Spinal muscular atrophy: Classification, aetiology, and treatment of spinal deformity in children and adolescents

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KEYWORDS
Spinal muscular atrophy; Spinal deformity; Neuromuscular scoliosis; Surgical treatment; Paralytic scoliosis

Summary
Spinal muscular atrophy is a hereditary neurological condition, which presents with symmetrical limb and trunk weakness. Spine deformity is the most frequent orthopaedic manifestation of the disease in patients who survive beyond the first year of life. Scoliosis in this group of severely disabled children decreases their sitting tolerance, causes pain from impingement of the ribs against the pelvis, affects ambulatory ability, and creates further respiratory compromise accelerating their death. Spinal arthrodesis is the only treatment that has a well-documented positive impact in restoring trunk balance and preserving function. This is associated with significant technical challenges and a high rate of life-threatening complications. A comprehensive review of the condition and a strategy for treating spinal deformity are presented in this paper.

Introduction
Spinal muscular atrophy (SMA) is an uncommon hereditary condition of autosomal recessive inheritance, which affects the anterior horn cells of the spinal cord and the neurons of the lower bulbar nuclei. It manifests clinically with symmetrical limb and trunk weakness affecting the proximal more than the distal trunk muscles and the lower more than the upper limbs. Sensation and mental function are not impaired. Fasciculation of the tongue and the deltoid are often seen, as well as a fine tremor affecting the hands. This tremor resolves during relaxation or sleep. The development of scoliosis, joint contractures and dislocation of the hip are the most common orthopaedic problems associated with the condition. The natural history of SMA is premature death,
which is caused by respiratory failure as the consequence of a gradually deteriorating respiratory muscular function.

The purpose of this article is twofold. Firstly, to provide a comprehensive overview on SMA, which is helpful to orthopaedists involved in caring for these children, including recent advancements in classification, understanding of the aetiology, and principles of management of the condition. The focus of the paper is, however, to investigate the characteristics of spinal deformity that develops in this group of patients and to describe a strategy for treatment. We believe that it is important to increase awareness of the complexity of these deformities in regard to their specific surgical considerations, as well as the coexistence of a multitude of associated medical problems that occur in this particular patient population and can jeopardize an inherently challenging surgical procedure.

Classification

There is no universally accepted classification for SMA. Currently, there is more than one classification systems in use with each one of these systems applying different terms to the various types of the condition (Table 1). A recent attempt at resolving this problem by the international SMA collaboration has led to the definition of specific diagnostic criteria and the development of a new classification system, which is based on the age at presentation of symptoms, functional abilities and life expectancy (Tables 1 and 2).

The classification system most widely used by orthopaedic surgeons is largely historical, but it correlates well with disease severity. In 1893, Hoffmann and in 1894, Werdnig described SMA for the first time in the literature. Kugelberg and Welander described in 1956 a similar condition of later onset that was also less progressive in nature. According to this classification, type I, acute Werdnig–Hoffmann or acute infantile SMA is characterized by generalized muscular weakness and hypotonia that has its onset before the age of 6 months. This is the most severe form of SMA with the earliest onset of symptoms and most rapid progression. In 30% of cases the onset is prenatal. There may be a history of poor foetal movement and crying is of low volume and not sustained following delivery. These children are never able to walk or sit unaided. They have lack of head control and often bulbar paralysis. Voluntary movement of their extremities may be limited to their fingers and toes and there is often fasciculation of the tongue and a fine tremor affecting the hands. Spinal reflexes are globally absent. Facial expression has been described as bland with a striking facial resemblance among patients due to muscle weakness. The cranial nerves may be also involved. Nasogastric feeding may be required as these children may suffer from dysphagia. As the intercostal muscles are affected, respiration depends mainly on the diaphragm. Respiratory movements are paradoxical with severe collapse of the ribs and a bell-shaped lower thorax. Progressive respiratory insufficiency can lead to recurrent chest infections and atelectasis that ultimately result in early death from pulmonary failure. The prognosis is poor and life expectancy ranges from 6 months to 2 years. As a consequence, orthopaedic intervention is rarely required.

Type II, chronic Werdnig–Hoffmann or chronic infantile SMA has its onset before the age of 18 months and is less severe than type I. Muscle weakness is not generalized at first and there is less disease involvement. After initial presentation, neuromuscular function in these patients may remain static for long periods before eventual progression. Most commonly the weakness begins in the proximal lower limb muscles (gluteal and quadriceps). The legs are affected earlier than the arms. The upper limbs are also involved but

| Table 1 Classification systems and clinical features of SMA. |
|-----------------------------|-----------------------------|-----------------------------|-----------------------------|
| **Historical**               | **SMA Collaboration**       | **Evans et al.**             | **Dubowitz**                |
| Type I, Werdnig–Hoffmann, Acute Infantile | Type I | Group I | Mild |
| Age of onset before 6 months | Generalized muscle weakness | Onset < 6 months | Never able to sit | Could stand unaided at same stage in life |
| Type II, Werdnig–Hoffmann, Chronic Infantile | Type II | Group II | Intermediate |
| Age at onset 2 months to 2 years | Lower limbs and proximal muscles affected first | Onset < 18 months | Can sit but are unable to walk | Could sit at some stage but was never able to stand unsupported |
| Unable to walk unaided | Unable to run or climb stairs | Never stand live beyond 2 years | Group III | Have limited walking ability |
| Kugelberg–Wellander, Chronic Childhood, Juvenile | Type III | Onset > 18 months | Stand alone Live into adulthood |
| Age at onset over 2 years | May walk until adolescence | May survive to fifth decade | Group IV | Severe |
| | | | | Walk, run and climb stairs | Was never able to sit unsupported |
this typically occurs later. A fine tremor of the hands and fasciculation of the tongue may be seen but it is less common than in type I. Fasciculation of the eyelids may be present. Respiratory problems develop after a variable initial asymptomatic period of time and thoracic deformity is less pronounced. Patients can typically sit if placed in position and may be able to stand with assistance. They have a waddling gait, increased lumbar lordosis, genu recurvatum, a protuberant abdomen and scoliosis. Prognosis in this form varies considerably and life expectancy ranges from 2 years to adulthood. Most patients live longer than 10 years and some may survive up to their fifth decade. The principal cause of death is respiratory compromise.

