Spinal Muscular Atrophy

What Is Spinal Muscular Atrophy?
Spinal Muscular Atrophy (SMA) refers to a group of diseases which affect the motor neurons of the spinal cord and brain stem. These cells are responsible for supplying electrical and chemical messages to muscle cells. Without the proper input from the motor neurons, muscle cells cannot function properly. The muscle cells atrophy and produce symptoms of muscle weakness. There are dozens of diseases which affect the motor neuron. SMA affects the voluntary muscles that are used for activities such as crawling, walking, head and neck control, and swallowing. Although SMA affects muscles throughout the body, the proximal muscles are often most severely affected. Weakness in the legs is generally greater than in the arms. Involvement of respiratory muscles can lead to an increased tendency for pneumonia and other lung issues. 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.

What Causes Spinal Muscular Atrophy?
SMA is an autosomal recessive genetic disease and is a relatively common “rare disorder” affecting approximately 1 in 6,000 births. It is considered to be one of the most prevalent genetic disorders and can strike anyone of any age, race or gender. In order to be affected both parents must be carriers and both must pass this gene on to their child. The likelihood of a child inheriting the disorder is 25%. Approximately about 1 in 40 people are genetic carriers.

The common forms of SMA are the result in a change in a gene located on chromosome #5 which creates a protein deficiency and then is responsible for the degeneration or premature death of the anterior horn cells. This protein deficiency has its most severe effect on motor neurons which send out nerve fibers to muscles throughout the body. As the child grows, their body is doubly stressed, first by the decrease in motor neurons and then by the increased demands on the nerve and muscle cells as they physically grow larger. The resulting muscle atrophy causes weakness and bone/spinal deformities that may lead to further loss of function, as well as additional compromise to the respiratory system.

Types of Spinal Muscular Atrophy
There are four types of SMA: Types I, II, III, and IV. The determination of the type is based upon the physical milestones the individual achieves. It is important to note that the course of the disease may be different for each individual.
**SMA Type I**

Type I is also called Werdnig-Hoffmann Disease. The diagnosis is usually made before 6 months of age and in the majority of cases it is made before 3 months of age. Some mothers even note decreased movement in the final months of pregnancy. Type I is the most severe form of SMA. Usually a child with Type I is never able to lift their head or accomplish the normal motor skills expected early in infancy. They generally have poor head control, and may not kick their legs vigorously, bear weight on their legs or be able to sit up unsupported. Swallowing and feeding become difficult and the child may have difficulties managing their own secretions. The tongue may atrophy and have rippling movements or fine tremors called fasciculations. There is weakness of the intercostal muscles and the chest is often smaller than usual. Since the strongest breathing muscle is the diaphragm, the infant appears to breathe with their stomach muscles and the chest may appear sunken. 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. Early morbidity and mortality are most commonly associated with bulbar dysfunction and pulmonary complications.

**SMA Type II**

The diagnosis of Type II is almost always made before 2 years of age, with the majority of cases diagnosed between 7 and 18 months of age. Clinical features include delayed motor skills. Children 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 with the assistance or bracing and/or a standing frame. They are at increased risk for complications from respiratory infections. They may also have weak intercostal 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. Swallowing problems are not usually characteristic of Type II, but vary from child to child. Some may have difficulty eating enough food by mouth to maintain their weight and grow, and a feeding tube may become necessary. They may have tongue fasciculations and manifest a fine tremor in the outstretched fingers. Scoliosis is almost always be present, resulting in need for spinal surgery or bracing during their clinical course. Decreased bone density can result in an increased susceptibility to fractures.

**SMA Type III**

Type III, referred to as Kugelberg-Welander or Juvenile SMA, is much more variable in age of onset, and children can present from around one thru 18 yrs. of age, although diagnosis prior to age 3 years is typical. This is the least deadly form of childhood-onset SMA. Clinically, they can stand alone and walk, but may show difficulty with walking at some point. 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. Most eventually need to use a wheelchair. Muscle aching and joint overuse symptoms are common. Fine tremors can be seen in the outstretched fingers but tongue fasciculations are seldom seen. Feeding or swallowing difficulties in childhood are very uncommon.

