Chapter 8
Physical Therapy and Orthotic Devices

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Introduction

Muscular dystrophies (MDs) are a group of genetically based neuromuscular disorders characterized by disease-specific patterns of progressive muscle weakness accompanied by postural compensations and the risk of progressive contracture, deformity, and compromised function which may be accompanied by involvement across numerous body systems [1–14]. Individual MDs differ in the genetic basis, the cause and site of pathology, specific clinical features, distribution and extent of involvement, natural history, and prognosis [15–18], the details of which have been covered in previous chapters.

Similarities in the clinical presentation of MDs have allowed the use of common principles of clinical management and intervention in the provision of optimal comprehensive care with the coordinated expertise of a multidisciplinary team [5, 6, 19, 20]. Comprehensive, anticipatory physical therapy (PT) management of MD is based upon an understanding of the pathokinesiology of each type of MD, an understanding of the progression of the pathokinesiology over time, individual evaluation within the context of each individual's life and goals, and provision of consistent, preventative management across the lifespan in order to minimize the clinical and functional impact of the diagnosis and to optimize quality of life [1–3, 5, 6, 11, 19–24].

Historically, physical therapists have worked with individuals with MDs to minimize the clinical impact of the cellular pathology, to prevent secondary

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complications, to promote and maintain the maximum level of function and functional independence, and to achieve and maintain the highest possible quality of life for all individuals in spite of the disease process and/or progression [6, 25–28].

We are now entering an exciting new era, in which the natural histories of neuromuscular disorders are changing and improving based on improved medical care and management, and in which actual disease modifying treatments are emerging (see Chapter 4). PT management may increasingly, and for the first time, have the opportunity to assist in contributing to improvement and recovery in individuals with muscle diseases in addition to using prospective anticipatory care to manage impairments and optimize function and participation. In this new era, it will remain important to understand and continue to use optimal principles of intervention in comprehensive, anticipatory, preventative management and to optimize the benefits of disease modifying treatments as they emerge.

Pathokinesiology

The underlying genetic basis and cellular pathology that characterize specific MDs differ, but each is typically characterized by a unique and genetically based progressive degeneration of muscle often accompanied by fibrosis and fatty infiltration that contributes to the development of contracture and deformity [29] (see Chapters 3–5). A self-perpetuating cycle of events in MDs has been described [14], in which imbalanced muscle weakness, compensatory movement patterns and postural habits, and the influence of gravity interact in the progression of disability [1, 2, 11, 14, 19, 30–32]. Weakness often progresses proximal to distal and is often first evident in muscles around the shoulder, trunk, and pelvic girdles, with patterns of muscle involvement specific and unique to each type of MD [1, 13, 15–18, 31, 33, 34]. As weakness increases, compensatory alterations are made in posture and movement to mechanically lock joints and substitute for lack of adequate muscle strength [11, 14, 19, 30, 31, 35]. The substitutions are effective in maximizing function but eventually lead to contracture and deformity that contribute to increasing weakness and disability [36, 37]. In addition to weakness that occurs due to actual muscle degeneration, weakness may also seem to "progress" in proportion to growth, as has also been described in other disorders characterized by weakness [38, 39]. The compromising impact and effect of gravity increases in magnitude with increased size as the muscles are less able to cope with an increase in mass, and during periods of rapid growth in which contracture can progress more rapidly.

Effective intervention is that which is focused on breaking this self-perpetuating cycle of events whenever possible so that strength can be maximally maintained, contracture and deformity can be minimized, and compensations can be used to maximize function without leading to increases in disability [14, 19].

The key to management of most neuromuscular diseases is in their predictability [1]. Muscle weakness progresses in specific and well-known orders and patterns [1, 13, 30, 31, 40–43]. Predictable compensations are used to cope with this increasing weakness [1, 33, 34, 44–46]. Specific muscle tightness, contracture, and deformity can result and occurs in predictable sequences without intervention [5, 6, 36, 40, 46] (Table 8.1). This predictability is a double-edged sword. On the one hand, the predictability is evidence of a progression that cannot yet be stopped. On the other hand, knowledge of the predictable progression empowers the multidisciplinary team, and the family, to plan ahead and intervene with prospective, preventative, anticipatory management. Many of the devastating secondary effects of the intrinsic myopathic process can be minimized with comprehensive, ongoing, anticipatory, and preventative management that maintains the highest possible quality of life despite disease progression [6, 55]. Multidisciplinary guidelines supporting this approach are available for increasing numbers of neuromuscular disorders [5, 6, 56, 57].

Physical Therapy Assessment

Assessment must be ongoing and comprehensive so that intervention can be timely and anticipatory [6, 37]. Specific areas of weakness, tightness, and compensation should be identified in order to allow intervention that optimizes and protects muscle integrity and function, prevents contracture and deformity, and provides for effective adaptive functioning and participation to the greatest extent possible [6]. Assessment and intervention should occur across the ICF (the World Health Organization International Classification of Function [58]) and across the lifespan [5, 6, 55, 59] and should include impairment level measures, functional measures, and measures of participation, while considering the context and environmental factors of the individual [60]. Assessment and management of musculoskeletal and cardiorespiratory involvement and function requires a multidisciplinary team [5, 6] (Table 8.2).

