

# LIMB CONTRACTURES IN PROGRESSIVE NEUROMUSCULAR DISEASE AND THE ROLE OF STRETCHING, ORTHOTICS, AND SURGERY

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Limb contractures are common impairments in selected progressive neuromuscular disease (NMD) conditions and can lead to such increased disabilities as worsening motor performance, decreased mobility, loss of ADL skills, and pain. This article reviews the pathogenesis of joint contractures in NMD, the prevalence of specific contractures among more common NMD conditions, and the role of optimal positioning, stretching, upright ambulation, orthotics and splinting, as well as surgery in the prevention and management of contractures.

## **PATHOGENESIS**

Contracture is defined as the lack of full active or passive range of motion (ROM) due to joint, muscle, or soft tissue limitation. Contractures may be arthrogenic, soft tissue, or myogenic in nature, and a combination of intrinsic structural changes of muscle and extrinsic factors leads to myogenic contractures in selected neuromuscular disease conditions. These factors include the following:

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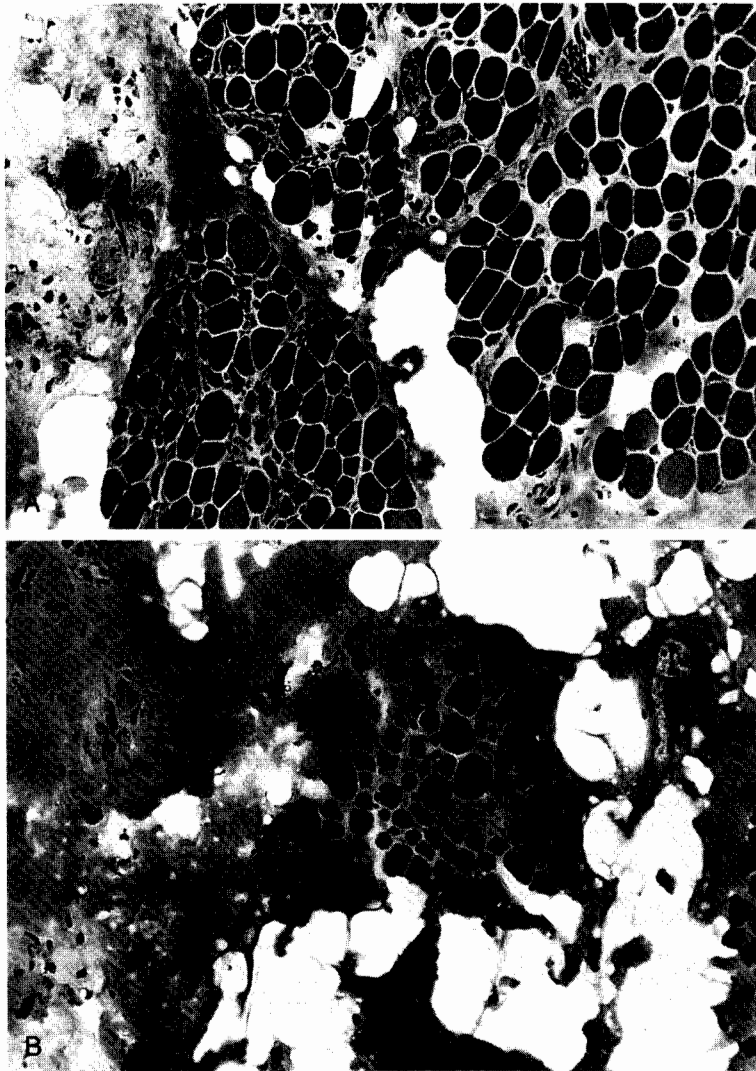
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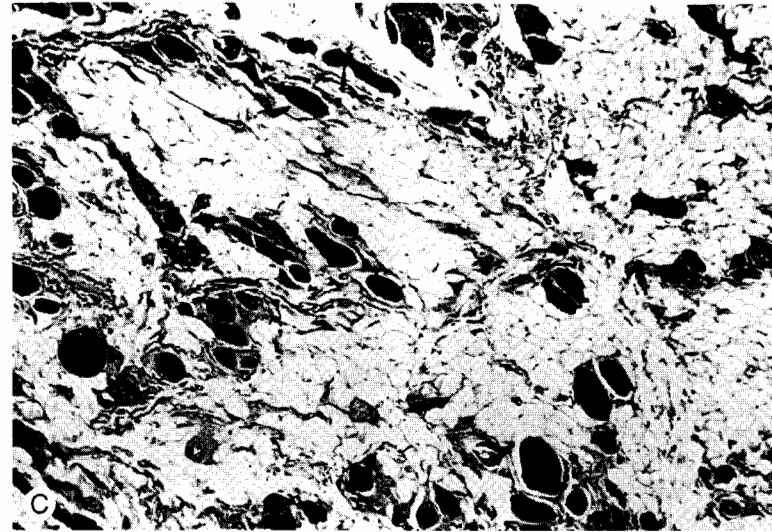
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**Figure 1.** Muscle biopsy specimens of the vastus lateralis in Duchenne muscular dystrophy (DMD). *A*, Three-year-old with both dystrophic and hypertrophied fibers, regenerating fibers with central nuclei, inflammatory cells, and fibrosis with fat infiltration. *B*, Nine-year-old with Duchenne muscular dystrophy and more extensive loss of muscle fiber, with dystrophic fibers, hypertrophied fibers, and central nucleated regenerating fibers. Note the more extensive fibrosis and fat infiltration.

*Illustration continued on opposite page*

**Degree of Fibrosis and Fatty Tissue Infiltration.** Intrinsic muscle tissue alterations in dystrophic myopathies place these patients at high risk for contracture formation. The most significant histologic changes are those of muscle fiber loss, abnormal residual dystrophic muscle



**Figure 1 (Continued).** *C*, Post-mortem muscle biopsy specimen in a 19-year-old with end-stage Duchenne muscular dystrophy. Note the paucity of muscle fibers and tremendous replacement of muscle fibers by fat and connective tissues. Hematoxylin and eosin stains.

fibers, segmental necrosis of muscle fibers, and increased amounts of lipocytes and fibrosis. For example, histologic sections of Duchenne muscular dystrophy patients aged 3, 9, and 19 years are shown in Figure 1A–C. Replacement of functioning muscle fibers with collagen and fatty tissue in concert with chronically shortened resting muscle length results in contracture formation. The collagen fibers of endomysium, perimysium, and epimysium undergo rearrangement and proliferation, causing muscle fibrosis and limitation of joint ROM. The inflammatory myopathies result in replacement of muscle fibers by increasing amounts of collagen and connective tissue in association with lymphocytic infiltration. Neurogenic atrophy typically results in a diminished degree of fibrosis and fatty tissue infiltration, creating different intrinsic tissue characteristics and lower risk of severe contracture formation during brief periods of immobilization.