Type III, Kugelberg–Welander, chronic childhood, or juvenile SMA has a later onset and better prognosis. The disease manifests clinically after the age of 18 months and usually before the age of 10 years. The onset of muscle weakness is gradual and typically follows a slowly progressive course. The affected children achieve normal or slightly delayed motor milestones. These patients are usually able to walk, run and climb stairs until late childhood and sometimes into adolescence. On clinical examination, they have atrophic proximal lower limb muscles with characteristic pseudohypertrophy of the gastrocnemius, which is spared. A positive Gower sign can also be elicited due to marked weakness in the pelvic girdle. Their ability to walk may remain for many years after the initial diagnosis and often continues into the third decade of life. Survivorship can extend to the fifth decade.

More recently, Evans et al.\(^9\) described a functional classification system, which has been based on the maximum physical function achieved by the affected patients rather than the age of onset or age at diagnosis of the condition (Table 1). This classification only applies to the chronic Werdnig–Hoffmann, and the Kugelberg–Welander forms of SMA and divides the children into four groups: Group I. Patients never develop the ability to sit independently and have poor head control. Group II. Patients have head control and can sit but are not able to walk. Group III. Patients can pull themselves up and walk in a limited fashion frequently requiring orthoses. Group IV. Patients develop the ability to walk, climb stair and run before the onset of weakness.

Dubowitz developed another classification system in 1978 according to the severity of the disease and incorporated three types (Table 1).\(^10\) Mild when the child has been able to stand unaided at some stage in life. Intermediate when the child has been able to sit without support at some stage in life. Severe when the child has never been able to sit without support.

### Epidemiology

Pearn et al.\(^11\) reported an incidence of 1 in 25,000 live births for SMA type I in the United Kingdom, making it one of the most common causes of genetically determined neonatal death. This translates to a gene frequency of about 1 in 160 and a carrier frequency of 1 in 80.\(^12\) Epidemiological data from Canada, Finland, Hungary, and Norway have recorded an incidence of SMA type I ranging from 1 in 25,000 to 1 in 15,000 live births.\(^13–16\) The incidence in types II and III SMA account for similar numbers of patients.\(^17,18\)

As anticipated for a genetically transmitted disease, the reported incidence of SMA in population groups where consanguineous marriages are a frequent practice is significantly higher. In Dammam Saudi Arabia, an incidence of the condition of up to 1 in 518 live births has been described.\(^19\) The disease is even more common among the Karaite community in Israel with an increased incidence of 1 in 400.\(^20\)

### Aetiology and pathogenesis

The complete pathogenesis of SMA is still undetermined; however, the genetic cause of many cases has been attributed to two genes. The identification of the Survival of Motor Neuron 1 (SMN1 or telomeric SMN\(^1\)) gene on the long arm of chromosome 5 (5q11.2–13.3) was first described by Brzustowicz et al. in 1990.\(^21\) The SMN1 gene lies within an inverted duplication which contains an almost identical copy of the gene (labelled SMN2 or centromeric SMN\(^2\)).\(^22\) The majority of SMA patients (98.7%) with the autosomal recessive form of proximal SMA lack SMN1 but carry at least one copy of SMN2, which only partially functions and is unable to compensate completely for the lack of SMN1.\(^23\) Both SMN1 and SMN2 encode for the survival motor neuron (SMN) protein.\(^24\) SMA develops as a consequence of an insufficient amount of the SMN protein.

The level of SMN protein expressed from SMN2 varies between patients, accounting for different disease phenotypes, and also shows a variable expression between different human tissues. High levels of SMN protein are expressed in the spinal cord, brain, liver and kidney.\(^24\) In SMA patients the tissue that shows the most significant change in SMN expression is the spinal cord. SMN expression is reduced 100-fold in the spinal cord of SMA type I patients when compared to non-SMA controls. The SMN protein is likely to have many functions but one of them is essential for cell survival. The loss of SMN protein has a catastrophic effect on the motor neuron cell, resulting in cell death. Exactly which metabolic processes are disrupted remains unknown. Examination of the spinal cord reveals loss of anterior horn cells (motor neurons) and secondary changes occur in the roots and nerves as a consequence of Wallerian degeneration.\(^25\) Denervation of the associated muscle groups leads to muscular atrophy and the clinical

### Table 2 Criteria for the diagnosis of SMA.\(^4\)

<table>
<thead>
<tr>
<th>Criteria for diagnosis</th>
<th>Exclusion criteria</th>
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<tr>
<td>1. Muscle weakness</td>
<td>1. CNS dysfunction</td>
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<tr>
<td>Symmetrical</td>
<td>2. Arthrogryposis</td>
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<tr>
<td>Proximal &gt; distal</td>
<td>3. Involvement of other neurologic systems or other organs (i.e., hearing, cardiac or vision)</td>
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<td>Legs &gt; arms</td>
<td>4. Sensory loss</td>
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<tr>
<td>Trunk involved</td>
<td>5. Eye muscle weakness</td>
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<tr>
<td>2. Denervation demonstrated</td>
<td>6. Marked facial weakness</td>
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<td>By EMG</td>
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<td>By muscle biopsy</td>
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<td>Clinical fasciculations</td>
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\(^4\) Hoffman, and the Kugelberg–Welander forms of SMA and divides the children into four groups: Group I. Patients never develop the ability to sit independently and have poor head control. Group II. Patients have head control and can sit but are not able to walk. Group III. Patients can pull themselves up and walk in a limited fashion frequently requiring orthoses. Group IV. Patients develop the ability to walk, climb stair and run before the onset of weakness.
presentation of SMA. Clinically evident sensory functional loss is rarely detected in these patients; however, microscopical changes in the dorsal spinal columns have been previously reported.26

Apart from the autosomal recessive form of SMA there are other genetically distinct forms that have been described including an autosomal dominant form,27–29 an X-linked type of SMA,30–32 distal SMA,33 and SMA with associated respiratory distress.34

SMA is considered by most investigators to be a progressive neuromuscular condition; however, the cause of this observed functional deterioration is not clear. In a prospective study using reproducible quantitative measurements, Russman et al.35,36 have examined muscle strength over a period ranging from 2 to 6 years in 73 patients with an age range of 4 to 57 years. Muscle strength was ‘an order of magnitude’ less in SMA patients (total muscle score 17.1 kg in patients less than 15 years of age; 21.1 kg in those aged more than 15 years) when compared to controls (total muscle score 101.7 and 229.5 kg respectively). However, with time no measurable loss in muscle strength was detected in individual patients with SMA. When comparing patients and controls over the age of 15 years to those aged less than 15 years, the authors found a 200% increase in total muscle score in the control group and only a 25% increase in the SMA group. It has been suggested that the functional loss observed in patients with SMA over the years may be due to secondary factors, such as the development of scoliosis and hip subluxation or may be related to medical complications rather than be attributed to a continued functional motor unit loss.4,35,36

Diagnosis

The diagnosis of SMA is primarily based on the clinical features and can also be supported by a positive family history. The diagnostic criteria for the condition have been described by an international SMA collaboration (Table 2).4

The diagnostic studies include muscle and nerve biopsy, electromyography, and conduction velocity.

