Understanding Spinal Muscular Atrophy (SMA)
This booklet is intended to serve as a source of information and support for children, adults, and families living with Spinal Muscular Atrophy (SMA).
Spinal muscular atrophy (SMA) refers to a group of inherited diseases of the motor nerves that cause muscle weakness and atrophy (wasting). The motor nerves arise from the spinal cord and control the muscles that are used for activities such as breathing, crawling, walking, head and neck control, and swallowing. SMA is a rare disorder affecting approximately 1 out of every 10,000 individuals worldwide.

SMA affects muscles throughout the body. In the most common types, weakness in the legs is generally greater than in the arms. Sometimes feeding, swallowing, and respiratory function (e.g., breathing, coughing, and clearing secretions) can be affected. When the muscles used for breathing and coughing are affected and weakened, this can lead to an increased risk for pneumonia and other respiratory infections, as well as breathing difficulty during sleep. The brain's cognitive functions and the ability to feel objects and pain are not affected. People with SMA are generally grouped into one of four types (I, II, III, IV) based on their highest level of motor function or ability.
SMA is an autosomal recessive genetic disease. About 1 out of every 50 people are genetic carriers of the disease (meaning that they carry the mutated gene but do not have SMA). In order for a child to be affected by SMA, usually both parents are carriers of the abnormal gene and pass this gene on to their child. Thus, the child has two abnormal copies of the gene, one from each parent, and this is termed a recessive genetic disease. When both parents are carriers, the possibility of a child inheriting the disorder is 1 in 4, or 25%, with each pregnancy.

SMA is caused by a missing or abnormal (mutated) gene known as survival motor neuron gene 1 (SMN1). In a healthy person, this gene produces a protein in the body called survival motor neuron (SMN) protein. In a person with mutated genes, this protein is absent or significantly decreased, and causes severe problems for 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, nerve cells may shrink and eventually die without this protein, resulting in muscle weakness. As a child with SMA grows, it is difficult for his/her weakened muscles to keep up with the demands of daily activities. The resulting weakness can also lead to bone and spine changes that may cause breathing problems and further loss of function.
There are four types of SMA: type I, II, III, and 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 affected individual. Although SMA is not generally regarded as a progressive disease, people affected with SMA typically lose function over time as muscles continue to weaken. Loss of function may occur gradually or more rapidly in the context of a growth spurt or illness. The reasons why muscle weakness and subsequent loss of function occur at different rates in different individuals remain poorly understood. It has been observed that individuals with SMA may be very stable in terms of their abilities for prolonged periods of time, often years, although the almost universal tendency is for continued loss of body function as they grow older.

**TYPE I**

SMA type I is also called Werdnig-Hoffmann Disease. The diagnosis of children with this type is usually made before 6 months of age, depending on the severity of their disease. Usually children with SMA type I have poor head control and are not able to accomplish developmentally-expected motor skills. The hallmark feature of SMA type I is that children are unable to sit or stand without help. They will require equipment such as strollers or wheelchairs for mobility. Swallowing and feeding will become difficult for children with this type of SMA and they will eventually lose the ability to swallow safely without aspirating (choking or inhaling secretions and food particles into the lungs). Children may eventually require a feeding tube to supplement their nutrition which permits the administration of liquid food into their stomachs. Families have different opinions and can make personal decisions about whether to place a feeding tube or not. If families chose to have a feeding tube placed, they may choose to do so before swallowing becomes difficult, however the timing for this procedure is also an individual preference.

**Please see the booklet entitled Caring Choices for Families of Infants Newly Diagnosed with SMA type I for more information on individual decision making.**
Children with SMA type I will experience weakness of the muscles used for breathing, those that help expand the chest and fill the lungs with air. The chest is smaller than usual in these children and they begin to breathe using primarily their stomach muscles (belly breathing). The lungs do not fully develop due to this type of breathing, and coughing becomes very weak. It may be difficult for children to get enough air into their lungs while sleeping, and eventually throughout the day. Many of these children will require equipment to help them breathe; again this is based on what each family chooses. Weakness also leads to problems with the spine (scoliosis) and hips (displacement) that may lead to further loss of ability/function. Severe curvature of the spine can be treated with a brace and in some instances surgery. Bones become weak and may break easily, and several different therapeutic supports are available to help position children with SMA type I comfortably.

**TYPE II**

The diagnosis of SMA type II is almost always made before two years of age. Children with this type have delayed motor milestones and display a range of physical abilities. The hallmark feature of SMA type II is the ability to maintain a seated position unsupported, however, some children may initially require assistance getting into that seated position. With assistance and bracing, these children may be able to stand, but are unable to walk and require a wheelchair to get around.
Children with SMA type II usually do not have swallowing problems, but this can vary from child to child. Some children may have difficulty eating enough food by mouth to maintain their weight and grow, and a feeding tube may become necessary if the family decides this is best. Children with SMA type II may also develop weakness of the muscles used for breathing and experience difficulty coughing. Some of these children may require equipment to help them breathe easier at night. Weakness also leads to problems with the spine (scoliosis) and hips (displacement) that may lead to further loss of function. Severe curvature of the spine can be treated with a brace and in many instances surgery. Bones become weak and may break easily, and a variety of therapeutic supports are available to help position children with SMA type II comfortably.

TYPE III

SMA type III is also known as Kugelberg-Welander disease, or Juvenile Spinal Muscular Atrophy. It is typically diagnosed by 3 years of age, but can be diagnosed as late as the teenage years. The hallmark feature of SMA type III is the ability to stand and walk independently. Affected individuals may have difficulty walking, running, and climbing stairs as they get older; some will lose the ability to walk independently in childhood, while others may remain ambulatory into adolescence or adulthood. Problems with the spine (scoliosis) may develop at various rates and ages. Swallowing and coughing difficulties, along with breathing difficulty at night, may occur but do so less commonly and later in the disease course than in
SMA type II. Children and adults with SMA type III are at risk of becoming overweight, as they are not usually able to be extremely physically active. Fine shaking of the fingers and hands (tremors) can be seen in this type of SMA, and symptoms of joint aches and overuse frequently occur. Curvature of the spine may occur and can be treated with a brace and in some instances surgery. As in SMA type II, bones become weak and may break easily, and a variety of therapeutic supports are available to help position individuals with SMA type III comfortably and maintain mobility.

