Families of SMA Canada Society

Support. Research. Hope.

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FSMACS is committed to serving you and your family.

A Guide To Understanding Spinal Muscular Atrophy

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This booklet is intended to serve as a source of information and support for children, teens, and adults with Spinal Muscular Atrophy (SMA). It was created by Families of Spinal Muscular Atrophy Canada Society (FSMACS). FSMACS is dedicated solely to eradicating SMA by promoting and supporting research, supporting families, and educating the public and medical community about SMA.

For more information about FSMACS please call us toll free at 1-855-824-1277 or visit our website at www.curesma.ca.

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How Families of SMA Canada Society Can Help

Families of SMA Canada Society is a resource for unbiased support. We do not advocate any specific choices or decisions; we are here for anyone who wishes to talk through their options. All choices related to SMA are highly personal and need to reflect your own personal values.

As caring parents and professionals, we offer support and understanding when it is most needed. By phone and networking , Families of SMA Canada Society staff and members are here for you.

Our newsletter keeps families and professionals up to date on the latest research , technology, and ideas for day to day coping. Families of SMA Canada Society also has an email list that families can sign up for to receive the latest in research updates and clinical trials.

Annual Conferences allow families and professionals hands-on techniques and family to family support, while also giving children a great opportunity to make new friends and have a great time!

Most of all, Families of SMA Canada Society can offer friendship and hope. We are one of the largest private funders of SMA research. We support families by supplying information, newly diagnosed kits and packages as well as support to families on their journey of caring for a member with Spinal Muscular Atrophy.

I Am a Carrier of the SMA Gene, What Can I Do?

You may have requested this information because you have been diagnosed as a carrier through a blood test. If you find you are a carrier of the SMA gene, Families of SMA Canada Society recommends that you seek the advice of a genetic counselor. This counselor can help you to better understand the risks and chances of having an affected child. The genetic counselor will take a complete family history , which will include any diseases, deaths, causes of death, stillbirths, and miscarriages of each family member. If you have already given birth to a child with SMA, the counselor can discuss your options that you may want to consider regarding future pregnancies.

The information presently available allows prenatal testing with 98% reliability. These are individual decisions and very personal, it is important that both parents discuss their feelings before making a decision.

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Table of Contents

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a motor neuron disease. The motor neurons affect the voluntary muscles that are used for activities such as crawling, walking, head and neck control, and swallowing. It is a relatively common "rare disorder"; approximately 1 in 6000 babies born are affected and about 1 in 40 people are carriers.

SMA affects muscles throughout the body although the proximal muscles (those closest to the trunk of one's body-i.e. shoulders, hips, and back) are most severely affected. Weakness in the legs is generally greater than in the arms. Sometimes feeding and swallowing can be affected. Involvement of respiratory muscles (muscles involved in breathing and coughing) can lead to an increased tendency for pneumonia and other lung problems. Sensation and the ability to feel are not affected. Intellectual activity is normal and it is often observed that patients with SMA are unusually bright and sociable. Patients are generally grouped into one of four categories; Type I, Type II, Type III and Type IV, depending on severity, age of onset and reaching key motor function milestones. The course of the disease may be different for each child.

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Prognosis, What Does it Mean?
What Are We to Expect?

Type II (and some Type III) Continued

If your child has frequent colds and difficulty coughing you may want to inquire about the cough assist machine. The Cough Assist ™ is discussed in detail in the Type I section. Diet, as with any growing child, is very important. Your child's diet deserves careful consideration. Excessive weight can make mobility more difficult. Constant contact with your physician and nutritionist is very important in this aspect of maintenance.

Type III

Because children with Type III walk at some point unassisted, it is important that they be monitored so that any difficulty may be detected at an early stage. The use of a walker and bracing may become necessary. The use of a lightweight manual wheelchair may be considered for distance as well as an electric scooter or other motorized chair. Physical and occupational therapists should be consulted. Diet should also be monitored.

Type IV Adult Onset

As an adult you are aware of your weaknesses and limitations. You should work together with your physician, physical and occupational therapists to work out the best possible program for you. As with Types I, II, and III, diet and nutrition are an important factor in your well being.

Prognosis, What Does it Mean?
What Are We to Expect?

Type II (and some Type III)

It is important to get your child upright at the earliest possible age. Standing is important in development. It allows for better respiratory function, improved bowel function, and encourages greater mobility. Getting your child in an upright position may sometimes require advocacy on the part of the parents to encourage the physician to write a prescription for standing aids.

There are several options to consider when choosing an appropriate standing aid. One option is a standing frame and/or parapodium. For added mobility and independence, a standing wheelchair is ideal. A child as young as thirteen months can use this. Bracing is also an option. Reciprocating Gait Orthosis (RGO's) and weight bearing Knee Ankle Foot Orthosis (KAFO's) have been found to work for children with Type II, and these children have been able to take some steps. Use of the appropriate type of assistive device or walker with braces is important and various options should be explored with your therapist.

