Neuromuscular Scoliosis: An Overview

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Contents

Key Points

- Progressive neurologic and muscle diseases can cause progressive and severe scoliosis.
- These diseases commonly affect the pulmonary and cardiac systems, which need to be considered when managing the scoliosis.
- Early intervention in Duchenne muscular dystrophy is recommended to optimize cardiac and pulmonary function.
- Pelvic obliquity is commonly associated with scoliosis, and in many cases, pelvic fixation is recommended.
- Segmental fixation is recommended in the osteopenic bone commonly found in patients with neuromuscular scoliosis.
- Different methods of treating early-onset neuromuscular scoliosis are being evaluated including growing rods (traditional growing rods [TGR] and magnetically controlled growing rods [MCGR]), vertically expandable prosthetic titanium ribs (VEPTR), and Shilla technique, but continued studies are needed to determine their true efficacy.

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12.1 Introduction

Scoliosis is a common finding among patients with neurologic or muscular diseases (Table 12.1). The spinal deformity in this population is often progressive and resistant to

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Table 12.1 Common diagnoses associated with neuromuscular scoliosis

Cerebral palsy[a]
Spinal muscular atrophy[a]
Myelomeningocele[a]
Duchenne muscular dystrophy
Arthrogryposis multiplex congenita
Central core disease
Nemaline myopathy
Friedrich ataxia
Spinal cord injury
Myotubular myopathy
Rett syndrome

[a]Will be discussed in more detail in subsequent chapters

nonoperative management. Progressive deformity can lead to a variety of challenges in the care of these patients, including difficulty with sitting balance or a decline in cardiopulmonary function. Surgical management often requires a multidisciplinary approach for preoperative medical optimization. Risks of surgery are generally higher in this patient population than in idiopathic scoliosis. However, appropriate planning and achievement of a balanced spine over a well-positioned pelvis can lead to good outcomes in the majority of patients.

Some common causes of neuromuscular scoliosis will be discussed in other chapters. However, there are many nuances in the care, both operative and nonoperative of other common neuromuscular scoliosis etiologies, such as Duchenne muscular dystrophy (DMD), arthrogryposis, Rett syndrome, and congenital myopathies. It is important to consider all the evidence in treating patients with specific neuromuscular conditions.

12.2 Duchenne Muscular Dystrophy

Duchenne muscular dystrophy (DMD) is an inherited X-linked recessive disorder isolated to the dystrophin gene causing an absence in the protein dystrophin [1]. DMD is usually first diagnosed by 5 years of age. Initial concerns by parents include delayed walking, clumsiness, or flat feet. It has been suggested to screen any male not walking by 18 months for DMD [2]. Later concerns by parents, usually at 4 or 5 years of age, are the inability to keep up with peers or increased difficulty climbing up stairs. Other clinical findings seen on examination include pseudohypertrophy of the calves, proximal muscle weakness, Achilles and iliotibial band contractures, and a positive Gowers' sign.

In the evaluation of DMD, the initial laboratory test evaluates serum levels of creatine phosphokinase (CPK). The diagnosis is then confirmed by genetic testing. In the remaining one-third of patients, a muscle biopsy is needed to specifically assess the quantity and quality of dystrophin present.

12.2.1 Spinal Deformity

Spinal deformity is the most critical orthopedic issue for the patient with DMD. The incidence of scoliosis is approximately 95%. In patients not medically treated, the onset of spinal deformity usually occurs at the same time that patients lose the ability to walk between the ages of 10 and 14 years. Early onset scoliosis (EOS) is exceedingly rare in patients with DMD. The risk of progression of scoliosis is also very high. Smith et al. reviewed the natural history of 51 patients with DMD and scoliosis, who had no surgical treatment and were followed until their death [3]. Seventeen of these patients had curves greater than 90° (33%). The mean rate of progression was 2.1° per month. In many cases, the curves continued to progress until the rib cage contacted the ilium.

The spinal deformity associated with DMD differs from the deformity seen in adolescent idiopathic scoliosis (AIS) as the rate of progression is greater [4, 5]. Unlike the typical hypokyphotic or lordotic patient with AIS, most progressive scoliosis in DMD patients is kyphotic in the sagittal plane. Wilkins and Gibson suggested two types of spinal deformity in DMD [6]. The more stable deformity is associated with an extended position, while the unstable pattern is characterized by a progressive kyphosis [6, 7]. Oda et al. also utilized sagittal alignment to help differentiate the deformity in DMD into three types, recommending surgery for the kyphotic deformities [8].

Considering that scoliosis typically develops once the patient becomes wheelchair bound, screening is not required while the patient is ambulatory. However, once the patient is unable to walk, radiographic screening should occur every 6 months.

12.2.2 Medical Considerations

In addition to the orthopedic manifestations, there are considerable medical complications associated with DMD. Of these, the problem that is most concerning for the spine surgeon is progressive worsening of pulmonary function. Muscle weakness, contractures, and spinal deformity result in a restrictive disease pattern. This progressive decline typically occurs in the second decade of life, worsens with increasing age, and ultimately leads to the patient's death [9–11].

Kurz et al. demonstrated that age and curve severity negatively affect pulmonary function [4]. Peak forced vital capacity (FVC) occurred at the same time that patients became unable to stand. Each year following then resulted in a FVC decline of 4%. If the patient developed scoliosis, an additional decline of 4% occurred for every 10° of thoracic scoliosis. A study by Yamashita et al. also supported the relationship of scoliosis and decreased pulmonary function [12].

Since age and thoracic scoliosis were found to be the best predictors of pulmonary decline, Kurz et al. recommended early surgical intervention in the DMD patient [4]. Others have also made similar recommendations. Galasko et al. demonstrated slightly improved survival and maintenance of FVC for the first 36 months postoperatively in those patients who underwent surgery [13]. Rideau et al. found static vital capacity at 2 years in five surgically treated DMD patients [14]. Velasco et al. supported spinal stabilization, demonstrating a significant decrease in the rate of respiratory decline postoperatively compared with presurgery rates [15].

Some authors have contradicted the positive effects of surgery on pulmonary function. Their studies found no significant difference between the surgical and nonsurgical group in terms of declining respiratory function [16–18]. Kennedy et al. demonstrated a similar decline in FVC of 3–5% per year in both operative and nonoperative patients [19]. The criticism of this study was that the surgical patients had severe scoliosis with pulmonary function too poor to benefit from surgery [20]. A 2020 study by Farber et al. demonstrated a loss of mean FVC from preoperative to postoperative testing in seven patients with DMD by 0.36 L [21]. Saito et al. suggested that more direct measures of respiratory muscle strength, such as maximal inspiratory pressure, maximal expiratory pressure, or sniff nasal inspiratory pressure, may more sensitively demonstrate respiratory function benefit from scoliosis surgery in DMD [22]. Chua et al. evaluated 29 patients with more than 10 years of follow-up and found that the apical vertebrae of scoliosis did not demonstrate a significant effect on the pulmonary function, nor did scoliosis surgery decreased the frequency of chest infections [23]. A Cochrane Review by Cheuk et al. was unable to give an evidence-based recommendation regarding the effect of surgery on pulmonary function since no randomized controlled clinical trials were performed [24].

Prior to any spinal surgery, preoperative pulmonary function tests should be performed. Common postoperative problems encountered include prolonged intubation and the need for permanent tracheotomy. Recently, studies have suggested that with aggressive postoperative pulmonary management, patients with low FVC can successfully undergo spinal fusion [25, 26]. Of the 45 patients prospectively collected, Harper et al. found no difference in outcomes between patients with a FVC greater than 30% compared with those less than 30%, and that the use of BIPAP postoperatively, even in those without a low FVC, may decrease the time of ventilatory support without increase in re-intubation rate. We recommend that if spinal fusion is being considered, early intervention should be performed before further decline in pulmonary function. A short duration of ventilatory assistance followed by early extubation and aggressive pulmonary management minimizes the risk of atelectasis and pneumonia.