Physical Therapy Intervention

Prevention of Contracture and Deformity

With weakness and compensation there may be no way to eliminate a compensatory pattern of movement without eliminating the function it serves, but it is important to try to find compensations that pose less of a risk of contracture and deformity and to try to avoid development of the contractures that contribute to the self-perpetuating evolution of weakness/contracture/functional loss [6, 14, 30, 31, 35]. The effects of chronic positioning, the unopposed influence of gravity, and imbalanced muscle

Table 8.1 Patterns of skeletal muscle weakness, compensation, and resultant risk of secondary contracture and deformity in representative diagnoses: Duchenne/Becker Muscular Dystrophy (DMD/BMD), Facioscapulohumeral muscular dystrophy (FSHD) and Emery-Dreifuss Muscular Dystrophy (EDMD) [2, 13, 19, 24, 30, 31, 47–50]

DMD/BMD [2, 3, 6, 19, 30, 31, 33, 44] DMD/BMD weakness, as detailed in Chapter 4, tends to be symmetrical		
Characteristic weakness (early stage)	*Characteristic compensations/posture/patterns of movement (early stage)*	*Tightness (early stage)*
– Hip extensors (*gluteus maximus*) – Ankle dorsiflexors (*anterior tibialis*) – Hip abductors (*gluteus medius*) – Hip adductors – Abdominals – Neck flexors (*sternocleidomastoid*) – Shoulder depressors and extensors (*lower trap/latissimus*) – Shoulder abductors (*deltoids*) – Elbow extensors (*triceps*)	– Increased lumbar lordosis (posterior trunk lean) to keep force line behind hip joint (initially see *less* anterior pelvic tilt as hyperextension at hip joint in stance as long as quadriceps are strong enough to counteract moment into knee flexion) – Lack of heelstrike – Increased hip flexion during swing to clear foot – Foot may be pronated and everted – May see "hip waddling gait" as do not get adequate forward weight shift – Increased UE abduction and lateral trunk sway – Cadence decreases – Gower's maneuver – Neck and UE weakness not usually noticeable functionally but apparent with testing	May see emerging tightness in: – Plantarflexors – Hip flexors – Iliotibial bands
Characteristic weakness (middle stage)	*Characteristic compensations/posture/patterns of movement (middle stage)*	*Tightness (middle stage)*
– Weakness progresses in muscles listed above – Quadriceps weakness = key to gait deterioration – Ankle everters (peroneals)	– Must get line of gravity simultaneously in front of knee joint and behind hip joint—uses: • Anterior pelvic tilt • Diminished hip extension in stance • Base of support widens: • Balance • Increased ankle plantarflexion and equinus positioning—to give torque that opposes knee flexion – Begin to see increased falling – Also get inversion with posterior tibialis relatively stronger—leads to unstable subtalar joint and more falling due to "twisting of the ankle"—although most falling is due to weakness in quadriceps and "knee buckling"	**Tightness** develops in: – Iliotibial band and tensor fascia lata – Hip flexors – Hamstrings – Gastrocsoleus – Posterior tibialis – **Important:** two-joint muscles get tight first

DMD/BMD [2, 3, 6, 19, 30, 31, 33, 44] DMD/BMD weakness, as detailed in Chapter 4, tends to be symmetrical		
Characteristic weakness (later stage)	*Characteristic compensations/posture/patterns of movement (later stages)*	*Tightness (later stages)*
– Weakness continues to progress in muscles listed above and becomes profound – UE weakness becomes more significant functionally and is imbalanced: – Elbow extension weaker than flexion – Forearm supination weaker than pronation – Wrist and finger extension weaker than flexion – Neck extensors, hamstrings, posterior tibialis are relatively spared until quite late in the disease – Distal hand function is relatively preserved, at least in long flexors but may be functionally compromised by lack of proximal stability and/or scoliosis requiring use of hands for sitting stability	– Prior to loss of ambulation, most compensations are used to maintain an upright posture and facilitate ambulation – After loss of ambulation, compensatory movements are primarily used to: • Achieve support and stability in sitting • Assist UE function – Compensatory movements include: • Leaning for stability • Contralateral trunk leaning during UE function to substitute for shoulder girdle weakness in arm lifting (deltoid weakness in abduction) • Backward leaning/lurching to compensate for deltoid weakness in forward flexion and biceps weakness in elbow flexion – Leading with head (especially using neck extensors) to shift weight and compensate for weak trunk musculature – Using mouth to grab fingers and move arm to substitute for proximal UE musculature – Pivoting forearm on elbow to substitute for elbow flexors	– Accelerated development of LE contractures – Beginning development of UE contractures – Tightness into elbow flexion and pronation – Tightness in wrist and finger flexors +/or extensors, lumbricals – Cervical spinal extensors and rotators ***Scoliosis**: the development of scoliosis is a major complication of the late or non-ambulatory stage, with natural history changing (decreasing) with steroids

(continued)

Table 8.1 (continued)

FSHD [4, 13, 47, 48, 51] FSHD clinical presentation much more variable than other diagnoses, as detailed in Chapter 3, with asymmetrical weakness not necessarily related to handedness

Characteristic/possible weakness	*Functional impact/compensation*	*Tightness/pain*
– Facial muscles: Orbicularis oculi Orbicularis oris Zygomaticus – Scapular muscles – Serratus anterior – Middle and lower trapezius – Horizontal abductor – Pectoralis major – Humeral muscles: (biceps brachii) – Abdominal weakness—lower weaker than upper (Beevor sign) – Hip extensors – Knee flexors – Anterior tibialis	– Scapular winging – Compensatory use of momentum and distal strength to move arms – Difficult closing eyes – Difficulty pursing lips, drinking with straw, whistling – Decreased ability to show facial expressiveness – Anterior pelvic tilt – Lumbar lordosis – Foot drop – Spinal asymmetry—with risk of scoliosis and tendency towards rigid spine in some	**Tightness** can develop in: – Spinal musculature – Neck musculature – Hip flexors – Plantarflexors – Shoulder girdle musculature **Scoliosis** is a risk as well as excessive spinal extension and sever lumbar lordosis **Muscle pain** can be a prominent feature

EDMD [52–54] Contractures are present early in relation to weakness, as detailed in Chapter 5, with less correlation to cycles of weakness and compensation