**Static Positioning and Lack of Full Active and Passive Range of Motion.** Weakness and inability to achieve active joint mobilization throughout the full allowable range is the single most frequent factor contributing to the occurrence of fixed contractures. For example, less than antigravity elbow extension strength places an individual at significant risk for an elbow flexion contracture. The position in which a joint is immobilized affects the number of sarcomeres present in a muscle; immobilization in a shortened position may cause muscle fibers to lose 40% of their sarcomeres.<sup>49</sup> Furthermore, in dystrophic myopathies, fibrosis of muscles in a resting position increases the tendency to form fixed contracture in the position of immobilization.

**Imbalance of Agonist and Antagonist Muscle Strength Across the Joint.** Although strength imbalance of flexor and extensor muscle groups across a joint has not been shown to be a factor leading to deformity in Duchenne muscular dystrophy,<sup>36</sup> such strength asymmetries appear to be important determinants of progressive contractures and deformity in more focal neuropathic entities, such as Charcot-Marie-Tooth (CMT)/hereditary motor sensory neuropathy. For example, in CMT, more pathologic involvement of the peroneal nerve compared with the tibial nerve may lead to asymmetry of strength, with greater plantar flexion than dorsiflexion strength and greater inversion than eversion strength. This, in combination with intrinsic foot muscle weakness, leads to equinovarus foot deformities. The absolute muscle strength is directly proportional to the physiologic cross-sectional area of muscle fiber.<sup>4, 60</sup> In Duchenne muscular dystrophy, the ankle plantar flexors are stronger than the ankle dorsiflexors, and ankle inverters are stronger than ankle everters.<sup>36</sup> With the ankle positioned in either plantar flexion or dorsiflexion, the cross-sectional area of the ankle inverters is greater than the cross-sectional area of the ankle evertors. This muscle imbalance may predispose Duchenne muscular dystrophy patients to develop hindfoot varus and ankle equinus contractures over time.<sup>36</sup> In addition, deformities such as hip dislocation can be produced in congenital myopathy, congenital muscular dystrophy, Duchenne muscular dystrophy, and spinal muscular atrophy because of a combination of imbalanced muscle strength around the hip joint, hypotonia, and hip flexion positioning predisposing to subluxation and frank dislocation of the femoral head.

**Lack of Upright Weight Bearing and Static Positioning in Sitting.**

Transition to wheelchair reliance with reduced time spent in upright standing places individuals with NMD conditions at risk for ankle plantar flexion, knee flexion, hip flexion, and elbow flexion contractures.<sup>36</sup> This tendency toward contracture formation is particularly problematic in NMD patients with less than antigravity knee extension or ankle dorsiflexion strength.

**Compensatory Postural Changes Used to Biomechanically Stabilize Joints for Upright Standing.** NMD patients with proximal muscle weakness of the hip and knee extensors frequently will exhibit lumbar lordosis, diminished stance phase knee flexion, and equinus posturing at the ankle during ambulation. The equinus posturing at the ankle leads to diminished heel strike and increased forefoot contact at the initial stance phase of gait. This is a compensatory measure to keep the weight line and ground reaction force line anterior to the knee (creating a stabilizing knee extension moment) and posterior to the hip joint (creating a stabilizing hip extension moment). This places individuals with such compensatory gait at risk for mild ankle equinus contractures (Fig. 2) before the onset of wheelchair reliance.<sup>36</sup>

**Functional Anatomy of Muscles and Joints.** Specific muscle groups at particular risk for contracture formation include two joint or multi-joint muscle groups in which the origin and insertion crosses multiple joints. These include the gastrocnemius, hamstrings, rectus femoris, ten-



**Figure 2.** Nine-year-old boy with Duchenne muscular dystrophy exhibiting postural compensations to maintain standing. Note the lumbar lordosis, anterior pelvic tilt, and equinus posturing at the ankle. Mild ankle plantar flexion contractures are present prior to transition to the wheelchair.

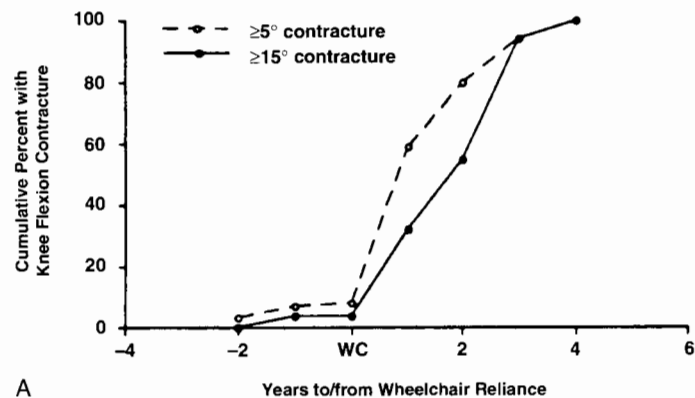
sor fasciae latae, iliopsoas, pectoralis major, biceps brachii, and long finger flexors (e.g., flexor digitorum sublimis and profundus). Generally, the position in which these muscle groups obtain maximal stretch are either not functional positions or they are positions rarely obtained over the course of the day. Muscles develop their greatest active force generation at their resting length, a point where there is minimal passive tension. Thus, placement of a muscle on passive stretch compromises the total tension produced and often places the limb in a generally nonfunctional position. Maximal stretch of biarticular/two-joint muscles requires a "counter-current shift" of agonist and antagonist muscle groups. For example, a straight leg raise with hip flexion and knee extension lengthens the hamstrings, whereas the rectus femoris is maximally shortened across both joints in this position. There are rare functional activities that bring the hip and knee joints into this position, and both muscle groups contract suboptimally along their length tension curves in this joint position. Similarly, maximal stretch to the rectus

femoris requires extension at the hip joint and flexion at the knee joint—a position that maximally shortens the hamstrings. This is, similarly, a nonfunctional position of the joint. Thus, maximal stretch to multijoint muscle groups requires *passive* range of motion, particularly if weakness makes the achievement of such joint positions impossible actively.

