

Understanding Spinal Muscular Atrophy: A Comprehensive Guide

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This booklet is to serve as a source of information and support to those involved with children and adults with Spinal Muscular Atrophy (SMA).

Families of SMA was founded by a group of parents of children with SMA Type II. They all continue to be actively involved working to raise money to distribute educational materials, provide patient services and support research, which will lead to a treatment and cure. Research funded by Families of SMA has aided in the discovery of the SMA chromosome, the location of the SMA gene, and the isolation of protein produced by the SMA gene.

Whether you have a family member or close friend, your interest in this is probably based on the fact that you or someone you care about is awaiting diagnosis of SMA or has been diagnosed with SMA.

We understand the concern that you might have upon hearing the term, Werdnig-Hoffmann, Kugelberg-Welander, Spinal Muscular Atrophy. We've all heard of leukemia, AIDS and cystic fibrosis but Spinal Muscular Atrophy is an unknown. It is still an unknown despite the fact that 1 in 40 people are carriers, and 1 in every 6,000 live births is affected which is why so much research is being conducted to discover its cause(s) and cure.

What Is Spinal Muscular Atrophy?

Spinal Muscular Atrophy (SMA) is a disease of the anterior horn cells. Anterior horn cells are located in the spinal cord. SMA affects the voluntary muscles for activities such as crawling, walking, head and neck control and swallowing.

It mainly affects the proximal muscles, or in other words the muscles closest to the point of origin, in this case those closest to the trunk of one's body. Weakness in the legs is generally greater than weakness in the arms. Some abnormal movements of the tongue, called tongue fasciculations may be present in patients with Type I and some patients with Type II. The senses/feelings are normal as is intellectual activity. In fact it is often observed that patients with SMA are unusually bright and sociable.

Type I Acute (Severe)

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 before 3 months, there may be lack of fetal movement in the final months of pregnancy.

Usually a child with Type I, (Werdnig-Hoffmann) is never able to lift his/her head or accomplish normal physical milestones. Swallowing and feeding may be difficult and the child may show some difficulties with their own secretions. There is a general weakness in the intercostals and accessory respiratory muscles (the muscles situated between the ribs). The chest may appear concave (sunken in) due to the diaphragmatic (tummy) breathing.

Please note: Although diagnosis may be made before 6 months of age it does not necessarily follow the same course of severity for each patient.

Type II (Chronic)

Diagnosis of Type II 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 although they are usually unable to come to a sitting position without assistance. At some point they may be able to stand. This is most often accomplished with the aid of bracing and/or parapodium/standing frame. Feeding and swallowing problems are not usually characteristic of Type II although in some patients this can occur and a feeding tube may become necessary. Tongue fasciculations are less often found in children with Type II but a fine tremor in the outstretched fingers is common. Children with Type II are also diaphragmatic breathers.

Type III (Mild)

Diagnosis of Type III, often referred to as Kugelberg-Welander or Juvenile Spinal Muscular Atrophy, is made sometime after 18 months of age and as late as adolescence. The patient with Type III can stand alone and walk, but may show difficulty with walking and/or getting up from a sitting or bent over position. With Type III, a fine tremor can be seen in the outstretched fingers but tongue fasciculations are seldom seen. Patients with Spinal Muscular Atrophy Types I, II and III will lose function over time. The explanation for this loss is unclear based on recent research.

Type IV (Adult Onset)

Typically in the adult form symptoms begin after age 35. It is very rare for Spinal Muscular Atrophy to begin between the ages of 18 and 30. Adult SMA is 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.

Adult Onset X-Linked SMA

This form also known as Kennedy's Syndrome or Bulbo-Spinal Muscular Atrophy, occurs only in males, although 50% of female offspring are carriers. This form of SMA is associated with a mutation in the gene that codes for part of the androgen receptor and therefore these male patients often have breast enlargement known as gynecomastia. Also noticeably affected are the facial and tongue muscles. Like all forms of SMA the course of the disease is variable, but in general tends to be slowly progressive or nonprogressive.