Patients with DMD should also undergo a preoperative cardiac evaluation including echocardiogram. Cardiac manifestations include cardiomyopathy and conduction abnormalities [16, 27, 28]. In those patients with severely reduced cardiac function that cannot be controlled pharmacologically, surgery may not be an option.

Similarly to other myopathies, there is an increased risk of malignant hyperthermia in DMD [29, 30]. In extreme cases, patients have died intraoperatively from sudden cardiac arrest. Typically, anesthesiologists refrain from using anesthetics that trigger malignant hyperthermia in this population. Awareness of the risk will maximize the preparedness of the entire team for these medically complicated patients. Additionally, Duckworth et al. demonstrated that patients with DMD had an increased overall rate of postoperative complications than those with other neuromuscular condition, including deep wound infection and hepatotoxicity, which appeared to be unique to DMD patients [31].

12.2.3 Nonsurgical Management of Scoliosis

Spinal deformity in DMD rarely develops in the ambulatory patient. Therefore, close screening of these patients should begin when the patient begins using a wheelchair fulltime. In those rare cases when scoliosis develops in an ambulatory patient, bracing should not be utilized. Evidence suggests that bracing in this situation is ineffective and may decrease the ability to walk [20]. For the nonambulatory scoliosis patient, bracing has also been discouraged. Previously published reports have shown that while there is a decrease in the rate of progression, orthotics do not prevent the development of severe scoliosis [2, 32].

Since Drachman et al. demonstrated positive outcomes with the use of steroids for the treatment of DMD, there has been much effort to investigate their effects on scoliosis [33]. Corticosteroids have been found to stabilize muscle strength in DMD for a period of time [34]. Shapiro et al. studied 88 patients who were not treated with steroids at the time they became wheelchair-dependent and noted that scoliosis was found in 85 of 88 patients (97%), with a major curve greater than 30 degrees in 75% of involved patients. Sagittal plane deformity was seen in 37 of 60 patients(62%) [35]. A recent Cochrane Review found evidence to support the use of steroids to improve muscle strength and function in the short term (6–24 months) [36]. However, it is not completely clear whether this has an effect on the progression of scoliosis. Some studies have suggested that similar to bracing, steroids can delay or limit the progression of scoliosis [37, 38–42]. A previous prospective study compared 30 DMD patients treated with deflazacort with 24 matched control patients, and while they suggested that steroids slowed the progression of scoliosis, they were unable to demonstrate the pre-

vention of spinal deformity requiring surgery [38]. More recently, Lebel et al. compared long-term follow-up of ambulatory DMD patients receiving deflazacort versus those not receiving the glucocorticoid [43]. At long-term follow-up, they found that 20% in the deflazacort group had developed scoliosis compared to 92% in the nontreatment group. Koeks et al. published a study of 5345 patients with DMD from a global database and reported that patients treated with corticosteroids were less likely to require scoliosis surgery with statistical significance [44]. A recent multicenter clinical trial by McDonald et al. compared treatment with either deflazacort or prednisone in ambulatory patients with DMD over 48 weeks and found a lower rate of functional decline in patients treated with deflazacort [45]. The use of steroids, however, has to be balanced with the potential complications, including weight gain, behavioral problems, fracture, glucose intolerance, gastrointestinal symptoms, skin changes, and cataracts [34, 36, 46] (Fig. 12.1). Future directions for medical management of DMD include gene therapy aimed at restoring the expression of dystrophin [47].

12.2.4 Surgical Management of Scoliosis

Given that early EOS is rare in DMD, posterior spinal fusion (PSF) and segmental spinal instrumentation (SSI) are the

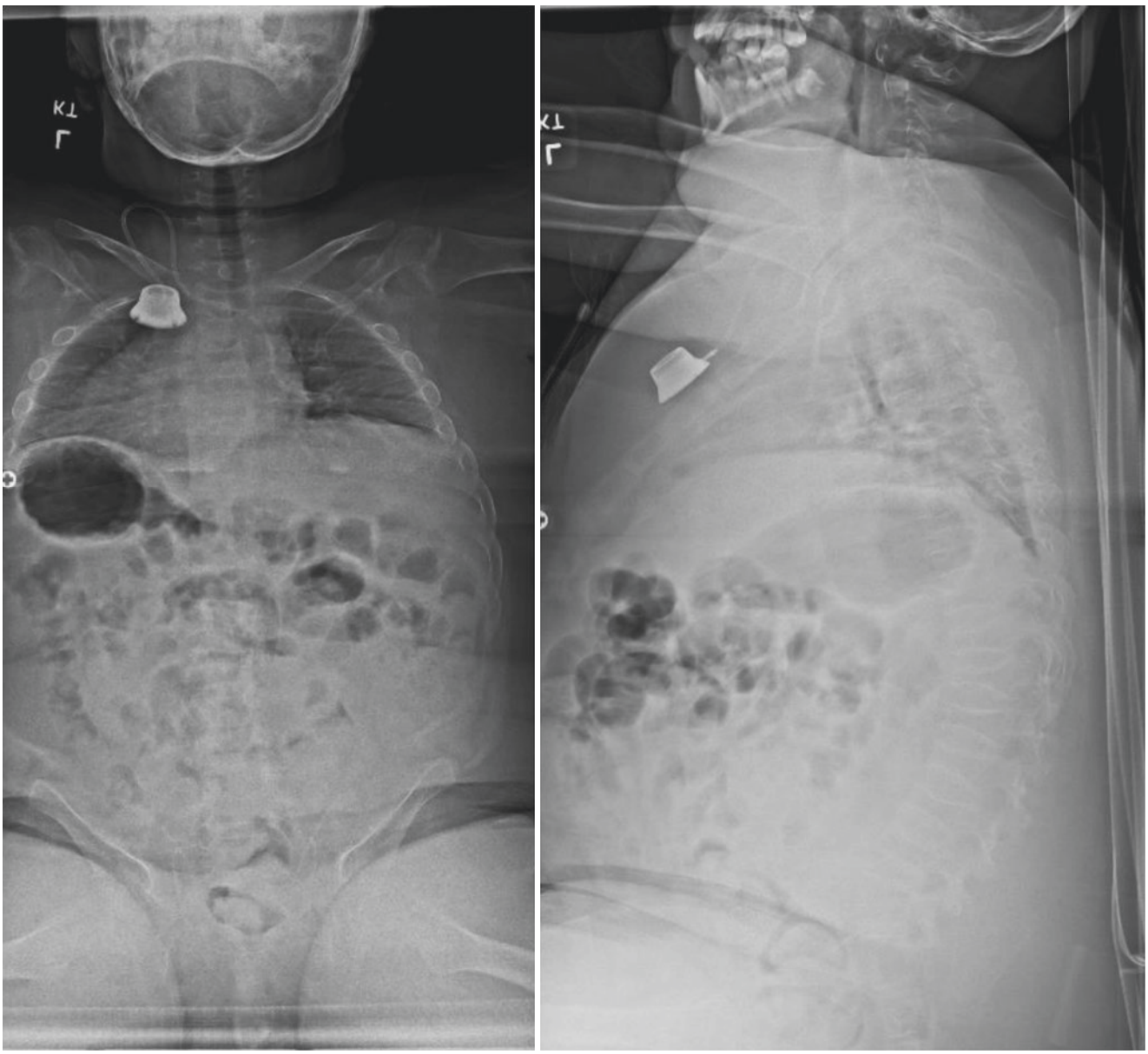

Fig. 12.1 A 14-year-old male with DMD with multiple chronic compression fractures secondary to chronic steroid use

standard surgical treatments for scoliosis in DMD. For a patient with documented progressive scoliosis that can tolerate surgery, there is little controversy over the need for surgical stabilization. The goals are to maintain sitting balance and patient mobility and minimize the effect of scoliosis on pulmonary function. Due to the high likelihood of developing scoliosis, some authors have suggested proceeding with surgery when patients lose the ability to ambulate [3]. Choi et al. recently demonstrated as well that even after loss of ambulation, there is a period of maintained flexibility with full reducibility of the curve remaining possible, but that the curve does become structural over time [48]. Most authors recommend surgery with radiographic evidence of scoliosis with major curves of 20–30° [20, 49–51].