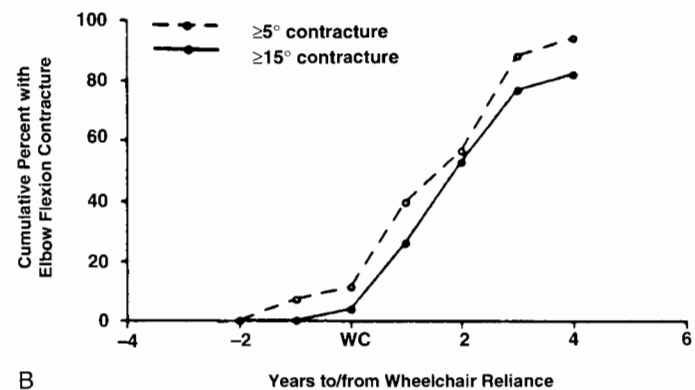
## CONTRACTURES IN SPECIFIC NMD CONDITIONS

### Duchenne Muscular Dystrophy

Significant joint contractures have been found in nearly all Duchenne muscular dystrophy (DMD) children older than age 13.<sup>14, 31, 36</sup>



A



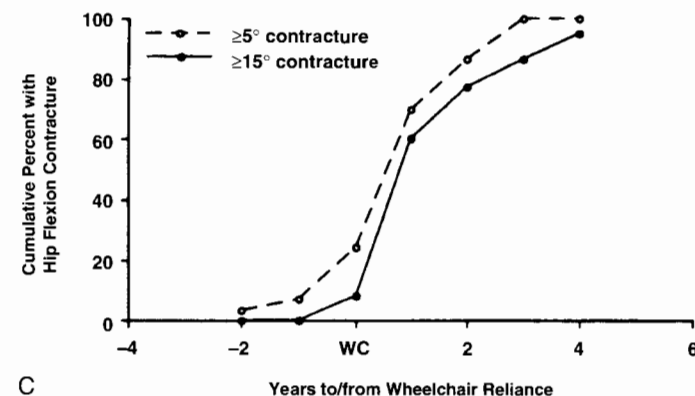
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**Figure 3.** Cumulative percentages of DMD subjects with  $\geq 5^\circ$  contractures (dashed line) and  $\geq 15^\circ$  contractures (solid line) versus years to and from consistent wheelchair reliance: A, knee flexion contractures; B, elbow flexion contractures.

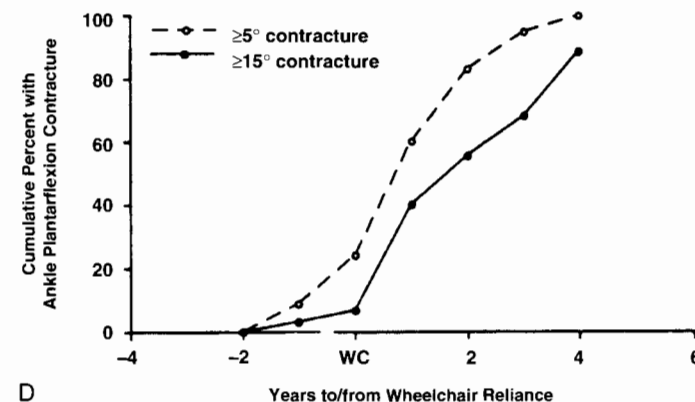
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The most common contractures include ankle plantar flexion, knee flexion, hip flexion, iliotibial band, elbow flexion, and wrist flexion contractures.<sup>36</sup> Significant contractures have been shown to be rare in DMD before age 9 for all joints.<sup>36</sup> There is no association between muscle imbalance around a specific joint (defined as grade 1 or greater difference in flexor and extensor strength) and the frequency or severity of contractures involving the hip, knee, ankle, wrist, and elbow in DMD.<sup>36</sup> Flexion contractures have been shown to be rare in those with  $\geq$  grade 3 extensor strength about a joint, an expected finding because of the definition of a grade 3 muscle on manual muscle testing (MMT). For those DMD subjects with less than antigravity strength about a joint, there is low correlation between the MMT strength of these specific muscle groups and the severity of joint contracture.<sup>36</sup>

The presence of lower extremity contractures in DMD has been shown to be strongly related to onset of wheelchair reliance (Fig. 3).



C

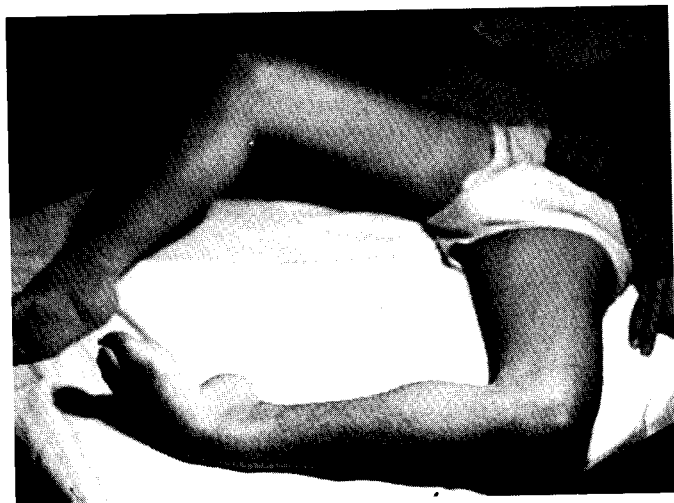


D

**Figure 3 (Continued).** C, Hip flexion contractures; D, ankle plantar flexion contractures. (From McDonald CM, Abresch RT, Carter GT, et al: Profiles of neuromuscular diseases: Duchenne muscular dystrophy. *Am J Phys Med Rehabil* 74:S70-S92, 1995; with permission.)

Lower extremity contractures were rare while DMD subjects were still upright, but developed soon after they assumed a sitting position in a wheelchair for most of the day. The occurrence of elbow flexion contractures also appears to be directly related to prolonged static positioning of the limb, and these contractures develop soon after wheelchair reliance.

The relationship between wheelchair reliance and hip and knee flexion contractures has been noted by others.<sup>3, 45, 58</sup> Brooke and colleagues<sup>13</sup> noted contractures of the iliotibial bands, hip flexor muscles, and the heel cords in most DMD patients by 6 years of age. Their subjects developed limitations of knee, elbow, and wrist extension about 2 years later. However, these early observed contractures were relatively mild. Given the tremendous replacement of muscle by fibrotic tissue in DMD subjects it is not surprising that a muscle with less than antigravity extension strength, statically positioned in flexion, would develop a flexion contracture (subsequent to wheelchair reliance, as seen in Fig. 4). Although 20% of DMD subjects in our study developed ankle plantar flexion contractures of  $\geq 5^\circ$  before wheelchair reliance, the lack of lower extremity weight-bearing likely contributed to the rapid acceleration in severity of these contractures after transition to wheelchair usage (Fig. 5). Ankle plantar flexion contractures were not likely a significant cause of wheelchair reliance, as  $< 10\%$  of our subjects had plantar flexion contractures of  $\geq 15^\circ$  before their transition to a wheelchair.<sup>36</sup> Natural history data suggest that weakness is the major cause of loss of ambulation in DMD, not contracture formation.<sup>36</sup>



**Figure 4.** Thirteen-year-old boy with DMD and severe hip flexion, knee flexion, and ankle equinovarus contractures developed subsequent to wheelchair reliance.



