PHYSICAL THERAPY MANAGEMENT

OF

DYSTROPHINOPATHIES

(Duchenne and Becker Muscular Dystrophy)

Laura E. Case, PT, DPT, MS, PCS
(case0001@mc.duke.edu)

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I. Introduction

II. Dystrophinopathies
- A. Pathology
- B. Pathokinesiology
  - 1. musculoskeletal involvement
    - a. ambulatory stages
      - - early
      - - transitional
    - b. non-ambulatory stage
    - c. scoliosis
  - 3. respiratory involvement
  - 4. cardiac involvement

III. Physical therapy management

- A. overview
- B. assessment
- C. intervention strategies
- D. stretching, positioning, & prevention of deformity
- E. strengthening
- F. respiratory program
- G. prolongation of ambulation
- H. spinal management
- I. mobility
- J. functional independence
- K. comfort

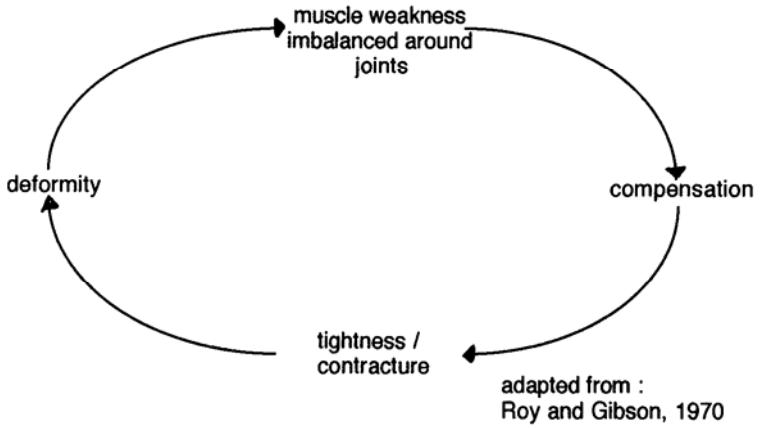
Dedicated to Ray Bowser, who enriched the lives of all who knew him; who taught me, or pushed me to learn, much of what I know about muscular dystrophy; and who lived his life with dignity, wisdom, and a sparkle in his eyes.

INTRODUCTION

Remarkable advances in molecular biology and genetics raise hopes for finding cures for many of the genetically determined neuromuscular disorders. As researchers work to find cures, clinicians continue to work with individuals with neuromuscular disorders to minimize the clinical impact of the cellular pathology of each disorder (e.g. the myopathic process that occurs with dystrophin deficiency), to prevent secondary 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 If quality of life is the focus for all individuals as we wait for a cure, 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 management, based on a thorough understanding of the pathophysiology and pathokinesiology of the diagnosis can offer individuals with dystrophinopathies a higher quality of life and can minimize the clinical and functional impact of the diagnosis. 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 for protection of the future that unfolds for that individual, and in order to help individuals stay on the best possible condition to make use of cures as they are found.

Dystrophinopathies are progressive neuromuscular disorders characterized on a cellular level by genetically based dystrophin deficiency and characterized clinically by progressive muscle degeneration. The pathokinesiology of disease progression is well understood in terms of cycles of progressive weakness, postural compensation, development of contracture / deformity, loss of function, and respiratory/cardiac compromise[1-11]. Identification of the genetic defect responsible for dystrophinopathies and the protein for which the gene is responsible, has been followed by greatly expanded, but still incomplete, understanding of the pathophysiology of dystrophin deficiency and by beginning success in the use of vectors to transport the gene into muscle cells with resultant expression of dystrophin, the use of antisense oligonucleotides to repair mutations, and other treatments that may significantly change the course of DMD/BMD and may one day offer a cure. Once believed to be different clinical entities, Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are now known to be variations on the continuum of a dystrophinopathy caused by different amounts / types of damage to the same gene locus.

PATHOKINESIOLOGY OF DISEASE PROGRESSION



A self-perpetuating circle of events was described in DMD by Roy and Gibson[12] in which imbalanced muscle weakness, compensatory movement patterns and postural habits, and the influence of gravity interact in the progression of disability. Weakness progresses proximal to distal and is first evident in muscles around the shoulder and pelvic girdles. In addition to weakness that occurs due to actual muscle degeneration, weakness may also seem to "progress" in proportion to growth. 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. As weakness increases, compensatory alterations are made in posture and movement to mechanically lock joints and substitute for lack of adequate muscle strength. The substitutions are effective in maximizing function but eventually lead to muscle tightness, contracture and deformity that feed into increasing weakness and disability. Effective intervention is that which is focused on breaking the vicious circle of events whenever possible so that strength is maximally maintained, contracture and deformity are minimized, and compensations can be used to maximize function without leading to increases in disability.