With the development of segmental spinal instrumentation (SSI) by Luque, there have been major improvements in the surgical stabilization of DMD spinal deformity [52–54]. SSI has improved fixation in otherwise osteopenic bone and has minimized the need for prolonged immobilization.

There is little controversy where the fusion should begin, typically in the upper thoracic spine at T2 [11, 20, 50]. Stopping short of this may allow for cephalic progression of the curve due to progressive trunk and neck muscle weakness, causing the patient to lose head control. The ideal caudal extent of the fusion, however, continues to be debated. Fixation to the pelvis is technically more demanding, and increases both operative time and the potential risk of complications [55, 56]. Some studies have suggested that spinal fixation to L5 was sufficient in the early-treated patients and minimal pelvic obliquity [50, 51]. Other studies have recommended fusing to the pelvis at the initial time of surgical intervention [57–63]. Patients are healthiest at the first surgery. Any attempts to later fuse to the pelvis in those that have progressive pelvic obliquity will pose a greater risk with their worsening medical condition. Alman and Kim reported on 48 DMD patients who underwent spinal fusion [57]. Thirty-eight patients with less than 10° of pelvic obliquity and 40° curvature underwent fusion and instrumentation to L5. Of these patients, 32 had progression of their pelvic obliquity. They found that curves with an apex below L1 were at the greatest risk of progression. Therefore, Alman and Kim recommended fusion to the pelvis for all curves with an apex below L1. Spinal fusion in the thoracic and lumbar spine has traditionally been achieved with the use of sublaminar wires. With advancements made in instrumentation, some have chosen to use hooks or pedicle screws for the stabilization of the deformity. Recent studies involving pedicle screw fixation have demonstrated improved major curve correction, as well as improved and maintained pelvic obliquity in patients with DMD [64–68]. Another study reported improved patient function, sitting balance, and quality of life with pedicle screw constructs [69].

Selection of implants is related to surgeon preference, cost, deformity, and patient anatomy and is beyond the scope of this chapter. Currently, our preferred technique is to utilize pedicle screws throughout the construct. Density of implants will be determined by bone density, deformity magnitude, and spinal stiffness (Fig. 12.2).

There are similar choices for instrumentation to the pelvis. Options include the Galveston techniques with either Luque or Unit Rods, Dunn-McCarthy technique with an S-rod, sacral screw, and iliac screw fixation [56, 58, 70–72]. Each has advantages and disadvantages. The Galveston technique is subject to loosening and migration of the rod [11]. In addition, the Galveston technique sometimes requires complex three-dimensional contouring to fit the altered pelvic anatomy. Iliac screws, on the other hand, are placed individually into each iliac wing and then connected to the rod through connectors. A recent study by Peelle et al. demonstrated equal effectiveness in controlling pelvic obliquity between the Galveston technique and iliac screw fixation [72]. Our current preferred method is to utilize sacral-alar-iliac fixation as described by Sponseller et al. when instrumenting to the pelvis in DMD patients [73].

Another important consideration in the preoperative planning for scoliosis surgery is the risk of significant blood loss. Of all pediatric spine surgeries, Duchenne muscular dystrophy has demonstrated, on average, to have the highest mean blood loss [74, 75]. This is important considering the poor cardiac reserve in this population. These patients require a large exposure from the upper thoracic spine to the lower lumbar spine or pelvis. Proper positioning is crucial to avoid abdominal compression which can exacerbate blood loss [76]. The paraspinal muscles can be difficult to elevate subperiosteally. Dysfunction of vascular smooth muscle as well as decreased platelet adhesion is thought to contribute to increased blood loss [74, 77, 78]. Besides diligent hemostasis intraoperatively, the use of antifibrinolytics may help to minimize blood loss. Shapiro et al. retrospectively evaluated the use of tranexamic acid (TXA) in 20 DMD patients and compared them with 36 control patients [79]. Tranexamic acid was found to reduce intraoperative blood loss and the need for homologous transfusion. Another option which has been studied for adolescent idiopathic scoliosis but not DMD includes the use of aminocaproic acid [80–82]. Vitale et al. investigated the efficacy of preoperative erythropoietin on hematocrit and transfusion rates in neuromuscular patients [83]. They found no clinical benefit in their treatment group. Our current practice is to work with anesthesia preoperatively to ensure that TXA is administered during surgery, and to utilize a cell saver system to maximize autologous transfusion in order to minimize usage of homologous blood products. In those patients who have evidence of preoperative platelet dysfunctions, we will transfuse the patients with

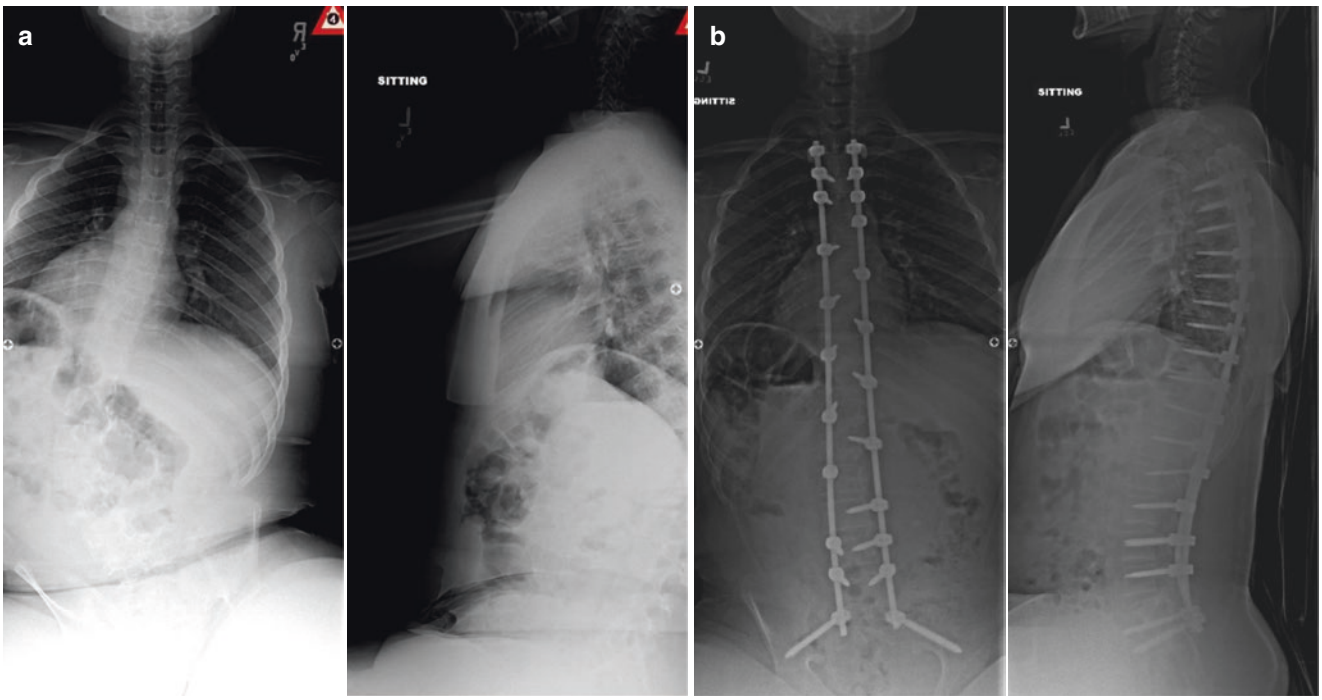

Fig. 12.2 (**a**) Preoperative AP and lateral radiographs of a 16-year-old male with DMD and thoracolumbar scoliosis. (**b**) Two-year postoperative radiographs demonstrating stabilization of the scoliosis with a posterior spinal fusion and segmental spinal instrumentation with intermittent pedicle screws and SAI pelvic screws

platelets before the start of the procedure. Postoperatively, hemoglobin levels are monitored closely to ensure that cardiac function is not overly stressed.