TYPE IV (ADULT ONSET)

In the adult form of SMA, mild to moderate symptoms usually begin in the second or third decade of life, typically after the age of 35, although they may occur as early as 18 in some cases. Adult onset SMA is much less common than the other forms. It is typically characterized by mild motor impairment such as muscle weakness, tremor, and twitching, with or without respiratory problems. Weakness is gradual and the muscles used for swallowing and breathing are rarely affected in SMA type IV. Life expectancy is normal and therapeutic supports are available to help maintain optimal function for individuals with SMA type IV.

SMA types I through IV are discussed in detail later in this booklet.
### Spinal Muscular Atrophy Respiratory Distress (SMARD)

SMARD is very rare and has been identified as a variant of SMA type I. Children with this type of SMA present similarly to infants with SMA type I and the diagnosis is usually made very early on in life, if at all. Infants with SMARD experience symptoms of severe respiratory distress due to extreme weakness in the muscles used for breathing; the arms and nearby muscles are affected more than other muscles. SMARD differs from SMA type I in that the upper spinal cord is affected more than the lower spinal cord. The specific location of the genetic mutation for SMARD has been identified on gene *IGHMBP2* and thus it can be determined through genetic testing when infants/children show signs of SMARD.

### Spinal Muscular Atrophy Type V/Distal Hereditary Motor Neuropathy

SMA type V is very rare and only a few cases have been reported. It is an autosomal dominant genetic disease, meaning that only one copy of the abnormal gene needs to be present (from one parent) for the disease to occur, and it is caused by mutations in the *BSCL2* and *GARS* genes. Type V affects nerve cells in the spinal cord, and muscle weakness occurs in the hands and feet only. The defining characteristic, which usually occurs first and may be brought on by exposure to cold temperatures, is cramps or weakness and wasting of the muscles of the hand, specifically on the thumb side of the index finger and in the palm at the base of the thumb. A high arch of the foot (pes cavus) is also common and some individuals may develop trouble walking (gait abnormalities). Symptoms usually begin during adolescence, but may occur from infancy through the mid-thirties. People with this disorder have normal life expectancies.

### Kennedy’s Disease

Kennedy's Disease is a rare form of SMA that affects only males. It is an X-linked autosomal recessive disease (the mother carries the defective gene on one of her X chromosomes); each male child born to a mother with the defective gene has a 50% chance of having the disease, and daughters born have a 50% chance of being a carrier of the disease. Early symptoms include slight shaking (tremor) of the outstretched hands, muscle cramps with exertion, and muscle twitching visible under the skin (fasciculations). Limb weakness usually begins in the shoulder or pelvic region, and may eventually spread to the facial and tongue muscles leading to difficulty swallowing (dysphagia), difficulty speaking (dysarthria), and aspiration pneumonia. Individuals with Kennedy’s Disease may require a wheelchair during later stages of the disease. Some males may experience breast enlargement (gynecomastia), low sperm count/infertility, and/or non-insulin dependent diabetes mellitus. Disease onset usually occurs between the ages of 20 and 40, although it has been diagnosed from adolescence up to age 70. People with this disorder have normal life expectancies.
Diagnosing Spinal Muscular Atrophy

SMA is often diagnosed on a clinical basis by how the child appears physically. The diagnosis may be suspected when children are noted to be weak or to have a delay in their developmental milestones, such as holding their head up, rolling over, sitting independently, standing, or walking later than would be expected. After a thorough medical history is reviewed and a physical exam is performed, a primary care provider may order genetic testing through a blood sample, or a child may be referred to a neurologist who will also perform an examination and then order genetic testing (again through a blood sample) to confirm the diagnosis.

Occasionally, an electromyography (EMG) or a muscle biopsy may be performed, or a creatine kinase (CK) level ordered. If it’s necessary, EMG is a test that measures the electrical activity of muscle. Sometimes this test is performed to help distinguish SMA from other disorders of nerve or muscle that may look similar to SMA. Small recording electrodes (needles) are inserted into the individual’s muscles, usually the arms and thighs, while an electrical pattern is observed and recorded. A muscle biopsy is a procedure in which a small sample of muscle is obtained in an operating room for further examination in a laboratory, again to help distinguish SMA from other muscle diseases. Results from an EMG or a muscle biopsy may be useful in instances when the diagnosis of SMA remains uncertain.

A CK level is measured through a blood sample and an elevated level indicates muscle disease. While this is not usually found in SMA, it may be helpful to distinguish SMA from other forms of muscle disease.
Individuals have two genes called survival motor neuron (SMN) 1 and 2. Researchers have identified the SMN1 gene as the primary producer of the SMN protein. In approximately 95% of patients with SMA, the SMN1 gene is missing, and in some cases, the SMN1 gene may be present but is altered, abnormal, or somehow damaged. The SMN2 gene is similar to SMN1, but does not produce as much SMN protein, or the right kind of protein, as the SMN1 gene. The presence of SMN2 is necessary for survival (as individuals cannot live without either SMN1 or SMN2) and is sometimes referred to as the “rescue gene.”

Genetic testing looks at genetic material in a patient’s blood sample to determine if the SMN1 gene is missing or damaged, thereby confirming the diagnosis of SMA. Currently, more specific genetic testing can also be done to identify the number of copies of the SMN2 gene present, although this is not done routinely.