Characteristic weakness	*Compensatory/resultant posture*	*Tightness*
– Slowly progressive muscle weakness in humero-peroneal pattern: • Initially proximal in upper extremities • Initially distal in lower extremities • Progresses to proximal limb-girdle pattern including vastii muscles, hamstrings, and adductors – Selective early relative sparing of lateral gastrocnemius, and longer sparing of rectus femoris in EDMD2	– Posture of increasing spinal extension, elbow flexion, plantarflexion – Lateral trunk lean during ambulation – Use of compensatory support mechanisms to maintain head control – Use of compensatory mechanisms and momentum to optimize hand use	– Early contracture of: • Elbow flexors • Cervical spinal extensors • Plantarflexors – Eventual tightness throughout spinal extensors at all levels

Table 8.2 Assessment tools across the ICF

Impairment	Function	Disability measures	Quality of life/ participation/activity	Person reported outcomes
– Passive ranges of motion and measures of muscle extensibility	Upper and lower extremity functional scales [13, 44, 61]	– PEDI (pediatric evaluation of disability index) [62]	– Peds QL [63]	– Fatigue scales [64–66]
– Identification of risks of tightness, contracture, and deformity	Timed functional tests [67, 68]	– Functional independence measure (FIM, WeeFIM) [69, 70]	– PODCI [71–76]	– Rate of perceived exertion [77–79]
– Muscle strength testing [80, 81] • Manual muscle testing • Dynamometry • Computerized quantitative muscle assessment	GSGC (gait, stairs, Gowers, chair) [82, 83] North Star Ambulatory Assessment [88–93] Motor function measure (MFM) [95] Modified Hammersmith functional motor scale and extend [98, 99] Quick motor function test (QMFT) [100] Egan klassification (EK) [103] Alberta infant motor scales [104] Gross motor function measure (GMFM) [105] Peabody developmental motor scales-2 [106] Bruinincks-Oserestky test of motor proficiency-2 (BOT-2) [107] PUL [108] Endurance: 6 min walk test [109] Observational gait analysis and kinematic analysis of movement, function, and compensatory patterns of movement	– Rotterdam handicap scale [84]	– Canadian occupational performance measure (COPM) [85, 86] – Child Health Questionnaire [94] – Activities scale for kids (ASK) [96, 97] – Activity monitors [101, 102]	– Borg dyspnea scale [78, 87]

[a]Pain should be reported using age-appropriate pain scales [110–118]

activity around joints contribute to the development of hypoextensibility (tightness) and contracture [2, 6, 11, 30, 35, 119]. Positioning for function and for management of the musculoskeletal system should be offered [6, 21, 23, 120–122].

Stretching: Prevention/minimization of contracture requires sufficient daily elongation of musculature and daily movement through more complete ranges of motion than the individual with MD typically uses actively and independently [1, 2, 6, 11, 21, 36, 44, 45, 119, 123]; these may be achieved through preventative stretching and varied positioning, facilitation of movement and position changes, use of therapeutic interventions including passive and active elongation, daily range of motion/stretching, the appropriate use of orthotic intervention, splinting, serial casting, power positioning components on mobility devices, participation in aquatics and cycling/assisted cycling and other forms of submaximal active movement and participation, and the use of adaptive equipment for positioning and prolonged passive elongation including the use of standers and stand-and-drive mobility devices [5, 6, 11, 19, 21, 23, 24, 37, 44, 45, 55, 119, 123–127]. A stretching program should begin early in the course of the disease and is more effective and more easily established as part of the daily routine if it is begun before muscle tightness/contracture is established and before stretching is uncomfortable. A preventative stretching program should address structures known to be at risk for tightness based on natural history of the specific diagnosis, as well as any structures identified by individual assessment to be at risk for contracture [6]. Direct and skilled physical therapy techniques of muscle elongation, joint mobilization, gentle manual traction, and use of modalities and other manual therapy techniques to increase joint mobility and muscle elongation should be included as appropriate for individual patients based on recommendations after individual physical therapy evaluation [128] (Table 8.3).

Orthotic intervention/adaptive equipment: Orthotic intervention may be recommended for function and/or for assistance in management of the musculoskeletal system and may include consideration of many different choices, configurations, and materials, for upper and lower extremities, trunk, and neck. Lower extremity orthotic intervention may include consideration of ankle–foot orthoses (AFOs or "short leg orthoses"), knee–ankle–foot orthoses (KAFOs or "long leg braces"), knee extension splints, inframalleolar orthoses ("foot orthoses"), or other types and configurations of orthoses, with control of varying degrees of freedom depending on specific diagnosis and individual assessment [6, 21, 23, 123, 129]. Upper extremity splinting, orthotic intervention, and support may include splinting for stretching or support of function [130] and is increasingly including exploration of exoskeletons and robotics to increase functional use of hands in the presence of proximal weakness [131, 132].

Use of lower extremity orthotic intervention and adaptive equipment for *function* during walking typically depends upon the distribution of weakness and the required use of compensatory patterns of movement for function. In the presence of relatively greater proximal weakness in individuals who are independently ambulatory, such as in Duchenne muscular dystrophy (DMD), the use of AFOs during walking is not typically recommended. This is because AFOs tend to compromise ambulation by

Table 8.3 Muscles/joints/tissue commonly at risk for tightness in MDs (specifics depend on specific diagnosis)[a]

Lower extremities	Upper extremities	Spine
Muscles/soft tissue:	*Muscles/soft tissue*:	Spinal extensors (including cervical)
Hip flexors	Shoulder musculature	Intercostals
Iliotibial bands	Elbow flexors	Risk of:
Hamstrings	Forearm pronators	– Scoliosis
Plantarflexors, especially gastrocnemius	Wrist and finger flexors and/or extensors	– Excessive kyphosis
Posterior tibialis	Lumbricals	– Excessive lordosis
Plantar fascia		– Pelvic asymmetries and mal-alignment
Two joint muscles get tight first	Two joint muscles get tight first	Excessive anterior pelvic tilt
Joints—risk of contracture into:	*Joints—risk of contracture into*:	Excessive posterior pelvic tilt
Hip flexion	Elbow flexion	Lateral pelvic tilt
Knee flexion	Wrist flexion (or extension)	Horizontal pelvic rotation
Ankle plantarflexion	Flexion at isolated finger joints (PIP, DIP) Extension at isolated finger joints (PIP, DIP)	

