Muscular Dystrophy Canada
let's make muscles move

Spinal Muscular Atrophy (SMA)

What Is Spinal Muscular Atrophy?

QuickFACTS:

What: Spinal Muscular Atrophy
Who: Anyone
When: At any age
Where: Motor nerve cells
Why: Various genetic causes

Muscular Dystrophy Canada's Exhaustive List Of SMA-Related Disorders

Childhood-onset spinal muscular atrophy:

- SMA Type I, or infantile progressive spinal muscular atrophy
- SMA Type I RD, or Diaphragmatic SMA with respiratory distress
- SMA Type II, or intermediate progressive spinal muscular atrophy
- SMA Type III, or juvenile progressive spinal muscular atrophy
- SMAX2, or lethal infantile spinal muscular atrophy X-Linked
- SMAL, or congenital nonprogressive SMA of lower limbs

Adult-onset spinal muscular atrophy:

- SMA Type IV, or adult progressive spinal muscular atrophy
- SMAD1, or Spinal muscular atrophy (SMA) with upper limb predominance
- SMAX1, X-linked spinal bulbar muscular atrophy

Spinal muscular atrophy is the name given to a group of genetic muscle-wasting disorders. SMA affects the nerve cells that control voluntary muscle. These nerve cells are called motor neurons and SMA causes them to die off. Without motor neurons, the brain cannot deliver signals to the muscles. Unable to fully use them, the muscles of a person with SMA will waste away. The larger muscles closer to the body - such as the hip and shoulder muscles - may be more affected than the smaller muscles of the hands and feet, and usually the legs are weaker than the arms. Muscles in the face and throat may experience weakness as well. Depending on what type of SMA a person has, there may also be a loss of reflexes. In the more serious cases of the disorder, the muscles responsible for breathing and swallowing may be severely affected.

The term SMA is used to describe a wide spectrum of disabilities with great variation in age of onset and severity of weakness. In the past, researchers and physicians have classified SMA in a variety of ways.

Are There Other Symptoms?

Depending on the course of SMA development, loss of reflexes or presence of tremor in the hands and fingers may be noted. Problems such as scoliosis (curvature of the spine) may occur because the normal growth of the skeletal system relies to an extent on normally functioning muscles. Intelligence and sensation (the senses of heat, pain, touch, etc.) are not affected in any of the forms of SMA.

In many types of SMA, the respiratory muscles are

affected in some way. Respiratory difficulties, including an increase in the frequency and severity of chest infections and episodes of choking and respiratory distress may be experienced and can be quite serious.

Who Can Be Affected By Spinal Muscular Atrophy?

Most types of SMA can affect either males or females. The age of onset depends on the type of SMA involved. SBMA almost always affects adult males.

Is Spinal Muscular Atrophy Anyone's Fault?

No. Spinal muscular atrophy is a genetic disorder. It is often seen in families with no previous history of the disorder

Is It Contagious?

No. Genetic disorders are not contagious.

How Is SMA Inherited?

Spinal muscular atrophy is passed from generation to generation.

What About the Less Common Forms of SMA?

SMA Type IV and some other rare cases of SMA may be transmitted through other patterns of inheritance as well, so it is important for parents and families to seek genetic counselling for clarification of how the disorder is transmitted within their family.

What Causes Spinal Muscular Atrophy?

The body is composed of hundreds of thousands of small units known as cells (for example brain cells, kidney cells, etc.). Movement of the body is actually started in the brain where nerve impulses carrying messages are conducted down the spinal cord and then out to the muscles. Cells called motor neurons, situated in the spinal cord, send signals to the muscles to tell them to move. These cells are in a part of the spinal cord known as the anterior horn, hence their other name - the anterior horn cells. We are born with more of these cells than are required. As part of the normal development of the body, a certain number of the motor neurons die back. In SMA, loss of motor neurons means that some voluntary muscles lose their connection to the spinal cord. Without this connection, such muscles are unable to function, leading to a weakening of the limbs and chest wall movement.