This figure illustrates the three types of SMN1 mutations: deletions, gene conversion of SMN1 to SMN2, and single nucleotide point mutations. (a) Xs indicate a deletion. A deletion removes part or all of the SMN1 gene. (b) In the case of gene conversion, the SMN1 gene has been converted to an SMN2-like gene (indicated by the nucleotide change to T). These two types of mutations (deletions and gene conversion events) are the most frequent types found in SMN1. About 95% of 5q-SMA patients have these two types of mutation, and these mutations are easily detected by the current diagnostic test for SMA as they both result in the loss of SMN1 exon 7. (c) Point mutations can also be found in the SMN1 gene, but at a much lower frequency than the other two types of mutations. Shown here are the locations of point mutations that have been found in the SMN1 gene. They are labeled A through T. About 5% of 5q-SMA patients have a deletion or gene conversion mutation on one chromosome and a point mutation on the other chromosome. An individual with this combination of mutations (point mutation with either a deletion or conversion mutation) will not be diagnosed as having SMA using the SMA diagnostic test as only one copy of the SMN1 gene is gone. Rather, this person will look like a carrier using the quantitative carrier test, even though they are symptomatic for SMA.
Samples must be sent out to a specific diagnostic genetic laboratory for this kind of testing, if the neurologist thinks it is necessary.

The numbers of copies of SMN2 an individual has is somewhat related to how severely that individual will be affected by SMA. Because the SMN1 gene is missing, the greater the number of SMN2 copies present, the more SMN protein is produced and the greater likelihood that more motor neurons will remain healthy and productive. Individuals with only 1 or 2 copies of the SMN2 gene will typically have the most severe expressions of the disease, whereas three or more copies of the SMN2 gene will typically mean a less severe expression. It must be noted, however, that the number of copies of SMN2 does not reliably predict what type of SMA an individual will have or how weak their muscles will become. Therefore, even after genetic testing results are reviewed by the SMA team, it is nearly impossible for any clinician to predict exactly how SMA will affect a given individual.

Please see the booklet entitled The Genetics of SMA for more information on genetics.
Each type of SMA varies among individual patients. This is crucial to remember when considering different aspects of an individual’s care. No two children will be exactly the same and thus treatment and care plans for each family should be tailored to meet their individual needs.

It is also important to remember that the brains of children with SMA are not affected at all and therefore their cognitive abilities remain normal. Individuals with SMA are usually very intelligent and children should be encouraged to participate in as many age- and developmentally-appropriate activities as possible, with adaptations made whenever necessary. It is essential that children with SMA are assisted in reaching their utmost potential in school, at home, and in their communities.

Ideally a team of clinicians that specializes in SMA and its specific complications should follow your child. Several hospitals throughout the country have designated clinics staffed with interdisciplinary team members who have expertise in caring for individuals with SMA and their families. Clinicians from different specialties (e.g., neurology and pulmonology) may gather at one time and see individuals and families together. In some healthcare centers, care may be provided by clinicians in different settings at different times. Either way, it is important that individuals with SMA and their loved ones feel comfortable with the care they are receiving. They should feel free to ask questions and receive honest answers from a team that they trust who can guide them in making decisions that are best for their children and families.

Interdisciplinary teams caring for people with SMA are often comprised of neurologists, nurses/nurse practitioners, pulmonologists/intensivists, orthopedics, genetic counselors, physical and/or occupational therapists, nutritionists, and social workers. Individuals with SMA and their family members work together with these interdisciplinary teams to create goals and personalized plans of care that best meet their needs as they change throughout the journey with SMA.
Having difficult conversations and making well-supported decisions before an emergency occurs will help families to be better prepared and to avoid making rash decisions at a stressful time. The World Health Organization (WHO) defines palliative care as “improving quality of life of patients facing life-threatening illnesses, and their families, through the prevention and relief of suffering by early identification and treatment of pain and other problems, whether physical, psychological, social or spiritual.” Palliative care prepares families for these situations and should be provided along with whatever treatment options families choose. The palliative care team can help families develop specific goals that are in alignment with their values and what works best overall for their child and family. Some families may choose invasive measures or non-invasive measures of support, as described throughout this booklet, and some may choose comfort measures alone. Some families may opt for more or fewer options than other families, but all families make choices based on love. Every child and family with SMA can benefit from, and should receive, palliative care throughout the course of their illness, independent of the choices they make.

Parents and individuals with SMA have rights! No one should travel this journey alone without support. Most SMA or other interdisciplinary healthcare teams can provide you with the services necessary to assist you in meeting all of your needs. Don’t be afraid to say NO if something doesn’t seem right to you. Don’t be intimidated or afraid to ask questions. If you forget to ask something, or feel overwhelmed, call your healthcare provider or contact Cure SMA for suggestions.

Please see the booklet entitled Caring Choices for Families of Infants Newly Diagnosed with SMA type I for more information on individual decision making.
Most children with SMA type I are diagnosed in infancy and much can be done to assist in their cognitive, physical, and emotional health from an early age. Members of the team caring for children with SMA type I will discuss with families the options for therapy, as well as equipment specific to what each child needs.

**THERAPEUTIC RESOURCES**

A social worker or other team member can help families work with health insurance companies to ensure that children receive the services they need. Young children are usually eligible for different types of therapies (i.e., physical, occupational, speech), often through Early Intervention or programs administered at the local community level.

**PHYSICAL AND OCCUPATIONAL THERAPY**

Instructions for range of motion exercises and other ideas for physical/occupational therapy initiated by licensed therapists are important no matter how young the child. Using balloons and feathers as toys makes for wonderful stimulation and offers children a feeling of independence and accomplishment. Games that encourage the act of reaching are another form of physical or occupational therapy that can be very helpful. A physical/occupational therapist can also suggest ideal seating systems and/or leg braces that will promote the greatest comfort and maximum mobility for children. Because the care of children with SMA differs from the care of many other children receiving physical and/or occupational therapy, it may be helpful for a child's community-based physical or occupational therapist to be in contact with the therapists from the SMA team for specific instruction and/or suggestions.

Aqua therapy can be very helpful for children with SMA, as the buoyancy of the water allows movement of the arms and legs that otherwise may not be possible due to muscle weakness. Be sure that the water temperature is at least 90°F and that the child is positioned so that his or her head does not go into or under the water, in an effort to avoid episodes of aspiration (getting fluid into his/her lungs).
NUTRITIONAL AND FEEDING NEEDS

Infants and young children with SMA type I must get adequate nutrition for proper brain growth and development. Children’s dietary needs should be discussed with a nutritionist knowledgeable about SMA who can recommend adjustments to feeds (e.g., formula, breast milk, cereal) to meet specific caloric needs (some formulas are better than others for children with SMA). At different times in the disease course, and sometimes in the context of a respiratory or other illness, young children may experience difficulty feeding and require a change in feeding or supplemental nutrition.