[a]Any muscles, joints, soft tissue or structures may be at risk for tightness, contracture, deformity based on individual assessment of typical/chronic posture, function, muscle imbalance, compensatory patterns of movement, and the influence of gravity

limiting the use of compensations needed for walking, such as toe-walking or intermittent toe-walking, may compromise proximal compensations needed to keep the line of gravity behind the hip and in front of the knee to maintain ambulation, and may make it more difficult to get up from the floor, with the added weight of AFOs further compromising function [6, 129]. In other types of MD characterized by relatively greater distal than proximal weakness, or in which more global weakness is present, such as in some types of congenital myopathy, AFOs may assist in providing distal stability. This can be beneficial during standing and/or ambulation as long as AFOs are lightweight enough and offer optimal support without unnecessarily compromising function or movement that is necessary for function. Newer, ultra-lightweight carbon fiber AFOs used in conjunction with lower profile orthotic intervention at the foot and ankle may offer lightweight support without compromising function in those with greater distal than proximal weakness. This can potentially offer dorsiflexion assist during swing to prevent "foot drop" and "steppage gait" and potentially provide some floor-reaction support of knee extension during stance and may decrease fatigue. Ankle height or supramalleolar orthoses (SMOs) are not typically helpful because they add weight that challenges active dorsiflexion (typically weak in MDs) without adding dorsiflexion assist. However, these could be considered in the rare situation in which weakness is extremely mild, with good strength in anterior tibialis, but with poor medial–lateral alignment that requires more support than an inframalleolar orthosis. KAFOs may be useful in children with greater weakness

throughout lower extremities in the absence of the ability to support weight-bearing independently. This approach has been shown to extend walking for several years in some individuals with DMD when independent walking becomes too difficult because of inability to support weight through lower extremities without support and/or inability to maintain biomechanical positioning to mechanically lock joints in support of weight-bearing and ambulation [46, 55, 121, 133–136]. Braced ambulation with KAFOs may be therapeutic rather than functional across settings [46] and is most often used in combination with motorized mobility for functional, safe, independent mobility in settings in which braced ambulation is not functional or does not allow optimal participation.

Use of lower extremity orthotic intervention and adaptive equipment for *musculoskeletal management* (to prevent contracture and deformity) may include the use of AFOs [6, 21, 123], KAFOs [23, 55, 133], thigh binders, splints, serial casting [126, 127], or other positioning devices at night [21] or in the evening or during any portions of the day when they will not unduly interfere with function [6]. The use of AFOs at night has been shown to minimize the progression of plantarflexion contractures [21] and is recommended if tolerated. AFOs used at night need to be comfortable and should be custom molded and lightweight enough not to unduly restrict bed mobility. A bed or foot tent can hold the blanket up off of the feet to avoid the feet getting tangled in the sheets. Adjustable angle orthoses can sometimes be used to provide differing amounts of stretch at different times of the day, or gradually increasing elongation for comfort. The use of ankle height or SMOs may be helpful for those using a wheelchair full time, in order to assist in maintaining optimal medial–lateral alignment if the footplate of the chair can be successfully used to limit excessive plantarflexion. The number of hours per day that a muscle is in a lengthened vs. shortened position will influence the development or prevention of contracture. Standard recommendations for prevention of progressive contracture support the maintenance of a lengthened position for six of every 24 h [137]. The use of standers and stand-and-drive motorized mobility devices is recommended for providing prolonged passive elongation into simultaneous hip and knee extension in an upright weight-bearing for optimizing and maintaining joint range of motion, providing muscle elongation over multiple joints, and optimizing bone integrity, if tolerated [20, 55, 133, 134, 138, 139].

Prevention/minimization of spinal deformity typically involves: promotion of symmetry through the vertebral column and pelvis; support of appropriate amounts of extension and flexion at specific levels of the vertebral column; maintenance of flexibility; support of optimal posture; and minimization of the asymmetrical deforming forces of compensatory patterns of movements used for function (in most neuromuscular disorders) or intrinsic to diagnosis (such as in FSH). The progression of spinal deformity in neuromuscular disorders has been well studied, and understanding of the individual pathokinesiology in each diagnosis and detailed assessment and management of the interaction between components in each individual are critical. The development and progression of scoliosis has been most extensively studied in DMD, which can be used as a model to understand the pathokinesiology, and can inform conservative treatment. The natural history of scoliosis

is changing with the use of steroids in DMD, with scoliosis appearing later, and with less devastating progression [140].

Scoliosis in ambulatory individuals with DMD has been studied [141, 142] and is characterized by a flexible, functional scoliosis related to asymmetrical lower extremity position/contracture, pelvic obliquity, asymmetrical realignment of shoulders, head, and upper extremities [35, 143, 144]. Fixed spinal asymmetry is typically minimized spontaneously in ambulatory individuals by prolonged, protective spinal hyperextension and locking of posterior intervertebral facet joints at lumbar and lumbosacral levels, and alternating torso shift and lateral trunk elongation [11, 35, 145].

Historically, prolongation of ambulation by management of lower extremity contracture and the use of long leg braces appeared to slow the development of scoliosis in some [146], likely via prolongation of protective spinal hyperextension maybe through the adolescent growth spurt, and continued torso shift and lateral trunk elongation over symmetrical lower extremities [35, 145, 147, 148]. Factors that have appeared to influence whether or not scoliosis appears prior to final loss of ambulation included: the age at which walking ceases; intervention used or not used to prolong ambulation; and final gait pattern [146].

