

SPINAL MUSCULAR ATROPHY

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What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a genetic, motor neuron disorder. Lower motor neurons are nerve cells in the spinal cord which send messages to the muscles of our bodies. These however, degenerate in SMA, causing muscles to weaken and waste away (atrophy). SMA can affect a wide range of muscles throughout the body with the muscles closest to our trunk, such as shoulders, hips and back, being the most affected.

SMA does not affect neurons of the brain and therefore intelligence is not compromised, and it is often noted that individuals are bright and sociable. Sensations, such as touch, temperature and pain, also remain undisturbed.

Of these rare neuromuscular disorders, SMA is one of the more common. Approximately 1 every 6,000 babies are affected and about 1 in 40 are identified as genetic carriers.

What are the Features of Spinal Muscular Atrophy?

SMA can be classified into several different types based upon the achievement of physical milestones. It is important to remember though, that SMA differs for every individual and therefore one's condition may not correspond exclusively to one particular type. The implications and management of SMA varies for all individuals, so it is important to discuss all concerns and options of care with your specialist.

Type I

Alternative names: Werdnig-Hoffmann disease or infantile progressive SMA.

Onset: Diagnosis is usually made before 6 months of age and occurs more frequently in males than females.

Symptoms and characteristics: Unable to sit unaided, lack of motor development, severe muscle weakness, poor head control, difficulty swallowing, feeding and breathing.

Prognosis: 50% of infants do not survive beyond 2 years of age

Inheritance: Autosomal recessive.

It is not possible to predict survival, therefore providing effective supportive and therapeutic management is suggested for all infants. Diaphragmatic breathers require comprehensive respiratory support. This is essential and ventilatory assistance may be required at times. An

increased risk of contracting pneumonia is apparent and it is important that family members are aware of the signs to ensure early medical assistance. Chest physiotherapy is helpful and sometimes regular suctioning of the airways to keep them clear.

Normal feeding can be difficult and time consuming, trying to ensure infants do not draw food or fluid into their lungs. Assisted feeding, via a tube into the stomach, for more severe cases may be recommended.

It is beneficial to use stimulation for cognitive, physical and emotional development, especially games that encourage the use of motor skills.

Supportive seating in push chairs and cars will be necessary, and motorized wheelchairs may be an option, as is assisted standing in standing frames.

Type II

Alternative names: Intermediate SMA.

Onset: Between 7 and 18 months.

Symptoms and characteristics: Weakness in arms, legs, upper and lower torso, unable to stand unaided, involuntary muscle contractions (fasciculations) of the tongue and small tremors in outstretched fingers (mini polymyoclonus) may be present.

Prognosis: Respiratory and swallowing problems may develop and survival is dependent on the severity of these.

Inheritance: Autosomal recessive.

It is important to get your child standing at the earliest possible age, as this allows for better respiratory function, bowel function, and encourages greater mobility and normal skeletal development. Standing aids may be helpful. Regular exercise and stretching programmes, with the help of a physiotherapist, to maintain muscle strength and flexibility, and to prevent the development of contractures (chronic loss of joint motion) are also necessary.

Respiratory problems in diaphragmatic breathers can arise and must be monitored. Chest physiotherapy can be beneficial.

Scoliosis, an abnormal curvature of the spine, can become an issue in SMA children and can impair respiration. Spinal bracing may be required, and in more severe cases spinal fusion surgery. An orthopaedic specialist is essential in monitoring the scoliosis.

A wheelchair is likely to be needed and the choice of wheelchair and seating support can be assessed by an occupational and/or seating therapist. Manual or motorised wheelchairs are available options and can

be introduced from as early as 1 year old, allowing exploration and development of spatial awareness.

These children tend to be advanced in intellectual development so offering adequate stimulation and learning experiences is essential for their advancement.

Type III

Alternative names: Kugelberg-Welander Disease, juvenile SMA.

Onset: 1-15 years.

Symptoms and characteristics: Weakness mostly in legs, hips, shoulders and less so in arms, minor walking difficulties, respiratory muscles are sometimes affected, and small tremors in outstretched fingers may be present.

Prognosis: Life span is generally not affected, though a wheelchair may be required in later life.