12.2.5 Long-Term Outcomes

As previously discussed, there is a controversy whether scoliosis surgery improves pulmonary function in the DMD patient. A recent Cochrane Review by Cheuk et al. was unable to provide an evidence-based recommendation for scoliosis surgery in DMD [24]. Their reasoning was the lack of randomized clinical trials. Of the 49 relevant studies addressing the outcomes of scoliosis surgery, none met their inclusion criteria for review.

Studies have suggested that spinal fusion does benefit patients beyond pulmonary function [84–87]. Bridwell et al. sent questionnaires to 33 patients with DMD evaluating function, self-image, cosmesis, pain, quality of life, and satisfaction [84]. Patients reported benefits in all categories with the highest ratings in cosmesis, quality of life, and satisfaction. Granata et al. and Takaso et al. found that sitting position, aesthetic improvement, and quality of life were all improved following spinal fusion [69, 85]. More than 90% of their patients/parents would give their consent again for surgery.

12.2.6 Duchenne Muscular Dystrophy Summary

Spinal deformity commonly affects the patient with DMD. Treatment of this deformity is complicated by progressive muscle weakness and deteriorating pulmonary function. Current literature suggests that surgical management of the deformity can maintain upright sitting posture, improve quality of life, and positively affect short-term pulmonary function. Unfortunately, a lack of randomized controlled trials has prevented formal Level-1 recommendations. If surgery is considered, however, it should be performed early when the patient is at his or her maximal health. In addition, if there is more than mild pelvic obliquity, one should consider including the pelvis in the instrumentation and fusion.

12.3 Arthrogryposis Multiplex Congenita

Arthrogryposis or "arthrogryposis multiplex congenita" (AMC) is a heterogeneous group of diseases with a similar phenotype of multiple congenital joint contractures [88, 89]. Currently, there are more than 150 subtypes that are believed to result from a failure of normal movement in utero. The etiology for this lack of movement may be myopathic, neu-

ropathic, or secondary to connective tissue abnormalities [90]. Amyoplasia is the term used to describe the more classic disease entity seen in orthopaedics. These patients have a dysgenesis of anterior horn cells resulting in replacement of muscle with adipose and fibrous tissue [91].

Patients with arthrogryposis multiplex congenita (AMC) have significant musculoskeletal deformities secondary to the contractures. The majority of patients have all four limbs involved (84%) [88]. Severe equinovarus feet (clubfeet), hip dislocations (unilateral or bilateral), and scoliosis are commonly seen. Nonorthopedic abnormalities include inguinal hernias, abdominal wall defects, gastroschisis, bowel atresia, hypoplasia of the labial folds, and cryptorchidism [88]. According to their long-term functional outcome study on 177 patients with AMC from 15 different countries, Nouraei et al. reported that 75% of respondents lived independently of family members, reported lower physical function scores than the general population, but similar or higher scores for other quality of life domains of the SF-36 [92]. In 2021 Verhofste et al. compared 35 AMC EOS patients with 112 matched idiopathic EOS patients from the Pediatric Spine Study Group [93]. All patients had ≥2 years postoperative growth friendly implant insertion and were compared for changes in spinal deformity and early onset scoliosis questionnaire (EOS-24) results. At the last follow-up, major curve correction and T1-S1 growth were comparable. AMC patients had poorer EOSQ-24 scores. Complications increased with longer follow-up in nonambulatory patients.

Similar to patients with other neuromuscular disorders, pulmonary function should be closely monitored perioperatively in patients with AMC. Li et al. have described risk factors for pulmonary dysfunction in patients with AMC, including both scoliosis and BMI as independent risk factors [94].

12.3.1 Spinal Deformity

The incidence of scoliosis in AMC is reportedly found between 30% and 67% [95, 96] (Fig. 12.3). The deformities are similar to other neuromuscular conditions with lumbar and thoracolumbar curves predominating [97, 98]. The curves are frequently stiff. Progression of the deformity can be rapid, up to 6.5° per year [98]. The earlier the presentation of scoliosis, the more severe the curve may become and the more likely it will be associated with pelvic obliquity. Increased lordosis is also frequently seen.

Scoliosis is typically refractory to orthotic management [97, 98]. Patients with AMC will frequently develop scoliosis early in life. There is minimal literature that has evaluated the treatment of EOS in these patients. Recently, Astur et al. and the Chest Wall Spinal Deformity Study Group evaluated ten children with AMC who underwent treatment with the use of the vertical expandable prosthetic titanium rib (VEPTR) device and found it to be an effective treatment method in these patients [99]. Using this rib-based distraction device, they obtained 37% correction of scoliosis and 29% correction of kyphosis. They also found improved thoracic volume. Six complications occurred in four patients in a total of 62 procedures performed. Proximal junctional kyphosis appeared to remain a problem; however, in this cohort, reoperation rates were reported to be as high as 28.6% at 4 years for patients with AMC [100]. Other than

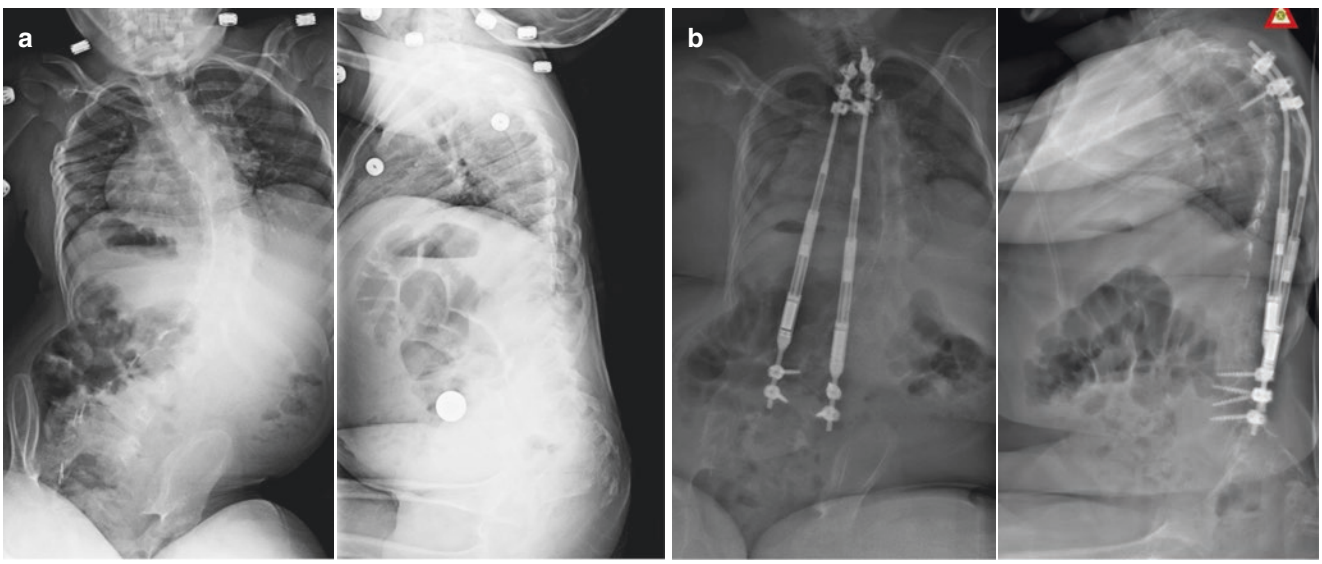

Fig. 12.3 (**a**) A 5-year-old male with progressive kyphoscoliosis secondary to AMC. (**b**) Patient was treated with magnetically controlled growing rods (MCGR)

this series, few studies have evaluated growing spine techniques in patients with AMC.