SMA Type IV (Adult Onset)
In Type IV, symptoms typically begin after age 35. It is rare for SMA to begin between the ages of 18 and 30. Adult onset SMA is much less common and is defined as onset of weakness after 18 years of age, and most cases occur after age 35. It is typically characterized by an insidious onset and very slow progression. The bulbar muscles (swallowing and respiratory function), are rarely affected. Motor impairment is mild without respiratory or gastrointestinal problems.

Within each SMA type, sub classifications have been proposed and can add to prognostic significance. For example, only 22% of patients with type 3a, with onset of symptoms before age 3 years, were still ambulatory at age 40 years, whereas 58.7% of the patients with type 3b, with onset after age 3 years, were still walking by age 40 years.

Diagnosing Spinal Muscular Atrophy
Clinical Symptoms
The initial step in diagnosing SMA begins with concerns about a children’s strength and gross motor abilities. These concerns usually occur early in life in children with Type I and II, whereas children with type III may not show any clinical symptoms for many years. Many other neuromuscular diseases can present with clinical symptoms identical to SMA. Some of these alternative diagnoses require different diagnostic tests and may warrant different forms of treatment. Typically, those with Type I and II will exhibit the most dramatic weakness in the proximal muscles of the legs and arms. Tongue fasciculations are a very important clinical sign and often guides the physician to the diagnosis of SMA. Most lose their deep tendon reflexes.

Genetic Testing
SMA is diagnosed primarily through a blood test identifying the SMN1 gene, in conjunction with a history and physical. Normally, there are two genes called Survival Motor Neuron 1 and 2. In approximately 95% of patients with SMA have an absence of the SMN gene sequence, which is present in normal individuals. Sometimes the gene is not missing, but mutated. The numbers of copies of SMN2, a near identical 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. Type is generally determined from the clinical examination of the degree of weakness and ability to achieve major motor milestones such as sitting independently or walking.
Electromyography Testing (EMG) for Spinal Muscular Atrophy
With genetic testing, the EMG is no longer commonly used. If it is, it consists of two parts. A small electrical stimulus is administered to the nerves of the arm and legs to determine how quickly electrical messages are carried by the motor and sensory nerves. This is necessary to differentiate some forms of nerve disease from SMA. The second part requires the insertion of a very fine electrical probe into several muscles. Characteristic abnormalities show that the muscle has lost nerve supply because of the malfunction of the motor neuron. These findings are called “abnormalities of denervation” and are found in all children with SMA.

Muscle Biopsy Tests for Spinal Muscular Atrophy
This is not commonly used with the availability of genetic testing. If used, the examination of muscle tissue is used to confirm the diagnosis of SMA. When the genetic investigations are not confirmatory, muscle biopsy is absolutely essential.

Prognosis
The SMN1 gene is the primary manufacturer of the SMN Protein. It is the absence/defect of this gene that causes SMA. 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 will typically mean a less severe expression.

SMA Treatment
Each Spinal Muscular Atrophy patient is an individual. The physical and emotional impact of this diagnosis on the patient and family are as different as people themselves. There are some problems, however, which can be anticipated and addressed before they become a threat.

Pulmonary Care
The key respiratory problems in SMA are:
1. Impaired cough resulting in poor clearance of lower airway secretions
2. Hypoventilation during sleep
3. Chest wall and lung underdevelopment
4. Recurrent infections that exacerbate muscle weakness

Pulmonary disease is the major cause of morbidity and mortality in Types I and II and may occur in a small numbers in Type III. Without respiratory support, infants who are unable to sit, usually die before the age of 2. Pulmonary compromise is caused by a combination of
inspiratory and expiratory muscle weakness. The diaphragm is relatively spared. In nonsitters, the result is a bell-shaped chest with sternal depression. In older sitters and walkers, respiratory function may be compromised further by scoliosis. Swallowing dysfunction and reflux are important contributors to pulmonary morbidity. Individuals tend to progress to daytime respiratory failure via a sequence of recurrent chest infections, nocturnal oxygen desaturation, nocturnal hypoventilation, and then daytime hypercarbia.

**Respiratory Support**
Children with Type I and II are especially vulnerable to respiratory complications. A regular program of respiratory therapy and breathing exercises may help. Precautions to avoid illness should be taken with yearly influenza immunization. The most emotionally charged issue is whether or not to initiate aggressive mechanical support. Although the medical options are reasonably simple, the emotional turmoil of placing a child on a ventilator is painful for parents. Less invasive means of dealing with respiratory problems include chest physiotherapy (CPT) to help children get through their URI’s. Because of difficulty with coughing, a simple cold may result in saliva and mucous obstructing an airway.