Muscle biopsy can be used to assist diagnosis and should be performed on moderately affected muscles using an open technique. A careful, atraumatic surgical technique is necessary to obtain a muscle sample (preferably from the vastus lateralis) and maintain its length and orientation. Local anaesthetic can be utilized but must spare the muscle. Compromised areas of musculotendinous junctions, or sites of previous scarring due to injections, recent trauma or electromyography needle application should be avoided. Histological samples show evidence of loss and atrophy of muscle fibres within normal groups of fibre bundles and no evidence of a primary myopathy.37,38 There is proliferation of perimysial connective tissue, and groups of giant fibres (type I) can also be seen. In neonates, perimysial fibrosis and a non-specific variation in muscle fibre diameter are the only histological features present. In adults with SMA, the same histochemical pattern of involvement is preserved with the addition of a progressive denervation and necrosis of muscle fibres occurring within groups of hypertrophic fibre bundles.

Electromyography may be used to support the diagnosis of SMA.38 Abnormal findings on this study increase in frequency and magnitude in direct relation to the severity and duration of the disease. Fibrillation potentials following denervation of muscle are the most accurate diagnostic features. Spontaneous motor unit activity occurs frequently, is present at all ages, and is specific for the condition. Conduction velocity in motor units is often normal. However, it can be decreased in severely affected children and increased in children with less extensive involvement.3,39,40 Evidence of a muscle tremor defect may be present on the electrocardiogram.35

Genetic testing for autosomal recessive SMA using a PCR-based DNA test is now possible.41 Liu et al.42 used magnetic resonance imaging to investigate the appearance of affected muscles in 17 patients with SMA (3 severe, 9 intermediate and 5 mild according to the Dubowitz classification). In the severe form, the authors found significant atrophy of all the affected muscles of the thigh and calf. In the intermediate form, ragged atrophy of the muscle bundles of the thigh and calf with selective sparing of the adductor longus muscle was observed. In the mild form, fatty infiltration of muscle bundles and increased intermuscular fat planes was detected.

The differential diagnosis should include all conditions that could manifest as a hypotonic baby. SMA should be distinguished from congenital myopathies where the electromyographic studies show myopathic findings. Hypotonic cerebral palsy or mixed forms with predominant flaccid paralysis have to be excluded. Neuromuscular conditions characterized by generalized muscle weakness, such as congenital muscular dystrophy or Duchenne muscular dystrophy also have to be ruled out.

Orthopaedic problems

Orthopaedic manifestations of the condition that may require treatment can occur primarily in patients with SMA type II or III who have a longer life expectancy. Scoliosis and hip subluxation or dislocation are the two most common orthopaedic problems encountered. In the study by Evans et al.9 scoliosis affected all patients with moderate to severe disease. Progressive hip subluxation and dislocation occurred in up to 50% of patients with more severe disease often after the age of 5 years.9

Hip and knee contractures may develop in children with SMA as a consequence of immobility. This poor lower limb function may accelerate the progression of the spinal deformity.43 Walking may be promoted by the early provision of lower limb orthoses and the use of a walking frame. The regular use of a standing frame can also retain some degree of muscular function in non-ambulatory children without severe fixed hip and knee contractures.

Patients with SMA type I are significantly hypotonic and have considerable respiratory and feeding difficulties leading to an early death. Orthopaedic treatment is rarely required and is mostly limited to range-of-motion exercises in order to prevent fixed joint contractures.44

In patients with SMA type II, subluxation and subsequent dislocation of the hip are caused by muscle imbalance and the inability to walk. The presence of muscular imbalance and the lack of mobility frequently produce a valgus deformity of the proximal femur, which progresses to
gradual displacement of the femoral head from the acetabulum. Unilateral hip subluxation results in pelvic obliquity that can contribute to the development of scoliosis. Passive stretching exercises and soft tissue releases may be required to treat hip deformity. Shapiro and Speccht have recommended performing a proximal femoral varus osteotomy to maintain hip alignment and restore a good sitting balance. Early attention should be paid to the hips in children where a long life expectancy is predicted, even though the incidence of re-operation is high in these patients. Other investigators have reported poor results following hip osteotomies in this patient population, or minimal clinical symptoms when no treatment has been offered.

In patients with SMA type III who are able to walk and have developed acetabular dysplasia with valgus deformity of the proximal femur, surgical reconstruction of the hip on the femoral side may further affect their poor abductor muscle strength and lead to loss of walking function. Periacetabular osteotomy has been recommended as an alternative to proximal femoral osteotomy in these patients in an attempt to restore congruity and containment of the hip joint and prevent further weakening of the hip abductors.

### Spinal deformity

**Epidemiology:** Spinal deformity in patients with SMA who survive early childhood constitutes a particularly common problem. There have been several previous studies to calculate the incidence of deformities of the spine in this group of patients. However, it is not likely that these studies have included the whole population of patients affected with SMA. A large number of children with SMA type I do not survive long enough for orthopaedic problems to require treatment and are not included in any of these reports. The development of spinal deformity should be anticipated in practically all patients who will have a life expectancy beyond the first few years of their life.

The reported incidence of scoliosis varies according to the length of clinical follow-up and the type of SMA; it ranges from 60% to almost 100%. In contrast, Benady recorded a relatively low incidence of scoliosis of 54% in a group of 46 patients with SMA. Seventeen of these 25 patients developed a curvature before the age of 4 years. Among the patients without scoliosis there were children with less severe SMA and a favourable expected survivorship; it is, therefore, likely that at least some of these patients will develop scoliosis in later life.