Genes are the basic functional units of heredity that tell the cells and tissues of the body what specialized functions they will perform. They are found in the nucleus of the cell as tiny segments of DNA on structures known as chromosomes. There are thousands of genes located on the 23 pairs of

What Is Spinal Muscular Atrophy Type I?

Also known as infantile progressive spinal muscular atrophy, this disorder usually appears within the first six months after a child is born. Weak from birth, the child will probably be unable to sit independently. This weakness is also apparent in the chest muscles leading to symptoms such as poor cough, poor feeding, and an increased breathing rate. Children with SMA Type I often have more frequent chest infections. The disorder is almost always fatal within two years and is considered to be the most common genetic cause of infant mortality.

Age of Onset: usually around the time of birth

Prognosis: poor

Symptoms:

1. Neonatal hypotonia.
2. Generalized muscle weakness is severe, with some infants being almost completely immobile.
3. Weakened muscles affect crying, sucking, breathing and swallowing.
4. Frequent chest infections.
5. Survival beyond the third year of life, while occasionally seen, is rare.

chromosomes inside each of the billions of cells within the human body.

(For more information on genes and genetics see the Info.MDC sheet "What is Genetics: Genetics, Genetic Testing, and Gene Therapy") The gene associated with SMA - that when deleted causes breakdown of the brain/muscle connection - has been found to be the survival motor neuron gene located on chromosome 5.

The significant majority of cases of SMA are caused by the deletion or mutation of the SMN (survival motor neuron) gene located on chromosome 5. Two versions of this gene exist: SMN1 and SMN2. These genes differ by only five basepairs, and these five basepairs are considered translationally silent. The SMN protein is ordinarily involved with the metabolism of a compound known as RNA.

How do these genes lead to SMA? It has been found that almost all cases of SMA are the result of missing or mutated copies of the SMN1 gene, but that the severity of the disorder then is related to the total copies of SMN2 in the nucleus. In other words, the absence of SMA1 causes SMA, while the quantity of SMA2 modulates the severity of the disorder.

In X-linked spinal bulbar muscular atrophy (SBMA), the disorder is caused by a mutation on the androgen receptor gene found on the X chromosome. In this disorder, the genetic mutation appears to involve unstable trinucleotide repeats in a gene that is responsible for the production of (androgen) male hormones receptor proteins. In general, the greater the number of repeats, the greater the severity of the disease. In individuals who are not affected, there are between 11 and 31 repeats of cytosine, adenine and guanine (CAG). Alternatively, in those who have SBMA, the number of repeats has been found to be between 40 and 60. It is believed that the number of repeats tends to increase in subsequent generations (genetic anticipation).

At the present time, SMA Type IV is less common and less clearly understood than other types, but it is also felt to be the result of a genetic error.

What Is Spinal Muscular Atrophy Type II?

Also known as intermediate SMA, this disorder can be recognized within the first year to eighteen months of life. It is not as severe as infantile SMA. Weakness is slower to appear than in SMA Type I, but the symptoms are very similar. In fact, it may be impossible to tell the difference between these two forms in the first year. The child is often able to sit independently, but usually unable to stand or walk on their own. The spine and also the respiratory muscles can be involved in the disorder.

Age of Onset: 6 months to 3 years

Prognosis: depends on the degree of respiratory involvement

Symptoms:

1. Possibility of neonatal hypotonia
2. Frequent respiratory infections may complicate the course of this disorder.
3. Life expectancy may be shortened.
4. Motor milestones delayed
5. Scoliosis

How Is Spinal Muscular Atrophy Diagnosed?

A diagnosis of spinal muscular atrophy is usually made by a physician. A family history and a complete physical examination may provide the doctor with information about an individual's history of muscle wasting (atrophy) and weakness in the trunk (upper body), arms and legs. It is important to determine whether the muscle weakness is due to SMA, other early childhood conditions or another neuromuscular disease. Physicians use diagnostic tests to aid in making an accurate diagnosis.

In Type I (infantile) SMA, genetic studies are often not considered until after the death of the affected child, when the opportunity to obtain a DNA sample may be more difficult. It is important that physicians and others responsible for the child's care communicate to all involved the necessity of a blood sample from the affected child in order to carry out prenatal diagnosis in later pregnancies. Arrangements to obtain and store DNA samples can be made through any Canadian Genetics Centre. The same approach can be used for prenatal diagnosis of the milder types of SMA, if desired.