The clinical course of DMD is changing with the use of steroids[13-25] with the potential to delay / prevent / minimize the secondary musculoskeletal impairments that have been part of the natural history. The principles of management of these impairments remain important, nonetheless, in order to work towards the highest levels of musculoskeletal status and function for each individual at each step along the way – with the hopes that greater success in management will be achieved for more individuals with less need for aggressive intervention.

The key to management of DMD, and in fact most neuromuscular disease, is it's predictability. Clinically, DMD is one of the most predictable neuromuscular disorders found in pediatrics. Muscle weakness progresses in a specific and well known order. Predictable compensations are used to cope with this increasing weakness. Specific muscle tightness, contracture, and deformity can result and occurs in predictable sequences without intervention. This predictability is a double-edged sword. On the one hand, the predictability is evidence of that which we cannot change. But on the other hand, knowledge of the predictable progression empowers us to plan ahead and treat with prospective, preventative, anticipatory management. Many of the devastating secondary effects of the intrinsic dystrophic process can be minimized with comprehensive, ongoing, anticipatory, and preventative management that maintains the highest possible quality of life despite disease progression.

DUCHENNE MUSCULAR DYSTROPHY:

Disease progression in DMD is often divided into 3 stages: early, transitional, and late or non-ambulatory. Comprehensive management of Duchenne must be based on a thorough knowledge of the progression of and interaction between weakness, compensation, and tightness/deformity at each stage.

EARLY STAGE

weakness in:

- hip extensors (gluteus maximus)
- ankle dorsiflexors
- hip abductors (gluteus medius)
- hip adductors
- abdominals
- neck flexors (sternocleidomastoid)
- shoulder depressors and extensors (lower trap & latissimus)
- shoulder abductors (deltoids)
- elbow extensors (triceps)

compensations:

- * ↑ 'd lumbar lordosis (posterior trunk lean) to keep force line behind hip joint (initially see less anterior pelvic tilt as hyperextension is used at hip joint in stance as long as quadriceps are strong enough to counteract force moment into knee flexion)
- lack of heelstrike
- ↑ 'd 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
- ↑ 'd upper extremity (UE) abduction and lateral trunk sway
- cadence ↓ 's
- Gower's maneuver

* neck and UE weakness not usually noticeable functionally but apparent with testing

Typical early picture is to see "clumsiness", delayed achievement of motor milestones of crawling and/or walking, and "flat feet".

TRANSITIONAL STAGE

weakness : - progresses in muscles listed under early stage
** quadriceps weakness - ** key to real gait deterioration **
- ankle everters (peroneals)

compensations:
** must get line of gravity simultaneously in front of knee joint and behind hip joint - use: - anterior pelvic tilt
- diminished hip extension in stance
- base of support widens:
- balance
- tight iliotibial bands
- ↑ 'd 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 of 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 to remember that 2 joint muscles get tight first ****

functional losses: in activities of elevation against gravity
- ability to rise from the floor.
- stairclimbing.
- rising from a chair.

maximally compensated gait:

- standing and walking way up on toes with wide base of support and extreme lumbar lordosis.
- extreme lateral trunk lurch and UE abduction
- neck used as final adjustment mechanism with extreme flexion with chin tucked

** may be asymmetrical with child standing with weight on one foot and balancing with other in extreme plantarflexion.

** the time period in which maximally compensated gait is used is the last time that the use of long leg braces, with or without lower extremity surgery, can be considered in order to prolong ambulation.

LATE OR NON-AMBULATORY STAGE

weakness: - continues to progress relentlessly in muscles listed previously and becomes profound.
- UE weakness becomes more significant functionally and is imbalanced:
- elbow extension weaker than flexion.
- forearm supination weaker than pronation.
- wrist & 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

compensations:
- prior to the loss of ambulation, most compensations are used to maintain an upright posture and facilitate ambulation.
- after the 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.

tightness: -accelerated development of LE contractures
- beginning development of UE contractures:
- tightness into elbow flexion and pronation.
- tightness in wrist and finger flexors.
- may not be significant contractures around shoulder girdle.
- tightness in neck (cervical) extensors

***scoliosis**: the development of scoliosis is a major complication of the late, non-ambulatory, or "wheelchair" stage

functional losses in: - UE abilities - sitting ability
- head control
- ADL - bathrooming, constipation, sleeping comfort

SCOLIOSIS

Considered a major complication of the "wheelchair" stage, scoliosis has been described during the ambulatory stage. Scoliosis that emerges during the ambulatory stage is a flexible, functional scoliosis related to the final, asymmetrical, maximally compensated gait pattern used prior to the loss of ambulation. Fixed scoliosis during the ambulatory stage is minimized spontaneously by protective spinal hyperextension and lateral trunk lurching and cannot be interfered with without compromising ambulatory status.