**Characteristics of the spinal deformity:** The collapsing long C-shaped scoliosis, typically seen in neuromuscular conditions, represents the most frequent pattern of spinal decompensation in children with SMA and is due to the inability of the trunk muscles to support the spine in the upright position. The curvature may involve a large segment of the spine with the apex located often in the thoracolumbar junction, extending into the pelvis and leading to the development of significant trunk and pelvic imbalance. Mertini et al. described the pattern of scoliosis in 40 patients with SMA. A single curve developed in 33 of these patients, with the most common type being a right thoracolumbar scoliosis (67%; 22 patients). An associated thoracic or thoracolumbar hyperkyphosis can occur in 30% of the patients. Primary thoracic hyperkyphosis and lumbar hyperlordosis have also been described with or without coronal spinal deformity.

The patient age at onset of scoliosis ranges from birth to adolescence. The age of onset, severity and progression of the scoliosis are related to the type of SMA and the extent of neurological involvement. The more severely affected the child, the higher the possibility of developing scoliosis at an earlier age and the greater the anticipated curve progression. The presence of familial involvement may also affect the severity and progression of the curve.

Riddick et al. reported on 36 patients with SMA whose ages ranged from 2 to 35 years and found an incidence of spinal deformity of 86%. Twenty-eight patients had a scoliosis or kyphoscoliosis, which was most commonly thoracolumbar (17 patients), and three patients had a thoracic hyperkyphosis. The average age at onset of the deformity was 6 years, ranging from 1 week to 12 years.

**Natural history:** The evolution of the scoliosis in this group of patients is characterized by an early onset with rapid progression and poor response to orthotic management, especially during the adolescent growth spurt. Scoliotic curvatures demonstrate a particularly higher incidence in children with more severe disease, directly proportionate to the extent of neuromuscular impairment and inversely proportionate to the ambulatory function.

The spinal deformity develops as a consequence of the generalized muscle weakness and the poor ambulatory function. Previous studies have reported an association between the poor level of motor function and the development of scoliosis in this group of children. The limited ability for these patients to mobilize is directly related to the extent of neurological disorder. Patients with SMA and mild scoliosis who lost ambulatory capacity and became wheelchair-bound showed rapid deterioration of their scoliosis.

In addition, scoliosis developed as soon as a previously ambulatory patient became wheelchair dependent. Aprin et al. reported in his group of SMA children an intervening time between loss of walking ability and the development of scoliosis of 2 years. Marked curve progression is also associated with periods of increased growth velocity.

Granata et al. studied the natural history of spinal deformity in 63 patients with mild and intermediate SMA. Twenty-five of 32 patients with mild and all but one patient with intermediate SMA had developed a scoliosis. The mean age at onset of scoliosis was 4.3 years for the intermediate group and 9.9 years for the group with mild SMA. A single curve developed in 74% of patients and this was most commonly thoracolumbar convex to the right. The authors found a linear correlation between the development of scoliosis and age in patients with the intermediate form and in those with the mild form of SMA who could not walk.

Rodillo et al. reviewed 63 patients, 37 with intermediate and 26 with mild SMA whose ages ranged from 2 to 26 years. In the intermediate form, all patients developed a scoliosis, which had an early onset and rapid deterioration. In the mild form, only 30% of the patients had a scoliosis that was progressive during puberty in those patients who had lost ambulatory function.
Hensinger et al.\textsuperscript{48} reported on 50 patients with SMA; 29 patients were found to have a significant scoliosis (58\%) and two patients had an increased lumbar lordosis without scoliosis. The average age of onset of the spinal curvature was 7.6 years ranging from birth to 13 years. In three patients, the scoliosis developed after loss of walking ability. In four cases the scoliosis was considered the cause of decreased ambulatory function.

Schwentker and Gibson\textsuperscript{37} reviewed the records of 130 patients with SMA and identified scoliosis as their major orthopaedic problem. Fifty patients were re-examined. Seven of 15 ambulatory patients had a scoliosis as opposed to 28 of 35 non-ambulators. The scoliosis in wheelchair-bound patients was more severe leading the authors to correlate the inability to walk with the severity of the scoliosis. They suggested that loss of ambulatory function was related to muscular weakness due to the underlying neurological disease rather than related to the spinal deformity. The pattern of scoliosis was analysed in 30 patients. Twenty-six were single curves, most commonly right thoracolumbar, and four were double curves.

Evans et al.\textsuperscript{9} reviewed 54 patients and reported an incidence of spinal deformity of 92\%. Thirty-nine patients had a single and eight had a double scoliosis. There were three patients with primary sagittal plane deformity. In this study, the more severe the inherent muscle weakness, the earlier the onset of the scoliosis and the more progressive the deformity. In patients with mild neurological involvement, progression of the scoliosis coincided with loss of ambulatory function.

**Functional impairment:** The development of a pathological spinal curve in addition to the underlying neurological disorder may significantly restrict the patient’s functional capacities and increase the need for nursing care. Ambulatory children may become confined to a wheelchair as the deformity progresses. Wheelchair-dependent patients gradually lose their ability to sit due to the severe trunk and pelvic decompensation and become hand-dependent sitters. The imbalance of the spine and the pelvis can result in the development of pressure sores. As the scoliosis progresses, it has an adverse effect on the respiratory function which also deteriorates and increases the risk for life-threatening complications.\textsuperscript{10,50}

### Treatment of patients with SMA

The treatment of SMA focuses on the prevention and management of medical complications, most commonly associated with poor respiratory function, and early interventions to correct musculoskeletal deformities in children with a favourable life expectancy. Despite the recent advances in our understanding of the genetic basis of SMA it remains difficult to predict which children that develop SMA at a young age will survive long enough to necessitate orthopaedic treatment. However, due to significant improvements primarily in respiratory support, prolonged survival should be anticipated for many patients with mild to moderate disease. Bach\textsuperscript{56} has recently reported the benefit on lung development and prevention of chest wall deformities by the use of nocturnal positive pressure ventilation.

The identification of the SMN1 and SMN2 genes and their protein product as part of the pathogenesis of SMA has resulted in the exploration of novel genetic treatments for the condition. As patients with SMA lack the SMN1 gene but maintain the SMN2, which produces at least some SMN protein, one possible therapeutic target is upregulating expression of the SMN2 gene. A number of compounds have been examined in an attempt to increase the expression of the SMN2 gene including valproic acid, sodium butyrate, phenylbutyrate, aclarubicin, and the aminoglycosides tobramycin and amikacin with encouraging in vitro results.\textsuperscript{57–61} Valproic acid has been tested in patients with SMA and has shown to increase SMN production in fibroblasts.\textsuperscript{57}

### Treatment of spinal deformity

#### Conservative management

There is no non-operative measure that has a documented effect on preventing scoliosis progression or the final outcome of the spinal deformity in children with SMA. The goal of all conservative modalities is not to correct the deformity but instead to retain function as the curve continues to progress and delay surgery for a later stage.