Children with SMA type I usually lose their ability to chew and swallow food and water safely at different ages, depending on the severity of their disease. They may also have trouble managing their own secretions (saliva). If they continue to take nutrition and hydration orally, they are at high risk for aspirating (inhaling) food and water and developing respiratory problems if food and water reach the lungs (aspiration pneumonia). To prevent these complications, tube feeding is commonly offered to ensure that infants and children receive enough calories to grow and adequate fluid to remain hydrated.

There are a few options for tube feedings, including the following:

- **Nasogastric (NG) tube**: a small flexible tube is inserted through the nose down to the stomach [nasoduodenal (ND) or nasojejunal (NJ) tubes are placed farther, into two different areas of the small intestine beyond the stomach]
- **Gastrostomy (G) tube**: a small tube is placed surgically directly into the stomach from outside
• **Gastrostomy-jejunostomy tube (G-J) tube:** a small tube that is placed surgically, directly into the stomach and the small intestine (jejunum). Some children may also develop significant gastroesophageal reflux (a condition where food moves from the stomach back up toward the throat), and if this occurs, medication may be used to treat the symptoms, and the following procedure may be recommended to help treat both the inability to eat safely by mouth as well as the reflux:

• **G-tube and Nissen fundoplication (stomach wrap)**

While both options are invasive, they each have various advantages and disadvantages. NG tubes (or ND or NJ tubes) can be easily placed and easily removed, if necessary. They occasionally come out on their own and need to be replaced. G-tube or G-J tube placement is permanent and easy to maintain, but this does require surgery and anesthesia. The option that is best for a child is determined through individual discussions between each family and their SMA care team.

Please see the booklet entitled *Nutrition Basics* for more information on the nutritional needs of individuals with SMA.

**RESPIRATORY NEEDS**

Lung problems pose some of the greatest challenges to young children with SMA, and are variable and hard to predict. Infants can develop difficulty coughing (weak cough), which leads to an impaired ability to clear their upper and lower airway secretions. As they get older, they will develop difficulty breathing during sleep, and ultimately while awake. Due to poor development of the chest wall and lungs, young children may have trouble handling the normal colds of childhood. If children cannot clear secretions well, this can lead to pneumonia and respiratory failure. These infections can make already weak muscles around the lungs even weaker. Children with SMA type I do not develop more respiratory illnesses than healthy children of the same age, but any illnesses they do contract are more dangerous because of muscle and lung weakness.

Respiratory care and support helps with lung function and breathing. A pulmonary/respiratory specialist will work with families to develop respiratory goals and supportive measures that are specific to each child’s needs.

It is very important that children with SMA, family members, and anyone frequently around them receive immunizations to help prevent contagious diseases. Respiratory infections such as influenza, pneumonia, and whooping cough (pertussis) could have devastating consequences for a child with SMA. Fortunately, these are preventable with proper immunizations.
WHAT RESPIRATORY EQUIPMENT WILL CHILDREN NEED?

Individuals with SMA can benefit from the use of a In-Exsufflator Cough Machine (often called Cough Assist). The In-Exsufflator Cough Machine works by applying positive pressure into the airways to fill the lungs with air, and then rapidly shifts to negative pressure to pull the air out of the lungs. The rapid change in pressure from positive to negative produces a high flow from the lungs on exhalation that is similar to a cough. This technique, referred to as “mechanical insufflation-exsufflation,” avoids damage to the airways while also clearing the lungs of secretions (thick saliva and mucus). The device offers children greater comfort and quality of life without the use of invasive procedures and equipment.

Respiratory distress can be monitored by measuring the level of oxygen saturation in the blood using a tool called a pulse oximeter. A sensor is placed on the child’s finger or toe with a small clip or tape with a red light to monitor the oxygen saturation.

Children with SMA type I usually require breathing support while sleeping. During colds or illness, all infants/children will require additional breathing support. Various types of respiratory care approaches and equipment can be life-saving during an illness. There are several options to consider.
**WHAT IS NON-INVASIVE RESPIRATORY CARE?**

Non-invasive describes a care approach or intervention that does not cut into or penetrate the body. Breathing support is placed on or outside the body.

As mentioned above, almost all children with SMA type I need support to breathe and can benefit from a Bi-level Positive Airway Pressure (BiPAP) machine or mechanical ventilator while they sleep, starting at an early age.

BiPAP uses a mask over the nose to provide a higher volume of air into the lungs during inhalation, filling the lungs with more air than a child can breathe in on his/her own. The SMA team will explain in more detail about BiPAP if/when a child requires this form of breathing support.

Mechanical ventilators, or respirators, are more complex than BiPAP machines, but also allow for control of more variables (e.g., the specific size/volume of a breath, or the number of breaths per minute). They come in a variety of models. The nasal mask provides non-invasive respiratory care, whereas the others are more invasive forms of support (see below). The nasal mask does not work well for every child and family, for many reasons, and may become difficult to tolerate. If this is the case, or if a child needs breathing support 24 hours per day, the family should talk with their SMA team about other options for the child’s respiratory care.

**WHAT IS INVASIVE RESPIRATORY CARE?**

Invasive means that something penetrates into the body as part of the procedure. Needles, tubes inserted into the body, and surgery are all examples of invasive care.

Invasive respiratory care requires the use of mechanical ventilation, as described above. Mechanical ventilation can be delivered through a nasal mask, a mouthpiece while awake, a tube inserted into the throat, or through a tracheostomy tube, a surgical incision in the throat that allows for placement of a more permanent breathing tube (see left).