**Figure 5.** Eleven-year-old patient with DMD and severe ankle plantar flexion contractures 1 year after transition to the wheelchair. At time of transition to the wheelchair the ankle plantar flexion contracture measured less than  $5^\circ$ .

### Becker Muscular Dystrophy

Contractures have not been shown to be a significant problem in ambulatory patients with Becker Muscular Dystrophy (BMD).<sup>35</sup> In this study, 14 subjects had ROM testing of the major upper and lower extremity joints. A decrease in ROM by  $\geq 20^\circ$  was found in only one subject at the elbow and in two subjects at the wrists, hips, knees, and ankles. Both had been nonambulatory for several years.

### Emery Dreifuss Muscular Dystrophy

Emery Dreifuss muscular dystrophy (EMD) is an X-linked recessive, progressive dystrophic myopathy with a gene locus identified at xq28.<sup>11</sup> The muscle protein which is deficient in EMD has been termed "Emerin." The condition usually presents in adolescence or early adult life, and many clinical features may be seen in early childhood. Patients may have a selective scapulohumeroperoneal distribution weakness and often striking wasting of the upper arms, accentuated by sparing of the deltoids and forearm muscles. There may also be focal wasting of the calf muscles and biceps. An associated cardiomyopathy usually presents with arrhythmia, and may lead to sudden death in early adult life. The cardiomyopathy may progress to four-chamber dilated cardiomyopathy with complete heart block and ventricular arrhythmias.

A hallmark of EMD clinically is the early presence of contractures of the elbow flexors with limitation of full extension. Heel cord tightness may be present early in the disorder, concomitant with ankle dorsiflexion weakness and toe walking. Unlike DMD, the toe walking in EMD usually is secondary to ankle dorsiflexion weakness and not a compensa-

tory strategy to stabilize the knee in proximal limb weakness. Tightness of the cervical and lumbar spinal extensor muscles, resulting in limitation of neck and trunk flexion with inability to flex the chin to the sternum and to touch the toes, also has been reported in EMD.<sup>19</sup>

A dominantly inherited disorder with a similar phenotype has been reported.<sup>64</sup> Some reported cases of "rigid spine syndrome" may be EMD cases in view of the marked predominance of males in this disorder and the associated contractures reported at the elbows and ankles.<sup>19</sup>

### Congenital Muscular Dystrophy

Congenital muscular dystrophy (CMD) represents a group of congenital dystrophic myopathies which are non-X-linked. These disorders present in infancy with hypotonia and early presence of contractures. Fukuyama congenital muscular dystrophy is an autosomal recessive condition linked to chromosome 9q31-33 with abnormal expression of dystrophin-associated glycoproteins and central nervous system involvement. Other forms of congenital muscular dystrophy may result from merosin deficiency, which is an extracellular protein important for maintenance of sarcolemmal membrane stability in the muscle fiber.<sup>53</sup> The loci for merosin deficient congenital muscular dystrophy have been linked to chromosome 6q.<sup>53</sup> Subjects with merosin deficiency often are intellectually normal. A further subtype of congenital muscular dystrophy with normal merosin has been reported, but the genetic locus has not been established.

Subjects with congenital muscular dystrophy often exhibit early contractures, including equinovarus deformities, knee flexion contractures, hip flexion contractures, and tightness of the wrist flexors and long finger flexors (Fig. 6). These myopathies have been reported to be fairly slowly progressive or relatively static; however, the contractures can become more severe over time with prolonged static positioning and lack of adequate passive range of motion and splinting/positioning.<sup>19</sup>

### Severe Childhood Autosomal Recessive Muscular Dystrophy (SCARMMD)

Several forms of severe childhood autosomal recessive muscular dystrophy (SCARMMD) have been linked to chromosomes 13q12<sup>6, 10, 20</sup> and 17q12-q21.<sup>41</sup> These dystrophic myopathies are due to abnormal expression of transmembrane sarcoglycans, which are glycoproteins linked to both dystrophin on the intracellular side of the sarcolemmal membrane and merosin on the extracellular side. These patients have similar degrees of dystrophic muscle fiber changes, fatty infiltration, and fibrosis histologically, as seen in Duchenne muscular dystrophy. The patients demonstrate progressive, predominantly proximal weakness affecting the shoulder and pelvic girdle. In addition, they develop re-



**Figure 6.** Eighteen-month-old boy with congenital muscular dystrophy (Fukuyama type) and severe hip flexion, knee flexion, and equinovarus contractures.

strictive lung disease and cardiomyopathy. Progression tends to be slower than observed in DMD.<sup>19</sup> In a study of 13 SCARMMD subjects, a reduction in ROM by 20° or more was seen in 23% of the subjects at the elbow, 17% at the wrist, 27% at the hip, 23% at the knee, and 30% at the ankle.<sup>37</sup> As with other dystrophic myopathies, the severity of lower extremity contractures seems to increase with proportion of time spent in a wheelchair.

### Facioscapulohumeral Muscular Dystrophy

Facioscapulohumeral muscular dystrophy (FSH) is a slowly progressive dystrophic myopathy with predominant involvement of facial and shoulder girdle musculature. Over time, patients develop ankle dorsiflexion and eversion weakness and pelvic girdle weakness. The condition is autosomal dominant, with linkage to the chromosome 4q35 locus.<sup>55, 61, 65</sup> Contractures are relatively uncommon in FSH muscular dystrophy. In the most comprehensive study to date,<sup>32</sup> 22 subjects had ROM testing of major upper and lower extremity joints. Clinically significant loss of ROM (defined as a reduction in ROM > than 20%) was present in two subjects (9%) at the wrist and in one subject (5%) at the hip, knee, and ankle. This subject had been wheelchair-dependent for 15 years at the time of ROM testing.

### Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a term used to describe a varied group of inherited disorders characterized by weakness and muscle

wasting, secondary to degeneration of both anterior horn cells of the spinal cord and brain stem motor nuclei without pyramidal tract involvement. Three subtypes of autosomal recessive predominantly proximal SMA have been described, all linked to chromosome 5q. A common nomenclature subdivides SMA into types I, II, and III, based on age of onset and age at death, whereas the other approach classifies cases as severe, intermediate, and mild, based on ability to achieve independent sitting and independent standing and walking. SMA type I (severe form) was defined by the International Consortium on SMA as follows: Onset from birth to 6 months, no achievement of sitting without support, and death usually prior to the age 2 years. In SMA type II (intermediate form), onset is before 18 months, sitting usually is obtained, but standing and ambulation are never obtained, and death occurs above the age of 2 years, usually much later. In SMA type III (mild form), the onset is after the age of 18 months, patients develop the ability to stand and walk, and death is in adulthood.