**Seating support:** Appropriate seating adaptations should be considered as the cornerstone of managing patients with scoliosis before surgical correction is indicated. These supports can be built on the patient’s wheelchair, which becomes the primary seating device for those children over time. They include offset chest lateral rests, shoulder harnesses and straps, and can significantly improve sitting balance and maintain an upright posture. Extended head rests are essential in order to provide adequate support in patients with poor head and neck control.

**Curve control-orthoses:** Bracing has also proved to be ineffective in controlling the deformity and does not seem to change the natural history of the scoliosis. Moreover, the traditional rigid thoracolumbosacral orthosis (TLSO) applied in the treatment of patients with idiopathic scoliosis can be restrictive for children with SMA who have respiratory compromise, feedings disorders, and occasionally skin problems.\textsuperscript{46,49} Alternatively, a soft brace or a bi-valved brace could be used whenever the patient is not using his adapted wheelchair to improve sitting position and facilitate activities of daily living. Previous authors have suggested that bracing can be useful as a measure to delay curve progression and increase sitting tolerance in patients with SMA.\textsuperscript{37,46,54}

Schwentker et al.\textsuperscript{37} reported on 23 patients with SMA treated with spinal orthoses. In many patients, bracing started before the spinal deformity developed with the aim to support the child in the sitting position. In these children, the initiation of brace therapy did not prevent the development of scoliosis. In 18 patients the scoliosis occurred or progressed after the brace was applied. In only...
three patients with moderate curves the deformity remained static during the period of orthotic treatment.

Riddick et al.\textsuperscript{[54]} treated conservatively 20 patients with SMA by using Milwaukee braces, total-contact underarm or sitting support orthoses, and serial casting. Their results were mostly discouraging on preventing scoliosis progression but allowed for a lower rate of curve deterioration in certain patients.

Aprin et al.\textsuperscript{[9]} presented 15 patients with SMA and scoliosis that were treated with a spinal brace; brace therapy was found to be uniformly ineffective in controlling the deformity. Orthotic treatment was discontinued in five patients because of respiratory complications. In the remaining 10 patients, the scoliosis progressed relentlessly and required spinal arthrodesis.

Evans et al.\textsuperscript{[9]} reported on the comparative use of four different types of orthoses in patients with SMA. The trunk and lower extremity sitting orthosis has been described as a moulded posterior plastic shell, which controls the patient from axilla to toes and is being used to support the young severely affected children. This orthosis can be extended behind the head and may have a removable anterior component. The authors found this orthosis to be well tolerated by children with SMA providing them with satisfactory trunk support and allowing them to be more interactive with their surroundings. In the same study, the Milwaukee brace could only be fitted in patients with the mildest forms of SMA. One of these children developed severe dental deformities as a consequence of the chin rest. A plastazote lined total-contact orthosis was used in 12 patients and was associated with rapid progression of the deformity. The thoracic suspension orthosis, described as a moulded body jacket suspended from a wheelchair was used in five patients with a good response in three of these children.

The stage at which bracing should be attempted still remains debatable. Schwentker et al.\textsuperscript{[37]} applied a brace before any spinal deformity developed. It has been suggested that the success of brace treatment is more likely in patients who can stand and walk more than in those who are non-ambulators.\textsuperscript{9,62} However, compliance can be more challenging in children who still retain good ambulatory function. Shapiro et al.\textsuperscript{[63]} proposed that bracing should be considered when the scoliotic curvature reaches 15–20° in order to slow progression of the deformity.

In our opinion, a bi-valved brace can be used in young patients with SMA and a mild scoliosis, which retains flexibility; the aim is to improve sitting balance when the patient is not using his adapted wheelchair, potentially delay deterioration of the deformity, allow further spinal growth, and postpone surgical treatment for a later age. Orthotic management in these children should be closely monitored and the underarm brace discontinued if respiratory compromise or chest wall deformities develop and if the patient’s compliance is poor. If the deformity progresses to more than approximately 50° or if it becomes rigid we believe that spinal arthrodesis should be considered.

**Surgical management**

*Spinal arthrodesis* with the use of instrumentation is the only treatment that has a well-established positive effect in children and adolescents with SMA. Rapid progression of the scoliosis and decompensation of the trunk leads to loss of function and cardiorespiratory impairment usually in an unexpectedly short period of time.\textsuperscript{9,12}

**Indications:** Spinal fusion is recommended in the presence of documented scoliosis progression and a curve size of between 40° and 60° in children ideally 10 years of age or older, especially if there is a recorded deterioration in their functional skills or their pulmonary capacities.\textsuperscript{9,55,63} At this size of deformity, an isolated posterior spinal arthrodesis should be sufficient to achieve satisfactory correction of the deformity without the need for additional anterior surgery or preoperative spinal traction. Other investigators have suggested earlier fusion for curves of approximately 35° or greater.\textsuperscript{51}

If the scoliosis is left untreated for too long a severe and fixed curvature develops and pulmonary function is likely to become further compromised. A combined anterior release and posterior instrumented spinal fusion will then be required to maximize flexibility of the curve and allow for increased correction; this is associated with significantly higher perioperative risks compared to the posterior-only procedure. Therefore, we believe that spine surgery should not be delayed even in the expense of growth if this delay will result in the necessity for an anterior release, which will cause substantial increase in the rate of perioperative life-threatening complications.

**Principles of surgery:** Restoration of coronal and sagittal trunk balance is considered equally if not more important than the percentage of scoliosis correction in patients with SMA. The spinal arthrodesis should be extended from the upper thoracic region, in order to prevent the development of recurrent proximal kyphosis, to the pelvis, especially if pelvic obliquity is present, if the curve involves the sacrum, or if the patient has poor sitting balance (Fig. 1). Progression of the deformity following short fusions both above and below the instrumented levels has been previously reported by Aprin and Dorr.\textsuperscript{9,64}

Long fusions to include the lumbosacral articulation are indicated even in the ambulatory children with SMA who have developed significant pelvic obliquity. Deterioration of the scoliosis and pelvic imbalance in these patients will lead to a gradual decline in the ambulatory function. It has been the authors’ experience that extension of the spinal arthrodesis to the sacrum does not significantly affect walking ability in most children, as long as a good coronal and sagittal spinal alignment has been achieved and the patients are mobilized with intensive physical therapy in the immediate postoperative period.