The use of a light weight manual wheelchair can be an exciting addition for the SMA child. It can provide mobility, independence, and a taste of adventure, while still allowing them to use some of their own strength. However, it should be understood that for true independence and mobility , a power wheelchair is necessary.

Scoliosis (curvature of the spine) occurs at some point in essentially all children with SMA Type I and Type II and some Type III. The degree of the scoliosis will be a factor in deciding how to treat it. Because scoliosis can restrict breathing and pulmonary function, necessary precautions should be taken early. Options for managing scoliosis are: custom seating systems, seating aids and a body jacket. Later, spinal fusion surgery may need to be considered.

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What Causes Spinal Muscular Atrophy?

SMA is an autosomal recessive genetic disease. In order for a child to be affected by SMA, both parents must be carriers of the abnormal gene and both must pass this gene on to their child. If both parents are carriers, the likelihood of a child inheriting the disorder is 25% or 1 in 4.

An individual with SMA has a missing or mutated gene (SMN1, or Survival Motor Neuron 1) that produces a protein in the body called SMN protein. The protein deficiency has its most severe affect on motor neurons. Motor neurons are cells in the spinal cord which send out nerve fibers to muscles throughout the body. SMN protein is critical to the survival and health of motor neurons, without this protein nerve cells may atrophy, shrink and eventually die, resulting in muscle weakness.

As a child with SMA grows, their bodies are doubly stressed, first by the decrease in motor neurons, and then by the increased demands on nerve and muscle cells as their bodies grow larger. The resulting muscle atrophy can cause weakness, and bone and spinal deformities that may lead to further loss of function, as well as additional compromise of the respiratory (breathing) system.

Type I

Type I SMA is also called Werdnig-Hoffmann Disease. The diagnosis of children with this type is usually made before 6 months of age and in the majority of cases the diagnosis is made before 3 months of age. Some mothers even note decreased movement in the final months of their pregnancy.

Usually a child with Type I is never able to lift their head or accomplish the normal motor skills expected early on in infancy. They generally have poor head control, and may not kick their legs as vigorously as they should, or bear weight on their legs. They do not achieve the ability to sit up unsupported. Swallowing and feeding my be difficult and are usually affected at some point, and the child may show some difficulties managing their own secretions. The tongue may show atrophy and rippling movements or fine tremors, also called fasciculations. There is weakness of the intercostal muscles (the muscles between the ribs) that help expand the chest, and the chest is often smaller than usual. The strongest breathing muscle in an SMA patient is the diaphragm. As a result the patient appears to breath with their stomach muscles. The chest may appear concave (sunken in) due to the diaphramic (tummy) breathing. Also due to this type of breathing, the lungs may not fully develop, the cough is very weak, and it may be difficult to take deep enough breaths while sleeping to maintain normal oxygen and carbon dioxide levels.

Prognosis, What Does it Mean? What Are We to Expect?

Type I (and some Type II) Continued

3. (Continued) A tracheostomy tube bypasses the mouth and vocal chords and goes directly from the skin to the trachea (wind pipe). A Respirator or ventilator is connected to the endotracheal tube or tracheostomy tube.

Consult your physicians and respiratory therapists or Families of SMA Canada Society for more literature.

It is important to understand your rights when it comes to making life-sustaining decisions for your child. Be sure that both parents discuss their feelings about this very delicate topic. It is a decision that can not be made lightly and all options should be covered. Talking to a counselor in the Department of Social Services at your hospital may be helpful. Once your decision has been reached, be sure that you put it in writing, and that all necessary medical personnel and family members are aware of your wishes. This is your decision, one you have reached with great care and anguish, and under no circumstance should you allow others to judge you or place their values upon you. You are never alone. Families of SMA Canada Society is just a phone call away.

Prognosis, What Does it Mean?
What Are We to Expect?

Type I (and some Type II) Continued

1. **BiPAP** (Bilevel Positive Airway Pressure) uses a nasal mask with a cap, which fits over the head to hold it in place over the nose . BiPAP provides a higher volume of air into the lungs during inhalation and inflates the lung greater than what the person can do on their own. During exhalation, the BiPAP pressure drops so that air can passively leave the lungs. The BiPAP machine can sense when the person is taking a breath and give the breath in synchrony with the individual. A respiratory rate is also set so that the BiPAP gives a minimum number of breaths per minute. The person can breath above that rate and the Bi-PAP will deliver more breaths. CPAP (continuous positive air pressure) should never be used in patients with SMA.