In older children and adolescents, PSF and SSI remain the standard and appear to be effective in preventing progression of the scoliosis. However, correction of the curves appears to be modest, about 35% [97]. Yingsakmongkol and Kumar reported slightly increased correction (44%) with a combined anterior and posterior fusion [95]. These series are dated, however, and do not assess surgical outcomes with current segmental instrumentation. In some cases, instrumentation was not used. If pelvic obliquity is present, fusion to the pelvis should be attempted. Evaluation for hip contractures should be evaluated preoperatively in the event that the final spinopelvic position impacts sitting ability [101]. Care should also be taken when positioning patients. Their stiff joints and osteopenia place them at increased risk of developing pathologic fracture.

12.4 Rett Syndrome

First described in 1966, Rett syndrome is a progressive neurologic disorder that affects one in 20,000 females [102, 103]. Patients initially appear normal at birth but then proceed through four stages of deterioration. The first stage typically has an onset between 6 and 18 months with developmental stagnation. The second stage (1–3 years of age) is characterized by lost language skills and autistic behaviors. In the third stage (2–10 years of age), patients may have seizures, exhibit some intellectual disability, and have repetitive hand motions. In the fourth stage, patients develop spasticity and muscle wasting. Scoliosis is most likely to present in this final stage.

12.4.1 Spinal Deformity

The musculoskeletal manifestations of Rett syndrome include lower extremity contractures, coxa valga, and scoliosis [103, 104]. The spinal deformity is similar to other neuromuscular diseases with a long C-shaped curve being the most common [105, 106]. However, patients can also present with a single thoracic or double major curve. Large curves are frequently associated with pelvic obliquity. As a patient gets older, so does the prevalence of scoliosis, particularly if the ability to ambulate is lost [107, 108]. Curve progression has been suggested to be more rapid than in idiopathic scoliosis or other neuromuscular scoliosis. Lidstrom et al. demonstrated greater that 15° per year of progression in the final stage of Rett syndrome [109]. For this reason, it has been recommended that patients are evaluated every 6 months following 5 years of age [110].

Bracing has been found to be largely unsuccessful in preventing the progression of scoliosis [105, 106, 110]. It, however, can be used to delay the need for surgical intervention to allow for more truncal growth. Posterior spinal fusion and segmental spinal instrumentation are the treatment of choice for the progressive scoliosis. In those patients who are nonambulatory, it is recommended to fuse from the upper thoracic spine to the pelvis to prevent delayed decompensation or pelvic obliquity. Ambulation is possible in patients with Rett syndrome and can be positively affected by surgery. Harrison et al. demonstrated no loss of ambulation in all five patients who walked preoperatively, and improvements in some patients [106]. Overall, PSF and SSI are successful in halting curve progression and improving spinal balance in the sitting and walking patient. Improvement in activities of daily living has been seen following spinal fusion in Rett syndrome patients, and data from Downs et al. have even suggested an increased life expectancy following spinal fusion [111, 112]. However, perioperative medical complications can be high, especially pulmonary (63%) and gastrointestinal (37%) in some series [113, 114]. Recent data suggest that perioperative pulmonary complications may be higher in patients with Rett syndrome even compared with those with other neuromuscular diseases [115, 116].

12.5 Congenital Myopathies

Congenital myopathies are a heterogeneous group of disorders characterized by weakness and hypotonia from birth [117]. Typically, the diseases have similar clinical findings but are classified based on histologic and microscopic findings. Central core disease, nemaline myopathy, and myotubular myopathy are just a few of the multiple congenital myopathies wherein scoliosis has been described [118–122]. They are genetically transmitted and can have variable penetrance.

12.5.1 Spinal Deformity

The musculoskeletal abnormalities associated with these disorders include congenital hip instability, foot deformities, other joint contractures, as well as scoliosis [121]. The curves are similar to other neuromuscular curves with a long, thoracolumbar shape. Kyphosis can also be associated with the deformity. As scoliosis progresses, it often becomes rigid. Rigid spine syndrome, as described by Dubowitz, has often been associated with these diseases and other congenital muscular dystrophies [123, 124].

If the patients present early and have flexible curves, scoliosis may be amenable to management with an orthosis. Those patients who fail bracing or present with large and

rigid curves, should undergo spinal fusion. Similar consideration as with other neuromuscular scoliosis must be given to the health and age of the patient. Poor pulmonary function has been associated with congenital myopathies [122]. At a minimum, patients should undergo preoperative pulmonary function testing. These patients are also at increased risk of developing malignant hyperthermia [118]. The anesthesiologist should be made aware of this before the day of surgery so that adequate preparation can be done.

Depending on the severity of the disease, patients may present with EOS. Those that demonstrate progression with the use of an orthosis may require surgical treatment with growth friendly surgery. However, there has only been one study adequately evaluating the short term results of "growing rods" in these patients [93]. Those patients that present later in life do well with a posterior spinal instrumentation and fusion. Anterior release can be done for the large rigid curve if the patient can tolerate the exposure. In the nonambulatory patient with pelvic obliquity, the fusion should be extended to the pelvis.

Similar principles to other neuromuscular diseases should be followed when treating patients with congenital myopathies. Posterior fusion is the treatment of choice. The need for traction or fusion to the pelvis should be determined on an individual basis. Depending on bone quality, a brace can be used postoperatively to support the instrumentation. Particular attention, however, has to be made toward the increased risk of hyperthermia.

12.6 Conclusion

Neuromuscular scoliosis represents spinal deformity secondary to neurologic or muscular disease of various etiologies, each with their unique management challenges. The progressive deformity can lead to multi-system downstream effects, which should be considered during the management of the scoliosis. Nonoperative management is less effective at preventing progression than in idiopathic curves. Surgical management usually requires long-segment fusion to the pelvis; however, less-invasive and growth-friendly strategies are being utilized but will require additional studies to determine long-term efficacy.

References

1. Kunkel LM, Hejtmancik JF, Caskey CT, Speer A, Monaco AP, Middlesworth W, et al. Analysis of deletions in DNA from patients with Becker and Duchenne muscular dystrophy. Nature. 1986;322(6074):73–7.
2. Sussman M. Duchenne muscular dystrophy. J Am Acad Orthop Surg. 2002;10(2):138–51.
3. Smith AD, Koreska J, Moseley CF. Progression of scoliosis in Duchenne muscular dystrophy. J Bone Joint Surg Am. 1989;71(7):1066–74.
4. Kurz LT, Mubarak SJ, Schultz P, Park SM, Leach J. Correlation of scoliosis and pulmonary function in Duchenne muscular dystrophy. J Pediatr Orthop. 1983;3(3):347–53.
5. Hsu JD. The natural history of spine curvature progression in the nonambulatory Duchenne muscular dystrophy patient. Spine. 1983;8(7):771–5.
6. Wilkins KE, Gibson DA. The patterns of spinal deformity in Duchenne muscular dystrophy. J Bone Joint Surg Am. 1976;58(1):24–32.
7. Gibson DA, Wilkins KE. The management of spinal deformities in Duchenne muscular dystrophy. A new concept of spinal bracing. Clin Orthop Relat Res. 1975;108:41–51.
8. Oda T, Shimizu N, Yonenobu K, Ono K, Nabeshima T, Kyoh S. Longitudinal study of spinal deformity in Duchenne muscular dystrophy. J Pediatr Orthop. 1993;13(4):478–88.
9. Inkley SR, Oldenburg FC, Vignos PJ Jr. Pulmonary function in Duchenne muscular dystrophy related to stage of disease. Am J Med. 1974;56(3):297–306.
10. Hapke EJ, Meek JC, Jacobs J. Pulmonary function in progressive muscular dystrophy. Chest. 1972;61(1):41–7.
11. Karol LA. Scoliosis in patients with Duchenne muscular dystrophy. J Bone Joint Surg Am. 2007;89(Suppl 1):155–62.
12. Yamashita T, Kanaya K, Yokogushi K, Ishikawa Y, Minami R. Correlation between progression of spinal deformity and pulmonary function in Duchenne muscular dystrophy. J Pediatr Orthop. 2001;21(1):113–6.
13. Galasko CS, Delaney C, Morris P. Spinal stabilisation in Duchenne muscular dystrophy. J Bone Joint Surg (Br). 1992;74(2):210–4.
14. Rideau Y, Glorion B, Delaubier A, Tarle O, Bach J. The treatment of scoliosis in Duchenne muscular dystrophy. Muscle Nerve. 1984;7(4):281–6.
15. Velasco MV, Colin AA, Zurakowski D, Darras BT, Shapiro F. Posterior spinal fusion for scoliosis in duchenne muscular dystrophy diminishes the rate of respiratory decline. Spine. 2007;32(4):459–65.
16. Shapiro F, Sethna N, Colan S, Wohl ME, Specht L. Spinal fusion in Duchenne muscular dystrophy: a multidisciplinary approach. Muscle Nerve. 1992;15(5):604–14.
17. Miller F, Moseley CF, Koreska J, Levison H. Pulmonary function and scoliosis in Duchenne dystrophy. J Pediatr Orthop. 1988;8(2):133–7.
18. Miller RG, Chalmers AC, Dao H, Filler-Katz A, Bost F. The effect of spine fusion on respiratory function in Duchenne muscular dystrophy. Neurology. 1991;41(1):38–40.
19. Kennedy JD, Staples AJ, Brook PD, Parsons DW, Sutherland AD, Martin AJ, et al. Effect of spinal surgery on lung function in Duchenne muscular dystrophy. Thorax. 1995;50(11):1173–8.
20. Hahn GV, Mubarak SJ. Muscular dystrophy. In: Weinstein SL, editor. The pediatric spine principles and practice. Philadelphia: Lippincott Williams & Wilkins; 2001. p. 819–32.
21. Farber HJ, Phillips WA, Kocab KL, Hanson DS, Heydemann JA, Dahl BT, et al. Impact of scoliosis surgery on pulmonary function in patients with muscular dystrophies and spinal muscular atrophy. Pediatr Pulmonol. 2020;55(4):1037–42.
22. Saito W, Mizuno K, Inoue G, Imura T, Nakazawa T, Miyagi M, et al. Perioperative evaluation of respiratory muscle strength after scoliosis correction in patients with Duchenne Muscular Dystrophy. Asian Spine J. 2017;11:787–92.
23. Chua K, Tan CY, Chen Z, Wong HK, Lee EH, Tay SK, et al. Long-term follow-up of pulmonary function and scoliosis in patients with Duchenne's Muscular Dystrophy and spinal muscular atrophy. J Pediatr Orthop. 2016;36(1):63–9.