Scoliosis in ambulatory stage:

- flexible, functional scoliosis related to:

    - asymmetrical LE position/contracture
    - pelvic obliquity
    - asymmetrical realignment of shoulders, head, UE

** fixed spinal asymmetry is minimized spontaneously at this stage by:

    - prolonged, protective spinal hyperextension and locking of posterior intervertebral facet joints at lumbar and lumbosacral levels.
    - alternating torso shift and lateral trunk elongation.

- with prolongation of ambulation by LE surgery and bracing, may get slower development of scoliosis via:

    - prolongation of protective spinal hyperextension maybe through adolescent growth spurt.
    - continued torso shift and lateral trunk elongation over symmetrical lower extremities.

** factors that influence whether or not scoliosis appears prior to final loss of ambulation:

    - age at which walking ceases.
    - intervention used or not used to prolong ambulation.
    - final gait pattern.

- generally agreed upon is that:

    - these curves are not fixed.
    - they are functionally necessary for ambulation.
    - they should not be corrected or ambulation will be lost.
    - attempts should be made to minimize long term effects of asymmetry with stretching, positioning, etc...while allowing compensations necessary for function.

Scoliosis in non-ambulatory period:

Scoliosis as a significant problem in Duchenne muscular dystrophy is that which either begins or develops more rapidly as ambulation is lost and full time use of a wheelchair begins. It is one of the most serious and disabling complications of Duchenne. Scoliosis can progress to a level of incapacitating severity that compromises pulmonary function, sitting ability, upper extremity function, comfort, and cosmetic integrity. The progression of scoliosis is variable, however, and final deformity ranges from mild in some individuals to severe in others. 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 in all individuals with Duchenne muscular dystrophy the catastrophic progression to severe deformity.

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.

factors that make the spine vulnerable:

- <u>severe symmetrical weakness in trunk musculature</u>:

    - decreases spinal support and stability
    - without external support, the spine is vulnerable to external forces it cannot oppose

- <u>rapid vertebral growth during adolescent growth spurt</u>:

    - 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).

- <u>loss of protective spinal hyperextension</u>:

    - spinal hyperextension is decreased or eliminated when the boys begin to sit full time:
    - posterior intervertebral facet joints are unlocked and allow more lateral flexion (bending) and rotation.
    - stretching of posterior spinal ligaments with kyphosis
    - 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:

- 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 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, a curve with convexity towards the side of dominance can develop.

- pelvic position:

    - posterior pelvic tilt - can further exacerbate an asymmetrical loss of spinal hyperextension by asymmetrically tightness in hamstrings

    - pelvic obliquity (lateral pelvic tilt)

    - pelvic rotation (in horizontal plane)

        **pelvic rotation and obliquity can be present in sitting from:
        - pre-existing asymmetry of soft tissue contracture around hips & pelvis (example: hip flexors, iliotibial bands)
        - asymmetrical pelvic position in the absence of asymmetrical contracture, from:
            - sling seat
            - poorly fitting wheelchair
            * any unstable sitting surface

    - lower extremity position:

        hips can have a direct effect on pelvis, then spine, as described above:
        - asymmetrical hip flexor +/or iliotibial band tightness or contracture
        - tight hamstrings leading to posterior pelvic tilt & kyphosis

        foot/ankle asymmetrical contracture into equinovarus from unopposed posterior tibialis & gastrocsoleus - tighter side pushes pelvis back into posterior horizontal pelvic rotation on same side.