2. **Negative Pressure Ventilation** refers to providing breaths into the lungs using a large chamber or tank that encircles the chest similar to the old Iron Lung. The chamber is connected to a vacuum pump that takes the air out of the chamber and, as a result , the chest wall expands to bring air into the lungs. A Port-A-Lung is an example of a negative pressure ventilator. It can be set to deliver a specific number of breaths per minute and a vacuum pressure.

3. **Mechanical Ventilators or Respirators** come in variety of different models . Mechanical Ventilators are more complex , but also allow for control of more variables. The Ventilator can be set to deliver a specific size of a breath at a set number of breaths per minute. Mechanical Ventilation can be delivered with a nose mask, mouthpiece while awake, or through a tracheostomy tube. A tracheostomy tube is placement of a surgical hole in the neck to the large airway (trachea) that a tube can be inserted.

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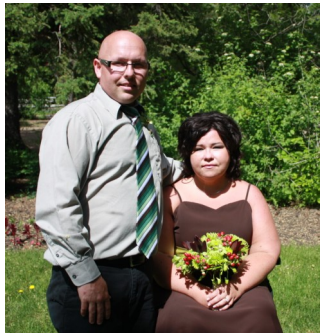
Type II

The diagnosis of Type II SMA is almost always made before 2 years of age, with the majority of cases diagnosed by 15 months. Children with this type may sit unsupported when placed in a seated position, although they are often unable to come to a sitting position without assistance. At some point they may be able to stand. This is accomplished with the aid of assistance or bracing and/or a parapodium standing frame. Swallowing problems are not usually characteristic of Type II but vary from child to child. Some patients may have difficulty eating enough food by mouth to maintain their weight and grow, therefore a feeding tube may become necessary. Children with Type II SMA frequently have tongue fasciculations and manifest a fine tremor in the outstretched fingers. Children with Type II also have weak intercostal muscle and are diaphramic breathers. They have difficulty coughing and may have difficulty taking deep enough breaths while they sleep to maintain normal oxygen levels and carbon dioxide levels. Scoliosis is almost uniformly present as these children grow, resulting in need for spinal surgery or bracing at some point in their clinical course. Decreased bone density can result in an increased susceptibility to fractures. Causes for concern include the respiratory system, as once weakened, it never fully recovers.

Type III

The diagnosis of Type III, often referred to as Kugelberg-Welander or Juvenile Spinal Muscular Atrophy, is much more variable in age of onset, and children can present from around 1 year of age or even as late as adolescent, although diagnosis prior to age 3 years is typical. The patient with Type II can stand alone and walk, but may show difficulty with walking at some point in their clinical course. Early motor milestones are often normal. However, once they begin walking, they may fall more frequently, have difficulty in getting up from the floor, or a bent over position, and ma be unable to run. With Type III, fine tremor can be seen in the outstretched finger but tongue fasciculations are seldom seen. Feeding or swallowing difficulties in childhood are very uncommon. Type III individuals can sometimes lose the ability to walk later in childhood, adolescence, or even adulthood, often in association with growth spurts or illness.



Type IV (Adult Onset)

In the adult form, symptoms typically begin after age 35. It is rare for Spinal Muscular Atrophy to begin between the ages of 18 and 30. Adult onset SMA is much less common than the other forms. It is defined as onset of weakness after 18 years of age, as most cases reported as type 4 have occurred after the age of 35. It is typically characterized by insidious onset and very slow progression. The bulbar muscles, those muscles used for swallowing a respiratory functions, are rarely affected in Type IV.

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Prognosis, What Does it Mean? What Are We to Expect?

Type I (and some Type II) Continued

Because a child with SMA Type I has difficulty coughing, contacting respiratory therapist is very important so you can be instructed in chest physiotherapy (CPT). CPT is a method of clearing the lungs of accumulated mucus by using body positioning and clapping on the chest to assist in loosening secretions. Saliva can settle in the nasopharnyx causing a faint gurgling sound. Often the secretions or mucus need to be removed by the use of a suction machine. Blowing raspberries and bubbles encourag1es respiratory strength.

Individuals with SMA can also benefit from the use of a cough assist machine. The Cough Assist ™ achieves this by applying a positive pressure to the airways to inflate the lungs, and then rapidly shifts to negative pressure to pull the air out of the lungs. The rapid shift in pressure produces a high expiratory flow from the lungs, simulating a cough. This technique, referred to as “mechanical insufflation-exsufflation,” avoids airway damage while clearing the lungs of secretions. The device offers patients greater comfort and quality of life without the use of invasive procedures and equipment. Patients as young as four months have been able to use the Cough Assist successfully.