24. Cheuk DK, Wong V, Wraige E, Baxter P, Cole A, N'Diaye T, et al. Surgery for scoliosis in Duchenne muscular dystrophy. Cochrane Database Syst Rev. 2007;1:CD005375.
25. Harper CM, Ambler G, Edge G. The prognostic value of preoperative predicted forced vital capacity in corrective spinal surgery for Duchenne's muscular dystrophy. Anaesthesia. 2004;59(12):1160–2.
26. Marsh A, Edge G, Lehovsky J. Spinal fusion in patients with Duchenne's muscular dystrophy and a low forced vital capacity. Eur Spine J. 2003;12(5):507–12.
27. Finsterer J. Cardiopulmonary support in duchenne muscular dystrophy. Lung. 2006;184(4):205–15.
28. Reid JM, Appleton PJ. A case of ventricular fibrillation in the prone position during back stabilisation surgery in a boy with Duchenne's muscular dystrophy. Anaesthesia. 1999;54(4): 364–7.
29. Wedel DJ. Malignant hyperthermia and neuromuscular disease. Neuromuscul Disord. 1992;2(3):157–64.
30. Heiman-Patterson TD, Natter HM, Rosenberg HR, Fletcher JE, Tahmoush AJ. Malignant hyperthermia susceptibility in X-linked muscle dystrophies. Pediatr Neurol. 1986;2(6):356–8.
31. Duckworth AD, Mitchell MJ, Tsirikos AI. Incidence and risk factors for post-operative complications after scoliosis surgery in patients with Duchenne muscular dystrophy. Bone Joint J. 2014;96-B(7):943–9.
32. Seeger BR, Sutherland AD, Clark MS. Orthotic management of scoliosis in Duchenne muscular dystrophy. Arch Phys Med Rehabil. 1984;65(2):83–6.
33. Drachman DB, Toyka KV, Myer E. Prednisone in Duchenne muscular dystrophy. Lancet. 1974;2(7894):1409–12.
34. Angelini C. The role of corticosteroids in muscular dystrophy: a critical appraisal. Muscle Nerve. 2007;36(4):424–35.
35. Shapiro F, Zurakowski D, Bui T, Darras BT. Progression of spinal deformity in wheelchair-dependent patients with Duchenne muscular dystrophy who are not treated with steroids. Bone Joint J. 2014;96-B(1):100–5.
36. Manzur AY, Kuntzer T, Pike M, Swan A. Glucocorticoid corticosteroids for Duchenne muscular dystrophy. Cochrane Database Syst Rev. 2004;(2):CD003725.
37. Yilmaz O, Karaduman A, Topaloglu H. Prednisolone therapy in Duchenne muscular dystrophy prolongs ambulation and prevents scoliosis. Eur J Neurol. 2004;11(8):541–4.
38. Alman BA, Raza SN, Biggar WD. Steroid treatment and the development of scoliosis in males with duchenne muscular dystrophy. J Bone Joint Surg Am. 2004;86-A(3):519–24.
39. Kinali M, Main M, Eliahoo J, Messina S, Knight R, Lehovsky J, et al. Predictive factors for the development of scoliosis in Duchenne muscular dystrophy. Eur J Paediatr Neurol. 2007;11(3):160–6.
40. King WM, Ruttencutter R, Nagaraja HN, Matkovic V, Landoll J, Hoyle C, et al. Orthopedic outcomes of long-term daily corticosteroid treatment in Duchenne muscular dystrophy. Neurology. 2007;68(19):1607–13.
41. Sanzarello I, Merlini L, Traina F, Rosa MA, Faldini C. Corticosteroid treatment impact on spinal deformity in Duchenne Muscular Dystrophy. Int Sch Res Notices. 2014;2014:965235.
42. Harvey A, Baker L, Williams K. Non-surgical prevention and management of scoliosis for children with Duchenne muscular dystrophy: what is the evidence? J Paediatr Child Health. 2013;50(10):E3.
43. Lebel DE, Corston JA, McAdam LC, Bigger WD, Alman BA. Glucocorticoid treatment for the prevention of scoliosis in children with Duchenne muscular dystrophy: long-term follow-up. J Bone Joint Surg Am. 2013;95(12):1057–61.
44. Koeks Z, Bladen CL, Salgado D, Van Zwet E, Pogoryelova O, McMacken G, et al. Clinical outcomes in Duchenne Muscular Dystrophy: a study of 5345 patients from the TREAT-NMD DMD global database. J Neuromusc Dis. 2017;4(4):293–306.
45. Mcdonald CM, Sajeev G, Yao Z, Mcdonnell E, Elfring G, Souza M, et al. Deflazacort vs prednisone treatment for Duchenne muscular dystrophy: a meta-analysis of disease progression rates in recent multicenter clinical trials. Muscle Nerve. 2019;61(1):26–35.
46. Mcdonald DGM, Kinali M, Gallagher AC, Mercuri E, Muntoni F, Frcpch HR, et al. Fracture prevalence in Duchenne muscular dystrophy. Dev Med Child Neurol. 2007;44(10):695–8.
47. King WM, Ruttencutter R, Nagaraja HN, Matkovic V, Landoll J, Hoyle C, et al. Orthopedic outcomes of long-term daily corticosteroid treatment in Duchenne muscular dystrophy. Neurology. 2007;68(19):1607–13.
48. Choi YA, Shin HI, Shin HI. Scoliosis in Duchenne muscular dystrophy children is fully reducible in the initial stage, and becomes structural over time. BMC Musculoskelet Disord. 2019;20:277.
49. Heller KD, Wirtz DC, Siebert CH, Forst R. Spinal stabilization in Duchenne muscular dystrophy:principles of treatment and record of 31 operative treated cases. J Pediatr Orthop B. 2001;10(1):18–24.
50. Mubarak SJ, Morin WD, Leach J. Spinal fusion in Duchenne muscular dystrophy—fixation and fusion to the sacropelvis? J Pediatr Orthop. 1993;13(6):752–7.
51. Sussman MD. Advantage of early spinal stabilization and fusion in patients with Duchenne muscular dystrophy. J Pediatr Orthop. 1984;4(5):532–7.
52. Luque ER. The anatomic basis and development of segmental spinal instrumentation. Spine. 1982;7(3):256–9.
53. Luque ER. Segmental spinal instrumentation for correction of scoliosis. Clin Orthop Relat Res. 1982;163:192–8.
54. Sarwark J, Sarwahi V. New strategies and decision making in the management of neuromuscular scoliosis. Orthop Clin N Am. 2007;38(4):485–96. v
55. Sengupta DK, Mehdian SH, McConnel JR, Eisenstein SM, Webb JK. Pelvic or lumbar fixation for the surgical management of scoliosis in duchenne muscular dystrophy. Spine. 2002;27(18): 2072–9.
56. Allen BL Jr, Ferguson RL. The Galveston technique of pelvic fixation with L-rod instrumentation of the spine. Spine. 1984;9(4):388–94.
57. Alman BA, Kim HK. Pelvic obliquity after fusion of the spine in Duchenne muscular dystrophy. J Bone Joint Surg (Br). 1999;81(5):821–4.
58. Brook PD, Kennedy JD, Stern LM, Sutherland AD, Foster BK. Spinal fusion in Duchenne's muscular dystrophy. J Pediatr Orthop. 1996;16(3):324–31.
59. Gaine WJ, Lim J, Stephenson W, Galasko CS. Progression of scoliosis after spinal fusion in Duchenne's muscular dystrophy. J Bone Joint Surg (Br). 2004;86(4):550–5.
60. Bentley G, Haddad F, Bull TM, Seingry D. The treatment of scoliosis in muscular dystrophy using modified Luque and Harrington-Luque instrumentation. J Bone Joint Surg (Br). 2001;83(1):22–8.
61. Ramirez N, Richards BS, Warren PD, Williams GR. Complications after posterior spinal fusion in Duchenne's muscular dystrophy. J Pediatr Orthop. 1997;17(1):109–14.
62. Myung KS, Lee C, Skaggs DL. Early pelvic fixation failure in neuromuscular scoliosis. J Pediatr Orthop. 2015;35(3):258–65.
63. Bui T, Shapiro F. Posterior spinal fusion to sacrum in non-ambulatory hypotonic neuromuscular patients: sacral rod/bone graft onlay method. J Child Orthop. 2014;8(3):229–36.
64. Takaso M, Nakazawa T, Imura T, Okada T, Toyama M, Ueno M, et al. Two-year results for scoliosis secondary to Duchenne mus-