Initially, invasive respiratory care uses a breathing tube [endotracheal tube (ETT)], which is inserted through the mouth and throat, down into the upper trachea. The tube is then connected outside to a mechanical ventilator. Long-term use of endotracheal tubes, however, can cause damage by irritating the mouth and throat if not taken out within a few weeks. When ventilator support is required for extended periods of time, it becomes important for the family and SMA team to have discussions about what the next steps or options might be, and what is best for each child and his/her family.
One option to consider is the placement of a tracheostomy tube. A tracheotomy is a surgical procedure in which a hole is made in the front part of the neck into the large airway (trachea or windpipe), through which a tube can be inserted to help with breathing. A tracheostomy tube bypasses the mouth and vocal cords and goes directly from outside the skin of the neck into the trachea. A respirator or ventilator is connected to the tracheostomy tube. The word tracheostomy is used to describe the creation of the actual opening (stoma) in the neck, but tracheotomy and tracheostomy are interchangeable in this context.

It is important for families to understand their rights when it comes to making life-prolonging decisions for their children, and to understand that these are very personal decisions in which there is no “right” answer. Members of a hospital’s pediatric palliative care team can assist a family in making the decision that is best for them. Once a decision has been reached, it is important to make certain that all necessary medical personnel (and any other individuals with whom a family wants to share this decision) are fully aware of their wishes. It is also important to know that families may change their mind regarding these choices at different points throughout the journey with SMA, and they should always feel free to talk with their SMA care team at any time.

Please see the booklet entitled Breathing Basics for more information on the respiratory needs of individuals with SMA.
SMA Type II (and Some Type III) – What You Should Know

In all types of SMA, there are many things that can be done to assist with the cognitive, physical, and emotional health of individuals living with the disease. Members of the SMA team will discuss with families the options for therapy and equipment that are specific to what each child needs.

**THERAPEUTIC RESOURCES**

Children with SMA type II can benefit from physical and occupational therapy as well as aquatherapy, and in some cases hippotherapy (therapeutic horse-riding). Therapy is usually provided first through Early Intervention, and then through the school system, depending on the child’s age. Many children also receive additional therapy after school or at home. It can be helpful for parents or other caretakers to provide stretching and range-of-motion (ROM) exercises at home. Your physical/occupational therapist can suggest ideal seating systems and/or leg braces that will be most helpful to promote the comfort and maximum mobility of your child. Because the care of children with SMA differs from the care of many other children receiving physical and/or occupational therapy, it may be helpful for a child’s community-based physical or occupational therapist to be in contact with the therapists from the SMA care team for specific instruction and/or suggestions.

Children with SMA type II may be able to sit unsupported but may first need assistance to get into a seated position, as previously mentioned. With assistance and bracing they may be able to stand, but are usually unable to walk.

It is important for SMA-affected children to be in an upright position at the earliest possible age. Standing is important in development as it allows for better respiratory function, improved bowel function, and greater mobility. Getting children into an upright position as much as possible, or tolerated, throughout the day is extremely beneficial. This may require advocacy on the part of the parents to encourage the use of standing aids in the classroom as well as at home.
There are several options to consider when choosing the appropriate standing aid:

- A standing frame and/or parapodium.
- A standing wheelchair is ideal for added mobility and independence. A child as young as 13 months can use this type of device.
- Bracing: Reciprocating Gait Orthosis (RGO’s) and weight bearing Knee Ankle Foot Orthosis (KAFO’s) usually work well for children with SMA type II, and in some instances, 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 the SMA team’s physical/occupational therapist.

The use of a lightweight manual wheelchair can be an exciting option for a child with SMA type II. It can provide mobility, independence, and a taste of adventure, while allowing them to use some of their own upper body strength. However, it should be understood that for true independence and mobility, and in cases when arm muscles are weak, a power wheelchair will be necessary. There are several different types of power wheelchairs available, and currently children with SMA are beginning to use them at two years of age. The physical therapist from the SMA care team can help families choose the wheelchair that is right for their child.

Weakness is usually greater in the legs than in the arms in SMA type II; however some children may benefit from wrist or hand splints as well. This will be recommended by the therapist on the SMA care team when the need arises.

Scoliosis (curvature of the spine) occurs at some point in virtually all children with SMA types I and II, and some with SMA 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 treatment measures should be implemented early. Options for managing scoliosis include custom seating systems, seating aids, and a body jacket. Later, spinal surgery may need to be considered.
NUTRITIONAL AND EATING NEEDS

The nutritional needs of a growing child are very important and proper dietary planning is essential in SMA type II. Children with SMA type II usually do not have swallowing problems, but this varies from child to child. Some children may have difficulty eating enough food by mouth to maintain their weight and growth, and a feeding tube may become necessary if the family decides this is best. Some children with type II may also become overweight, and excessive weight can make mobility more difficult. It is essential to discuss each child’s specific dietary needs with the dietician/nutritionist from the SMA team who is knowledgeable regarding the specific nutritional needs of children with SMA type II.

Please see the booklet entitled Nutrition Basics for more information on the nutritional needs of individuals with SMA.

RESPIRATORY NEEDS

Children and adolescents with SMA type II may develop weakness of the muscles used for breathing and experience weakened coughing. Some individuals may require equipment to help them breathe easier such as a CoughAssist™ device or a BiPAP machine for nighttime use, especially during times of illness. In some cases, respiratory function may decrease further, making more invasive respiratory supports a consideration.

Please see the booklet entitled Breathing Basics for more information on the respiratory needs of individuals with SMA.
SMA Type III—What You Should Know

THERAPEUTIC RESOURCES

Individuals with SMA type III can benefit from physical and occupational therapy and a specific plan should be developed by the physical/occupational therapist from the SMA care team that is tailored to meet each individual’s specific needs, which may vary greatly.

Individuals with SMA type III have the ability to stand and walk independently. Fine shaking of the fingers and hands (tremors) can be seen, and symptoms of joint aches and overuse frequently occur. Difficulty walking, running, and climbing stairs may develop as individuals get older; some will lose the ability to walk independently in childhood, while others may remain ambulatory (able to walk) until adolescence or adulthood. As mentioned previously, the use of a walker and bracing may become necessary and recommendations for appropriate equipment can be made by the therapist on the SMA team. The use of a lightweight manual wheelchair may be considered for distance, as well as an electric scooter or other motorized chair, which will also be advised by the SMA team as needed.

Problems with the spine (scoliosis) may develop at various rates and ages, and bracing and surgery may also be considered as options when appropriate.