Some children benefit from supplemental oxygen (O2) most often at night. Before initiating O2 treatment, it must be determined that the brain is not using low concentrations of O2 in the blood to determine how rapidly a child must breathe. Some of the normal regulation of breathing may be altered in those with chronic neuromuscular diseases. The decision to use supplemental O2 should be preceded by an evaluation by a pulmonary specialist.

Although the standard means of mechanical ventilation involves a tracheostomy, other forms of support may be used. This includes negative pressure ventilators and external positive airway pressure support systems. The use of a Bi-PAP has been especially as it provides the benefits of a ventilator. The decision to use any or all of these forms of respiratory support are emotionally charged and highly personal.

**Assessment and Monitoring**
The frequency of ongoing assessment depends on the clinical status and rate of progression of disease. Suggested frequency of evaluation is every 3 to 6 months, less often in stable walkers, and more frequently in clinically unstable nonsitters.

**Nonsitters**
Recommendations for respiratory assessment include evaluation of cough effectiveness, observation of breathing, and monitoring gas exchange. The majority of nonsitters with SMA may be too weak or too young to perform pulmonary function testing. Therefore, the most
useful evaluation of respiratory muscle function may be observation of cough ability. The physical examination also provides an assessment of respiratory status including respiratory rate, work of breathing, and presence of paradoxical breathing, chest wall shape, and color. Gas exchange monitoring, including pulse oximetry, can be used as a spot check during the day for hypoxemia and as a guide to direct airway clearance. Overnight pulse oximetry with chart recording can be used to screen for nocturnal hypoxemia.

End-tidal carbon dioxide (CO2), transcutaneous CO2, and serum bicarbonate measurement are also important assessment tools. However, serum bicarbonate may give a false sense of reassurance, as normal values may exist despite significant respiratory compromise during sleep. End-tidal CO2 and transcutaneous CO2 can be used to assess for sleep-related hypoventilation. The onset of hypoventilation is insidious, and patients may be clinically asymptomatic. Initially, hypoventilation will occur in sleep but as deterioration progresses, daytime respiratory function will be impacted. Polysomnography is a diagnostic tool during which respiration and sleep state are continuously monitored, and thus identifies the presence and severity of sleep-disordered breathing. It is useful in nonsitters, even in children without obvious symptoms, and can be used to initiate and titrate respiratory support. Additional screening tests include a baseline chest x-ray to provide an initial reference point and for comparison during respiratory deterioration. Formal evaluation of swallowing is indicated in those with unexplained respiratory deterioration and recurring pneumonia. Arterial blood gases for routine monitoring of respiratory function are not recommended.

Sitters
Recommendations of respiratory assessment for sitters are similar to nonsitters and include physical examination and pulmonary function tests (PFT). Sitters should be evaluated for presence and severity of scoliosis. Additional recommended assessments include forced vital capacity and lung volume measurements during PFT’s, assessment of sleep-disordered breathing, and pulse oximetry monitoring. Less important assessments include blood gas, CO2 monitoring, and chest x-ray. A routine swallow study is not recommended for sitters unless clinically indicated.

Walkers
In general, walkers have relatively preserved pulmonary function until late into their disease. Recommendations for routine assessment include complete PFT’s including spirometry, lung volumes, and respiratory muscle function tests. In addition, cough effectiveness and the physical examination are an important routine assessments. Further evaluation should be directed by clinical symptoms and indications.
Anticipatory Respiratory Care
Providing families with information about options for care and anticipating future needs are crucial to respiratory management of SMA. Nonsitters are the most fragile group, and early discussions should include the option of noninvasive ventilation and secretion management because of the rapid progression of the disease. In addition, anticipatory guidance and education for chronic care, illness management, and perioperative care should be provided. Day-to-day management should include deviations from the child’s baseline, routine cough and secretion management, understanding hypoventilation, and intervention. Illness management includes rapid access to specialty medical care providers, airway clearance and secretion management, respiratory support, nutrition and hydration management, and a low threshold to start antibiotics. Routine immunizations, including influenza vaccine, pneumococcus vaccine, and respiratory syncytial virus prophylaxis, are recommended.