Diagnosing Spinal Muscular Atrophy

It is important that we all understand the tests that we must endure. This is especially important when it comes to our children. After having gone through this testing we often do not remember to ask for an explanation of all of the tests. If and when we did ask, we weren't clearheaded enough to hear or understand the explanation. We, as parents, have gathered information to answer these initial questions and explain as simply as possible their diagnostic uses. Up until 1995 there were 3 major lab tests used for diagnosis, as well as the clinical exam.

As recently as the fall of 1995, probes that detect deletions in Types I, II, and III SMA were reported. One of these probes is for a gene called Survival Motor Neuron (SMN) and detects the absence of gene sequences in approximately 90-94% of SMA patients and is not absent in normal individuals. This information makes this SMN gene test very useful for the diagnosis of SMA.

However the defect in this gene cannot be used to indicate the severity of the disease. It is believed that a blood test to screen for SMN deletion is all that is necessary to make the diagnosis. It may not be necessary to perform the EMG and the muscle biopsy.

The results may show that there is no deletion of the SMN gene. If this is the case than a muscle biopsy and/or EMG would be necessary to confirm the diagnosis.

(1) Serum Enzymes

This is a regular blood test. The enzyme most commonly studied is CPK (creatine-phosphokinase). In Type I (Werdnig-Hoffmann) this enzyme tends to be normal, but moderate elevation may occur in the milder forms.

(2) EMG (Electromyography)

This test measures the electrical activity of muscle. In this procedure small needles are inserted into the patient's muscles, usually the arms and thighs, while an electrical pattern is observed and recorded. The readout is similar to that of an EKG or lie detector. In addition, a nerve conduction velocity (NVC) may also be performed. In this test the response of a nerve to an electrical stimulus is measured. When performing this test on a child, if at all possible, it should be performed by a doctor experienced in dealing with children. Also be sure to bring lots of things with which to keep your child occupied. Hold your child on your lap during the procedure, as it is a tremendous help in making an unpleasant procedure somewhat bearable. Ask your doctor whether your child/the patient should be given a mild pain killer or sedative prior to the test.

(3) Muscle Biopsy

This is a surgical procedure where an incision, approximately 3 inches long, is made and a small section of muscle is removed, usually from the thigh. The biopsy is used to check for degeneration. Although many doctors may persuade you of the necessity of a general anesthetic, this procedure can be done with a local anesthetic. It is an especially important point when dealing with children who may be suffering from a neuromuscular disorder and may have weak respiratory function.

PLEASE NOTE: THERE IS AN ALTERNATIVE TO THE BIOPSY, IT IS A PROCEDURE KNOWN AS A NEEDLE BIOPSY. INSTEAD OF A 2 to 3 INCH INCISION, ONLY A SMALL NICK IN THE SKIN IS NECESSARY. BE SURE TO INQUIRE ABOUT THIS PROCEDURE.

No matter what, understand that you as a parent or as a patient 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 S.M.A. for suggestions.

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

Each type of SMA has variability among individual patients. Please keep this in mind when reading this section.

Type I Acute

In the acute type of this disease the bulbar muscles are often affected, and this may make feeding and swallowing extremely difficult. Breathing is often labored due to reduced strength of the chest muscles, and most breathing can be seen in the abdominal areas, with the chest appearing sunken in. Because of increasing overall weakness or repeated respiratory infections, the prognosis is poor. Death in the majority of children with Type I S.M.A. usually occurs by 2 years of age.

AGAIN IT IS IMPORTANT TO NOTE THE OVERLAP OF TYPES I AND II.

Type II Intermediate

Because of the range of progression seen in patients with Type II it is hard to tell how fast, if at all, the weakness will progress. Some children may learn to walk with the aid of bracing and may survive into adulthood. However, others, due to weakened chest and respiratory muscles may become increasingly weak with probable respiratory infections such as pneumonia. There are many cases in which the initial progressive weakness may remain the same, or there may be periods of worsening followed by long periods of stability. With such variables, age of death can vary greatly. It can take place as early as 3 years as in Type I or not until adulthood.