Contractures are problematic in SMA II patients and SMA III patients who have lost ambulation. One study found reductions in ROM by  $> 20^\circ$  among 22% to 50% of SMA II subjects, depending on the joint.<sup>16</sup> Hip, knee, and wrist contractures were most common. Lower extremity contractures have been found to be rare in ambulatory SMA patients.<sup>16, 29</sup> Patients with SMA perceive their elbow flexion contractures (Fig. 7) to hinder one or more daily functions, and the contractures were reported to be associated with greater discomfort.<sup>63</sup>

### Amyotrophic Lateral Sclerosis

Amyotrophic lateral sclerosis (ALS) is a rapidly progressive motor neuron disorder that results in profound appendicular, bulbar, and respiratory muscle weakness, but only mild joint contractures.<sup>31</sup>

In one study, only 26% of ALS subjects had ankle plantar flexion contractures, 13% had shoulder contractures, and only 20% had contractures of any joint measuring  $\geq 20^\circ$  by goniometry.<sup>31</sup> The low prevalence and mild severity of contractures in ALS is likely due to: (1) the short disease duration and relatively short time between wheelchair reliance and death; and (2) the neurogenic nature of the muscle wasting, with less severe fibrosis and fatty tissue infiltration.

### Myotonic Muscular Dystrophy

Myotonic muscular dystrophy (MMD) is an autosomal dominant disorder with a gene locus on 19q13.1.<sup>5, 12, 15, 25, 33</sup> Heterogeneous onset is explained by abnormal expansion of trinucleotide CTG repeat within an unstable area of the chromosome.<sup>18</sup> Those with "congenital" myotonic muscular dystrophy presenting in infancy may have 1000 to 2000 CTG repeats or more. In these children, a congenital club foot or talipes



**Figure 7.** Ten-year-old boy with spinal muscular atrophy type II and severe elbow flexion, hip flexion, and ankle plantar flexion contractures. Left wrist flexion positioning with ulnar deviation is controlled by a wrist splint. Scoliosis is present and sitting posture is improved with a thoracolumbosacral orthosis (TLSO). Note the severe diffuse atrophy of muscle groups.

equinovarus is a fairly common deformity. In patients with noncongenital myotonic muscular dystrophy, contractures at the wrist, ankle, and elbows are relatively uncommon and mild.<sup>30</sup>

### Limb Girdle Muscular Dystrophy

A heterogeneous group of muscular dystrophies predominantly affecting pelvic girdle and shoulder girdle musculature has been referred to as limb girdle muscular dystrophy (LGMD). The genetic heterogeneity of these disorders is becoming increasingly appreciated, with several subtypes showing both dominant inheritance<sup>50</sup> and seven documented subtypes with recessive inheritance.<sup>9, 8, 18, 66</sup> In autosomal dominant limb girdle muscular dystrophy, reduction in ROM by  $20^\circ$  or more was seen in 20% of subjects at the wrist, 10% at the hip, 10% at the knee, 20% at the ankle, and none at the elbow.<sup>37</sup> In autosomal recessive limb girdle muscular dystrophy, reduction in ROM by  $20^\circ$  or more was seen in 5%

of the subjects at the elbow, 5% at the wrist, 5% at the hip, 5% at the knee, and none at the ankle.<sup>37</sup> In general, contractures in these populations were evident only in those with long-standing wheelchair reliance.

### Hereditary Motor Sensory Neuropathy

Multiple subtypes of hereditary motor sensory neuropathy (HMSN) exist, as described elsewhere in this issue,<sup>18</sup> with genetic heterogeneity among both the primarily demyelinating forms as well as the primarily axonal forms. Distal muscle wasting tends to be more frequently evident in the axonal forms owing to neurogenic atrophy from axonal loss and denervation. Among primarily demyelinating forms, distal wasting may at times take decades to develop. In one study of 53 subjects,<sup>17</sup> reduction in ROM by 20° or more was seen in 9% at the ankle, 8% at the knee, 2% at the elbow, 14% at the hip, and 19% at the wrist. Focal wasting of intrinsic foot and hand musculature is a common finding in HMSN patients, and the most common foot and ankle deformity has been equinovarus deformity. A cavus foot with hammer toe deformities also is common, with and without an equinus contracture or hindfoot varus. The wasting of intrinsic foot musculature often proceeds the wasting of interossei, hypothenar, and thenar muscles of the hand.

### Miscellaneous Conditions Leading to Congenital Joint Rigidity (Arthrogryposis)

Arthrogryposis is a symptom complex characterized by congenital rigidity of the joints, and is not a specific diagnostic entity. By definition, arthrogryposis involves multiple joints, with the distal joints more often affected than the proximal. The feet, ankles, hands, and wrists are most commonly affected. A variety of central nervous system disorders, such as chromosomal syndromes, developmental disorders, and congenital malformations of the central nervous system may result in arthrogryposis. Focal embryologic anterior horn cell disorders also may result in arthrogryposis. Peripheral neuropathy such as congenital hypomyelination neuropathy or hereditary motor sensory neuropathy type III may be associated with arthrogryposis. Both transient neonatal and congenital forms of myasthenia have been described with congenital joint contractures. Of the nondystrophic congenital myopathies, X-linked myotubular myopathy is perhaps the condition most commonly associated with arthrogryposis. Finally, a primary focal amyoplasia leading to strength imbalance around joints also may result in congenital contractures.

## MANAGEMENT OF CONTRACTURES

Contractures in progressive NMD conditions should be managed with the following concepts in mind:

1. Prevention of contractures requires early diagnosis and initiation of physical medicine approaches such as passive ROM and splinting while contractures are still mild.
2. Contractures are inevitable in some NMD conditions, such as DMD.
3. Advanced contractures become fixed and show little response to stretching programs.
4. A major rationale for controlling contractures of the lower extremity is to minimize the adverse effect of contractures on independent ambulation. However, the major cause of wheelchair reliance in NMD is generally weakness, not contracture formation.
5. Static positioning of both upper and lower extremity joints in patients with weak musculature is the most important cause of contracture formation.
6. Passive stretching for control of lower limb contractures is most successful in ambulatory patients with early mild joint contractures.
7. Upper extremity contractures may not negatively impact function if they are mild.
8. Joint range of motion should be monitored regularly by physical therapists and occupational therapists using objective goniometric measurement.

### Management of Lower Extremity Contractures in Patients with Proximal Weakness

Four principal physical therapy modalities must be regularly carried out to prevent or delay the development of lower extremity contractures for those at risk for musculoskeletal deformity. These include: (1) regularly prescribed periods of daily standing and walking; (2) passive stretching of muscles and joints; (3) positioning of the leg to promote extension and oppose joint flexion when the patient is non-weight-bearing through the lower extremities; and (4) splinting, which is a useful measure for the prevention or delay of ankle contracture.<sup>44</sup>

A minimum of 2 to 3 hours of daily standing and walking is necessary in addition to passive stretching for the control of contracture formation.<sup>59</sup> Decreases in daily standing time must be identified promptly and measures implemented to promote upright weight bearing. For patients who are marginally ambulatory or those only recently wheelchair reliant, a standing table or standing frame with attachable desk top surface may be very helpful for the continued prevention of hip flexion, knee flexion, and ankle plantar flexion contractures. Such

standing frames may be stationary or mobile. It is essential that the patient weight-bear with equal weight distribution between the right and left sides. Such a balanced stance helps prevent asymmetric contractures of heel cords, pelvic obliquity with adduction and abduction contractures, and flexion contractures about the hip and knee on the non-weight-bearing side.

