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1. Introduction

1.1 Overview and incidence

Spinal muscular atrophy (SMA) is a neuromuscular disorder characterized by degeneration of alpha motor neurons resulting in hypotonia, progressive muscular weakness and atrophy.\(^{30}\) Spinal muscular atrophy is one of the leading hereditary causes of infant mortality,\(^ {31}\) it comprises the second most common fatal progressive diseases after cystic fibrosis.\(^ {28}\) Spinal muscular atrophy is the most common neuromuscular disease in childhood after Duchenne muscular dystrophy with an estimated incidence of 1 per 5,000 to 10,000 live births.\(^ {4,11}\)

2. Pathogenesis

Spinal muscular atrophy is known to be genetic disorder that is inherited as an autosomal recessive disorder but some dominant or X-linked traits are reported. The pathological basis of SMA is abnormality of the large anterior horn cells in the spinal cord caused by deletion or mutation of the Survival Motor Neuron-1 (SMN1) gene located at chromosome region 5q.\(^ {1}\) Absence of all or part of the SMN has been detected in 98% of patients with SMA\(^ {10,13}\) and results in reduction of the full length protein necessary for proper function of the anterior horn cells.\(^ {10,13}\) The decreased level of the SMN protein results in selective death of spinal motor neurons,\(^ {31}\) with the severity of the disease being inversely proportional to the amount of the SMN in the anterior horn cells.\(^ {1}\) The severity of SMA ranges from total paralysis and need for ventilatory support to relatively mild muscle weakness.\(^ {1,32}\)

3. Clinical manifestation and classification

SMA is manifested by various clinical features that cause a variety of debilitating symptoms. Muscle weakness is a hallmark feature of SMA and patients with SMA are among the weakest and most hypotonic seen in any muscle clinic.\(^ {13}\) Clinically, the disease is characterized by progressive symmetrical muscle weakness, which starts proximally and moves distally, with the proximal muscles being more affected than the distal muscles.\(^ {5}\) Muscle weakness is associated with muscle atrophy, hypotonia, absence or marked decrease of deep tendon reflexes, fasciculation of the tongue, and tremors of the hand.\(^ {5,20}\) Patients with SMA have normal intellectual function. Contractures and spinal deformity have been reported to be common impairments. Pulmonary infections and restrictive lung disease are
the most serious complications in SMA. In general, the clinical course of SMA is highly variable, and it is more of a continuous spectrum with the age of onset from birth to adulthood, and the age of death from infancy to normal life expectancy. The severity of the diseases and clinical manifestations show a continuous range from the very severe to very mild forms of the disease. With age, muscle weakness increases, and the symptoms progress and patients lose their functions over time. The progression of the disease process varies both between and within types. Current evidence suggests that maximum function achieved is more closely related to life expectancy than age at onset. Based on the age at onset, clinical presentation, and the maximum functional level achieved, SMA is usually classified into the following broad types.

3.1 Type I SMA (Werdnig-Hoffmann disease)

Type I SMA is the most severe form of SMA, it is also known as Werdnig-Hoffman disease. The age of onset is typically from birth or in the first 6 months of life and the child never developing independent sitting. Werdnig-Hoffmann disease is characterized by severe generalized muscle weakness and hypotonia. Infants typically have significant wasting, and weakness in the limbs and trunk and present with decreased movements, especially against gravity. Most infants present with lack of head control and are never able to roll from supine or to pull to sitting. Significant oral motor weakness results in difficulties in sucking and swallowing and makes feeding progressively more difficult. Weakness of the intercostal muscles results in limited respiratory function, and breathing is usually entirely diaphragmatic resulting in development of abnormal breathing patterns and respiratory complications. The severity of respiratory complications is generally proportional to the weakness. Early morbidity and mortality are commonly associated with pulmonary complications, and death occurs during the first 2 to 3 years of life.

3.2 Type II SMA (Intermediate form)

Children with the intermediate form have an onset between 6 and 18 months of age. They are able to sit and may develop ability to stand but they are unable to walk independently. Some of the less involved children are able to walk with braces or assistive devices at some point of their life. Children with type II exhibit symptoms of weakness similar to type I SMA but with much less severe degree. Distal muscle weakness is less severe than proximal muscle weakness and starts later in the course of the disease. There is a delay in the acquisition of motor skills, with the majority of those children sit at the age of 12 months of age. As the disease progresses, children exhibit more weakness and regressed gross motor development. Feeding and swallowing are not difficult. Contractures are common including scoliosis or kyphoscoliosis. Early onset and rapidly progressing scoliosis is uniformly present; severity of scoliosis increases as the disease progresses and may require bracing and/or spinal fusion. Pulmonary complications are pervasive especially with scoliosis and as the disease progresses. Ventilatory assistance is common in later stages of the disease.