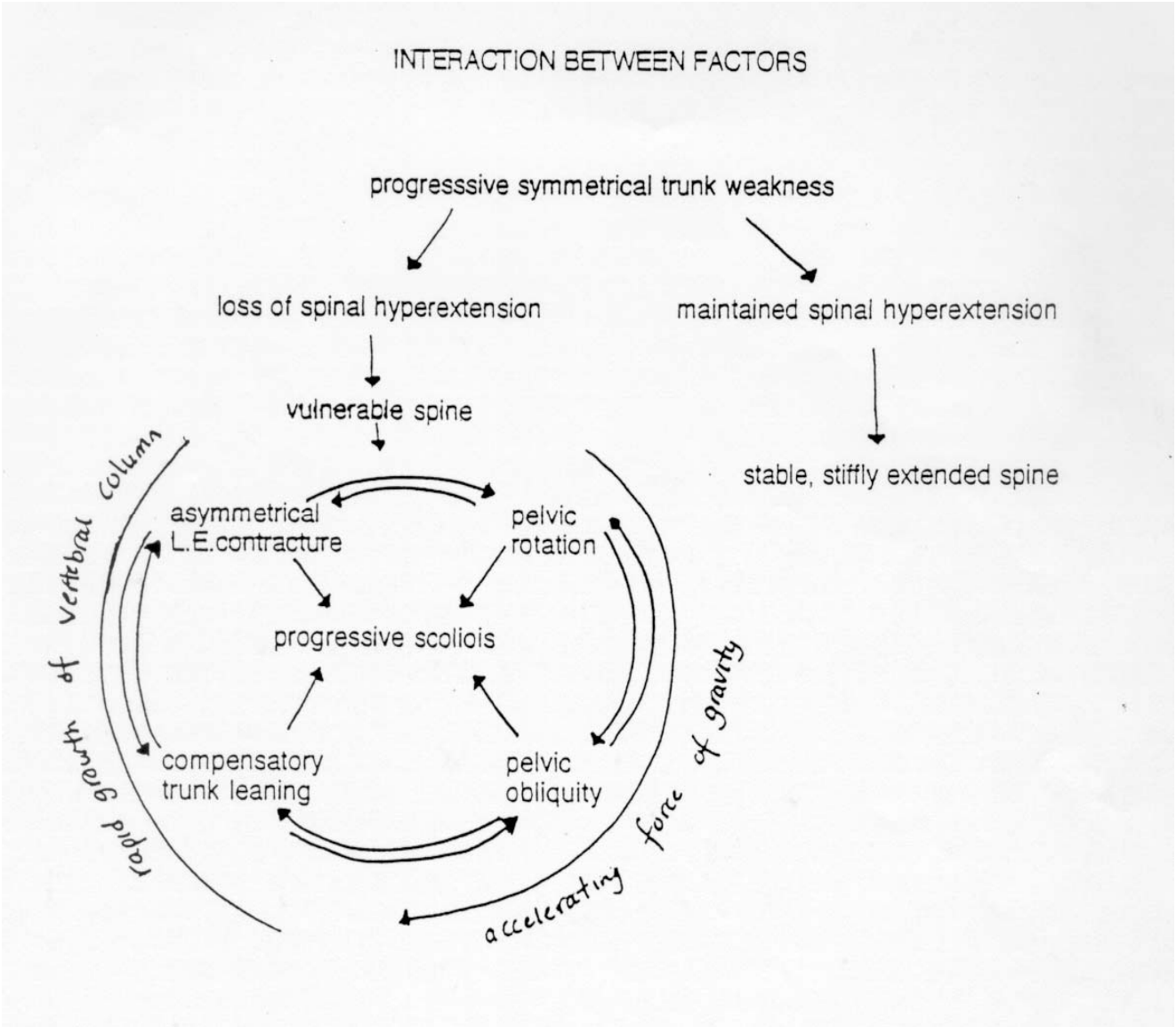
The deforming force of **gravity** on the vertebral column increases in the presence of asymmetrical spinal-pelvic alignment that compromises the simple mechanical ability of the vertebral column to withstand the force of gravity. In addition, the resultant unequal distribution of weight on epiphyseal 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 is lost.
- particular vulnerability is present in those who lose protective spinal hyperextension

** initiating factor that is imposed upon spine with potential to cause asymmetry and progressive scoliosis may be 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.



typical patterns of progression:

- two typical pathways described by Wilkins & Gibson[26]:

    - "unstable pathway" to severe scoliosis
    - "stable pathway" to hyperextension without scoliosis

- three distinct clinical courses described by Rideau[27]:

    - rapidly progressive with severe scoliosis
    - moderately rapid progression with varying amounts of scoliosis
    - more slowly progressive without scoliosis

- "natural" progression described by Hsu[28]

impact of scoliosis:

- respiratory

- sitting ability - scoliosis compromises sitting more than does weakness[28]:

    with curves > 40 degrees, cannot sit without propping on arms.
    with curves > 80 degrees, cannot tolerate sitting for long periods.

- UE function - can be compromised by having to prop on upper extremities:

    * since weakness progresses proximal to distal, hand function should remain relatively well preserved until late in the disease and should be maximized as an area of greatest residual function.

    * premature loss of effective hand function because of scoliosis is unnecessarily disabling.

- comfort

- cosmesis

OSTEOPOROSIS / RISK OF FRACTURE

Osteoporosis and increase risk of fracture has been reported. Osteoporosis in the long bones of ambulatory individuals with DMD has been correlated with the level of muscle weakness[29] and is present long before ambulation is lost[30, 31]. Osteoporosis in the long bones of individuals with DMD decreases significantly after the loss of ambulation, is lower in long bones than in the vertebral column[31] is worse in the lower extremities than in the upper extremities, and is exacerbated by the use of steroids[32]. Long bone fracture in individuals with DMD occurs most frequently with a fall[29] but can also occur without a history of trauma[33]. Individuals with DMD show varied levels of decrease in BMD in the vertebral column while ambulatory[31, 32] with rapid losses in vertebral BMD once ambulation is lost[31] and with increased rate of verebral compression fracture with the use of steroids[34-36].