Passive stretching to maintain or improve range of motion is an enormously important component of the program to prevent contractures. Such passive range of motion has been documented to be efficacious in slowing the development of contractures in DMD.<sup>1, 2, 26, 44, 46, 59, 62</sup> A program of passive stretching should be started as early as possible in the course of neuromuscular disease and become part of a child's or adult's regular morning and evening routine. Proper technique is essential for passive stretching to be effective. With each stretch, the position should be held for a slow count of 15, and each exercise should be repeated 10 to 15 times during a session.<sup>58</sup> Stretching should be performed slowly and gently; an overly strenuous stretch may cause discomfort and reduce cooperation. Written instructional materials should be provided to the patient and family as a supplement to verbal instructions and demonstrations by the physical therapist. The specific anatomic focus of stretching exercises prescribed for lower extremity contractures will vary with the type of neuromuscular disease. For example, in younger DMD patients, passive stretching should be focused on: (1) gastrocnemius-soleus; (2) hamstrings; (3) iliotibial band.

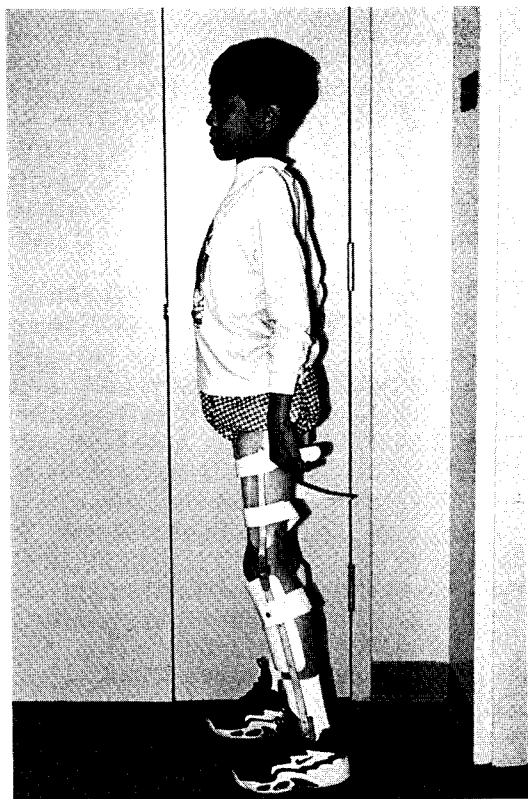
Lower extremity positioning may be a useful adjunct for preventing contracture formation. The limb is placed in a resting position that opposes the tendency for flexion contractures to develop. The prone-lying position is an effective method to stretch the hip flexors. Proper instruction in the stretching of the iliotibial bands, which contribute to hip flexion tightness, is of enormous importance in individuals with dystrophic myopathies and proximal weakness, such as DMD, BMD, SCARMD, and LGMD.

Splinting is another adjunctive measure used to slow the development of contractures in neuromuscular diseases. Short leg ankle-foot orthotics (AFOs) or nighttime resting splints have been used to maintain a 90° angle of the foot relative to the tibia. Some authors indicate that such splinting is effective for reducing heel cord contractures in DMD<sup>44</sup>; however, other investigators do not believe that short-leg orthotics change the natural history of heel cord contracture formation.<sup>23, 57, 58</sup> Any splinting must be used in conjunction with passive stretching and/or standing or ambulation. Splinting for prevention of equinus deformity generally has been used at night. However, AFOs may be worn while in the wheelchair throughout the day. Long leg KAFO orthotics, which immobilize the knee in extension, may be worn for ambulation, for standing, or as a night resting brace; however, in most patients with tight hamstrings they generally are not well-tolerated. Stretching, positioning, and splinting have limited roles in the management of hip and knee contractures in patients who use a wheelchair primarily.

Progressive resistive exercises to strengthen the agonist muscles have limited roles in the prevention and management of contractures. This is primarily because the extensor muscles generally are exceedingly weak (less than grade 3) in a significant contracture. Static positioning of the joint is generally a more important determinant of contracture formation than muscle imbalance of flexors and extensors across the joint.

### **Orthoses and Orthopaedic Surgery in Neuromuscular Diseases with Proximal Lower Extremity Weakness**

McDonald and colleagues<sup>36</sup> have reported the best predictors of imminent wheelchair reliance in DMD to be knee extension strength of 3- or less and a time to ambulate 30 feet of  $\geq 12$  seconds. Bracing and surgical management of contractures for prolonged ambulation in NMD conditions with proximal weakness, such as DMD, was first reported by Spencer and Vignos in 1962.<sup>51</sup> These authors stressed the importance of orthopaedic surgical contracture releases at the time of bracing. A number of principles should be emphasized for these populations. First, with an appropriate therapy program, equinovarus contractures generally are absent or very mild in DMD at the time walking ability ceases.<sup>36</sup> In addition, hip and knee flexion contractures are also absent or extremely mild in ambulatory DMD patients at the time of bracing.<sup>38, 44, 56</sup> The wide-based Trendelenburg gait exhibited by these patients with gluteus medius weakness places the hip in an abducted position, leading to iliotibial band contractures. The late phase of ambulation often is associated with more marked joint contractures involving the iliotibial bands and heel cords, because DMD patients spend more time sitting and less time standing. The release of contractures at both the heel cord and iliotibial band generally is necessary to obtain successful KAFO bracing.<sup>21, 22, 48, 51</sup> Other authors have reported bracing of DMD patients without surgical release of the iliotibial bands.<sup>27, 47</sup> Hip and knee flexion contractures generally are not severe enough to interfere with bracing at time of transition to wheelchair.<sup>36</sup> The iliotibial band contractures are released with a low Young fasciotomy and a high Ober fasciotomy. The ankle deformity may be corrected with either a tendo achillis lengthening (TAL) or a TAL combined with surgical transfer of the posterior tibialis muscle tendon to the dorsum of the foot. The posterior tibialis tendon transfer corrects the equinovarus deformity but prolongs the time in a cast and recovery time, and it increases the risks of prolonged sitting. The orthopaedic surgical release of these contractures allows the DMD patient to be braced in lightweight polypropylene KAFOs with solid ankle set at 90°, drop-lock knee joints, and ischial weight-bearing polypropylene upper thigh component (Fig. 8). The ischial seating gives a more upright standing posture with reduction in lordosis. DMD patients who are braced may or may not require a walker for additional support. At times, DMD patients who have had excellent home stretch-



**Figure 8.** Twelve-year-old boy with DMD who has achieved prolonged ambulation with bilateral knee ankle foot orthotics (KAFOs). Quadriceps strength is grade 2 bilaterally and time to walk 30 feet was 12 seconds more than 1 year prior indicating an impact on time to wheelchair by the bracing. The patient did not require orthopedic surgery to allow bracing.

ing programs can be placed immediately into KAFO bracing without surgical tenotomy.