In the small cohort of patients with milder neurological involvement (usually type III SMA) and good functional skills where the scoliosis is associated with minimal or no pelvic imbalance we are performing spinal arthrodesis from the upper thoracic to the lower lumbar spine (T1–T2 to L4–L5 level) sparing the lumbar sacral joint in order to maintain some degree of spinal flexibility. In these less severely affected patients, who most commonly have a well-preserved walking ability, the iliac crest has good bone quality and can be used as bone graft.\textsuperscript{65} The use of autograft bone harvested from the iliac crest in these children increases the rate of fusion and prevents the development of a non-union. The absence of significant osteopenia in the
vertebral bodies also allows for the use of third generation instrumentation systems with segmental pedicle screw fixation as opposed to sublaminar wires to correct the spinal deformity (Fig. 2).

Children with SMA and severe neurological disease occasionally have considerable weakness in their neck muscles. This weakness can be aggravated following spinal fusion to T1 or T2 and the patients may develop significant

Figure 1  Preoperative anteroposterior (A) and lateral (B) radiographs of the spine of a boy aged 13 years and 8 months with a left thoracolumbar scoliosis measuring 79° with 39° of associated pelvic obliquity (black line) and increased thoracolumbar kyphosis. The patient underwent a posterior spinal arthrodesis using segmental instrumentation and allograft bone extending from T1 to the sacrum with pelvic fixation. This resulted in a very satisfactory correction of his scoliosis, which measured 22° at 2 years follow-up, levelling of the pelvis and restoration of a normal sagittal balance (C,D).
neck pain. The way to overcome these symptoms is to engage these children post-surgery on a regular physical therapy programme, which should focus on strengthening their neck muscles. Occasionally, the use of a supportive soft neck collar as a temporary measure can alleviate the neck discomfort. In extreme cases, where the child has a scoliosis and very poor head control, extension of the spinal arthrodesis proximally to the occiput can be considered. However, that increases the risk of pseudarthrosis and requires an instrumentation system with tapered rods and occipital plate fixation.

Anterior Surgery: Anterior spinal release can be used to allow for better scoliosis correction in patients with rigid deformities. However, the severe risk of consequent respiratory compromise following a thoracotomy or a thoracoabdominal approach to the spine suggests that an anterior spine arthrodesis should only be attempted in patients with mild disease and good preoperative pulmonary function. The need for postoperative tracheostomy in this group of children has been previously related to the use of an anterior spinal operation. As the severity of the scoliosis is directly associated with the degree of neurological involvement, it is unlikely that patients with significant curves, who would be candidates for an anterior release, will have sufficiently preserved respiratory reserves to survive this procedure.
Anterior spinal instrumentation alone provides stabilization of a short segment of the spine and is, therefore, not indicated in patients with SMA. An isolated anterior instrumented fusion cannot correct pelvic obliquity and has a high risk of proximal and distal add-on or residual deformity. Anterior instrumentation can be also used after the multi-level discectomies as the first stage of a combined anterior and posterior spinal fusion; this is then followed by an instrumented posterior spinal arthrodesis. In the vast majority of our patients we do not use anterior instrumentation when we are performing the anterior release. We have found that in these severe deformities, a greater degree of correction can be achieved with the use of segmental posterior instrumentation following a circumferential spinal release, which includes complete anterior discectomies and anullectomies, as well as extensive posterior facetectomies and capsullectomies at every level from T1 to the sacrum.

Preoperative assessment: Surgery to correct scoliosis in children with SMA is a significant undertaking and carries considerable risks. These patients may have a number of concomitant medical problems, which will need to be addressed before surgical management is considered. A multi-disciplinary approach will be required and the expert input from a wide group of specialists is indispensable to optimize the patients’ medical condition prior to surgery and minimize the potential for complications. The spine surgeon should not only limit his role to performing the operation; he needs to recognize that much of the success of the surgical procedure depends on a good coordination of the actions of the medical, nursing and therapist teams. In our experience, the involvement of dedicated anaesthetists, paediatric neurologists, cardiac and respiratory physicians, gastroenterologists, dieticians, physiotherapists, occupational therapists, and intensive nursing care in the perioperative management of these children is essential for a favourable outcome. A thorough preoperative evaluation of the patients is routinely performed, including monitoring of their respiratory capacity, cardiac function, immune system, coagulation mechanisms, nutritional status, urinary system and their overall level of functional deficiencies.

Respiratory: The major limiting factor for scoliosis surgery in children with SMA is their respiratory function. Pulmonary function at the time of surgery may already be significantly impaired and this is an essential prognostic parameter for postoperative respiratory complications. \(^{52,67}\) Patients with a vital capacity of 50% or less of predicted value are at risk of developing pulmonary edema or atelectasis. \(^{67}\) In the same study, patients with a vital capacity of more than 60% did not have respiratory complications after scoliosis surgery. \(^{65}\) Patients with a vital capacity of less than 25% are at significant risk of developing life-threatening pulmonary complications. \(^{64}\) In this case, the risk of death involved in the procedure possibly outweighs the gains from correcting the deformity and these patients are not considered by most surgeons as adequate surgical candidates.

Rodillo et al. \(^{50}\) reported on treating surgically patients with SMA and a vital capacity of less than 35% of predicted with a good outcome. Over the last 2 years we have performed posterior spinal fusion in a number of children with SMA whose vital capacity was as low as 20% of predicted with encouraging results (Figs. 3 and 4). Our patients were kept intubated for 24–72 h following surgery and were then placed electively on assistive ventilation through a C-pap or bi-pap machine for a period varying from a few days to a few weeks. With the provision of positive pressure ventilation, aggressive respiratory physical therapy to clear secretions and prevent the development of chest infections, and patient mobilization on their adapted wheelchair soon after surgery all our patients survived the procedure and had a very satisfactory outcome. Most patients with a vital capacity of 25% or above of predicted did not require assistive ventilation long-term. We believe that scoliosis surgery should not be attempted in patients with respiratory function of less than 20% who are at the greatest risk of postoperative death. \(^{69}\)

The preoperative application of a halo-femoral traction has been suggested as a measure to achieve improvement of the respiratory function (less than 10%) in children with SMA. \(^{69}\) However, this should be weighed against the associated morbidity and the poor patient compliance. We do not perform halo-femoral traction at our institution as we have not found it useful and young patients do not tolerate it well.

