerebellar ataxia is first suspected with the onset of symptoms (see Symptoms). Atrophy (wasting) of the cerebellum is a typical finding in patients with SCA and can be detected with a brain scan. Medical assessment also involves an extensive examination by a neurologist. Genetic testing is another important part of the final diagnosis, as symptoms among the different types of spinocerebellar ataxia are similar. The Division of Human Genetics at the University of Cape Town currently tests for SCA1, SCA2, SCA3, SCA6 and SCA7.

What is the genetic fault that causes SCA?

Spinocerebellar ataxia is caused by a genetic fault that involves the repetition of a particular DNA region in the responsible gene, over and over again. The scientists refer to this as a 'triplet repeat'.

What are the symptoms of SCA?

Each type of SCA (e.g. SCA1, SCA2 etc) is the result of different faults in different genes, but overall the different types share some symptoms. The age at which symptoms will first appear, varies in each case and the longer the 'triplet repeat', the earlier in life the symptoms will start. Normally first symptoms appear in adulthood but they can also occur in childhood or very late in life.

Individuals with SCA initially develop poor coordination of movement (ataxia). This results in a wide-based, uncoordinated, unsteady way of walking, abnormalities in hand or eye movements, speech difficulties and later on difficulty in swallowing. The symptoms typically worsen over time. In some SCA sub-types (SCA1, SCA2, SCA3) there may be progressive muscle weakness and occasionally some intellectual deterioration.

How is SCA inherited?

Spinocerebellar ataxias are inherited as a dominant trait, which means that any parent who carries the fault in the gene will have a 50 percent chance of having an affected child (see Autosomal Dominant sheet). However, if a child does not inherit the gene it cannot pass it on to its own children. The chain of inheritance is then broken.

How Common is SCA?

The incidence of SCA differs from population to population but in general it is thought to be approximately one to five per 100,000 people.

In South Africa, the most common types of SCA are SCA1, SCA2 and SCA7. The SCA7 mutations have so far only been found in families of Black ethnic origin.

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Is there any treatment or cure available for SCA?

Not at present. There is also no treatment to slow the progression of the disease. Treatment is aimed at supporting the patient and his/her family. Orthopaedic aids and appliances may be prescribed and speech and occupational therapists may give advise when speech and swallowing difficulties become a problem. A physiotherapist can give assistance on how to perform chest physiotherapy when chest muscles become affected during the later stages of the disease.

Testing

How does the test work?

The Division of Human Genetics at the UCT provides genetic testing for people affected with SCA (diagnostic testing) and for their family members if they are over 18 years old (predictive testing). This test has been shown to be highly reliable in determining if a person has inherited the mutated (faulty) genes that cause SCA1, SCA2, SCA3, SCA6 and SCA7.

SCA Predictive Testing Protocol

The predictive test for SCA allows testing of family members that may be at risk before any clinical signs present. A programme or protocol has been recommended by doctors and geneticists based on their experiences in dealing with individuals and families at risk for the disease. If, after careful consideration, you decide to take the test, you will be requested to come to the Groote Schuur Hospital Neuro- Genetic Clinic or the Department of Human Genetics on at least four occasions to see the doctors involved in running the programme.

The full protocol for predictive testing is available at the following website:
http://www.humangenetics.uct.ac.za

How soon will I have the results?

Results will be available within approximately 4 weeks of the test . The results will be given to you personally via your general practitioner, neurologist or by the staff of the Division of Human Genetics at the University of Cape Town.

Genetic Counselling

As this is a genetic condition, genetic counselling is strongly recommended. Genetic counselling provides information on the condition, its inheritance pattern, risks to other family members and the prognosis. The Division of Human Genetics at the University of Cape Town (UCT) can be contacted in this regard.

Further information on SCA can be found at:

http://groups.msn.com/ATAXIASOUTHAFRICA/ yourwebpage1.msnw

http://www.geneclinics.org/profiles.

Contact details:

A dedicated Neurogenetic Clinic is held at Groote Schuur Hospital on the first Thursday of every month. Contact for an appointment.

Tel: (021) 404 6235 or 406 6304

Support Groups:

Spinal Cerebellar Ataxia Association (S.C.A.A.)
Mr. A. Andrews (Secretary)
No. 47 Reddy Ave.
Grassy Park
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