RESPIRATORY INVOLVEMENT

Respiratory insufficiency is the major cause of death in DMD with 90% of patient death related to chronic respiratory failure and its complications. 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 present.

Involvement includes:

Less effective breathing due to muscle weakness:

- Weakness progresses in intercostal muscles, abdominal muscles, and neck flexors with relative sparing of diaphragm until later in the disease. ** Relative sparing of the diaphragm is important because the diaphragm is responsible for 70% of quiet respiration. It is the primary muscle of respiration and can be strengthened.

- A diaphragmatic pattern of breathing is 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. Coughing becomes difficult when forced vital capacity is < than 1.25 liters an/or when peak flow cough is < than 200-160 liters/minute (Michelle Eagle, PT, PhD – personal communication & presentation 7-03 at Parent Project Muscular Dystrophy Conference)

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: The respiratory insufficiency of DMD may be compounded by scoliosis when present.

**** see consensus statement and guidelines:**
Finder, JD, Birnkrant, D, Carl, J, Farber, HJ, Gozal, D, Iannaccone, ST, Kovesi, T, Kravitz, RM, Panitch, H, Schramm, C, Schroth, M, Sharma, G, Sievers, L, Silverstri, JM, Sterni, L: Respiratory Care of the Patient with Duchenne Muscular Dystrophy. American Journal of Respiratory and Critical Care Medicine Vol 170, 456-465, 2004 by

CARDIAC INVOLVEMENT

Cardiac muscle is affected by the dystrophic process. Myocardial fibrosis may occur, primarily involving the free wall of the left ventricle. Cardiac involvement may be 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, dilated cardiomyopathy.

Cardiac involvement across the spectrum of muscular dystrophies may also include AV block, atrial paralysis, atrial fibrillation or flutter, ventricular arrhythmia, conduction defects, reduced ejection fraction.

Cardiac involvement in Becker muscular dystrophy 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.

Carriers may be have cardiac manifestations and should be assessed and followed.

**** see consensus statements regarding management of cardiac involvement in muscular dystrophy **:**

Cardiovascular Health Supervision for Individuals Affected by Duchenne or Becker muscular dystrophy, Pediatrics Vol 16 No 6, 2005

Bushby, K., Muntoni, F., and Bourke, J.P. (2003). 107th ENMC International Workshop: The management of cardiac involvement in muscular dystrophy and myotonic dystrophy. 7th-9th of June 2002, Naarden, the Netherlands - Neuromuscular Disorders 13(2003) 166-172

PHYSICAL THERAPY MANAGEMENT

ASSESSMENT

Assessment must be ongoing and comprehensive so that intervention can be timely and anticipatory. Specific areas of weakness, tightness, and compensation must be identified in order to allow intervention that maximizes strength, prevents deformity, and provides for effective adaptive functioning to the greatest extent possible.

- range of motion (ROM)
- postural alignment
- spine: standard assessment plus:
- presence or absence of functional compensatory leaning.
- influence of pelvic and LE positioning/contracture in all planes in sitting and standing.
- tendency towards hyperextension vs kyphosis.
- positioning and support required in wheelchair, if applicable.
- developmental/functional status:
- assessment in all positions and transitions between positions.
- compensatory movement patterns and resultant risks.
- activities of elevation, antigravity function.
- manual muscle test (MMT).
- respiratory status:
- vital capacity
- patterns of breathing
- cough
- knowledge of pulmonary hygiene
- gait/mobility status:
- assessment of gait including patterns used to compensate for weakness, asymmetries, influence of tightness/contracture
- functional mobility in all situations
- endurance
- wheelchair or mobility status/planning:
  manual wheelchair
  motorized device
- fine motor status
- adaptive equipment, including wheelchair (see spinal management for specifics of assessment of wheelchair and wheelchair positioning)
- orthotics/casting/bracing
- weight
- functional independence and caregiver/home status
- activities of daily living (ADL)
- functioning at home and at school
- typewriters, computers
- environmental control
- support system for child and caregiver
- physical environment and accessibility
- transfers
- comfort (example// sleeping)
- child and family's understanding of diagnosis
- child and caregiver's inputs and concerns

INTERVENTION STRATEGIES

Physical therapy in early care:

- assessment
- education of family
- prevention of deformity
- maximization of strength and functional capabilities
- establishment of therapy programs and home programs
- intervention to maintain ambulation (if appropriate)

Physical therapy in middle stage care (following loss of ambulation):

- continuation of early stage program, plus:
- aggressive spinal program
- wheelchair management
- adaptive functioning
- transfers/body mechanics emphasis...home consults

Physical therapy in late stage care:

- continuation of above program, plus:
- consultation regarding fatigue levels, positioning and endurance in positions, comfort, maximizing of UE function and vocational/classroom adaptation.