Inheritance: Autosomal recessive.

Individuals with Type III SMA can typically stand and walk unaided, however may have difficulty at some point in the course of their condition, and may show difficulty when rising to an upright position and running. It is important to monitor these motor abilities to ensure any emerging difficulties are detected early and so needs can be catered for. Walking aids such as braces, walking sticks or frames may become necessary, or perhaps even a lightweight wheelchair or motorized scooter.

Physiotherapists should play a regular role in monitoring as well as in exercise and fitness programmes.

Type IV

Alternative names: Adult-onset SMA.

Onset: 18-50 years.

Symptoms and characteristics: Generalised muscle weakness and wasting, muscle twitches are common.

Prognosis: Symptoms remain relatively mild and has little impact, if any, on life span.

Inheritance: Autosomal recessive and dominant forms.

In Type IV SMA, individuals should be aware of their own weaknesses and limitations and work together with a physician, physiotherapist and occupational therapist to develop a programme that best caters for his or her needs.

What Causes Spinal Muscular Atrophy?

Most types of SMA are autosomal recessive disorders, however some are autosomal dominant disorders. For further information on genetics and how disorders are inherited, please refer to the *Genetics Factsheet*.

In SMA there is a defect on the gene SMN1 (survival motor neuron 1), located on chromosome 5. The absence of the associated protein, SMN, can have a severe effect on lower motor neurons, causing them to atrophy, shrink and eventually die. Consequently, the brain can no longer send messages to the muscles, impairing muscular activity.

During growth, there is an increased demand on the motor neurons and the muscle atrophy results in muscle weakness, a central feature of SMA. This may lead to bone and spinal deformities and further loss of function, including the compromise of the respiratory (breathing) system.

In some cases, the gene SMN2 (survival motor neuron 2) may be replicated up to four times. To some extent, the presence of additional SMN2 genes can help to replace the protein needed for the survival of motor neurons. As a result, individuals with more copies of this gene, tend to experience less severe symptoms in later life.

Diagnosis of Spinal Muscular Atrophy

Diagnosis usually commences after the identification of key characteristics of SMA. An individual's family history can also be an indicator that any symptoms are related to the presence of SMA. Several tests can be carried out to confirm the diagnosis:

- Muscle Biopsy – shows typical signs of damage and is easier to detect in older children
- Electromyography (EMG) – observes the electrical activity of muscles and its consistency with activity typical of SMA individuals
- DNA Testing – can identify the presence of the abnormal gene in the individual with SMA as well as in carriers.

Soon after a diagnosis of SMA 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 SMA 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 Muscular Atrophy

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

From an early stage, it is important to undergo regular exercise and stretching programmes, with the help of a physiotherapist, to maintain muscle strength and flexibility.

For all types of SMA, a good diet with plenty of fresh fruit and vegetables is very important in ensuring excessive weight does not impede mobility. Contact with a physician and/or a nutritionist is valuable for this.

Research into Spinal Muscular Atrophy

Research is being carried out to find the specific cause of the degeneration of the lower motor neurons in SMA 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. Sodium valproate has been shown to increase expression of SMN2 and thereby improve upon the severity of this condition.

Support for People with Spinal Muscular Atrophy

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

There is no reason why individuals with SMA should be disadvantaged in terms of receiving full education. For more information, request the Education Pack available from the MDA.

Disability should not hinder employment possibilities, though it is wise to choose a career that does not require 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.

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

Families of Spinal Muscular Atrophy NZ can also offer support. Their website www.smanz.org has additional information on the condition and research being carried out. They can be contacted on 0800 FSMANZ or by e-mail on newzealand@fsma.org

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 MDA USA website has an extensive site with plenty of further information on any muscular dystrophy conditions as well as research news.

www.fsma.org – website designed to support families whose children have SMA

Information in this fact sheet was primarily sourced from:

National Institute of Neurological Disorders and Stroke (2006) Spinal Muscular Atrophy Fact Sheet <www.ninds.nih.gov>

Families of SMA (2006) Understanding Spinal Muscular Atrophy. <www.fsma.org>

Muscular Dystrophy Association of Australia (2003) Spinal Muscular Atrophy. <www.mda.org.au>