Which Diagnostic Tests Are Performed?

1. A muscle biopsy is a test where a small piece of muscle tissue is surgically removed and examined under a microscope. This test may show changes in the muscle that are characteristic of SMA.

2. An electromyogram or EMG is a procedure in which small electrodes (needles) are placed into a muscle. The presence of a particular form of electrical activity in the muscles detected by this test suggests the possibility of SMA.

3. With recent advances in genetic research in SMA, it is now possible to perform prenatal (before birth) diagnosis for many couples who have had a child with SMA and who are having another child. This DNA based diagnosis is also replacing the muscle biopsy for the diagnosis of an infant or child with possible SMA in many centres. There are several Canadian laboratories that perform this test. Your genetic counsellor can give you up-to-date information.

What Is Spinal Muscular Atrophy Type III?

Juvenile SMA is a milder form of SMA that starts at a later age than either Type I or II. Although symptoms sometimes appear in a mild form as early as 1 year old, most children with this disorder are diagnosed between 2 and 12 years of age - and occasionally a diagnosis is not made until adulthood. Running, rising from the floor and climbing stairs become increasingly difficult as the child grows older. Also, walking is usually possible for many years after the diagnosis is made, though the gait may be affected

Age of Onset: usually 5 to 15 years or earlier

Prognosis: good; life expectancy is often normal

Symptoms:

1. Weakness may be noted in youth or even adulthood
2. Much more variable than either Types I or II
3. Motor difficulties are usually progressive, but may be marked with periods of stability.

Who Are The Members Of The Health Team?

Individuals with SMA or SBMA and their family may be referred to a neuromuscular clinic at a specialized hospital or rehabilitation centre in their province or a local team of health professionals may follow them.

Treatment will vary with age and with the severity of weakness, but the following health professionals will give you an idea of whom you may be in contact: The physician (neurologist, pediatrician, physiatrist and/or family physician) will guide the treatment program and will monitor the person's overall condition, including feeding and respiratory problems. Referrals may be made to other health professionals, such as a dietician, respirologist, or genetic counsellor.

The orthopedic surgeon will monitor bone and joint development, especially in the hips and spine. Children with SMA are at risk for curvature of the spine and may require back bracing or eventual surgery. A referral may be made to an orthotist for the provision of leg braces, standing frames or

walking devices.

The physiotherapist (PT) will teach parents chest physiotherapy techniques that will help prevent respiratory complications. Families will also be taught a stretching and exercise program in order to try to prevent joint contractures (abnormal shortening of muscle tissue) and to maintain the best possible muscle strength. The physiotherapist helps clients develop individual exercise programs that include the type and amount of activity that is best for them.

The occupational therapist (OT) can help clients learn how to adapt to physical limitations and attain their optimum level of achievement in activities of daily living. He/she may construct splints to be worn at night by the client to keep the joints in a good position. For the very young infant, the OT may use foam and straps for positioning the baby in a side-lying or sitting position to allow a better view of the family and the environment. As well, the OT assists with feeding problems, if they are present. For the older baby and child, the OT may choose a special seat to provide good posture and alignment. For all clients, the therapist will also be involved in choosing therapeutic equipment to enhance function at home, school or the workplace. There is a wide range of equipment available-from mobility aids (such as scooters and motorized wheelchairs) to self-care equipment (such as bathtub lifts) and technical aids (such as computers).

The OT/PT, in conjunction with an infant development worker, may have suggestions for activities to enhance intellectual development.

The role of the nurse, whether in the clinic or the community, is to work with the family to understand the disorder and its management. The social worker, in conjunction with the nurse, provides support to the individual and family coping with the effects of SMA.

Finally, SMA is a rare disorder. As a consequence, other families with children who have SMA can themselves be sources of much useful and practical information.

How Can Families Help?

Education and information about SMA and issues related to living with a disability are important in the

What Is X-Linked Spinal Bulbar Muscular Atrophy?