STRETCHING, POSITIONING, & PREVENTION OF DEFORMITY

With weakness and compensation there is often no way to eliminate a compensatory position without eliminating the function (for example, ambulation, UE function), but we must try to find compensations that pose less of a risk of deformity and we must try to avoid the contractures that contribute to the self perpetuating evolution of weakness/ contracture/ functional loss. With more severe weakness, positioning for function and for management of the musculoskeletal system should be offered. The effects of chronic positioning, the unopposed influence of gravity, and imbalanced muscle activity around joints contribute to the development of hypoextensibility (tightness) and are most successfully managed preventatively by support of adequate positioning augmented by daily range of motion / stretching, appropriate use of splinting, casting, orthotic intervention and adaptive equipment, and standing if possible.

In Duchenne muscular dystrophy, the most severe contractures occur in 2 joint muscles and those that have a postural function. Characteristic tightness over the course of the disease occurs in:

lower extremity:

iliotibial bands
tensor fascia lata
hip flexors
hamstrings
gastrocsoleus
posterior tibialis

upper extremity:

elbow flexors
forearm pronators
wrist/finger flexors

thorax

spine
(including cervical spinal extensors or “neck extensors” in older individuals)

- passive stretching - must be done daily and is best augmented by standing

  **active assistance - use whenever / wherever possible to counteract pushing

  - PNF contract/relax - use whenever / wherever possible

  - joint mobilization - patella (knee cap)
    - anterior/posterior movement of tibia on femur
    - traction - on all joints
    - elbow

  - myofascial release techniques

- modalities: may be used - for comfort
  - for a more effective stretch
  - for a more physiological stretch

- heat: increases comfort, increases plasticity in tissues but have to avoid excessive generalized heat that can cause fatigue & compromise strength

  - hot packs - localized use on:
    - knees
    - hands
  - after bath time at home

- casting/orthotics:
  - for use at night or during rests periods (e.g. when watching TV)
    - ankle foot orthoses (AFO's) for plantarflexor tightness
    - wrist / finger splints- for wrist/finger flexors
    - knee immobilizers or knee extension splints
    - serial casts

  - for function in certain circumstances:
    - ambulation
    - for support of wrist extension for hand function

- positioning: - prone lying
  - wheelchair positioning
  - Nada chair for stretching hamstrings

- standing: - long leg braces (KAFO's)
  - tilt tables, standing tables, hydraulic standers, standing wheelchairs

** standing may be instrumental in minimizing osteoporosis as well as minimizing lower extremity contracture, as has been shown in other diagnoses[37-41] and as has been recommended for consideration for individuals with DMD[35, 42].

*****A stretching program should begin early in the course of the disease. A stretching program 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 painful.*****

LET CHILD BE IN CHARGE OF THE STRETCH WHENEVER POSSIBLE

STRENGTHENING - DYSTROPHINOPATHIES

The role of muscle activity and the effects of exercise on the progression of muscle degeneration in dystrophinopathies are not yet fully understood because the role and mechanism of dystrophin deficiency itself in the complex mechanisms that lead to muscle degeneration are not fully understood. Cycles of degeneration and regeneration occur in the presence of dystrophin deficiency, but it is unclear what initiates and influences these cycles, and how these cycles are related to factors such as altered strength and stability of the cell membrane, increased permeability of the cell membrane, altered mechanisms of Ca++ regulation, calpain activity, ischemia, mast cell infiltration, satellite cell function and proliferative potential, basic fibroblast growth factor (bFGF) activity, activity of platelet derived growth factor (PDGF) receptors, endomysial and perimysial fibrosis, and eventual replacement of muscle by fibro-fatty connective tissue. The effect of active muscle contraction on integrity of the plasma membrane and the contractile unit in the presence of the structural, physiological, and biochemical vulnerability intrinsic to dystrophin deficiency is not completely clear and may relate to the strength and duration of contraction and the load imposed. The mechanism behind progressive failure of regeneration in human DMD and canine CXMD as opposed to a potentially greater retention of regenerative capacity of the mdx mouse is not fully understood. The potentially greater retention of regenerative capacity of the mdx mouse may be less than believed in the past as clinical musculoskeletal impairments seen in humans and in canines have been described in mice if they are followed longer[43].