Please see the thorough explanation of therapies and equipment described in the SMA type II - Therapeutic Resources section above for a review of mobility-assist devices.

NUTRITIONAL AND EATING NEEDS

Swallowing difficulty may occur in SMA type III, but less commonly and later in the disease course than in SMA type II. Children and adults with type III are at risk of becoming overweight as they age and are less able to be physically active. Dietary modifications should be made as necessary by the dietician/nutritionist on the SMA care team.

Please see the booklet entitled Nutrition Basics for more information on the nutritional needs of individuals with SMA.
RESPIRATORY NEEDS

Minor weakness of cough, along with breathing difficulty at night, may occur in some cases of SMA type III. Respiratory interventions as described in previous sections above (i.e. CoughAssist™ and/or BiPAP) may be recommended as needed.

Please see the booklet entitled **Breathing Basics** for more information on the respiratory needs of individuals with SMA.

Type IV: Adult Onset

As adults, individuals with SMA type IV are likely aware of their personal weaknesses and limitations. They should work together with members of the SMA care team, such as physical and occupational therapists and nutritionists, to design the best possible exercise and diet program to maintain health and well-being. Therapists can also assist individuals in obtaining the equipment needed to get around and accomplish activities of daily living at home. Individuals should only do as much as they feel comfortable doing, and should seek out the necessary adaptations at home, school, and/or work, as well as in their vehicles. Individuals may also consider getting assistance with specific activities if needed (e.g. cooking, showering, dressing) as certain muscles grow weaker over time. A service pet may also be helpful at some point in the journey with SMA type IV. As with types I, II, and III, diet and nutrition are important factors in maintaining overall health, and a nutritionist can help develop individualized dietary plans.
Cognitive/Academic Needs

It is important to once again note that SMA does not affect the brain or its development, and thus it does not affect an individual's ability to learn and succeed academically. From ages one to three, children will usually receive services through Early Intervention (EI) and will then have an Individualized Education Plan (IEP) or a 504 plan (named for Section 504 of the Rehabilitation Act and the Americans with Disabilities Act) that details the specific services and/or modifications and accommodations they will need in school after age three. Individuals with SMA usually receive physical and occupational therapy at school, as described above, and may be assigned a paraprofessional (individual teaching aide) or a one-to-one classroom aide to assist them in meeting their physical needs (e.g. getting around, lifting or moving objects in the classroom, using the bathroom). It is important to advocate for students with SMA to be placed in appropriate academic settings to optimize their intellectual growth. Their physical limitations should not limit them from receiving academic stimulation in a developmentally-appropriate environment. Classroom modifications may be necessary in some instances to accommodate their physical needs.
I Am a Carrier of the SMA Gene... What Can I Do?

This information may have been requested by someone identified as a carrier of SMA through a blood test. Cure SMA recommends that carriers of the SMA gene seek the advice of a genetic counselor, especially if they plan to have children, as s/he can help people to better understand the chances of, and risks associated with, having children with SMA. The genetic counselor will take a complete family medical history, and obtain information regarding any diseases, deaths, stillbirths and miscarriages experienced by each family member. A genetic counselor can also discuss options regarding prenatal testing, and can provide information to guide individuals in making the choices that are best for them. If someone has already given birth to a child with SMA, the counselor can discuss options to consider regarding any future pregnancies.

WHEN TWO CARRIERS HAVE A CHILD...

THERE ARE THREE POSSIBILITIES

- 25% CHANCE UNAFFECTED
- 50% CHANCE CARRIER
- 25% CHANCE SMA

1 IN 50 PEOPLE CARRY THE GENE

Please see the booklet entitled *The Genetics of SMA* for more information on genetics.
What are the key areas of SMA research?

**BASIC SCIENCE RESEARCH**
Basic science research looks at the fundamental building blocks of life, including molecules, proteins, cells and genes. Often referred to as “lab” or “bench” research, basic science research is carried out in a laboratory by researchers using microscopes and petri dishes. Other types of research, like translational research or clinical research, are based on the findings and clues offered by basic science research.

Basic science research plays a critical role in the discovery and testing of chemical or biological materials that have the potential to become SMA drugs and therapies; and the identification of existing drugs with potential in SMA. Testing at this stage is conducted on proteins, cells, and in living animals, but not in humans.

Critical questions in SMA biology, including what is going wrong in the body to cause SMA, are answered by basic science research. It gives researchers many seed ideas or clues that lead to more advanced research.

**DRUG RESEARCH—TRANSLATIONAL RESEARCH**
Drug research tests chemical and biological materials to see if they can be turned into drugs and therapies.

The National Institutes of Health (NIH) defines translational research as... “the process of applying discoveries generated during research in the laboratory, and in preclinical studies, to the development of trials and studies in humans.” This early phase is called the drug discovery phase. Researchers also work on drugs that are already approved for use in different diseases and conditions to see if they can help people with SMA.

Translational research means taking something that we learn in one area of research and applying it to another area of research; as in applying the clues from basic research to the later stages of SMA drug making. Researchers are translating basic research discoveries from the laboratory into new drugs and therapies that can be tested in clinical trials with human subjects.

**CLINICAL RESEARCH**
Clinical research is patient-oriented research, conducted with human subjects or human tissues. The investigator (the researcher) directly interacts with his subjects during the research project. Patient-oriented research can study a) the mechanisms (methods and parts of how something works) of human disease, b) therapeutic (used to treat disease) interventions, c) clinical trials, and d) the development of new technologies. (source: NIH)

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1 From the NIH Roadmap: http://nihroadmap.nih.gov/clinicalresearch/overview-translational.asp
How Cure SMA Can Help

If you are a family recently diagnosed with SMA, please e-mail infopack@curesma.org or call the Cure SMA National Office at 800.886.1762. We are available to support you and will overnight a comprehensive newly diagnosed informational packet on SMA to you. We will also send a Newly Diagnosed Care Package of toys for your child and a binder full of resources that is specific to your state. These Newly Diagnosed Care Packages are made possible through donations and ideas from other SMA parents.