cular dystrophy fused to lumbar 5 with segmental pedicle screw instrumentation. J Orthop Sci. 2010;15(2):171–7.
65. Takaso M, Nakazawa T, Imura T, Ueno M, Saito W, Shintani R, et al. Can the caudal extent of fusion in the surgical treatment of scoliosis in Duchenne muscular dystrophy be stopped at lumbar 5? Eur Spine J. 2010;19(5):787–96.
66. Modi HN, Suh S, Hong J, Cho J, Park J, Yang J. Treatment and complications in flaccid neuromuscular scoliosis (Duchenne muscular dystrophy and spinal muscular atrophy) with posterior-only pedicle screw instrumentation. Eur Spine J. 2010;19(3):384–93.
67. Modi HN, Suh S, Song H, Fernandez HM, Yang J. Treatment of neuromuscular scoliosis with posterior-only pedicle screw fixation. J Orthop Surg Res. 2008;3:23.
68. Scannell BP, Yaszay B, Bartley CE, Newton PO, Mubarak SJ. Surgical correction of scoliosis in patients with Duchenne Muscular Dystrophy: 30-year experience. J Pediatr Orthop. 2017;37(8):e464–9.
69. Takaso M, Nakazawa T, Imura T, Okada T, Fukushima K, Ueno M, et al. Surgical management of severe scoliosis with high risk pulmonary dysfunction in Duchenne muscular dystrophy: patient function, quality of life and satisfaction. Int Orthop. 2010;34(5):695–702.
70. McCarthy RE, Bruffett WL, McCullough FL. S rod fixation to the sacrum in patients with neuromuscular spinal deformities. Clin Orthop Relat Res. 1999;364:26–31.
71. Marchesi D, Arlet V, Stricker U, Aebi M. Modification of the original Luque technique in the treatment of Duchenne's neuromuscular scoliosis. J Pediatr Orthop. 1997;17(6):743–9.
72. Peelle MW, Lenke LG, Bridwell KH, Sides B. Comparison of pelvic fixation techniques in neuromuscular spinal deformity correction: Galveston rod versus iliac and lumbosacral screws. Spine. 2006;31(20):2392–8.
73. Sponseller P, Zimmerman R, Ko P, Pull Ter Gunne A, Mohamed A, Chang T, Kebaish K. Low profile pelvic fixation with the sacral alar iliac technique in the pediatric population improves results at two-year minimum follow-up. Spine. 2010;35(20):1887–92.
74. Shapiro F, Sethna N. Blood loss in pediatric spine surgery. Eur Spine J. 2004;13(Suppl 1):S6–S17.
75. Noordeen MH, Haddad FS, Muntoni F, Gobbi P, Hollyer JS, Bentley G. Blood loss in Duchenne muscular dystrophy: vascular smooth muscle dysfunction? J Pediatr Orthop B. 1999;8(3):212–5.
76. Archer JE, Gardner AC, Roper HP, Chikermane AA, Tatman AJ. Duchenne muscular dystrophy: the management of scoliosis. J Spine Surg. 2016;2(3):185–94.
77. Turturro F, Rocca B, Gumina S, De Cristofaro R, Mangiola F, Maggino N, et al. Impaired primary hemostasis with normal platelet function in Duchenne muscular dystrophy during highly-invasive spinal surgery. Neuromuscul Disord. 2005;15(8):532–40.
78. Labarque V, Freson K, Thys C, Wittevrongel C, Hoylaerts MF, Vos RD, et al. Increased Gs signalling in platelets and impaired collagen activation, due to a defect in the dystrophin gene, result in increased blood loss during spinal surgery. Hum Mol Genet. 2007;17(3):357–66.
79. Shapiro F, Zurakowski D, Sethna NF. Tranexamic acid diminishes intraoperative blood loss and transfusion in spinal fusions for duchenne muscular dystrophy scoliosis. Spine. 2007;32(20):2278–83.
80. Khoshhal K, Mukhtar I, Clark P, Jarvis J, Letts M, Splinter W. Efficacy of aprotinin in reducing blood loss in spinal fusion for idiopathic scoliosis. J Pediatr Orthop. 2003;23(5):661–4.
81. Florentino-Pineda I, Thompson GH, Poe-Kochert C, Huang RP, Haber LL, Blakemore LC. The effect of amicar on perioperative blood loss in idiopathic scoliosis: the results of a prospective, randomized double-blind study. Spine. 2004;29(3):233–8.
82. Cole JW, Murray DJ, Snider RJ, Bassett GS, Bridwell KH, Lenke LG. Aprotinin reduces blood loss during spinal surgery in children. Spine. 2003;28(21):2482–5.
83. Vitale M, Privitera D, Matsumoto H, Gomez J, Waters L, Hyman J, Roye D. Efficacy of preoperative erythropoietin administration in pediatric neuromuscular scoliosis patients. Spine. 2007;32(24):2662–7.
84. Bridwell KH, Baldus C, Iffrig TM, Lenke LG, Blank K. Process measures and patient/parent evaluation of surgical management of spinal deformities in patients with progressive flaccid neuromuscular scoliosis (Duchenne's muscular dystrophy and spinal muscular atrophy). Spine. 1999;24(13):1300–9.
85. Granata C, Merlini L, Cervellati S, Ballestrazzi A, Giannini S, Corbascio M, Lari S. Long-term results of spine surgery in Duchenne muscular dystrophy. Neuromuscul Disord. 1996;6(1):61–8.
86. Cervellati S, Bettini N, Moscato M, Gusella A, Dema E, Maresi R. Surgical treatment of spinal deformities in Duchenne muscular dystrophy: a long term follow-up study. Eur Spine J. 2004;13(5):441–8.
87. Suk KS, Lee BH, Lee HM. Functional outcomes in Duchenne Muscular Dystrophy scoliosis. J Bone Joint Surg. 2014;96(6):409–15.
88. Bernstein RM. Arthrogryposis and amyoplasia. J Am Acad Orthop Surg. 2002;10(6):417–24.
89. Hall JG. Arthrogryposis multiplex congenita: etiology, genetics, classification, diagnostic approach, and general aspects. J Pediatr Orthop B. 1997;6(3):159–66.
90. Banker BQ. Neuropathologic aspects of arthrogryposis multiplex congenita. Clin Orthop Relat Res. 1985;194:30–43.
91. Hall JG, Reed SD, Driscoll EP. Part I. Amyoplasia: a common, sporadic condition with congenital contractures. Am J Med Genet. 1983;15(4):571–90.
92. Nouraei H, Sawatzky B, Macgillivray M, Hall J. Long-term functional and mobility outcomes for individuals with arthrogryposis multiplex congenita. Am J Med Genet A. 2017;173(5):1270–8.
93. Verhofste BP, Emans JB, Miller PE, Birch CM, Thompson GH, Samdani AF, et al. J Bone Joint Surg Am. 2021;103(8):715–25.
94. Li Y, Sheng F, Xia C, Xu L, Qiu Y, Zhu Z. Risk factors of impaired pulmonary function in arthrogryposis multiplex congenital patients with concomitant scoliosis. Spine. 2018;43(8):E456–60.
95. Yingsakmongkol W, Kumar SJ. Scoliosis in arthrogryposis multiplex congenita: results after nonsurgical and surgical treatment. J Pediatr Orthop. 2000;20(5):656–61.
96. Sarwark JF, MacEwen GD, Scott CI Jr. Amyoplasia (a common form of arthrogryposis). J Bone Joint Surg Am. 1990;72(3):465–9.
97. Daher YH, Lonstein JE, Winter RB, Moe JH. Spinal deformities in patients with arthrogryposis. A review of 16 patients. Spine. 1985;10(7):609–13.
98. Herron LD, Westin GW, Dawson EG. Scoliosis in arthrogryposis multiplex congenita. J Bone Joint Surg Am. 1978;60(3):293–9.
99. Astur N, Flynn JM, Flynn JM, Ramirez N, Glotzbecker M, van Bosse HJ, et al. The efficacy of rib-based distraction with VEPTR in the treatment of early-onset scoliosis in patients with arthrogryposis. J Pediatr Orthop. 2014;34(1):8–13.
100. Paul JC, Lonner BS, Vira S, Feldman D, Errico TJ. Does reoperation risk vary for different types of pediatric scoliosis? J Pediatr Orthop. 2018;38(9):459–64.
101. Komolkin I, Ulrich EV, Agranovich OE, Bosse HJV. Treatment of scoliosis associated with arthrogryposis multiplex Congenita. J Pediatr Orthop. 2017;37(Suppl 1):S24–6.
102. Newton PO. Neuromuscular scoliosis. In: Herskowitz HN, et al., editors. The spine. Philadelphia: Saunders Elsevier; 2006. p. 535–64.