Although not all children diagnosed with Type II develop respiratory weakness, respiratory failure is usually the cause of death following a bout of pneumonia or other respiratory infection.

Type III Mild

Patients with Type III will again vary greatly. However the prognosis is very good. Often walking will be possible, or the patient will be fully functional for years before assistance is necessary. As with Type I and Type II respiratory infections should be presented and necessary precautions taken.

Type IV Adult Onset

There is nothing unusual or distinctive about the current management of the adult forms of Spinal Muscular Atrophy. Proper diagnosis, genetic counseling and appropriate physical therapy remain mainstays.

Taking The Diagnosis Home ... What Can We Do?

Our first reaction when the doctors told us we could take our child home was "Home?!" How could they expect us to go home with a child whom we had brought in "well," and now they are sending us home with a child who has a life-threatening disease? It is hard to accept that with so many modern medical advances there is so little help. The medical field has yet to find any drug, therapy or surgery to cure the Spinal Muscular Atrophy diseases.

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 and emotional health of your child. Using balloons and feathers as toys makes for wonderful stimulation and allows them that feeling of independence and accomplishment. Reaching games are a form of physical therapy that can be very helpful. Instructions in range of motion and other physical therapy ideas by a licensed physical therapist are important no matter how young the child. Your physical therapist can also suggest ideal seating systems that will be most helpful in the comfort and maximum mobility of your child.

Also getting in touch with a respiratory therapist is very important especially so you can be instructed in CPT (chest physiotherapy). CPT is a means of clearing the lungs of accumulated mucus, by using a series of procedures to assist in coughing. Saliva can settle in the nasopharynx causing a faint gurgling sound. Often the secretions or mucus can not be cleared with these noninvasive measures and the use of a suction machine may be necessary. Blowing raspberries, bubbles, anything encourages respiratory strength.

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).

Aspirating can also become a problem with children when eating. Sometimes the child may even aspirate his/her own secretions. As this becomes a problem loss of weight may also be noted and assistive feeding may be necessary.

Two possible options are:

- (1) Naso Gastric Tube: which is a tube inserted through the nose which goes directly into the stomach.
- (2) Gastric Tube: is a more permanent option. It is a surgical procedure to insert a tube or button directly into the stomach.

Although many doctors may try to persuade you of the necessity of a general anesthetic, this procedure can be done with a local anesthetic and an intravenous (IV) sedation drip.

It will be necessary to monitor respiratory distress by measuring the level of oxygen and to determine if oxygen from an outside source is necessary. The tool used to measure this is called a Biox-oximeter. This is a small clip which is usually placed on the patient's index finger to determine the oxygen saturation.

To help the child with breathing a ventilator can be used. There are several possibilities when considering ventilation.

- (1) Negative Pressure Ventilation can be achieved by placing the patient in a Port-A-Lung. This is a much smaller version of the old fashioned Iron Lung. It works by using external ventilation to create negative pressure to set the rate of breathing.
- (2) Bi-Pap (Biphasic Positive Airway Pressure). This ventilation unit uses a nasal mask with a cap, which fits over the head to hold it in place over the nose. A small hose is attached that feeds

oxygen. This unit allows maximum inspiration and expiration. A small alarm is also attached to detect for leaks.

For long term ventilation a tracheotomy is usually performed. There may be other options available. Consult your physicians and respiratory therapists or contact FAMILIES OF S.M.A. 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)

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. This includes getting the child upright at the earliest possible age.

Standing is important in the development of all children. It allows for better respiratory function, greater bowel function, and encourages greater mobility. Getting your child in an upright position can sometimes take pushing on the part of the parent to encourage the physician to write a prescription for standing aids.

There are several options to consider when choosing the appropriate standing aid. Among them are a standing frame and a 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 Gait Orthosis (RGO's) have been found to work for children with Type II, and these children have been able to take some steps.

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 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. Among these options are custom seating systems, seating aids and a body jacket. Later spinal fusion may need to be considered.