Nutritional: Malnutrition is a common finding in children with SMA. This can be attributed to the combination of a poor diet and a high metabolic demand due to recurrent chest infections or other medical illnesses. In addition, the neurological condition may alter normal metabolic pathways. \(^{70}\) Muscle function is a critical nutritional reserve for protein, carbohydrate and mineral metabolism. Reduction of muscle mass may lead to loss of this nutritional reserve and limit the patient’s ability to adjust to simple nutritional changes such as overnight fasting. Fatty acid metabolism may be affected and patients with SMA may have a lower dietary fat tolerance; conversely a high carbohydrate diet (the result of a low fat diet) may place additional stress on the respiratory system by increasing carbon dioxide production.

Poor nutrition predisposes to delayed wound healing and a poor immunological response to infection. Spine surgery should be postponed in the malnourished patient; supplemental nutrition in the preoperative period may be necessary to optimize a surgical candidate. Soon after the procedure, we place our patients on total parenteral alimentation or nutritional supplementation through a nasogastric tube until they can feed orally.

Bleeding: A considerable amount of intraoperative blood loss should be anticipated during scoliosis correction in children with SMA. Bone quality is often inherently poor, especially in the non-ambulatory patients because of disuse osteopenia. Poor bone quality increases the risk of intraoperative blood loss and instrumentation failure during spinal fusion. Preoperative management of osteoporosis with the administration of intravenous bisphosphonates might be a consideration, especially in non-ambulators with marked osteoporosis, to maximize their bone quality before spine surgery.

When these children are scheduled for spinal arthrodesis there must be at least one blood volume of blood typed and cross-matched. The use of a cell saver system during the procedure may limit the need for transfusion. Controlled
hypotensive anaesthesia, normovolemic haemodilution and meticulous haemostasis during tissue dissection are required to reduce blood loss. Early administration of fresh frozen plasma should be considered in the presence of a gradually increasing intraoperative blood loss, especially in patients with identified coagulopathies or those with severe neurological involvement. Recently, aprotinin has been reported to reduce blood loss during spinal surgery in this group of children.

During surgery, transfusion of a significant volume of packed red cells and clotting factors may be required. There are many problems associated with large-volume blood transfusions, which include anaphylaxis and lesser febrile reactions, hyperkalemia, hypocalcemia, and hypothermia. A consumptive coagulopathy (disseminated intravascular coagulation) may be the result of large volume transfusions and must be avoided with the appropriate administration of clotting products.

**Surgical considerations**

**Historical perspective:** The introduction of Harrington instrumentation provided a tremendous evolution in the operative management of these complex deformities, but the incidence of pseudarthrosis and consequent curve...
progression remained high. Harrington instrumentation was used in the surgical management of neuromuscular scoliosis until the late 1970s. The distraction techniques applied through the Harrington system failed to restore normal sagittal spinal balance and were often impractical in the soft bone of patients with SMA. In addition, postoperative immobilization of the spine in a cast was required and that could result in further compromise of the respiratory function.

In 1977, Luque developed the concept of segmental spinal fixation with the application of translational corrective forces through the use of multiple-levels sublaminar wires and two single rods. His technique achieved a wide distribution of forces over each vertebra, with an increased initial spinal stabilization and a low risk of instrumentation failure. This led to a greater degree of correction and decreased the risk of implant related complications. However, later reports indicated a high rate of pseudarthrosis associated with the Luque system of up to 10%, instrumentation-related complications up to 21%, and curve progression postoperatively in up to 30% of the cases.

Figure 4 Preoperative anteroposterior (A) and lateral (B) radiographs of the spine of a girl aged 11 years and 7 months with a very severe right long thoracic scoliosis measuring 115°, 21° of associated pelvic obliquity (black line) and increased thoracic kyphosis measuring 100°. Due to her poor respiratory function (vital capacity of 24% of predicted) the patient underwent a posterior spinal arthrodesis using segmental instrumentation and allograft bone extending from T2 to the sacrum with pelvic fixation. This resulted in a satisfactory correction of her scoliosis, which measured 53° at 2 years follow-up, marked reduction of her pelvic obliquity (black line), which measured 10°, and restoration of a normal sagittal balance (C,D). At discharge, the patient did not require assistive ventilation.
The Galveston technique of intramedullary placement of the rod in the iliac bed was developed by Allen and Ferguson, and accomplished a secure pelvic fixation. The combination of the Luque wiring technique and the Galveston intrailiac rod fixation resulted in fewer instrumentation-related complications. 

However, it was soon recognized that the two unconnected soft rods introduced by Luque were moving independently, which was the reason why they failed to provide adequate stabilization. The use of postoperative immobilization did not prove to resolve the problem. The Unit rod, a further development of the Luque–Galveston technique, accomplished the requirement for rigidly connected rods and has been widely used in North America in the treatment of neuromuscular scoliosis. This instrumentation technique has provided improved correction of the spinal deformity in both the coronal and sagittal planes, as well as balancing of the pelvis.

**Newer techniques:** More recently, newer instrumentation systems using primarily pedicle screws have been introduced in the management of neuromuscular scoliosis. These are based on the same principle of segmental fixation with pedicle screws or hooks instead of sublaminar wires and the alternative fixation of iliac bolts for lumbo-pelvic or sacroiliac plates for sacro-pelvic fixation. A common problem in the application of third generation instrumentation that uses pedicle hooks or screws in patients with SMA and marked associated osteopenia is poor vertebral fixation, which can limit the ability to perform corrective maneuvers and may increase significantly the risk of pseudarthrosis. It is our experience that in this group of children with considerably poor bone quality, the lamina provides the strongest point of fixation compared to the pedicle or the vertebral body and can withstand segmental translational forces applied through sublaminar wires that can achieve correction of the deformity and balancing of the spine in both the frontal and lateral planes. On the contrary, if repeat surgery is required, for example to address a non-union, it is technically considerably easier and safer to revise an instrumentation system that uses pedicle screws with or without hooks as opposed to sublaminar wires.

As these modern techniques become common and widely utilized, it is essential to establish benchmarks for degree of deformity correction and complication rate related to the technical aspects of the individual procedure. As third generation spinal instrumentation is being widely utilized, there is also a significant increase in implant cost; this has to be balanced against possible benefits obtained by the use of these latest techniques.

