

SPINAL BULBAR MUSCULAR ATROPHY

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What is Spinal Bulbar Muscular Atrophy?

Spinal Bulbar Muscular Atrophy (SBMA) is a genetic, motor neuron disorder, otherwise known as X-linked SBMA or Kennedy's disease. Motor neurons are nerve cells which send messages to the muscles of our bodies. Spinal motor neurons of the spinal cord primarily control the voluntary movement of our limbs and the bulbar motor neurons of the brain stem control the voluntary movement of muscles for facial expressions, speaking and swallowing. In SBMA, both of these groups of neurons degenerate, causing muscles to weaken and waste away (atrophy).

SBMA is an adult-onset disorder with symptoms appearing between 20 and 50 years of age. An estimated 1 in 40,000 individuals are affected worldwide. It tends to only affect men, although female carriers may express mild symptoms. Severity is variable among individuals and life span is generally unaffected.

What are the Features of Spinal Bulbar Muscular Atrophy?

SBMA can affect a wide range of muscles throughout the body. Early symptoms include tremors in outstretched fingers (polymyoclonus), muscle cramps, and involuntary facial muscle contractions (fasciculations).

Weakness of arms and legs usually begin in the pelvic or shoulder regions. Most individuals with SBMA retain mobility, however some individuals may be wheelchair-bound in later years.

Facial and tongue muscles can later weaken in the course of the disease resulting in difficulty swallowing (dysphagia) and slurring of speech (dysarthria). Recurrent aspiration pneumonia, caused by food traveling down the airways, occurs in approximately 10% of elderly individuals, and is the only life-threatening problem in SBMA.

Some men develop the excessive enlargement of breasts (gynecomastia), and it is possible to get breast reduction surgery to counter this. It is common for testicles to atrophy and to have a low sperm count or become infertile. Important to note however, is those who are capable of reproducing will have unaffected sons and unaffected daughter carriers.

A few affected males also experience degeneration of the dorsal root ganglia, a part of the spine responsible for passing on sensory information

to the brain. Possible implications of this are mild abnormalities in the sensory function of fingers and toes.

What Causes Spinal Bulbar Muscular Atrophy?

SBMA is an X-linked disorder and therefore primarily affects males. For further information on genetics and how disorders are inherited, please refer to the *Muscular Dystrophy Association Genetics Factsheet*.

In SBMA, there is a mutation on the androgen receptor gene, which produces the androgen receptor protein - responsible for controlling the activity of and response to male sex hormones (androgens). The androgen receptor protein is present in many cells, but more so in motor neurons. When androgens bind to the protein, the function of motor neurons in the spinal cord and brain stem are disrupted. These dysfunctional nerve cells eventually die, resulting in the inability to send messages to the associated muscles and affecting an individual's control over them.

The mutation of the gene can also lead to mild androgen insensitivity, which leads to the diminishment of some male characteristics, such as lower sperm count, and the accentuation of female characteristics, such as breast enlargement.

Diagnosis of Spinal Bulbar Muscular Atrophy

Diagnosis is primarily made through blood tests to check for elevated creatine phosphokinase (CPK), a characteristic of SBMA. Blood samples can also be DNA tested to identify the presence of the abnormal X gene in the individual, as well as in carriers.

Note that SBMA is often misdiagnosed as Spinal Muscular Atrophy or Lou Gehrig's disease due to the similarity of symptoms.

Soon after a diagnosis of SBMA in the family, it is essential that genetic counselling is arranged, for one or both of two issues. The first is the probability of Mum or Dad having the disorder, and the second is whether testing for SBMA in pregnancy can be offered and with what degree of accuracy.

Genetic counselling provides information about possible diagnostic tests, including prenatal testing. Genetic services in NZ are available and a referral can be made by the MDA.

Management of Spinal Bulbar Muscular Atrophy

As there is currently no cure for SBMA, treatment focuses on the prevention and management of symptoms.

It is important to ensure a healthy diet and to exercise regularly to ensure excessive weight does not impede mobility. Physiotherapists and occupational therapists should play an ongoing role in management, including the use of braces and walkers, to encourage mobility. Speech therapists are also beneficial for those suffering from dysarthria. Medication is available to reduce muscle cramps and tremors.

Research into Spinal Muscular Atrophy

Research is being carried out to find the specific cause of the degeneration of the motor neurons in SBMA and as the exact mechanisms become better understood, it will be possible to direct treatment strategies to the cause, rather than simply towards the symptoms.

Support for People with Spinal Muscular Atrophy

Support is available from the MDA who can offer information, support, advocacy and referrals to other providers. There is also a nationwide support network for those interested in meeting with others.

Disability should not hinder employment possibilities, though it is wise to choose a career that does not require heavy physical activity. Any individual has the right to equal pay and equal rights for employment. For more information contact the Employment Relations infoline on 0800 800 863 or visit www.ers.dol.govt.nz.

Due to the adult onset of SBMA, some individuals may already have an established career when diagnosed. It may be necessary to have an evaluation SBMA is likely to affect an individual's ability to perform his or her job. This is usually performed by a physiotherapist or occupational therapist.

The government promotes equal employment opportunities in private sector and can be contacted on (09) 525 3023 or visit www.eeotrust.org.nz

Workbridge provides a professional employment service for people with all types of disabilities and administers support funding on behalf of Work and Income. Contact on 0508 858 858 or visit www.workbridge.co.nz

More Information

Muscular Dystrophy Association can be contacted for further information, assistance, advice, support and referrals, on 0800 800 337 or by e-mail at info@mda.org.nz.

The Muscular Dystrophy Association Website also contains information on services available within NZ, our quarterly magazine, contacts, membership details, news and links to other sites - www.mda.org.nz

Further Resources

www.nzord.org.nz – the New Zealand Organisation for Rare Disorders website provides information on a number of rare disorders, a directory of support groups, practical advice, health and disability resources, research information, news and issues.

www.mdausa.org – the Muscular Dystrophy Association USA website has an extensive site with plenty of further information on any muscular dystrophy conditions as well as research news.

www.kennedysdisease.org – the Kennedy’s Disease Association website with information on the condition and current research.

Information in this fact sheet was primarily sourced from:

National Institute of Neurological Disorders and Stroke (2006) Kennedy’s Disease Fact Sheet <www.ninds.nih.gov>

Muscular Dystrophy Association USA (2001) Spinal Bulbar Muscular Atrophy. <www.mdausa.org>