It has generally been agreed that spinal curves during the ambulatory period are not usually "fixed" (i.e., rigid or inflexible), are functionally necessary for ambulation, and cannot be corrected without risking the loss of ambulation [35]. Attempts should be made, therefore, to minimize long-term effects of asymmetry with stretching, positioning, etc., while allowing compensations necessary for function. In individuals with intrinsic asymmetry of weakness, as has been identified in FSH, and extreme anterior pelvic tilt and lumbar lordosis, the use of a soft corset during ambulation may provide support that decreases pain and fatigue during ambulation without compromising compensations to the extent that ambulation is compromised.

Scoliosis in non-ambulatory individuals: Scoliosis as a significant problem in DMD and other neuromuscular disorders typically either begins or develops more rapidly as ambulation is lost and full time use of a wheelchair begins [35, 149]. It is one of the most serious and disabling complications of many neuromuscular disorders and has been studied extensively in DMD, with the understanding of the principles of progression and treatment gained in DMD useful in the management of all neuromuscular scoliosis [150]. Neuromuscular scoliosis can progress to a level of incapacitating severity that compromises pulmonary function, sitting ability, upper extremity function, comfort, and cosmetic integrity [11, 35]. The progression of scoliosis is variable, however, and final deformity ranges from mild in some individuals to severe in others [150]. The significance of the variability is in the opportunity it offers for effecting change and for making use of intervention that may prevent or minimize the development of scoliosis. Attempts at successful management must be based on a comprehensive understanding of the factors that contribute to the development of scoliosis. Aggressive conservative management must be coordinated with consideration of surgical options in order to prevent the catastrophic progression to severe deformity in all individuals with MDs.

Factors that contribute to the development of scoliosis can be divided into those factors that make the spine vulnerable and those factors that initiate asymmetry [151].

Factors That Make the Spine Vulnerable [151]:

- *Severe symmetrical weakness in trunk musculature* [150, 152]
  - Decreases spinal support and stability.
  - Without external support, the spine is vulnerable to external forces it cannot oppose.
- *Rapid vertebral growth during adolescent growth spurt* [152, 153]
  - Often coincides with, or follows, the loss of ambulation.
  - Increases vulnerability to potentially deforming forces (the musculoskeletal system is known to be more vulnerable to any deforming force during periods of rapid growth).
- *Loss of protective spinal hyperextension* [11, 19, 154, 155]
  - Spinal hyperextension is decreased or eliminated when individuals begin to sit full time [156].
  - Posterior intervertebral facet joints are unlocked and allow more lateral flexion (bending) and rotation [19, 150, 156].
  - Stretching of posterior spinal ligaments increases with kyphosis [150].
  - Can be exacerbated by posterior pelvic tilt caused by tight hamstrings and lower extremity alignment in sitting.

Asymmetrical forces imposed on the symmetrically weak and vulnerable spine [151]:

- *Compensatory movement patterns* used:
  - *For stability*—Tend to lean on one arm of the wheelchair, may lean forward also—tends to push that shoulder up.
  - *For upper extremity (UE) function*—Use lateral trunk flexion towards the contralateral (opposite) side when elevating or abducting one upper extremity, in order to substitute for weak shoulder muscles, with persistent leaning towards the non-dominant side, may contribute to development of a curve with convexity towards the side of dominance [152, 157].
- *Pelvic position*:
  - *Posterior pelvic tilt* [11, 150]
    - Can further exacerbate an asymmetrical loss of spinal hyperextension by asymmetrically tightness in hamstrings [150]
  - *Pelvic obliquity* (lateral pelvic tilt) [11, 35, 150]
  - *Pelvic rotation* (in horizontal plane) [150]
    - Pelvic rotation and obliquity can be present in sitting from:
      - *Preexisting asymmetry of soft tissue contracture* around hips and pelvis [35] (for example: hip flexors, iliotibial bands)

- *Asymmetrical pelvic position* in the absence of asymmetrical contracture, from [11, 150]
  - Sling seat
  - Poorly fitting wheelchair
  - Any unstable sitting surface

– *Lower extremity position* [30, 35]

- *Hips* can have a direct effect on pelvis, then spine, as described above:
  - Asymmetrical hip flexor and/or iliotibial band tightness or contracture
  - Tight hamstrings leading to posterior pelvic tilt and kyphosis
- *Foot/ankle* asymmetrical contracture into equinovarus from unopposed posterior tibialis and gastrocsoleus—tighter side pushes pelvis back into ipsilateral posterior horizontal pelvic rotation.

The deforming force of **gravity** on the vertebral column increases in the presence of asymmetrical spinal-pelvic alignment that compromises the simple mechanical ability of the vertebral column to withstand the force of gravity. In addition, the resultant unequal distribution of weight on epiphyseal growth plates increases the potential for an initial flexible scoliosis to become structural.

Interaction Between Factors

- Symmetrically weak and vulnerable spine is present in all individuals with DMD when ambulation ceases.
- Particular vulnerability is present in those who lose protective spinal hyperextension. This is the initiating factor that is imposed upon the spine with the potential to cause asymmetry and progressive scoliosis. It may include any one of previously described factors and may be different in each person.
- Once asymmetry is initiated, secondary asymmetries are established and spinal deformities can progress in a self-perpetuating circle of weakness, compensation, and contracture.

Management of the spine must be anticipatory and preventative with consideration across the continuum of intervention options, including stretching, positioning, external support, and surgical options, with coordination between the multidisciplinary team. The use and timing of anticipatory and preventative conservative measures is coordinated with ongoing assessment regarding the potential need for surgical stabilization to manage curves that progress in spite of conservative measures. Care must be taken to coordinate with the rest of the team, with particular coordination between PT, orthopedics, pulmonary medicine, and cardiology, as conservative measures are employed. This helps ensure that the window of opportunity for surgical spinal stabilization (which is dependent on the interaction between pulmonary, respiratory, and cardiology status) is not missed, if the individual will need surgical stabilization at some point (see Chapter 9).

