



Understanding Spinal Muscular Atrophy

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This booklet is intended to serve as a source of information and support for children and adults with Spinal Muscular Atrophy (SMA). It was created by Families of SMA (FSMA). FSMA is the largest international organization dedicated solely to eradicating SMA by promoting and supporting research, helping families cope through informational programs and support, and educating the public and the medical community about SMA. For more information about FSMA, visit <http://www.CureSMA.org> or call 1-800-886-1762.

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 genetic 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 often 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, based on certain key motor function milestones.

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. Although 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 Survival Motor Neuron (SMN) protein. This protein deficiency has its most severe affect on motor neurons. Motor neurons are nerve cells in the spinal cord which send out nerve fibers to muscles throughout the body. Since 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 the 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.

There are four types of SMA, SMA Type I, II, III, IV. The determination of the type of SMA is based upon the physical milestones achieved. It is important to note that the course of the disease may be different for each child.

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 of the final months of their pregnancy.

Usually a child with Type I is never able to lift his/her 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 may 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 breathe with their stomach muscles. The chest may appear concave (sunken in) due to the diaphragmatic (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.

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, and 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 intercostals muscles and are diaphragmatic 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.

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 a year of age or even as late as adolescence, although diagnosis prior to age 3 years is typical. The patient with Type III 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 sitting on the floor or a bent over position, and may be unable to run. With Type III, a fine tremor can be seen in the outstretched fingers 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, and most

cases reported as type IV have occurred after age 35. It is typically characterized by insidious onset and very slow progression. The bulbar muscles, those muscles used for swallowing and respiratory function, are rarely affected in Type IV.

Patients with SMA typically lose function over time. Loss of function can occur rapidly in the context of a 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 age.

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 numbers 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 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 performed 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.

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 likelihood that more motor neurons remain healthy and productive. Individuals with only 1 or 2 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 individual's 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 service departments that can give you a shoulder to lean on. Don't be afraid to say NO if something doesn't 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 for suggestions. In this context, it is also important that your child be followed by a physician who is familiar with SMA and its complications.

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 physical or occupational 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°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 for aspirating when eating. Sometimes the child may aspirate his/her own secretions. The child may choke while eating and may also 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 into the stomach.

Because a child with SMA Type I has difficulty coughing, contacting a 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 positioning and clapping on the chest to assist in loosening secretions. Saliva can settle in the nasopharynx 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 encourages respiratory strength.

Individuals with SMA can also benefit from the use of a cough assist machine. The CoughAssist™ 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 4 months have been able to use the CoughAssist™ 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 more breathing support, especially with colds. There are several options to consider.

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 breathe above that rate and the BiPAP 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 a variety of models. Mechanical ventilators are more complex, but also allow for control of more variables. The ventilator can be set to deliver a specific size 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 tracheotomy is placement of a surgical hole in the neck to the large airway (trachea) that a tube can be inserted. A tracheostomy tube bypasses the mouth and vocal cords 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 contact Families of SMA for 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 cannot 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 personal and family members are aware of your wishes. This is your decision, one you have reached with great care and anguish, and under no circumstances should you allow others to judge you or place their values upon you. You are never alone. Families of SMA is always just a phone call away.

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 the 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 13 months can use this. Bracing is also an option. Reciprocating GaitOrthosis (RGO's) and weight bearing knee ankle foot orthoses (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 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.

If your child has frequent colds and difficulty coughing you may want to inquire about a cough assist machine. The CoughAssist™ is discussed in more 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 a nutritionist is very important in this aspect of care.

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 light-weight 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 therapist 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.

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 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 with you options that you may want to consider regarding future pregnancies.

The information presently available allows for 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.

How Families of SMA Can Help

Families of SMA 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 can offer support and understanding when it is most needed. By phone and networking, Families of SMA staff and members are here for you.

A quarterly newsletter keeps families and professionals up to date on the latest in research, technology, and ideas for day-to-day coping. Families of SMA also has an e-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 the children a great opportunity to make new friends and have a great time.

Because Families of SMA understands the financial hardship of living with SMA, the organization maintains an equipment pool, which is available, free of charge, to members of Families of SMA.

Most of all Families of SMA can offer friendship and hope. As one of the largest private funders of SMA research, FSMA has funded over \$23 million for SMA research, and has allocated an additional \$12-\$15 million over the next three years.

Families of SMA has more than 24 chapters worldwide and more than 5,000 member families and is a founding member of the International Alliance for Spinal Muscular Atrophy, a coalition of International SMA groups working to find a cure for SMA. (www.iasma.net). Please contact us so that we can help you with any questions you may have or support you may need. We are all here for each other.

This information was written under the editorial supervision of FSMA's Medical Advisory Board.



Balancing Life's Tough Times™

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