Respiratory distress can be monitored by measuring the level of oxygen saturation in the blood using a tool called a pulse oximeter. A small clip or tape with a red light and a sensor is placed on the patient’s finger or toe to determine the oxygen saturation . Children with SMA Type I usually require breathing support while sleeping. Some children require additional breathing support especially with colds. There are several options to consider. ***(continued on page 14)***

Prognosis, What Does it Mean?
What Are We to Expect?

Type I (and some Type II)

While most children diagnosed with Type I are still infants there are a myriad of things that can be done to assist in the cognitive , physical, and emotional health of your child. Using balloons and feathers as toys makes for wonderful stimulation and allows them a feeling of independence and accomplishment. Reaching games are a form of occupational and physical therapy that can be very helpful. Instructions in range of motion and other physical/occupational therapy ideas by a licensed therapist are important no matter how young the child. Your physical/occupational therapist can also suggest ideal seating systems that will be most helpful in the comfort and maximum mobility of your child.

Water therapy can be very helpful as the buoyancy of the water allows movement of the arms and legs that may otherwise not be there. Be sure that the water temperature is at least 90 degrees F and that the child's head does not go under the water or into the water. You must watch so that the child has no possibility of aspirating (getting fluid into their lungs).

Children with SMA who have difficulty swallowing are at risk of aspirating when eating. Sometimes the child may aspirate on their own secretions. The child may choke while eating and also may experience weight loss as swallowing becomes more difficult. Assistive feeding may be necessary. Two possible options are:

1. Nasogastric Tube (NG tube): a surgically placed tube through the nose that goes directly into the stomach.
2. Gastrostomy Tube (G-Tube); a surgically placed tube through the skin that goes directly to the stomach.

Patients with SMA typically lose function over time. Loss of function can occur rapidly in the context of growth spurt or illness, or much more gradually. The explanation for this loss is unclear based on recent research . It has been observed that patients with SMA may often be very stable in terms of their functional abilities for prolonged periods of time, often years, although the almost universal tendency is for continued loss of function as they grow.

Diagnosing Spinal Muscular Atrophy

SMA is diagnosed primarily through a blood test which looks for the presence or absence of the SMN1 gene, in conjunction with a suggestive history and physical examination. Normally, individuals have two genes called Survival Motor Neuron 1 and 2. In approximately 95% of patients with SMA, there is an absence of the SMN gene sequence which is present in normal individuals. Sometimes the SMN1 gene is not missing, but mutated. The number of copies of SMN2, a near identical backup copy of the SMN1 gene, is related to the severity of the disease, but does not reliably predict a specific SMA type in a given individual. SMA type is generally determined from the clinical examination evaluating the child's degree of weakness and ability to achieve major motor milestones such as sitting independently or walking. Occasionally doctors may request a muscle biopsy or EMG, (electromyography) testing. Since the genetic blood test became available , a muscle biopsy is almost never indicated and is valuable mainly in cases where the blood DNA test is negative.

EMG measures the electrical activity of muscle. Sometimes this test is preformed to help distinguish other disorders of nerve or muscle which can mimic SMA. Small recording electrodes (needles) are inserted into the patient's muscles, usually the arms and thighs, while an electrical pattern is observed and recorded. In addition, a nerve conduction velocity test (NCV) is performed to help assess how well the nerves are functioning in response to an electrical stimulus. Small shocks are repeatedly administered to help assess nerve integrity and function. When performing this test on a child, if at all possible it should be performed by a doctor experienced in caring for children.

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Prognosis, What Does it Mean? What Are We to Expect?

Researchers have identified the SMN1 gene as the primary manufacturer of the SMN Protein. It is the absence/defect of this SMN1 gene that causes Spinal Muscular Atrophy. However there is another form of this gene called SMN2. The SMN2 gene is similar to SMN1, but does not produce as much protein, or the right kind of protein, as the SMN1 gene. One determination of prognosis is the number of copies of the SMN2 gene. The greater the number of SMN2 copies, the more SMN protein is produced and the greater the likelihood that more motor neurons remain healthy and productive. Individuals with only one or two copies of the SMN2 gene will typically have the most severe expressions of SMA. Three or more copies of the SMN2 gene will typically mean a less severe expression.

Each type of SMA has variability among individual patients. Please keep this in mind when considering an individuals care.

Raising a child with SMA should be no different than raising a child who is not affected. Do as many things as possible that are age appropriate. Many times this means making adaptations. It is very important that children with SMA are assisted in reaching their utmost potential.

It is important to understand that parents and patients have rights and that you are not alone. Most hospitals have Social Services departments that can give you a shoulder to lean on. Don't be afraid to say NO if something does not seem right. Don't be intimidated or afraid to ask questions. If you forget to ask something, call your doctor or contact Families of SMA Canada Society for suggestions. In this context, it is also important that your child be followed by a physician who is familiar with SMA and it's complications.