While DMD subjects are still ambulating independently they can use their ankle equinus posturing from the gastrocnemius soleus group to create a knee extension moment at foot contact, thus stabilizing the knee when the quadriceps muscle is weak. Several authors have cautioned against isolated heel cord tenotomies while DMD patients are still ambulating independently.<sup>21, 48, 52</sup> Tendo achillis lengthening without simultaneous bracing above the knee can lead to loss of walking ability unless the quadriceps are grade 4 or better. Overcorrection of the heel cord contracture in a DMD patient may result in immediate loss of the ability to walk.<sup>62</sup>

Continued braced ambulation complements the stretching program by maintaining the hip and knee in full extension and by preventing ankle equinus deformity through lower extremity weight-bearing at a

fixed 90° position provided by the AFO component of the KAFO brace. The duration of ambulation in DMD has been successfully prolonged by prompt surgery and bracing, immediately implemented following loss of independent ambulation. The gains in additional walking time have been variable, but generally between 2 and 5 years.<sup>21, 26, 40, 43, 51, 58, 67</sup>

Benefits include decreased severity of heel cord and knee flexion contractures at age 16.<sup>58</sup> The combined surgical heel cord release and posterior tibial tendon transfer have the benefit of keeping the foot and ankle in a good functional position after braced ambulation ceases, making shoe wear and foot positioning on the wheelchair leg rest better. Prolonged ambulation by lower extremity bracing in DMD has never been documented to be an independent factor in the prevention of scoliosis. No prospective studies evaluating long-term contracture progression in DMD subjects with and without surgical tenotomies for prolonged braced ambulation have been carried out.

Disadvantages of braced ambulation center around the excessive energy cost of braced ambulation and safety concerns in the event of falls. DMD subjects need gait training by physical therapy, and they need to be taught fall techniques with locked knees.

Another question regarding braced ambulation pertains to patient selection. Only some DMD patients accept the bracing and continue ambulating. It is theoretically possible that DMD patients who choose to continue ambulating in braces are on the less severe end of the spectrum (i.e., "outlier" DMD patients)—patients who show less severe scoliosis and less rapid loss of pulmonary function. Studies need not only randomize patients to surgical versus nonsurgical lower extremity treatment, but also control for such other variables as peak obtained forced vital capacity (FVC). Alternatively, information regarding timed motor performance and natural history could be used to evaluate the usefulness of surgical tenotomies and lower extremity bracing. For example, the author's data<sup>36</sup> demonstrate that 100% of subjects who ambulate 30 feet in  $\geq 12$  seconds transition to a wheelchair in less than 12 months. If such a population undergoes surgical tenotomy and lower extremity bracing and continues to ambulate for more than 12 months, then the surgical and orthotic intervention would be demonstrated to change the natural history regarding ambulation and time to wheelchair.

In our previous study,<sup>36</sup> ankle plantar flexion contractures were not likely a significant cause of wheelchair reliance because a small proportion ( $\leq 10\%$ ) of our subjects had plantar flexion contractures of  $\geq 15^\circ$  before they were transitioned to a wheelchair. Because of the relationship between contracture formation and wheelchair reliance, it is likely that orthopaedic contracture releases will allow a DMD patient to be braced in KAFOs for prolonged ambulation, but will not likely enable the patient to ambulate for a longer period without bracing, as proposed by Bach and McKeon.<sup>7</sup> In a recent randomized trial, Manzer and colleagues<sup>34</sup> showed no benefit from early surgical treatment of contractures in DMD (e.g., early posterior tibial tendon transfers with subsequent brace-free ambulation). These data and the findings of the author<sup>36</sup> support the notion that weakness is the major cause of loss of ambulation

in DMD, not contracture formation. Thus, the primary role of orthopaedic surgical tenotomies and posterior tibialis tendon transfers likely is the provision of optimal alignment for KAFO bracing. Little evidence supports the efficacy of early prophylactic lower extremity surgery in DMD.

### **Orthotic Management of NMD Patients with Distal Lower Extremity Weakness**

A number of NMD conditions may cause significant weakness of distal musculature. Generally, lower extremity strength loss predates upper extremity involvement in most, owing to peripheral nerve involvement (e.g., HMSN). Peroneal nerve innervated muscles (ankle dorsiflexors) often are clinically weaker than tibial nerve innervated muscles (ankle plantar flexors). Some myopathies may give isolated weakness of ankle dorsiflexors and everters, including FSH, scapuloperoneal distribution LGMD, MMD, Emery-Dreifuss muscular dystrophy, and distal myopathies (e.g., Maresbery-Griggs myopathy, Finnish tibial muscular dystrophy, and type II early adult onset autosomal recessive distal myopathy). Type I early adult onset autosomal recessive distal myopathy may present initially with dystrophic changes in the posterior compartment.

Ankle foot orthotics (AFOs) often are useful in patients with distal weakness. An AFO may have the following functions: (1) the plantar flexion stop substitutes for ankle dorsiflexion weakness during swing phase of gait; (2) the dorsiflexion stop substitutes for ankle plantar flexion weakness during stance phase; (3) the solid plate extended at or distal to the metatarsal heads combined with a dorsiflexion stop substitutes for ankle plantar flexion weakness at push-off; (4) extension of the AFO anterior to the malleoli provides medial-lateral stability, substituting for weakness of the ankle invertors or everters; (5) a solid ankle AFO helps prevent progressive deformity; and (6) a solid ankle AFO set at a neutral 90° angle helps provide knee stability if the quadriceps are weak.

A few NMD conditions present with distal upper extremity weakness, including Asian variant distal SMA and late-onset autosomal dominant distal myopathy of the Welander form. These and other NMD entities with distal upper extremity weakness later in the disease course may benefit from upper extremity orthotics to stabilize the wrist in a functional position, stabilize the thumb MP joint for opposition, or stabilize the MCP joints in intrinsic lumbrical weakness. Universal cuffs and other adapted devices may be very helpful, and these patients can benefit from regular occupational therapy evaluations.