Intervention described in the literature has included prolongation of ambulation, external support including bracing, specialized seating systems, wheelchair modifications, promotion of upper extremity symmetry, control of lower extremity position, and spinal surgery. Bracing of the spine in individuals with DMD has historically not been tolerated or successful but may have a role in other diagnoses and situations, especially in younger children with myopathies characterized by more profound trunk weakness at earlier ages. Orthotic intervention may include supportive garments, corsets, or spine jackets in younger children with some types of MD in order to support more vertical, symmetrical, and extended spinal alignment and more stable posture and stability in upright. Such interventions may assist in maintaining spinal symmetry, or providing some support which may be beneficial in ambulatory individuals in whom some support is helpful but must avoid excessive restriction of movement that may limit compensatory movement required for ambulation [158–160].

Optimal support and positioning in seating systems is critical in musculoskeletal management of the spine and extremities and must include maintenance of midline, symmetrical pelvic position with prevention of lateral pelvic tilt, horizontal pelvic rotation, and excessive anterior or posterior pelvic tilt; maintenance of a midline erect spine, and support of a symmetrical midline head position. Typical recommendations include: a solid seat and back; rigid lateral trunk supports; hip guides; adductors; a head rest and adequate upper extremity and foot and leg support; with power positioning components for power tilt, power recline, separately elevating power elevating leg rests, power adjustable seat height, and power standing [6]. Seating system components are needed for support for function, prevention of progressive contracture and deformity, and maintenance of skin integrity. Power positioning components are needed for function, for independent position change for prevention of contracture and deformity, for support of adequate frequency and duration of weight-bearing throughout the day, and for provision of independent weight shift and pressure relief throughout the day that is adequate to maintain skin integrity.

Physical therapy management of the spine in the individual with MD must involve ongoing evaluation and intervention. Ongoing evaluation must attend to the asymmetrical forces acting on the vulnerable spine and should include assessment of:

- Pelvic position
- Spinal alignment including
  - Medial–lateral alignment
  - Rotational tendencies
  - Amount of extension
  - Symmetry vs. asymmetry
- Lower extremity position and its effect on the spine
- Compensatory movement patterns and positioning

Goals of PT Management of the Spine

- Maintain ambulation and standing as long as possible
- Promote spinal extension in sitting except in diagnoses or situations characterized by excessive extension, such as EDMD or in rigid spine syndromes [145]

- Maintain maximal symmetry of positioning in wheelchair
- Limit use of compensatory movement patterns that lead to deformity
- Provide for UE function with symmetry
- Maintain flexibility

Suggestions for Wheelchair Management

- **Wheelchair support/positioning**—the individual's chair should fit well and provide support that achieves:
- *Sitting position* characterized by:
  - A level pelvis without obliquity or rotation
  - A straight, erect, midline spinal position
  - Elimination of kyphosis and encouragement of extension except in diagnoses or situations characterized by excessive extension, such as EDMD or in rigid spine syndromes
  - Symmetrical LE position with good foot placement (not too much plantarvarus) and without hip abduction
- *Sufficient trunk support* so that asymmetrical leaning is not necessary for maintenance of an upright position
- *Control of asymmetrical movement patterns*
- Specific recommendations for *wheelchair seating system components* include:
  - Solid seat attached to frame of chair
  - Solid back attached to frame of chair
  - Pelvic control in all planes:
    - Hip control blocks (hip guides)
    - Seat belt appropriately located and/or adapted
    - subASIS bar?
  - *Knee pads* to control abduction (adductor pads)
  - *Planar, rigid, lateral trunk supports*—appropriately located and *strong* enough to:
    - Prevent the need to lean laterally for stability
    - Stop compensatory lean for UE function
  - *Control of lower extremity position*—might include:
    - Foot plate appropriately located and angled
    - Ankle straps
    - Padded footrests or foot cradles
    - AFO's
    - Surgical correction of ankle–foot deformity
  - *Arm rests* appropriately located to encourage spinal extension rather than kyphosis
  - *Chest strap* (in older individual) in order to provide additional support that centers trunk and allows leaning into lordosis [161]

  - *Lumbar roll* as appropriate to encourage spinal extension
  - *Head support* (customized as needed)

- Power tilt-in-space, power recline, with separately elevating power elevating leg rests, for
  - Changes in position, maintenance of skin integrity
  - Opening up of hip and knee angles to assist in minimizing the development of contractures
- Power standing
- Power seat elevation (power adjustable seat height)

Control of Asymmetrical Compensatory Movement Patterns

- Evaluate during **all** functional activities (wheelchair driving or propulsion, writing, eating)
- **Stop compensatory lean**!
- Provide for function with symmetry—might include:
  - Relocation of wheelchair controls (joystick)
    - Closer to hand on wheelchair arm to prevent need for reaching
    - Use of non-dominant hand?
    - Alternate sides periodically?
    - Central location? (but this can compromise stability and increase need for leaning)
  - Raised desk/tray/table height—works very well to allow pivoting of arm on elbow
  - Overhead sling
  - Balanced forearm orthoses
  - Robotic/exoskeleton forearm support
  - Other adaptive equipment
- **Standing**—to assist in control of LE contracture and to encourage spinal extension as well as offering more general physiological benefits and increased function
- **Maintaining flexibility**
  - Elongation in prone, supine, or sidelying to maintain symmetrical lateral elongation and flexibility
  - Maximally preventing contractures in lower extremities
- **Parent/child education**
  - Educate individual and caretakers about symmetry vs. asymmetry and goals of spinal management as described above
  - Have individual monitor symmetry vs. asymmetry with visual feedback at mirror periodically, and when making changes in support or positioning to establish accurate “feel” of symmetry

It is important to stop and consciously reassess postural alignment at regular intervals—even as frequently every three months in addition to daily awareness.