If your child is having continuous upper respiratory infections you may want to inquire about an IPPB (Intermittent Positive Pressure Breathing) machine. The IPPB may assist with respiratory

function and help prevent the lungs from becoming stiff . IPPB is also helpful in eliminating secretions. Using an Incentive Spirometer daily allows you to measure lung capacity. When volumes are low it usually indicates an increase in mucus and /or a cold developing.

Diet as with any growing child is very important. Your child's diet deserves careful consideration. Excessive weight can make mobility more difficult. Good eating habits help contribute to strong minds and strong bodies. Constant contact with your physician and a 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 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 watched.

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. It is important to keep your body and mind healthy.

The diagnosis of Spinal Muscular Atrophy can be a frightening prospect, but it can also be looked on as a gift. Unfortunately in life, it seems to take something tragic or earth shattering to make us open our eyes to the joys of today. Having a child with SMA or being an adult with SMA gives life a different perspective. We become more aware of the simple pleasures, the seasons, the smiles, the tears. We become grateful for the simple things, getting a glass of water, taking a step, breathing. We learn to accept diversity and challenges with a new found enthusiasm and drive and we learn how many people there are who truly care.

What Causes Spinal Muscular Atrophy?

Spinal Muscular Atrophy is an autosomal recessive disease, which means that both parents must be carriers. Both parents must have the gene responsible and these genes must be passed onto their child. When a child has received this gene from each of its parents it will than be affected by SMA. Although both parents are carriers the likelihood of passing this gene along to a child and having an affected child is 25%, or 1 in 4.

Familial Forms (affecting other family members) of Spinal Muscular Atrophy in the older age group can occur as autosomal recessive, mutants or autosomal dominant. The genetic defects underlying these diseases make it necessary to be precise regarding the inheritance pattern in a particular family.

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

If you find you are a carrier of the SMA gene it is necessary that you seek the advice of a Genetic Counselor. This counselor will assist biological parents to better understand the risks and chances of having another affected child. The Genetic Counselor will take a complete family history which will include any diseases, deaths, causes of death, still births and miscarriages of each family

member. Using this information helps them to identify persons likely to carry a defective gene. Sometimes laboratory studies will follow.

The information presently available allows for prenatal testing with 98% reliability. The decision to have prenatal testing performed and the options available once the results of the testing are back can be difficult. These are individual decisions and very personal. It is important that both parents have discussed their feelings together and with their Genetic Counselor who will offer unbiased support in their decision. Often times a family may also wish to consult with their clergyman. Once a decision has been made it is important to be supportive of one another and to allow any necessary time to grieve. This is a difficult decision, one that has taken great anguish and thought. Under no circumstances should you allow others' values or judgment to affect you. Remember you are not alone. Families of SMA is only a phone call away.

Ongoing Research

The Indiana University Roster which is funded by Families of S.M.A. was instrumental in locating the chromosome. The gene has been identified and the protein discovered. Now through the collaborative efforts of the North American Spinal Muscular Atrophy Research Group, funded totally by Families of SMA, research moves forward to the next stages.

- * continued development of SMA mouse models
- * development of therapeutic strategy by either gene replacement or gene activation
- * clinical trials

Major researchers throughout the world are being funded by Families of S.M.A. in efforts to not only alleviate but to cure the Spinal Muscular Atrophy diseases. It is an exciting thought that as research progresses we will reach our goal for a treatment and cure in the not too distant future.

How Families of SMA Can Help

As caring parents and professionals who have experienced the day to day trials and tribulations of raising a child with Spinal Muscular Atrophy or have had to deal with the loss of a child or family member with Spinal Muscular Atrophy, we can offer support and understanding when it is most needed. By phone and networking Families of SMA volunteer staff and members are there when you need them or just a friendly ear to listen and share. The publishing of a quarterly newsletter keeps families and professionals up to date on the latest in research, technology, and day to day coping in regards to Spinal Muscular Atrophy.

The sponsoring of annual conferences allows 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 living with Spinal Muscular Atrophy can cause, they have a very large 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. 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.



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