**Surgical technique used by the authors**

As previously mentioned, anterior release is performed in very few selected patients with severe scoliosis and increased curve rigidity who retain satisfactory pulmonary function. We perform the anterior procedure in the lateral decubitus position, using a thoracic or thoracolumbar retroperitoneal approach, through a removed rib, depending on the apex of the deformity and the levels of anterior release that are required. The anterior approach allows for an extensive release of the anterior longitudinal ligament, complete annulectomy and discectomy, with the intention to provide angular and rotational mobility of the spinal segments, while at the same time enhancing anterior fusion in the excised disc spaces with the application of morselized rib graft. Anterior instrumentation is very rarely used.

Once the anterior procedure is accomplished, the patients who are in a good general medical condition to tolerate the second stage procedure under the same anaesthetic are immediately rolled into the prone position and a posterior fusion with instrumentation is then performed. For most of our patients we select to stage the anterior and posterior procedures under separate anaesthetic sessions. In this second group, after the anterior stage is completed, the patients are taken to the intensive care unit (ICU), where hyperalimentation and aggressive pulmonary care are initiated until the posterior surgery 7–8 days later. The chest drain is removed when the drainage is less than 100 ml/day, usually on the third or fourth day after surgery.

A posterior instrumentation system, which includes two independent rods and multi-level sublaminar wire fixation, is being used in all patients to apply translational corrective forces and provide segmental stability, with the spinal arthrodesis routinely extending from T1 or T2 to the sacrum with pelvic stabilization using the Galveston technique (Fig. 1). Distally, iliac screws are placed intramedullary in the iliac bed and these are attached to the bilateral rods using lateral connectors. Cross-connectors are bridging the two rods proximally and distally to create a rectangular frame and increase stability of the construct with the aim to prevent vertical movement between the two independent rods.

Decortication of the transverse processes and lateral laminae with extensive excision of the facet joints and their capsule is performed at every level and the posterior instrumentation is always supplemented by abundant allograft bone mixed with autogenous graft harvested from the spinous processes. Iliac crest bone is not obtained in these patients due to various degrees of osteopenia. Wound drains are not routinely placed and the lumbosacral fascia is closed meticulously in order to obliterate dead space and aid wound healing.

All patients receive prophylactic antibiotic treatment with the administration of a first-generation cephalosporin immediately before and for 24 h after surgery. Before anaesthesia induction, arterial and central venous lines are placed, and the central venous line is maintained until the second stage in patients who will require a two-stage procedure. A nasogastric tube is used to decompress the stomach and a Foley catheter to monitor urinary output. Cell-saver is used intraoperatively, and our patients receive homologous blood transfusions at the discretion of the anaesthetist. Spinal cord monitoring with the use of somatosensory evoked potentials is used during surgery in all patients. Motor-evoked potentials are recorded only in patients with ambulatory function.

**Postoperative care:** The most important concern after scoliosis surgery is respiratory care. Coughing ability decreases following the operation and this along with the restriction of the patient’s mobility increases significantly the risk of atelectasis and pulmonary infection. Patients with severe neurological involvement and a decreased vital
capacity often require ventilatory support (C-pap or bi-pap) or endotracheal intubation. Close monitoring by a respiratory physician is essential as patients may have to remain for several days or even weeks on non-invasive ventilation. If the previous measures prove ineffective, tracheostomy can be performed to provide pulmonary support for a longer time. Intensive respiratory physiotherapy and early mobilization of the patient are also fundamental to prevent pulmonary complications.

Nutritional supplementation is initiated soon after surgery in the form of nasogastric feedings or total parenteral alimentation in order to provide adequate dietary coverage until the patients can feed orally.

We do not use postoperative immobilization or external trunk support in our patients. The patients are mobilized early to an upright position in their wheelchair and are engaged in an intensive physical therapy programme. Their wheelchair is modified to adapt with the corrected seating posture. A reclining wheelchair has to be used for the initial mobilization following spinal instrumentation to the sacrum and the pelvis; an upright sitting position can be usually achieved gradually within the next few days.

Postoperative complications: Scoliosis surgery in children with SMA is associated with complication rates as high as 45%, which are more prevalent in older patients and in those children with more severe deformities. Patients with extensive neurological deficits can be particularly sensitive to analgesic medications, such as opioids, that suppress respiratory function. Respiratory insufficiency, atelectasis, and pulmonary infections are the most common complications following surgery. Other reported complications include rupture of the diaphragm, pulmonary embolism, acute gastric volvulus, and narrowing of the diameter of the chest as a consequence of cast immobilization.

Complications related to the technical aspects of the surgery include neurological damage to the spinal cord and the nerve roots, failure of the spinal instrumentation and subsequent pseudarthrosis, which can occur with an incidence ranging from 6% to 28% of the patients, wound infection, and postoperative urinary tract infection.

Outcome after spinal fusion: Spinal arthrodesis in children with inherent neurological impairment constitutes a major physical insult and can be associated with an initial decline in functional skills in some of these patients. This initial deterioration in motor activity can be reversed with intensive physical therapy and should be weighed against the loss of function that would occur if the deformity was left untreated due to further progression. In previous reports, the negative impact of the scoliosis surgery on function has improved 5 years after the operation. Other investigators have reported improvement in function following spinal fusion. Scoliosis correction has been associated with an 86% patient satisfaction rate; this was mainly due to a better sitting balance, which allows the patients to use their upper limbs for functional tasks other than supporting their trunk in the upright position.

Conclusions

The development of spinal deformity constitutes the most common musculoskeletal problem in SMA and should be anticipated in almost all the patients who survive beyond early childhood. The development of an abnormal spinal curvature in this group of severely disabled children decreases their sitting tolerance, creates pain from impingement of the ribs against the pelvis, and accelerates deterioration in respiratory function. Scoliosis surgery in paediatric patients with SMA who have severe neurological compromise and complex medical co-morbidities is associated with significant technical difficulties and an increased risk of life-threatening complications related primarily to their poor pulmonary capacity. However, there is a well-documented positive impact on these children by correcting their spinal deformity in performing activities of daily living. With recent advancements in medical management and a multidisciplinary approach, life expectancy for this population of patients can be longer than previously reported. Operative procedures to restore the balance of the spine and the pelvis have a definitive effect in improving the patients’ quality of life, preserving function, and prolonging their survivorship.

References