3.3 Type III SMA (Kugelberg-Welander disease)

Type III SMA, often referred to as Kugelberg-Welander disease or Juvenile-onset SMA, is the mild form of the disease. Children with type III SMA have an onset age typically after 18 months. This form is more variable in age of onset, although most diagnosis prior to age 3
years is typical, weakness in other mild cases may not be noticeable until late childhood. Patients are able to achieve independent walking and whilst some children may lose this ability in childhood, others maintain walking until adolescence or adulthood. Early motor milestones are often normal, including ability to walk, which is achieved at the normal age or slightly late. Although they are able to ambulate, they may show difficulty with walking at some point in their clinical course secondary to proximal muscle weakness. Walking is characterized by lack of balance, falls, increased lumbar lordosis, hyperextended knees or genu recurvatum, and excessive waddling. Muscle weakness mainly affects proximal muscles of the lower extremities and is less severe than types I and II SMA. Proximal muscle weakness often results in difficulty in stairs climbing, hopping, running and jumping. Gower’s’ maneuver may be present when getting up off the floor. Scoliosis and pulmonary complications are common in patients with type III SMA but less frequent and not severe as in patients with type II SMA. The incidence and severity of complications including scoliosis and pulmonary complications are related to the degree of muscle weakness and the functional status. Many patients with type III lose the ability to functionally ambulate as they get weaker during adulthood. Individuals with type III SMA usually have normal life expectancy.

3.4 SMA type IV (Adult-onset form)

Adult-onset SMA is a rare type. The onset of the disease is in the adulthood, typically in the third or fourth decades. The signs and symptoms are similar to those of type III SMA but the impairments and degree of disability are often mild. Life expectancy is normal.

3.5 Other forms of SMA

There are other very rare types of SMA disorders with similar symptoms but they are caused by different genes other than SMN1 and genetic mutation. Other forms of SMA include distal spinal muscular atrophy, characterized by distal muscle weakness; X-linked spinal and bulbar muscular atrophy (Kennedy disease), an X-linked adult onset form of SMA; childhood bulbar SMA (Fazio-londe disease), a progressive bulbar palsy; Hexosaminidase A deficiency, with variable neurologic findings, including progressive dystonia, spinocerebellar degeneration, and lower motor neuron disease; and Monomelic muscular atrophy, a cervical form of spinal muscular atrophy.

4. Diagnosis

Diagnosis of SMA is suspected on the bases of the clinical picture, muscle biopsy and electromyography. Genetic testing is the only definitive diagnostic test for patients with SMA. With the use of genetic testing, the role of EMG and muscle biopsy in confirming the diagnosis of SMA is limited. They can be used for the diagnosis of patients with SMA who present without homozygous deletion of the SMN gene. Genetic testing of SMA shows a deletion of the SMN gene on the fifth chromosome. EMG findings usually show a pattern of denervation including fibrillation potentials, positive sharp waves, and large amplitude, short duration actions potentials. Sensory nerve conduction velocities are normal with no marked decrease of motor nerve conduction velocities. Muscle biopsy provides evidence of muscle denervation with groups of small atrophic fibers with large hypertrophic fibers.
5. Prognosis
The course of SMA is relentlessly progressive. Prognosis varies according to the age of onset, type of SMA, and the maximum function achieved. The age at the time of the onset has the strongest relationship to prognosis. It appears that the earlier the onset of the disease, the faster the progression and the poorer the prognosis. The current prognosis for children with type I SMA is very poor, with the death usually occurs in the first two years of life as a result of respiratory failure caused by respiratory complications. Some children with SMA type I can survive beyond two years of age with the use of ventilator assistance. The prognosis of type II SMA is extremely variable. Patients with type II SMA have short life span; survival into adulthood is possible with aggressive respiratory care. Majority of patients with type III SMA remain independent in ambulation throughout adult life. Patients with Type III SMA are expected to have normal life expectancy. Patients with the onset begins before two years of age continue to ambulate until an average of twelve years of age. Patients with the onset after two years of age continue to ambulate throughout the adult life.