103. Guidera KJ, Borrelli J, Raney E, Thompson-Rangel T, Ogden JA. Orthopaedic manifestations of Rett syndrome. J Pediatr Orthop. 1991;11(2):204–8.
104. Loder RT, Lee CL, Richards BS. Orthopedic aspects of Rett syndrome: a multicenter review. J Pediatr Orthop. 1989;9(5):557–62.
105. Keret D, Bassett GS, Bunnell WP, Marks HG. Scoliosis in Rett syndrome. J Pediatr Orthop. 1988;8(2):138–42.
106. Harrison DJ, Webb PJ. Scoliosis in the Rett syndrome: natural history and treatment. Brain and Development. 1990;12(1):154–6.
107. Killian JT, Lane JB, Lee H-S, Skinner SA, Kaufmann WE, Glaze DG, et al. Scoliosis in Rett syndrome: progression, comorbidities, and predictors. Pediatr Neurol. 2017;70:20–5.
108. Downs J, Torode I, Wong K, Ellaway C, Elliott EJ, Christodoulou J, et al. The natural history of scoliosis in females with Rett syndrome. Spine. 2016;41(10):856–63.
109. Lidstrom J, Stokland E, Hagberg B. Scoliosis in Rett syndrome. Clinical and biological aspects. Spine. 1994;19(14):1632–5.
110. Bassett GS, Tolo VT. The incidence and natural history of scoliosis in Rett syndrome. Dev Med Child Neurol. 1990;32(11):963–6.
111. Downs J, Young D, de Klerk N, Bebbington A, Baikie G, Leonard H. Impact of scoliosis surgery on activities of daily living in females with Rett syndrome. J Pediatr Orthop. 2009;29(4):369–74.
112. Downs J, Torode I, Wong K, Ellaway C, Elliott EJ, Izatt MT, et al. Surgical fusion of early onset severe scoliosis increases survival in Rett syndrome: a cohort study. Dev Med Child Neurol. 2015;58(6):632–8.
113. Gabos PG, Inan M, Thacker M, Borkhu B. Spinal fusion for scoliosis in Rett syndrome with an emphasis on early postoperative complications. Spine. 2012;37(2):E90–4.
114. Karmaniolou I, Krishnan R, Galtrey E, Cleland S, Vijayaraghavan R. Perioperative management and outcome of patients with Rett syndrome undergoing scoliosis surgery: a retrospective review. J Anesth. 2015;29(4):492–8.
115. Cohen JL, Klyce W, Kudchadkar SR, Kotian RN, Sponseller PD. Respiratory complications after posterior spinal fusion for neuromuscular scoliosis. Spine. 2019;44(19):1396–402.
116. Downs J, Torode I, Wong K, Ellaway C, Elliott EJ, Christodoulou J, et al. The natural history of scoliosis in females with Rett syndrome. Spine. 2016;41(10):856–63.
117. Barnes MJ. Other neuromuscular deformities. In: Weinstein SL, editor. The pediatric spine principles and practice. Philadelphia: Lippincott Williams & Wilkins; 2001. p. 881–91.
118. Gamble JG, Rinsky LA, Lee JH. Orthopaedic aspects of central core disease. J Bone Joint Surg Am. 1988;70(7):1061–6.
119. Martinez BA, Lake BD. Childhood nemaline myopathy: a review of clinical presentation in relation to prognosis. Dev Med Child Neurol. 1987;29(6):815–20.
120. Cahill PJ, Rinella AS, Bielski RJ. Orthopaedic complications of myotubular myopathy. J Pediatr Orthop. 2007;27(1):98–103.
121. Shapiro F, Specht L. The diagnosis and orthopaedic treatment of inherited muscular diseases of childhood. J Bone Joint Surg Am. 1993;75(3):439–54.
122. Mertz KD, Jost B, Glatzel M, Min K. Progressive scoliosis in central core disease. Eur Spine J. 2005;14(9):900–5.
123. Arkader A, Hosalkar H, Dormans JP. Scoliosis correction in an adolescent with a rigid spine syndrome: case report. Spine. 2005;30(20):E623–8.
124. Flanigan KM, Kerr L, Bromberg MB, Leonard C, Tsuruda J, Zhang P, et al. Congenital muscular dystrophy with rigid spine syndrome: a clinical, pathological, radiological, and genetic study. Ann Neurol. 2000;47(2):152–61.