Acute Care Management
The goal of acute management is to reduce atelectasis and enhancing airway clearance by noninvasive respiratory support. Blood gas monitoring may be of benefit. Airway clearance with manual cough assist or mechanical insufflation-exsufflation, along with oral or airway suctioning, CPT, oximetry are important and recommended. Assisted cough techniques are preferred over deep suctioning and bronchoscopy.

Chronic Management
Essential to chronic management is discussion of the family’s goals, which includes balancing caring for the child at home for as long as possible, long-term survival, quality of life and comfort, and the availability of resources. Goals of chronic management are to normalize gas exchange, improve sleep quality, facilitate home care, reduce hospitalizations and intensive care unit care, and reduce the burden of illness on the family. There is insufficient evidence, but based on experience and consensus, early aggressive and proactive intervention may prolong life without compromising quality of life.

Airway Clearance
Airway clearance is very important in both acute and chronic management of all patients. Caregivers should learn to assist coughing in all patients with ineffective cough. These techniques include manually and mechanically assisted cough. Daily assisted cough is recommended in more severely affected patients. Secretion mobilization techniques are also helpful and include CPT and postural drainage. Oximetry should be used to provide feedback to guide therapy. Oral suctioning can assist in secretion management after assisted coughing.
**Respiratory Support**
For those with daytime hypercapnia, respiratory support is indicated. In children with sleep-disordered breathing, nocturnal noninvasive ventilation reduces symptoms of sleep disturbance, nocturnal sweating, and morning headaches and improves appetite and concentration. In general, noninvasive ventilation settings are individualized to achieve adequate inspiratory chest wall expansion and air entry and normalization of O2 saturation and end-tidal CO2 or transcutaneous CO2 measurements. Noninvasive ventilation should be combined with airway clearance techniques.

In nonsitters, care without ventilation support is an option. Noninvasive ventilation can be used palliatively to facilitate discharge from hospital to home and reduce work of breathing. Continuous positive airway pressure (CPAP) is an option in a nonsitter who is not synchronous with bilevel positive airway pressure (BiPAP) and can be used with the goal of transitioning to BiPAP. Use of BiPAP, even for short daytime periods, may improve chest wall and lung development and reduce ribcage and sternal deformity in nonsitters and sitters, resulting in possible beneficial effects on pulmonary function. Adult walkers may develop sleep disordered breathing or acute ventilatory failure at the time of a chest infection. Noninvasive ventilation is an appropriate intervention and may be required during sleep chronically.

**Perioperative Care**
Those with SMA are at high risk for post anesthesia complications, which may lead to prolonged intubation, nosocomial infections, tracheotomy, and death. Complications include upper airway obstruction, hypoventilation, and atelectasis from impaired cough and impaired mucociliary clearance due to anesthetic agents. Pain may exacerbate respiratory compromise. Respiratory status should be optimized before surgery. If respiratory function and/or sleep study are abnormal, nocturnal noninvasive ventilation and assisted coughing techniques may be indicated preoperatively.

Postoperative management should be determined by preoperative function and the type of surgery performed. Patients are encouraged to bring their personal devices, such as noninvasive ventilation and mechanical insufflation-exsufflation-E machines, to use in the postoperative period. Although O2 is used frequently in the postoperative setting, it must be applied with caution in the patient. Hypoxemia secondary to hypoventilation may be mistaken with hypoxemia due to other causes, such as mucus plugging and atelectasis. End-tidal CO2 or transcutaneous CO2 monitoring or arterial blood gas analysis will facilitate appropriate O2 use. Good pain control will aid in preventing hypoventilation secondary to splinting. Postop pain management should be titrated to promote airway clearance and minimize respiratory suppression. Increased respiratory support may be needed while controlling postop pain.
**Gastrointestinal and Nutritional Care**

The problems associated with gastrointestinal and nutritional complications include:

1. **Feeding and swallowing problems.** Bulbar dysfunction can result in feeding and swallowing difficulties and aspiration pneumonia. The severity is variable in patients with intermediate severity and rare in those who are mildly affected.

2. **Gastrointestinal dysfunction.** Dysmotility problems include constipation, delayed gastric emptying, and potentially life-threatening gastroesophageal reflux (GER).

3. **Growth and undernutrition/overnutrition problems.** Without management, growth failure is universal in nonsitters; excessive weight gain is more common in sitters and walkers.

4. **Respiratory problems.** Respiratory complications raises concern for gastrointestinal problems of aspiration and GER. Increased work of breathing may also result in increased energy expenditure.