6. Assessment
Since assessment is important for guiding clinical management and for evaluating therapeutic outcomes, thorough assessment of patients with SMA is essential. Assessment should include regular assessment pertinent to children with neuromuscular disorders such as assessment of posture, muscle strength, performance, range of motion, gait assessment, respiratory assessment, and quality of life measures. Assessment of functional status and level of disability using standardized outcome measures should be also included.

6.1 Joint integrity, range of motion and muscle flexibility
Active and passive joint range of motion can be assessed using goniometry. Functional range of motion, muscle length, and soft tissue flexibility should be assessed using standard methods. Assessment of joint integrity, range of motion and muscle flexibility should be done periodically to monitor development of contractures, particularly when the patient loses ability to ambulate.

6.2 Assessment of posture
Routine posture examination should be performed on a regular basis. Posture examination includes examination of resting posture and changes in posture that occur with movement. Examination for the presence of scoliosis is essential especially for patients who are wheelchair dependent.

6.3 Muscle strength
Assessing muscle strength in children can be difficult, because the results depend on the patient’s effort. Strength deficits in children with SMA can be assessed using manual muscle testing (MMT) or hand-held dynamometer. Manual muscle testing is the most
widely method to assess muscle strength in clinical practice. It is a reasonably easy and inexpensive in measuring muscle strength in patients with SMA, but it does not allow grading small changes in muscle strength. A handheld dynamometer can be used to quantify muscle strength. Using a dynamometer is easy and comfortable and allows for measuring small changes in strength over a continuous range.\textsuperscript{19}

### 6.4 Respiratory function

Pulmonary function tests are parts of the regular assessment in patients with SMA to monitor changes in respiratory status. Routine assessment of respiratory function includes complete pulmonary function tests, including spirometry, lung volumes, and respiratory muscle function tests.\textsuperscript{30} Assessments of cough effectiveness and breathing pattern are important for the non-ambulatory patients or patients who are too weak or too young to perform pulmonary function testing.\textsuperscript{30}

### 6.5 Gait

No disease-specific gait test or measure exists for patients with SMA. Description of gait deviations and safety and stability during walking should be included as part of routine gait analysis. The 10-Meter Walk test can be used to measure gait speed. The Six Minute Walk Test or the Two Minute Walk Test can be used to measure endurance during gait.

### 6.6 Measurement of functional status and quality of life

Examination of functional status including examination of functional mobility skills and activities of daily living is an important consideration. Several clinical outcome measures can be used to measure functional outcomes in patients with SMA including generic measures and disease-specific measures. Generic outcome measures commonly used in patients with SMA include the Gross Motor Function Measure, the Test of Infant Motor Performance, the Alberta Infant Motor Scale, the Wee Functional Independence Measure, the Motor Function Measure, and the EgenKlassifikation Scale. Disease-specific outcome measures include the Hammersmith Functional Motor Scale, the Modified Hammersmith Functional Motor Scale, the Children’s Hospital of Philadelphia Test of Strength in spinal muscular atrophy, the Infant Test for Neuromuscular Disease, and the Spinal Muscular Atrophy Functional Rating Scale. Selection of the outcome measures to be used is based on age of the patient, patient’s functional level, aspects of function being measured, ease of administration, and burden imposed on the patient. The Pediatric Quality of Life Inventory (PedsQL) instrument can be used to measure quality of life.

### 7. Management

No specific therapy is currently available for SMA. Treatment is usually supportive, and may include physical therapy, occupational therapy, nutrition, orthotic management, and possibly surgical intervention. Appropriate recommendations are made on the basis of each patient’s presentation and functional level.\textsuperscript{15} The most important goal in the management of the patients is to achieve maximal independent living with maximized mobility, and to
prevent the development of complications. Treatment focuses on prevention of complications of severe weakness including restrictive lung disease, orthopedic deformities, immobility, and psychosocial problems.

The multidisciplinary approach is important to assess and address the needs of the patient/family. The multidisciplinary team might consist of neurologist, pediatrician, physiatrist, physical therapist, occupational therapist, speech therapist, nutritionist, pulmonary specialist, orthotist, genetic counselor, social worker, and psychologist. Family education and patient/family centered care are important parts in the management of patients with SMA. Education should include the disease process, associated impairments and complications, physical limitations, functional abilities, prognosis, and expected outcomes.

7.1 Therapeutic exercises and strength training

The overall therapeutic goals are to achieve maximal independence in mobility, and to prevent and delay progression of complications. Exercise programs may help to improve and maintain range of motion, maintain mobility, and prevent or slow the progression of contractures, orthopedic deformities and respiratory failure.