The above spinal management plan outlines conservative measures that can be used in an attempt to prevent the progression of scoliosis in individuals with DMD. Close coordination with the rest of the medical team is important in identification of those individuals in whom conservative measures are not working so that more aggressive means, such as surgery, can be used for spinal management.

Spinal surgery is discussed in detail elsewhere (see Chapter 9).

Optimizing strength

Concern about whether or not strengthening activities hasten the progression of weakness in dystrophic muscles are longstanding and exist for many reasons, yet precise knowledge regarding what types of muscle activity may be detrimental or beneficial is limited [22, 172–187]. A certain amount of muscle activity has been assumed to be beneficial in preventing disuse atrophy, maintaining residual strength, providing or maintaining a potential trophic influence of active movement on muscle, and maintaining functional status and flexibility [174, 182, 183, 187]. Overwork weakness, however, should be avoided, as should exercise-induced damage [182, 183]. Eccentric muscle activity and maximal resistive exercise are believed to be detrimental to fragile muscles and should be avoided [183]. Submaximal aerobic exercise within the limits of pain and fatigue is generally supported, balanced by the use of energy conservation techniques for support of function and participation [174, 182, 183, 187] with respiratory muscle training supported by some with similar caution about overexertion [167, 188–192].

Managing/minimizing pain

This often involves assessment and correction of posture; assessment and correction of abnormal or excessive pressure imposed by abnormal posture, immobility, and abnormal weight-bearing with decreased ability to change positions; muscle tightness and/or over-lengthening, imbalanced muscle activity, and functional compensations; patterns and presence of overuse; fatigue; with consideration of other factors such as fracture and cardiac etiology important in settings of acute, new onset, or changing pain [193]. The use of energy conservation techniques and analysis of ergonomics during function are important in prevention and reduction of pain, as is the provision of appropriate adaptive equipment to support function, movement, position change, and pressure relieving surfaces for sitting and sleeping. More direct treatment for relief of pain should be coordinated by the multidisciplinary team and may include PT interventions using modalities of heat, cold, TENS, and massage [47].

Respiratory Management [162–164]

- Comprehensive evaluation and management by pulmonary medicine specialists is recommended [162, 163, 165, 166]
- Respiratory function can be compromised by a number of factors:
  - Progressive muscle weakness interacts with spinal/thoracic deformity to result in severe decline in pulmonary function.
  - Intrinsic lung disease is *not* typically present.
- Involvement typically includes:

Less effective breathing due to muscle weakness

- Weakness may present and progress in respiratory muscles including diaphragm, intercostal muscles, abdominal muscles, and accessory muscles of respiration such as neck flexors, depending on the specific diagnosis.
- A diaphragmatic pattern of breathing may be used with very little intercostal activity. This restricted pattern of breathing and increasing muscle weakness leads to an inability to expand and compress the lungs fully.
- Total lung capacity, vital capacity, and forced inspiratory and expiratory abilities decrease and residual volume increases.
- Progression:

Shallow breathing
↓
More rapid breathing (to get rid of CO_2)
↓
Less chest or lung volume/expansion
↓
Decreased breathing volume

- **Decreased lung expansion**: leads to little areas of collapse of lung tissue (i.e., atelectasis vulnerable to infection).
- **Decreased coughing ability**: due to weakness in abdominals and muscles of forced expiration as well as decreased ability to take a deep breath just before coughing. This leads to retention of secretions.
- **Restricted thoracic mobility** and stiffening of the chest wall result from fibrous replacement of the muscles of the thoracic wall as well as from restricted patterns of breathing and decreased lung movement. This leads to further decrease in lung mobility and expansion. It may be accompanied by ankylosing of the joints.
- **Impact of spinal deformity** on respiratory status: Respiratory insufficiency compounded by scoliosis when present.
- **Goals of interventions**:
  - Maintain chest wall mobility
  - Maintain strength and endurance in respiratory muscles as much as possible, possibly with submaximal exercise, especially when young (and also by providing them with sufficient rest with non-invasive ventilation as needed) [167].

- Establish and maintain most efficient breathing pattern possible
- Establish good pulmonary hygiene
- Coordinate with pulmonary team
- Support appropriate use of noninvasive inspiratory and expiratory aids

- **Suggestions**:
  - Inspiratory exercises/segmental breathing
    - To strengthen diaphragm gently, as appropriate, depending on diagnosis
    - For lung expansion and chest wall mobility
    - For more efficient breathing
  - Swimming
    - Breath control
    - Breathing patterns
    - Endurance
  - Practice coughing and use of mechanical assistance (manual assistance, ambu-bag)
  - GPB—glossopharyngeal breathing
  - Airway clearance techniques with postural drainage as necessary, with use of percussion or oscillatory vest
  - Periodic review of pulmonary hygiene techniques for at home
  - Spinal program to attempt to avoid potential further compromise of respiratory system by scoliosis
  - Inspiratory muscle aids: for example, nocturnal or daytime non-invasive ventilatory support
  - Expiratory muscle aids: for example, mechanical insufflation–exsufflation (MIE)—Cough Assist™
  - Coordination with team for anticipatory management regarding potential tracheostomy if necessary.

Cardiac [168]

Cardiac muscle can be affected by the dystrophic process and anticipatory, preventative, comprehensive evaluation and management by cardiology is recommended [168–171]. Myocardial fibrosis may occur, primarily involving the free wall of the left ventricle. Cardiac involvement may also be affected by respiratory status and by scoliosis that, if severe, can cause direct cardiac compression. Cardiac involvement is frequently progressive and may be eventually characterized by the ECG abnormalities, hypertrophic cardiomyopathy, and dilated cardiomyopathy [171].

Cardiac involvement across the spectrum of MDs may also include AV block, atrial paralysis, atrial fibrillation or flutter, ventricular arrhythmia, conduction defects, and reduced ejection fraction [171].