The fibrosis that occurs in DMD and CXMD but perhaps less in *mdx* muscle is not well understood, but may differ between species and could be significant in long term differences in regeneration. Fibrosis may present vascular and structural impediments to regeneration and may increase the protease activity of the calpain system by creating conditions of ischemia. The increased stiffness of fibrotic connective tissue may further tax remaining contractile units by increasing the load against which they must work. The proliferation of connective tissue occurs *prior* to extensive cell necrosis and is not simple scarring secondary to chronic tissue damage. Ongoing fibrosis and infiltration by connective tissue, primarily type III collagen, steadily accompanies degeneration. Abnormal distribution of, or response to, bFGF has been postulated to occur secondary to changes in membrane permeability and/or membrane bound signalling (Hoffman & Gorospe, Hardiman,& Partridge) with significant differences noted between DMD/CXMD and mdx muscle.

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 on muscle of active movement, and maintaining functional status and flexibility. Overwork weakness, however, must be avoided, as must exercise-induced damage. Concerns 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. Eccentric muscle activity and maximal resistive exercise are believed to be detrimental and should be avoided. Less is known about lower levels of activity. Some of the evidence regarding the damage of heavy exercise and eccentric contractions come from studies of normal muscle. Even in non-dystrophic skeletal muscle, very heavy exercise and/or compensatory hypertrophy results in fiber splitting, vacuoles, and necrosis with proliferation of connective tissue as well as fiber hypertrophy. Exercise in inadequately conditioned muscles can lead to injury of various degrees (Armstrong et al 1991). Early studies suggested that programs of long-term submaximal isokinetic exercise were beneficial if started early in the course of the disease when strength was at its maximum. Early

studies suggested that exercise did not appear to hasten weakness in young children or in young animal models although some evidence also seemed to suggest that the dominant upper extremity weakens first. Subsequent studies continued to suggest that gentle exercise does not hasten weakness in the human (Jackson et al 1987), or the *mdx* mouse, and that even more aggressive exercise does not cause exercise induced damage in the mouse model. However, studies of the canine model suggest vulnerability even to mild forms of exercise. Questions still remain about to what extent muscle activity in and of itself, may be damaging.

The identification of dystrophin's location as a link between the intracellular cytoskeleton and the external matrix suggests a mechanical, protective role in addition to the physiological and biochemical roles of the entire dystrophin-glycoprotein complex and associated parts of the muscle cell, all of which are assumed to be compromised in the dystrophin deficient muscle cell. The role of muscle activity in causing damage to the cell vs contributing a trophic influence is not yet known. Whether or not active movement has a role, positive or negative, in the fibrotic process is not clear.

Clear guidelines for the appropriate amount and type of exercise do not yet exist. Submaximal levels of isokinetic and aerobic exercise have been recommended in the past in order to prevent disuse atrophy and maintain residual strength while avoiding overwork weakness. These exercise programs were recommended to be started early in the course of the disease since exercise has been believed to be most effective in increasing / maintaining strength if it is initiated when residual strength is at its maximum.[44, 45]

*****long periods of rest and immobility should be avoided, even during periods of illness because functional losses can occur *****

possibilities for exercise:
- control of alignment for optimizing biomechanical advantage
- positioning and support for function
- movement facilitation
- PNF active assist
- swimming
- submaximal
- functional –
- trike riding – if not against too much resistance (i.e. not uphill)

fatigue: criteria for assessing fatigue is vague: if feel well rested after a good night's sleep, exercise level probably o.k.

particular areas in which strength is lost early:
- neck flexors
- abdominals
- shoulder girdle musculature : especially deltoids, latissimus, stabilizers
- pelvic girdle musculature: especially gluteals
- knee extensors
- ankle dorsiflexors

* specific areas indicated by individual evaluation

PROMOTION / PROLONGATION OF STANDING AND AMBULATION WITH KAFO'S (LONG LEG BRACES) (with or without lower extremity orthopedic surgery)

Controversy in the field has existed regarding whether or not to recommend orthopedic lower extremity surgery and lower extremity bracing as ambulation was about to be lost. In groups advocating the use of long leg braces, controversy was historically focused on identification of appropriate candidates and conditions, with a general consensus that this choice would not be made until ambulation was about to be lost. Subsequent controversy focused on timing of surgical intervention as well as candidacy, with some authors advocating early surgical intervention (Rideau, 1996) and other authors recommending more traditional methods of surgical intervention and bracing (Vignos, 1996).

pros:

- promote / prolong functional status in walking and ADL.
- more effective control of lower extremity contractures.
- delay/minimization of severe osteoporosis.
- easier weight control.
- improved cardiovascular/cardiopulmonary conditioning.
- easier transfers.
- delayed or diminished scoliosis.

cons:

- walking maintained is not necessarily functional
- effort required may impose excessive exercise, depending on amount of walking done and amount of effort required
- prolongs struggle and dependence vs freedom of wheelchair
- psychological damage of effort towards that which will eventually fail

Consensus has usually been that...in the right situation, *with an experienced team*, it can be beneficial for some children. Critical considerations include: dedication, motivation, and coping mechanisms of the child and family; skill, experience, and dedication of the team, especially if L.E. surgery is needed to maintain ambulation (pediatric orthopedic surgeon, anesthesiologist, P.T., orthotist); lack of excessive obesity; simultaneous emphasis on functional independence; appropriate timing; and *conscientious following of well developed and tested protocol for management*.**

Choices should not be made *between* ambulation and functional mobility - bracing for supported ambulation and the use of motorized mobility for functional independent mobility are often both appropriate.