Strengthening exercises have been shown to be effective to slow the deterioration of muscle weakness in patients with neuromuscular disorders. Strengthening exercises may prolong ambulation and delay dependence on wheelchair for mobility in patients with neuromuscular disorders including children with Duchenne muscular dystrophy. Strengthening exercises may be used to slow the deterioration in muscle strength. The role of strengthening exercises in SMA is not well established due to lack of clinical trials on the effects of exercise programs and lack of trials critical data to support appropriate exercise prescription. Appropriate exercise recommendations in SMA are based on the patient’s presentations and functional status, and therapists’ experience with similar neuromuscular conditions. Recommendations regarding strengthening of patients with SMA include precautions and guidelines from other degenerative muscle diseases.

In patients with neuromuscular diseases, excessive strengthening exercises may contribute to deterioration in muscle strength by increasing muscle degeneration. Therefore, excessively strenuous strengthening such as excessive resistive exercises, eccentric exercises, and maximal aerobic training should be avoided. Excessive fatigue and overwork weakness should be avoided, frequent rests and self-initiated rests should be given frequently especially for the weaker patients and patients with decreased respiratory functions. Positioning and support can be used to maximize biomechanical advantage and minimize the effects of gravity. Monitoring with oximetry is recommended particularly for patients with compromised respiratory functions. It is important to monitor responses to exercises such as fatigue, pain, and muscle soreness.

Strengthening exercises for SMA may include low-intensity strengthening exercises, and submaximal aerobic exercise. Since there is no evidence to support traditional strength training for patients with SMA, practicing functional activities and tasks of activities of daily living may be good recommendation for those individuals. These exercises and activities should be designed based on the age, developmental stage, and functional level.
Exercise strategies for young children and infants may include practicing developmental skills. This includes activities to facilitate head and trunk control; floor mobility skills such as rolling and creeping; facilitation of weight shift and weight bearing and transitions between positions; and facilitations of upright positions and skills such as sitting, standing and walking as appropriate. For the less involved and older individuals, exercise strategies include practice functional activities such as standing and walking, and gentle aerobic programs.

7.2 Participation in physical activities

Weakness and difficulty in moving independently in patients with SMA contribute to physical inactivity and limited participation in exercise programs. The consequences of physical inactivity are particularly detrimental, could contribute to secondary impairments and may lead to additional decline in functional status. Recent evidence suggests that engagement in physical activities helps improve physical functioning in children with disability. Participation in physical activities may promote physical functioning, quality of life, health, and well-being.

Participation in physical activity may include participation in recreational and sports activities. Recreational programs that can be beneficial include swimming, cycling, and riding when appropriate. Activities should be selected carefully with the goal of improving functional performance and daily activities, promoting aerobic fitness and preventing complications of inactivity. Activities should be selected based on the age, developmental skills, and functional abilities.

7.3 Aquatic exercises

Aquatic therapy or hydrotherapy is being used on an increasing basis and has been shown to be beneficial for children with SMA. The use of aquatic therapy in SMA may be related to the physical properties of water. The properties of water provide weight relief and postural support, facilitate antigravity movements allowing more freedom of movement, and provide an opportunity to perform activities that may be too difficult to accomplish on land. Aquatic exercises provide low-intensity strength training, walking and balance exercises, and aerobic training without the fear of fatigue or overwork.

7.4 Feeding and nutrition

Infants with type I SMA have poor oral motor control with sucking and chewing and tendency to get fatigued easily during feeding and swallowing. Lack of head control and head support may also affect swallowing. Those children may have difficulty getting enough nutrition and are at risk of aspiration. Some infants may require indwelling nasogastric tube to supplement oral feeding. Gastrostomy may be an option for some children to improve carer satisfaction and quality of life, and to avoid aspiration.

7.5 Management of contractures

Muscle contractures and orthopedic deformities are common complications among patients with SMA. Contractures and orthopedic deformities occur primarily in type II and type III
patients who have longer periods of muscle weakness. Contractures develop secondary to muscle weakness, muscle imbalance, lack of mobility, and poor posture and positioning. Development and severity of contractures are related to the severity of muscle weakness, the duration of muscle weakness, and immobility. Muscle contractures are common in muscles that cross two joints or more. Classic contractures are seen in iliotibial band, hip flexors, knee flexors and plantar flexors.