Also known as SBMA or Kennedy's disease, X-linked spinal bulbar atrophy is a relatively rare disorder that almost always affects men. Symptoms can occur from the late teens to middle age. Like SMA Type I, SBMA often affects the motor neurons that control the tongue and throat, resulting in difficulty with chewing and swallowing. In addition, people with SBMA experience varying degrees of muscle weakness and wasting, especially of the facial muscles.

Age of Onset: 15-59 years of age

Prognosis: good; life expectancy is not affected

The genetic mutation related to SBMA is in the androgen receptor gene that results in changes to male hormones (androgens). As a result, men affected with this disorder may show symptoms related to androgen insensitivity or deficiency such as breast development, testicular atrophy (wasting), reduced libido, impotence and infertility. These symptoms may become evident before muscle weakness is experienced. Androgen sensitivity is not expressed in all men with this disorder.

Many men with SBMA also experience fasciculations (small, local muscle contractions under the skin). Intelligence is normal. Although SBMA is slowly progressive, life expectancy is usually unaffected.

life of any person living with SMA. Parents are often their children's main source for information about the disorder. As difficult as it may be, questions need to be addressed openly and honestly. Adults affected with the disorder and their families may also have questions and concerns. Therapists, physicians and MDC Services personnel are available resources.

A balanced, nutritional diet is essential to achieve the maximum function of muscles. As well, excessive weight gain by people affected by spinal muscular atrophy is not recommended. The extra work that weakened muscles must do to lift excess weight only makes these muscles seem weaker than they actually are. Lots of fruit and fibre are important in the diet since constipation is also a common problem. Parents may need to provide their child with information about healthy nutrition and how it affects him/her. The ongoing support and understanding of family where nutrition is concerned is important.

SMA does not affect an individual's intelligence. Children should be encouraged by their parents and teachers to learn to the best of their ability. Providing a stimulating environment and challenging activities can help foster a life-long love of learning.
It is important to treat family members with SMA the same as others in the family. For example, children with SMA need discipline just as their siblings do. Chores that are physically suitable for them should be a part of their day. Extracurricular activities are as important for those with SMA as they are for anyone else and they should be included wherever possible.

People with SMA need and deserve to be as independent as possible. They need to experience the same sense of responsibility, competence and achievement, as do the other members of the family. The family provides the environment in which these attitudes and experiences are free to develop. Friendships are important, as well. Regular interaction and playtime with siblings and children in the neighbourhood and at school help a child learn necessary social and problem-solving skills. Parents can go a long way towards encouraging their child socially and participating as a family in social and recreational activities. Adults also benefit from a social network that is satisfying to them and understanding of their needs. Family and friends play an integral role in the emotional well-being of those with SMA. At school, encouraging the child with SMA to take part as normally as possible within the school environment is very important. Rules and subsequent consequences should be the same for all children within the classroom. Realistic expectations about academic performance and any modifications to a child's educational program need to be resolved as they become evident. Parents' relationships with teachers and school administration pave the way for open discussion and positive resolution of any problems that may arise. Information about the child's disorder will benefit all of the staff who work at the school. MDC can provide families with assistance, if desired, in keeping teachers up-to-date about SMA and related issues.

In the work environment, it is the responsibility of the adult with SMA to negotiate employment conditions with his/her superiors and co-workers.

Family members can provide support, encouragement and suggestions as specific needs are determined and instituted within the workplace.

All people with SMA may face frustration as they deal with physical and attitudinal barriers within the community where they live, work and go to school. While many adults may be comfortable advocating on their own behalf, children and adults can benefit from the ongoing support and empathy of their families.

How can I help?

Muscular Dystrophy Canada conducts year-round fund raising campaigns to support our diverse programs. Your gift will help the Association provide the dollars necessary to assist individuals living with neuromuscular disorders, and fund much needed medical research and educational information. Please make a gift through our National office or any Regional or Community Muscular Dystrophy Canada office.

All Muscular Dystrophy Canada Information Sheets are available on our website: www.muscle.ca

Ce feuillet d'information est aussi disponible en français.