Cure SMA is a non-profit organization and the largest worldwide network of families, clinicians, and research scientists working together to advance SMA research, support families, and educate the public and professional communities about SMA.

Cure SMA is a resource for unbiased support. We are here to help all individuals living with SMA and their loved ones, and do not advocate any specific choices or decisions. Parents and families make different choices regarding what is best for their families and children, consistent with their personal beliefs. Parents and other important family members should be able to discuss their feelings about these topics, and to ask questions of their SMA care team. Such decisions should not be made lightly and all options should be considered and weighed carefully. All choices related to SMA are highly personal and should reflect personal values, as well as what is best for each child, individual, and family. Families are never alone. Cure SMA is always just a phone call away.

As caring parents and professionals, we can offer support and understanding. Through phone and multimedia networking, the team at Cure SMA is here to support you, along with other families and friends affected by SMA. We provide invaluable support and services to individuals and families such as: medical information and equipment, support and resources, an annual conference, local community support through our vast volunteer network, regular publications, and web-based support.
We send SMA type I, type II, and type III care packages to all newly diagnosed families who contact Cure SMA. These care packages include a range of items such as informational materials, toys appropriate for individuals with each type of SMA, books, blankets, hand-made quilts, and DVD’s. Many ideas for the items in these Care Packages come from SMA parents themselves.

The Newly Diagnosed Binder is a program that was launched as a way to help families with information and resources specific to each state. These binders contain important information about SMA, including all of the SMA Care Series Booklets and a list of resources available to the family in their own state. This binder also acts as an organizational tool, where families can keep important medical information concerning their child, all in one location. Every family receives a binder in their Newly Diagnosed Care Package.

Cure SMA receives generous funding from donors to provide wagons for our Newly Diagnosed Care Package Program. We are thrilled to be able to provide Wagons to all newly diagnosed families once they contact Cure SMA. These wagons are beneficial for all types of SMA. For infants with type I, the wagons are wonderful, as these children often are required to lay flat once they lose muscle tone and cannot adequately support themselves. For families affected by type II and type III, these wagons are ideal for parents to be able to transport their children without the use of a stroller or wheelchair.

We also provide our members with a subscription to our Family Support Newsletter called Directions, and our publication dedicated to SMA research news and progress, called Compass.
Directions contains information on daily living, letters and stories from families, upcoming events, articles by doctors and researchers, and other resources.

Because Cure SMA understands the financial hardship of living with SMA, the organization also maintains an equipment pool, which is available free of charge to members of Cure SMA. Cure SMA has a widespread list of equipment suitable for SMA children in our Equipment Pool. Once the family is no longer in need of the item, we kindly ask them to return it so that we are able to loan it to another family. Some of the items in our Equipment Pool have been donated by other SMA families. Other items have been purchased with funding that Cure SMA receives specifically to purchase certain items for the Equipment Pool. Some of the items in the Cure SMA Equipment Pool include:

- Car beds (Infant Cosco Car Bed and the Hope Car Bed)
- EZ-on vests
- Bath chairs
- Medical strollers
- Manual wheelchairs
- Standers
- Therapy and positioning devices
THE ANNUAL SMA CONFERENCE

Every year, Cure SMA sponsors a conference to bring together the leading SMA researchers, clinicians, and families living with SMA. Cure SMA has been hosting the Annual SMA Conference since 1989. The weekend is filled with a wide variety of workshops, a memorable children’s program, a family fun fest and carnival, many opportunities to connect and interact with families and receive first hand updates from the researchers. Every year, we look forward to reuniting as a community at this conference and showing our support for others. As always, the family and the research conferences run alongside each other. This is the largest conference in the world for those affected by SMA, and also for those involved in providing support and care for SMA patients. There is no other program like it for those affected by SMA. The interactions between the researchers and families at this one conference are extremely special. The annual conference also provides these children an opportunity to make new friends and have a great time.

The Goals of the SMA Conference:
1) To allow networking between researchers, medical providers, patients and families
2) To educate researchers on the latest research advancements
3) To attract the best researchers to the SMA field and encourage collaborations
4) To educate patients and families on the basics of SMA
5) To update patients and families on the latest research and medical progress
CONTINUING MEDICAL EDUCATION CONFERENCE

Cure SMA has introduced a new component for medical professionals at the Annual SMA Conference. A conference is now held prior to the start of the Annual SMA conference and researcher conference. The CME conference, titled “Interdisciplinary Perspectives on Spinal Muscular Atrophy: Defining Your Role”, for medical professionals, is the first of its kind for Cure SMA. The conferences have previously covered two components of SMA: research and support. The research aspect covered scientific, research & clinical updates, while the support aspect included the family support workshops and family networking. The addition of this conference provides an exciting new third component of care to the Cure SMA Conferences by educating medical providers on SMA. Cure SMA partners with an accredited hospital who can provide continuing medical education credits for medical professionals, and offers a full day of lectures and educational materials to all attendees.

NEWLY DIAGNOSED CONFERENCE PROGRAM

Thanks to the generous funding provided by the Erin Trainor Memorial Fund, Cure SMA covers the registration fees for the Annual SMA Conference for all families newly diagnosed since the last conference. The mission of the Erin Trainor Memorial Fund (ETMF) is to generate substantial funds to be able to provide conference scholarships, allowing newly diagnosed SMA families/individuals the opportunity to attend the Annual SMA Conference. Our goal is to allow as many newly diagnosed SMA families as possible to attend and experience the benefits of this amazing conference. The meeting gives families the opportunity to gather critical care and daily living information early after diagnosis, learn directly from experienced SMA physicians, and network with other families. This program is automatically offered to all newly diagnosed families that contact Cure SMA.