Prevention and treatment of contractures are important issues in the management of patients with SMA. Management of contractures should begin before the contractures exist. Management of contractures includes combination of consistent program of range of motion exercises, positioning, regular stretching, and splinting. Muscle groups that are at risk of developing contractures should be targeted for stretching. Range of motion and stretching exercises can be used to preserve and increase flexibility. Active range of motion and stretching exercises can be used to maintain flexibility and prevent contractures in the ambulatory patients. In the non-ambulatory patients, regular range of motion program and passive stretching are used to prevent development and slow progression of contractures. Ankle foot orthoses and night splints can be used to maintain flexibility and range of motion. Positioning devices and custom fitted equipment can be used for positioning to provide low-intensity prolonged stretching. A tilt in space or recliner chairs can be used to allow easy positioning changes. Standing program provides low-intensity prolonged stretch that can be used for the non-ambulatory children.

7.6 Adaptive equipment and assistive devices

Patients with SMA frequently benefit from use of assistive and adaptive devices, with changing needs as their condition progresses. Adaptive equipment and assistive devices can be used to provide positioning, control contractures and deformities, and support function. The choice of assistive devices for patients with SMA is based on individual clinical decisions due to lack of definite intervention trials. The decision to use an assistive device should be a collaborative decision between the patient, family, orthopedic surgeon, physiatrist, and therapist.

7.6.1 Orthotics

Ankle foot orthoses (AFO) or night splint can be used to provide prolonged stretch to control the progression of plantar flexion contractures. Knee splints may be used to control hamstring flexibility and knee flexion contractures. Thigh binders can be used to control iliotibial band contractures. Assistive devices including braces, taping, AFO, knee-ankle-foot orthoses (KAFO), and hip-knee-ankle-foot orthoses (HKAFO) can be used to provide support and maintain joints alignments. Assistive devices may be used to facilitate stability, weight bearing and upright posture during standing and ambulation.

7.6.2 Positioning devices

Positioning devices can be used to provide support, and control contractures and deformities. Positioning devices can be custom fitted, special foam, or cushions. They allow easy positioning and stretching, and provide support. Head and trunk lateral support can be
used to accommodate for weak neck and trunk muscles and lack of head and trunk control, they can be used to assist in positioning and to maintain upright head and trunk during sitting.

7.6.3 Standing devices

Non-ambulatory patients with SMA may benefit from a standing program using standing frames or swivel walkers. Standing programs are used for non-ambulatory patients to prevent or reduce secondary impairments by maintaining muscle extensibility, preventing muscle and soft tissue contracture, promoting optimal musculoskeletal development, and to address the issue of reduced bone mineral density.27,29

7.6.4 Wheelchairs and seating systems

Because of the progressive weakness associated with SMA, many patients benefit from a wheeled mobility device as the primary means of locomotion. Wheelchair seating system deserves special considerations since many patients require a full-time use of the wheeled mobility device. The course and progression of the disease, presenting symptoms, degree of spinal deformity, and whether the patient is using mechanical ventilation should be taken into considerations when deciding on a mobility device. Manual wheelchairs allow the patients to maintain upper body strength and cardiovascular endurance. A power wheelchair should be considered when impairments prevent manual propulsion. Power wheelchairs enable patients to maintain a level of independence while moving within their environment and to compensate for mobility limitations.14 For young children who are not ambulatory, power mobility may be used to provide independent mobility at appropriate developmental age.25 Children as young as two years can independently propel wheelchair.14, 25

7.7 Management of scoliosis

Progressive weakness and reduced mobility associated with SMA place the patients at risk of contractures and scoliosis. Scoliosis is the most serious orthopedic problem seen in patients with SMA. Scoliosis develops earlier and progresses faster in non-ambulatory children than ambulatory children, scoliosis is seen in almost all children with type II SMA and majority of patients with type III, with the severity is less in type III SMA as compared to type II SMA. The incidence and severity of scoliosis increase with age and severity of muscle weakness, with the severity and progression of scoliosis increase once patients lose ambulation and become dependent on wheelchair for ambulation. Reduced respiratory function is common in patients with scoliosis. As muscle weakness progresses, the degree of scoliosis increases causing more discomfort and difficulty in positioning and respiration. Presence and degree of spinal deformity should be monitored periodically by examination and routine radiography, particularly for the non-ambulatory patients or as patient loses ambulation. Spinal x-rays are indicated once there is clinically detected scoliosis.32 Range of motion program and spinal positioning are important to provide comfort and slow the progression of spinal deformities. Adequate trunk supports on a wheelchair, and wheelchair modifications such as custom molding, gel or air cushions may be needed to provide maximum support, and comfort and may minimize the progression of spinal deformity. As
scoliosis progresses, external bracing such as thoraco-lumbo-sacral orthosis can be used to provide support and to apply forces to realign the vertebral column. External bracing is used to reduce, prevent, or slow the progression of scoliosis. The decision and timing of surgical intervention are based on degree of scoliosis, curve progression, pulmonary function, and bony maturity. Surgical correction of spinal fusion is indicated to prevent further progression and deterioration of scoliosis and respiratory function. The outcomes of spinal stabilization include improved sitting balance, endurance and cosmetics and slowed pulmonary progression. Intensive preoperative and postoperative physical therapy is required to prevent respiratory complications, and loss of strength or function after spinal fusion. Orthotic intervention, new wheelchair or wheelchair modification are likely to be required after the surgeries and should be included in the preoperative plan of care.