**Feeding and Swallowing Problems**

Feeding and swallowing difficulties are common in nonsitters and sitters but rarely in walkers. Symptoms of feeding difficulties include prolonged mealtime, fatigue with oral feeding, and choking or coughing during or after swallowing. The presence of recurrent pneumonias is a potential indicator of aspiration, which may be without evident choking or coughing. Problems in the preoral phase include limited mouth opening and difficulties in getting food to the mouth for self-feeding. In the oral phase, difficulties include weak bite force, reduced range of jaw motion limiting mouth opening, and increased fatigue of masticatory muscles. This affects biting and chewing abilities and can lead to prolonged mealtimes and fatigue, limiting sufficient intake. Masticatory and facial muscle weakness affects oral bolus control, chewing, and bolus propulsion, which contribute to reduced feeding efficiency. Difficulties with strength and efficiency are reported in the oral and pharyngeal phase of the swallow. Poor coordination of the swallow with airway closure can lead to penetration and aspiration of the airway. Poor head control may also be a factor in the development of feeding difficulties, precluding neck tuck or other compensatory postures to enhance the safety of swallowing.

**Management of Feeding and Swallowing Difficulties**

Treatment should aim at reducing the risk of aspiration during swallow and optimizing efficiency of feeding and promote enjoyable mealtimes. Changing food consistency and optimizing oral intake are appropriate treatment strategies. A semisolid diet can be used to compensate for poor chewing and reduce length of mealtimes. Thickened liquids may protect against aspiration of thin fluids. Preferably, this intervention would be evaluated objectively on videofluoroscopic swallow studies. Positioning and seating alterations and orthotic devices to enhance self-feeding ability may improve swallow safety and efficiency. Such interventions should be planned in liaison with an occupational therapist and/or physiotherapist.
There is no consensus regarding when to refer a patient for consideration for gastrostomy tube (GT) placement and whether one should supplement or replace oral feeding with tube feeding in a nonsymptomatic patient. However, optimal management requires proactive nutritional supplementation as soon as inadequate oral intake is recognized. It usually takes time to schedule GT placement. In the interim, supplementation via nasogastric (NG) or nasojejunal (NJ) feeding is desirable. Nasojejunal feeding may be preferable when GER with aspiration is a concern, especially when the patient is on ventilatory support. However, technical difficulty may prevent its feasibility. The presence of a NJ or NG tube may also result in a less-than-ideal mask fit when there is a need for the use of noninvasive ventilation such as BiPAP. Gastrostomy tube feeding is the optimal method of feeding when insufficient caloric intake or unsafe oral feeding is of concern.

**Gastrointestinal Dysfunction**
Children with SMA suffer from the following: GER, constipation, and abdominal distension and bloating. Gastroesophageal reflux is an important determinant of mortality and morbidity in patients with SMA. It can be associated with silent aspiration and results in pneumonias and, at times, life-threatening events. Frequent “spitting up” or vomiting after meals, complaints of chest or abdominal discomfort, bad breath, or obvious regurgitation of feeds may indicate GER. Some children may refuse feeds when they develop discomfort with swallowing, placing them at risk for under nutrition. High-fat foods delay gastric emptying and increases the risk of GER. Constipation is a frequently reported problem and is likely multifactorial (ie, abnormal gastrointestinal motility, reduced intake of dietary fiber, inadequate fluid intake, low muscle tone of the abdominal wall). Infrequent bowel movements can lead to abdominal distention and bloating.

**Management of Gastrointestinal Dysfunction**
Medical management of GER typically involves the use of acid neutralizers and/or inhibitors of acid secretion. This latter includes both histamine blockers and proton pump inhibitors. Short-term use is reasonable for symptomatic management. However, increasing evidence suggests that prolonged may be associated with a greater risk for gastroenteritis and pneumonia. When delayed gastric emptying or diminished motility is present, probiotics such as acidophilus or lactobacillus may help maintain a healthy gastrointestinal flora, particularly after antibiotic treatment. Gastrostomy tube feeding does not prevent GER. As a result, a fundoplication is commonly performed.