Cardiac involvement in Becker muscular dystrophy [171] is often out of proportion with skeletal muscle involvement, additionally taxed by increased level of gross motor activity, with cardiac transplantation a viable option in some cases. Emery Dreifuss muscular dystrophy (EDMD) is typically characterized by cardiac conduction defects [53]. Cardiac care of individuals in MDs is more anticipatory and preventative than in the past. With increased survival, pacemakers and defibrillators are beginning to be used for some individuals [169].

Carriers of DMD/BMD may have cardiac manifestations and should be assessed and followed [170].

Maintaining Function

- At every age, and every stage, age-appropriate function, participation in all aspects of life in which the individual is interested, and maximal independence should be supported.
- The bottom line should always be—"can he/she keep up with his/her peers?"
- Technology is the key to freedom in many situations.

Adaptive Equipment and Assistive Technology

- Mobility devices:
  - Manual, motorized, power assist, scooters
  - Custom seating
  - Power positioning components:
    - Power tilt
    - Power recline
    - Separately elevating power elevating leg rests
    - Powered seat elevation
    - Powered standing and powered stand and drive
- Cycles, power assist cycles
- Standers
- Power adjustable beds and pressure relieving mattresses
- Lifts and transfer devices, powered lift (including ceiling lifts, pivot lifts, stair lifts, powered patient lifts)
- Upper extremity supports (forearm supports, robotics elbow blocks to keep hand from sliding away from joystick)
- Mini-proportional joy sticks
- Computer access, infra-red environmental control, bluetooth, voice activation, eye gaze systems
- Internet access
- Environmental control units (infra-red and bluetooth)
- Prism glasses for reading in bed or with limited mobility in neck flexion
- Bidets

- Bath and shower chairs
- Respiratory equipment:
  - Cough assist
  - BiPAP and noninvasive ventilation
  - Vest
- Ramps, portable ramps, van lifts, vertical platform lifts
- Bathing and bathrooming equipment that fosters ease and independence
- Power operated beds
- Handheld devices (smart phones, tablets, etc.)
- Modified sports equipment

Sports/Adapted Sports

- Swimming, cycling, wheelchair/adapted sports, dance

Conclusion

Remarkable advances and progress in research raise hopes for finding treatments and cures for many of the genetically determined neuromuscular disorders. If quality of life is the focus for all individuals as we wait for more specific treatment and cures, effective intervention can be offered in many areas by using continually updated skills and resources, ingenuity, and a comprehensive understanding of each neuromuscular disorder. Comprehensive, anticipatory physical therapy (PT) management of MD is based upon an understanding of the pathokinesiology of each type of MD, an understanding of the progression of the pathokinesiology over time, individual evaluation within the context of each individual's life and goals, and provision of consistent, anticipatory, preventative management across the lifespan in order to minimize the clinical and functional impact of the diagnosis and to optimize quality of life. Optimal management is important for each individual not only for the sake of each day that is experienced as we wait for a cure but also for protection of the future that unfolds for that individual, and in order to help individuals stay in the best possible condition to make use of cures as they are found.

Helpful Websites

Information About Diagnoses

http://www.parentprojectmd.org
http://www.mdausa.org/
http://www.mdausa.org/disease/40list.html (list of diagnoses covered by Muscular Dystrophy Association (MDA)

http://www.fsma.org/
http://www.ninds.nih.gov/disorders/charcot_marie_tooth/detail_charcot_marie_tooth.htm
http://www.pompe.org.uk/
http://www.amda-pompe.org/
http://www.pompe.com/healthcare/pc_eng_hc_main.asp
https://www.genetests.org/
http://www.emedicine.com/neuro/topic668.htm
http://curecmd.org/
http://www.childmuscleweakness.org/

Adaptive Equipment/Assistive Technology/Orthotic Intervention

Orthotic Intervention

http://www.dafo.com/

Lifts, Bathing, and Bathrooming Equipment

http://www.arjo.com/
http://www.image-management.com/
http://www.surehands.com/
http://www.bfl-inc.com/index.php

Respiratory Care

www.coughassistt70.respironics.com
http://www.thevest.com/

Oximeters: http://www.pulseoximeter.org/

Car Seats for Fragile Infants

http://www.eztether.com/index.php/instructions/hope-car-bed

Standers

http://www.easystand.com/
http://mulhollandinc.com/products/rocket/
http://www.permobil.com

Wheelchairs/Mobility Devices

http://www.permobil.com
http://www.pridemobility.com/
http://www.dekaresearch.com/ibot.shtml
http://www.frankmobility.com/e-fix.php

Cycling

http://www.exnflex.com/

Other

http://www.portable-wheelchair-ramps.com/
http://www.ableplay.org/
http://accessiblelivingltd.com/portableramps.htm
http://www.aelseating.com/
http://www.easystand.com/
http://www.exnflex.com/
http://www.ezlock.net/
http://www.invacare.com/cgi-bin/imhqprd/index.jsp
http://www.kayeproducts.com/
http://www.adaptivemall.com/lectoilshowc.html
http://www.mulhollandinc.com/
http://www.ncatp.org/
http://www.portable-wheelchair-ramps.com/
http://www.pridemobility.com/
http://www.primeengineering.com/
http://www.quickie-wheelchairs.com/
http://www.rifton.com/index.htm
http://www.ezonpro.com/index.shtml
http://www.adaptivemall.com/tilslidrecba.html
http://www.duralife-usa.com/index.htm?group=5&content=2005
http://www.usatechguide.org/itemreview.php?itemid=846
http://www.permobilusa.com/templates/startpage.aspx?id=806
http://www.nadachair.com/
http://www.panthera.se/en/produkt_x.html
http://kinovarobotics.com/
http://www.pro-bed.com/
http://www.volker.co.uk/index.php

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