7.8 Management of respiratory complications

Lung diseases resulting from weakness associated with SMA are the most common and most serious complications of SMA. Respiratory impairments place the patient at risk for respiratory tract infections, and pulmonary insufficiency/failure, and are the major causes of morbidity and mortality.

The key respiratory problems in SMA are impaired cough with poor clearance of lower airway secretions; hypoventilation; chest wall and lung underdevelopment; and recurrent infections. Sleep-disordered breathing resulting from hypoventilation is common in SMA. Cardiopulmonary endurance reduces markedly once the child becomes wheelchair dependent. Increased weakness, decreased mobility level, and development of scoliosis are important factors to consider when assessing the respiratory function and progression of respiratory complications.

Patients with SMA should be evaluated by a respiratory care specialist. Routine evaluation of respiratory function including pulmonary function testing should be done on a regular bases. Pulmonary function testing with forced vital capacity can be used as a baseline and as a predictor of respiratory reserve. Pulse oximeters can be used at home as to indicate when the child is not ventilating properly.

Providing information about respiratory care and anticipated future needs is crucial to respiratory management of SMA. Patients with SMA and their families should learn how to monitor the respiratory function. Signs of respiratory insufficiency include deceased alertness, confusion, headache, pallor, and night-time restlessness.

Chest infections should be treated with antibiotics, postural drainage, chest physical therapy, and when appropriate, assisted ventilation. Patients and families should be educated on how to perform postural drainage techniques, assisted coughing, and breathing exercises.

Respiratory care for patients with SMA includes airway clearance techniques, respiratory exercises, chest physical therapy, and noninvasive ventilation, including intermittent positive pressure ventilation, bilevel positive airway pressure ventilation, and negative
pressure ventilation. Ventilatory assistance might be used for patients with respiratory failure.

7.9 Genetic counseling
Genetic counseling for patients or parents who wish to have another child is extremely important. SMA genetic testing can be used for carrier detection and detection of an affected fetus.

7.10 Vocational counseling
Some patients with SMA are limited to occupations that do not require physical demands. Vocational counseling and planning may be beneficial early during high school years to facilitate transition from school to postsecondary education. Vocational counseling may be necessary to help adjustment to work settings.

7.11 Psychological support
Counseling and psychological support are important strategies in the management of SMA. Anxiety and depression can greatly impact patients and families’ quality of life and their abilities to cope with the diseases and progressive changes in function and abilities. Formal counseling and psychological support should be available to assist patients and families coping with the severity and progression of the disease. Patients and families should be educated regarding the disease process and expectations and making sure that families are having the appropriate expectations for mobility and function.

8. Summary
Spinal muscular atrophy, a neuromuscular disorder, is one of the leading genetic causes of infant mortality. The disease is caused by deletion or mutation of the SMN1 gene and a reduction in the levels of functional SMN, resulting in selective death of spinal motor neurons. The type of SMA (I, II, III, or IV) is determined by the age of onset, the severity of symptoms, and the maximum function achieved. There are other rare types of SMA disorders with similar symptoms but they are caused by different genes other than SMN1 and genetic mutation. Spinal muscular atrophy is characterized by severe progressive muscle weakness, atrophy and hypotonia. Complications of muscle weakness include decreased mobility function, restrictive lung disease, contractures, orthopedic deformities and psychosocial problems. There is no cure for SMA. Treatment is usually supportive and focuses on management of the symptoms and preventing complications of muscle weakness. Pulmonary complication is a hallmark of the disease and is the main cause of death especially in type I and type II SMA. The prognosis and clinical course of SMA are highly variable, and they are more of a continuous spectrum with the age of death from infancy to normal life expectancy.

9. References