**Management of growth and under nutrition or over nutrition problems**
Children with SMA are at risk for growth failure or excessive weight gain. Growth failure is seen mostly in nonsitters and some sitters, whereas obesity is a problem of the stronger sitters and
walkers. Normal body mass indexes may not represent the ideal weights for children with SMA. Decreased activity will lead to reduced resting energy expenditure and increased risk of obesity. Routine history, physical examination, and monitoring of growth charts form the evaluation to identify growth failure or excess. The goal is to maintain each child on his or her own growth velocity. Growth curves followed over time are the most accurate indicator of nutritional status. Difficulty in obtaining accurate standing height measurements due to contractures or inability to stand may complicate growth monitoring. Recumbent length, segmental measurements, or arm span may be useful surrogate markers for linear growth in these children. Other methods for monitoring body composition include skinfold measurements, muscle circumference, or bioelectric impedance analysis. A 3-day dietary record is a simple and accurate tool that can help assess whether nutritional intake is adequate. Nutrient intake should meet the daily recommended intakes for age. Supplements to provide more than the dietary recommended intake for vitamin, mineral, protein, or fat should be discouraged. If an elemental formula is used, a dietitian should be involved to help ensure the child does not receive insufficient or excessive amounts of nutrients, to perform laboratory assessments as needed, and to monitor adequate growth. Children with SMA may have acceptable fat mass but may be perceived as underweight based on weight/height criteria because of their decreased lean body mass. This will result in inappropriate dietary recommendations that could lead to relative obesity. Those at risk for obesity should have growth parameters in the lower percentiles for weight/height and body mass index.

Management of Nutrition in the Acutely Sick

SMA patients are particularly vulnerable to catabolic and fasting states. Those with severe muscle wasting are more likely to develop hypoglycemia in the setting of fasting. Significant abnormalities are most likely in nonsitters and sitters, increasing their vulnerability for metabolic decompensation in the setting of a catabolic state. Thus, it is necessary to avoid prolonged fasting, particularly in the setting of acute illness, in all SMA patients. Nutritional intake should be optimized to meet full caloric needs within 4 to 6 hours after an admission for acute illness, via enteral feeding, parenteral feeding, or a combined approach. Prompt postop caloric supplementation is recommended to avoid muscle catabolism.

Orthopedic Care

Key Problems

Muscle weakness of varying severity limits motor function of trunk and upper/lower extremities, resulting in contracture formation, spinal deformity, limited mobility and activities of daily living, and increased risk of pain, osteopenia, and fractures. Evaluation includes range of motion, strength, function, seating and mobility, orthotics, and x-rays. Progressive weakness and immobility of the arms and legs may predispose to other orthopedic problems including
contractures. Physical therapists can instruct children and their families in range of motion techniques to help prevent contractures. Night splints of the ankles and wrist may also be useful in preventing contractures. Facilitated independent sitting or standing with a special chair or a standing frame may be an important part of the child’s daily therapy program.

Hip subluxation is a common comorbidity. As patients age, there is a significantly higher prevalence of kyphoscoliosis, difficulty coughing, joint contractures, and voice/speech problems in types I and II. In type III, there is also a significantly higher prevalence of fatigue and hypermobility of the hand. Scoliosis develops in more than 50% of children with SMA, most commonly in nonambulatory children or in those who lose the ability to walk.

**Evaluation**

Traditional measurements of strength are not possible in severely affected patients; thus, emphasis is on observation of function. Most children with SMA require help or supervision with bathing, dressing and assistance with mobility. Stairs present a major obstacle. Early and generalized joint contractures and scoliosis correlate with level of motor function and walking with support. Rolling by 5 years of age correlates with eventual walking, and inability to roll correlates with severe disease. Muscle strength can be quantified using muscle testing in children with Type II or III. Flexion contractures, which affect almost half of patients, are often noted during periods of inactivity and are considered intractable if greater than 45°. Activities of daily living are hampered, and contractures are perceived to be associated with disability. Pain increases in frequency and severity over time and correlates with decreased scores on quality-of-life indicators.

**Interventions**

In nonsitters, nutritional support, posture management, seating, contracture and pain management, therapy for activities of daily living and assistive equipment, wheelchairs for mobility, limb orthotics, and developmental therapies are important. In sitters, wheelchair mobility, contracture management, physical therapy, and occupational therapy are of highest value, with strong considerations for spine and limb orthotics and spine surgery. In walkers, the highest emphasis is on provision of physical therapy, occupational therapy, and wheelchair/mobility, although orthotics, scoliosis surgery, and pain management figured prominently.