CONFERENCE SCHOLARSHIP PROGRAM

Financial need scholarships are also available to assist SMA families in attending the conference, who do not qualify for the Newly Diagnosed Program. This program allows families who may not be able to financially afford attending the conference, the opportunity to apply to this program and receive financial aid, in the hopes that by waiving their registration fees, they may be able to attend the conference.
CURE SMA WEBSITE – WWW.CURESMA.ORG

The Cure SMA website provides vital up-to-date information and advice that families need on a variety of SMA-related issues. The website is here to help guide families through a wide variety of general information on SMA. This section is a wonderful resource for families. Some of the information available includes:

- Coping with the SMA diagnosis
- Advice and information on the basics of SMA, where we have links to our SMA Care Series Booklets in PDF format, quick facts on SMA and Frequently Asked Questions
- Education for SMA-affected individuals
- Cure SMA Publications
- Information on SMA medical care including respiratory care, physical and occupational therapy, and nutrition
- Adapting toys for SMA children
- Information on the Annual SMA Conference
- Links to Cure SMA chapter pages
- Resources for grief and loss
- Legislative information
- Links of information for adults with SMA

Cure SMA offers free informational booklets called SMA Care Series to all families, friends and professionals. These booklets are available on a variety of specific topics such as Genetics and Diagnosis, Respiratory Care, Nutrition and many more. SMA Care Series Booklets are also available in Spanish, as well as on our website for download.
Cure SMA will provide a Primary Care Provider Packet to any primary care provider who contacts Cure SMA or through a family submission. These packets are specifically designed for primary care providers who may not be familiar with Spinal Muscular Atrophy. We hope to encourage and spread SMA awareness throughout the medical community by sending this important information to providers.

The Cure SMA Care Provider Mailing List is a database containing our professional members. The database includes: pediatricians, neurologists, pulmonologists, PTs, OTs and many other medical professionals. We keep these providers up to date on the latest SMA information available, by mailing them all new and updated SMA Care Series Booklets. Our Care Provider Mailing List currently includes over 1,000 providers from all over the US. If you would like any of your family’s care providers added to this mailing list, please email info@curesma.org or call 800.886.1762.

The Cure SMA Facebook page is the best place to receive constant updates on anything and everything SMA related. It is also a wonderful place to network with many SMA families worldwide.

We are always available if you want to talk with others who understand. Cure SMA has numerous chapters throughout the US, as well as networking programs that link families together for mutual support. Please contact us at any time!

Please see the booklet entitled *Cure SMA Family Support and Patient Services* for more information on the ways in which Cure SMA offers support.
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About Cure SMA

Cure SMA is dedicated to the treatment and cure of spinal muscular atrophy (SMA)—a disease that takes away a person’s ability to walk, eat, or breathe. It is the number one genetic cause of death for infants.

Since 1984, we’ve directed and invested in comprehensive research that has shaped the scientific community’s understanding of SMA. We are currently on the verge of breakthroughs in treatment that will strengthen our children’s bodies, extend life, and lead to a cure.

We have deep expertise in every aspect of SMA—from the day-to-day realities to the nuances of care options—and until we have a cure, we’ll do everything we can to support children and families affected by the disease.

Learn more about how you can help us reach a treatment and cure at www.cureSMA.org.

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We’re on the Web!
www.cureSMA.org
Make a Difference and Get Involved

Cure SMA is about our community coming together to support each other. Our research and support programs are funded through the grassroots fundraising efforts of our SMA families.

Cure SMA is the leader in SMA research. We have funded over $57 million dollars so far directly for SMA research. Our successful results and progress from basic research to drug discovery programs to clinical trials provide real hope for families and patients. Over the last 30 years, Cure SMA has:

- Discovered a back-up gene, SMN2, for the disease
- Funded five multi-center clinical trials for existing drugs
- Funded research that led to the first ever clinical trial for a new drug designed specifically to treat SMA

Our research funding model is based on relying on independent and neutral research and medical experts to evaluate and recommend which specific projects are the best to fund. A key element here is the balance of different approaches using drugs and cells and genes. Also important is the track record of success at Cure SMA in getting all this research to move forward. We have world-class advisory groups, and also have experts on staff to guide and direct these research and clinical programs forward and make sure that your funds are used as effectively as possible.

Cure SMA has been involved in funding two-thirds of the ongoing novel drug programs for SMA.

We have been successful in implementing our model of early diversified investments followed by licensing programs to industrial partners for clinical development.

In recent years, Cure SMA has been successful in expanding basic research discoveries from the bench to the clinic. These innovative programs are enhancing our diverse research portfolio and advancing the progress being made toward growing the SMA drug development pipeline. Specific achievements include:

- Fifteen novel SMA therapeutic programs currently being conducted, up from two just ten years ago
- Over a dozen companies now investing in the SMA drug pipeline representing an increase in funding and resources from industry
- Five projects currently in varying stages of human clinical trials
While we celebrate these milestones, we direct our focus to funding more viable research projects in order to maximize the opportunity for success in finding a treatment and ultimately a cure for SMA.

**Cure SMA provides services to more than 70% of all families affected by SMA in the United States.**

Cure SMA is a balanced and broad organization and we are involved in great programs in many areas. In addition to research, Cure SMA funds critical family support and patient care programs. Cure SMA remains devoted to supporting the SMA community by providing information and care packages to all newly diagnosed families. We also offer an ongoing series of booklets on SMA Care, which are available to all SMA families and professionals in both English and Spanish. Cure SMA has a collection of equipment available free of charge suitable for SMA children. Cure SMA has been hosting the Annual SMA Conference for 25 years, which is the largest conference in the world for those affected by SMA. We are proud to offer an amazing support program for all newly diagnosed SMA families to attend our Annual SMA Conference through our Newly Diagnosed Scholarship Program. This program allows newly diagnosed families to attend the Annual Conference with all registration fees waived.

**It truly is because of all our families that we are able to fund all of these projects and make progress.** If you are interested in organizing a fundraiser to support these programs please contact the development department at 800.886.1762 or fundraising@curesma.org.

Cure SMA has support available and many tools and materials to help make your event a success. These tools consist of fundraising manuals, numerous templates for sponsorship packets, flyers, press releases plus all of our wonderful signage and promotional items. We also can provide your event with its own website allowing for easy online registration and fundraising. We support all types of events ranging from the Cure SMA signature Walk-n-Roll, to golf outings and fun comedy nights.

**Make a difference and join in.** Your efforts will help bring us closer to a treatment and make a profound difference in your local community and to the families everywhere affected by spinal muscular atrophy.