### **Orthotic Management of NMD Patients with Proximal Upper Extremity Weakness**

NMD patients with proximal shoulder girdle weakness have been managed with mobile arm supports; however, in the author's experi-

ence, these often are prescribed and fit but rarely are used consistently or accepted long term. An extension tray placed on the wheelchair lap tray or elevating hospital bedside table puts the patient's upper extremities in an elevated flexed position at the shoulder, allowing the hand to be raised to the mouth with a grade 2 to 2+ biceps. Alternatively, some patients choose to eat meals at a higher kitchen counter or at a bar-style counter.

### **Surgical Management of Foot and Ankle Deformities in NMD Patients with Distal Weakness**

Cavus feet are common in peripheral neuropathies such as HMSN type I and II and with Friedreich's ataxia. There are three forms of pes cavus, as determined by the orientation of the os calcis during stance: (1) pes cavovarus; (2) pes calcaneocavus; and (3) pes equinocavus. A pes cavovarus deformity, combined with decreased proprioception, causes difficulty in walking, lack of balance, and painful callosities. The peripheral neuropathy and intrinsic muscle weakness of the foot result in hyperextension at the metatarsophalangeal joints and flexion at the interphalangeal joints, with resultant claw toe deformities.

Treatment of the cavus foot depends on the patient's age, flexibility of the foot, bony deformity, and muscle imbalance. As soon as the diagnosis is confirmed, daily manipulation of the foot to resist the depression of the first metatarsal, stretching of the plantar fascia, and extension of the toes may be helpful. A nighttime or full-time ankle-foot orthosis in a neutral ankle position custom molded to the cavus foot deformity may decrease the tendency toward development of an equinus deformity and delay the onset of a fixed deformity. A supple foot can be managed nonoperatively by serial casting in a walking cast, followed by an AFO with a solid ankle in neutral position and a lateral heel wedge to minimize varus. If a fixed soft tissue or bony deformity ensues, orthopaedic surgery may be necessary to produce a plantigrade foot. In children, triple arthrodesis is contraindicated. A variety of soft tissue procedures may be considered (e.g., plantar fascia release, posterior tibialis tendon transfer to the dorsum of the foot, etc.), combined with bone surgery, including calcaneal osteotomy and dorsal osteotomy of the first metatarsal to correct a rigid plantar flexed first metatarsal. In the rigid, skeletally mature foot, more definitive procedures may be considered, including radical plantar fasciotomy release to relieve mid-foot cavus; transfer of the posterior tibialis tendon to the dorsum of the foot; transfer of the extensor hallucis longus tendon to the neck of the first metatarsal (i.e., Jones procedure) to reduce pronation of the forefoot; correction of claw toes by interphalangeal joint arthrodesis; and calcaneal and metatarsal osteotomies to correct a rigid cavus foot. A triple arthrodesis sometimes is considered as a salvage procedure for severe heel varus and severe midfoot deformity for achievement of hindfoot stability.

## Management of Upper Extremity Contractures

The primary focus in the literature has been on contractures of the lower extremities because contractures of the arm and hand cause little functional deficit until the late stages of some NMD conditions. Elbow flexion contractures in DMD occur soon after transition to the wheelchair, secondary to the static positioning of the arms in elbow flexion on the arm rests of the chair.<sup>36</sup> In DMD, deterioration in upper extremity function lags approximately 2 years behind the loss of lower extremity function. Other associated deformities in DMD include forearm pronator tightness and wrist flexion-ulnar deviation in the later stages of the disease. The force of gravity increases the tendency toward wrist flexion. The regular palm-down position of the hand increases the occurrence of forearm pronator contracture.<sup>58</sup> Prophylactic occupational therapy management of the wrist and hand is recommended in DMD to slow the development of contractures and to maintain fine motor skills. Positioning with a wrist flexion contracture limits finger flexion movements by shortening the muscle length of the long finger flexors. This contributes to reduced grip strength. Daily passive stretching of the wrist flexors and intrinsic and extrinsic muscle of the hand and wrist are recommended as are active range of motion exercises for the wrist and long finger extensors. Nighttime resting splints, which promote wrist extension, metacarpophalangeal extension, and proximal interphalangeal flexion, are recommended. Daytime positioning should emphasize wrist and finger extension, but any splinting should not compromise sensation or function.

Mild elbow flexion contractures of  $\leq 15^\circ$  are of no functional consequence to the patient using crutches or a wheelchair. Contractures of the elbows over  $30^\circ$  can interfere with the use of crutches in ambulatory patients with NMD. Severe elbow flexion contractures greater than  $60^\circ$  decrease distal upper extremity function and produce difficulty when dressing.<sup>58</sup> Passive stretching of the elbow flexors may be combined with passive stretch into forearm supination to help prevent contractures.

Shoulder contractures are less problematic in patients with profound proximal muscle weakness. Combined shoulder internal rotation-adduction contracture and elbow flexion deformity may interfere with self-feeding. Severe shoulder internal rotation deformities may complicate dressing, produce pain on passive range of motion, and cause pain during sleep.

## SUMMARY

Contractures are exceedingly common impairments in selected progressive NMD conditions, particularly those with excessive fibrosis and fatty infiltration into muscle (i.e., dystrophic myopathies) and more severe NMD conditions, resulting in significant weakness and wheelchair reliance, such as SMA. Less than antigravity strength produces an

inability to achieve full active range of motion. Static positioning of limbs (generally in flexion) and lack of weight bearing results in fixed contractures. This article has reviewed the prevalence and distribution of contractures in specific NMD conditions. Aggressive rehabilitation strategies, including stretching, positioning, splinting, upright weight bearing, and orthopaedic surgical management may help minimize the degree of disability in NMD patients with contractures.

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# SPINAL DEFORMITY IN PROGRESSIVE NEUROMUSCULAR DISEASE

## Natural History and Management

Dennis A. Hart, MD, and Craig M. McDonald, MD

Severe spinal deformity in progressive neuromuscular disease (NMD) leads to multiple problems, including poor sitting balance, difficulty with upright seating and positioning, pain, difficulty in attendant care, and potential exacerbation of underlying restrictive respiratory compromise. Severe scoliosis and pelvic obliquity can in some instances completely preclude upright sitting in a wheelchair. Populations at risk for scoliosis include Duchenne muscular dystrophy (DMD), severe childhood autosomal recessive muscular dystrophy (SCARMD), congenital muscular dystrophy (C-MD), facioscapulohumeral muscular dystrophy (FSH), congenital myotonic muscular dystrophy, spinal muscular atrophy II and III, and Friedreich's ataxia. This article reviews the prevalence, natural history, and management of spinal deformity in the progressive neuromuscular disorders.

### DUCHENNE MUSCULAR DYSTROPHY

#### Prevalence and Natural History

The reported ultimate prevalence of scoliosis in DMD varies from 33% to 100%.<sup>18, 26, 31, 41, 44, 50, 60, 61, 70, 71, 74, 77, 80, 89</sup> This marked variability

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