**Orthotics**

In selecting and fabricating an orthosis it is important that everyone work together to ensure that it’s fabricated and allows the wearer to meet their functional goal. Spinal orthoses may be used for postural support. When used, spinal orthoses should be fabricated with an abdominal
cutout to allow appropriate diaphragmatic excursion and access to gastrostomy tubes where present.

**Orthopedic Surgery**
Surgical correction of scoliosis should be considered based on the curve progression, pulmonary function, and bony maturity. Scoliosis surgery provides benefits in sitting balance, endurance, and cosmesis. Evidence suggests that earlier surgery results in better outcome. Beneficial effects on pulmonary function remain controversial, but the rate of pulmonary decline may be slowed. Intraoperatively, excessive bleeding may occur. Postoperatively, complications include loss of correction, pseudoarthrosis, and chest/wound infections. Careful consideration is warranted for patient who is ambulatory because altered function, balance, and respiration may result in loss of independent walking. Pelvic obliquity may require surgical fixation. Although there is a higher rate of hip subluxation in SMA, few are painful. Surgical reduction and osteotomy are frequently followed by redislocation. In most circumstances, this surgery is avoidable. Ankle and foot deformities make conventional shoes difficult to wear, and soft tissue releases may be considered. In walkers, if soft tissue releases are performed, rapid and aggressive physical therapy may improve outcome.

**Mobility**
Children with Type I and Type II virtually never achieve independent standing or walking. Their mobility invariably requires wheelchair assistance, because most children lack the upper body strength to propel a manual wheelchair, a power chair is the logical choice. This enables them to participate in household and outdoor activities with their family and peers.

**Palliative Care**
In the case of those with SMA, the appropriate goal of therapy may not be clear. Some therapies may be perceived as placing quality of life in conflict with duration of life, prolonging suffering rather than relieving the burden of disease. Optimal clinical should be mindful of potential conflict of therapeutic goals. This conflict is made more difficult by the need for surrogate decision makers for a dependent infant and the fact that parents, siblings and other relatives will be affected by and thus have some valid interest in care decisions.

A choice for or against interventional supportive care is not a single choice, nor must it be unchanging with circumstance. There are some interventions that are better done early so as not to constrain later potential assistance. For example, placement of a gastrostomy tube is better done relatively early; when associated risks are lower, to provide more stable and comfortable nutritional support later when feeding is more tenuous. Similarly, it is important to discuss and determine the appropriate response to potential life-threatening respiratory
insufficiency, as emergency resuscitation during times of crisis without prior respiratory support are associated with many more problems. If appropriate, other forms of noninvasive respiratory device that might reduce the potential for emergent respiratory support should be introduced according to increasing need. Whenever possible, caregivers should ideally permit sufficient time after diagnosis prior to discussing these difficult issues; in all cases, sufficient time, honest appraisal of the choices, openness to revisiting decisions made, and personal rapport are essential to these discussions. End-of-life care decisions need to be defined and neither delayed nor aggressively forced upon unsuspecting, grieving, and stunned parents. Care for patients with spinal muscular atrophy is often best accomplished with a multispecialty team approach, when possible. Successful teams have a point person who is mindful of the many needs and can obtain appropriate medical, social, and spiritual assistance as appropriate.

In addition, hospice referral or other provision for the specific issues regarding terminal care, grief, and bereavement support is important. In the circumstance of a choice against mechanical ventilatory support, appropriate provision for management of terminal dyspnea can be of comfort to the patient and family alike. Use of nebulized narcotics can avoid much of the concern that overdosing contributes to death and provide comfort to the patient.

**Resources**

**Mosby Skills**

Level 2 Career Development  
Level 3 Career Development  
Cough Physiotherapy: Postural Drainage  
Cough Assist Machine  
Suctioning: Nasopharyngeal  
Enteral Nutrition: Naso/Oralgastric Feeding Tube Insertion, Care and Removal (PSA)  
Enteral Nutrition: Nasogastric, Nasoduodenal, Nasojejunal Feedings (PSA)  
Gastric Residual Assessment (PSA)  
Gastrostomy and Gastrostomy-Jejunostomy Tubes: Care and Management (PSA)  
Gastrostomy Tube (GT) and Low-Profile GT: Removal and Reinsertion (PSA)  
Range of Motion Exercises (PSA)

**Organizations**

**Families of SMA**

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