Individuals already walking should be educated about making choices regarding bracing and surgery to maintain ambulation before a decision is at hand. Consideration of a major decision such as this should not have to occur at the last minute, in a crisis mode, when walking is about to be lost. The use of braces may evolve gradually as the need for stretching and support grows. Choices should be discussed openly in an ongoing manner as needs change.

RESPIRATORY PROGRAM

goals:

- to maintain chest wall mobility
- to maintain strength and endurance in respiratory muscles as much as possible (also by providing them with sufficient rest)
- to establish and maintain most efficient breathing pattern possible
- to establish good pulmonary hygiene
- to coordinate with pulmonary team
- to support appropriate use of noninvasive inspiratory and expiratory aids

suggestions:

- inspiratory exercises / segmental breathing
    - to strengthen diaphragm
    - for lung expansion and chest wall mobility
    - for more efficient breathing

- swimming
    - breath control
    - breathing patterns
    - endurance

- practice coughing, and use of mechanical assistance

- GPB - glossopharyngeal breathing

- 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:
    example: nocturnal or daytime IPPV (with volume ventilator, BiPAP)

- expiratory muscle aids:
    example: mechanical insufflation-exsufflation (MIE)
        ("Cough Assist" is common brand)

- coordination with team for anticipatory management regarding potential trach

SPINAL MANAGEMENT

intervention described in the literature:

- prolongation of ambulation
- external support:
    - bracing
    - specialized seating systems
    - wheelchair modifications
- promotion of upper extremity symmetry
- control of lower extremity position
- spinal surgery

Physical therapy management of the spine in the individual with Duchenne muscular dystrophy or spinal muscular atrophy 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 P.T. management of the spine:

** maintained ambulation and standing as long as possible
**promote spinal extension in sitting
** 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 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 hyperextension
    - 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 (adductors) to control abduction
  - should be “flip-down”, “swing-away”, or removable to allow use of urinal
- planar, rigid, lateral trunk supports - appropriately located and *strong* enough to:
  - prevent the need to lean laterally for stability
  - stop compensatory for UE function
- control of LE position - might include:
  - foot plate appropriately located and angled
  - ankle straps ?
  - padded footrests or foot cradles
  - polypropylene 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
- lumbar roll as appropriate to encourage spinal extension
- head support as needed – perhaps with lateral support in older individual

* power tilt-in-space, power recline mechanism with power elevating leg rests for older individuals - for changes in position, maintenance of skin integrity, opening up of hip and knee ankles to assist in minimizing the development of contractures

* power seat elevation for access to the environment

* power standing capability for independence in rising to stand

control of asymmetrical compensatory movement patterns

1. evaluate during **all** functional activities (wheelchair driving or propulsion, writing, eating)

2. **stop compensatory lean !**

3. provide for function with symmetry - might include:
   - relocation of wheel control box
     - 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 while sitting erect
- balanced forearm orthoses
- overhead sling
- other adaptive equipment

standing - to assist in control of LE contracture and to encourage spinal extension as well as offering more general physiological benefits
- hydraulic standers (examples: Easy Stand, Grand Stand)
- tilt tables
- standing wheelchairs (example: Permobile)

maintaining flexibility
- elongation in prone, supine, or sidelying to maintain symmetrical lateral elongation and flexibility
- maximally preventing contractures in LE's as described previously

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.

**** STOP**** every three months to reassess posture - asymmetry and malalignment can sneak up on you without anyone noticing. You have to make yourself stop and consciously reassess at regular intervals.

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:

Surgical intervention with spinal instrumentation now offers successful stabilization of the spine in individuals with DMD and is considered by many to be the conservative method of choice for spinal management. Segmental instrumentation allows stabilization with immediate post-operative mobilization (sitting usually re-initiated a few days after surgery) and without the need for post-operative external immobilization (i.e. no cast or body jacket after surgery). Most authors recommend careful monitoring of both the progression of scoliosis and respiratory status so that surgical stabilization can be offered for progressive curves while pulmonary status is still sufficient for tolerance of surgical intervention. Historical guidelines included consideration of surgical intervention as curves approach 40°, with some now recommending consideration of surgical intervention at 20°, while forced vital capacity (FVC) remains greater than 50%. If forced

vital capacity declines significantly below 50% while a spinal curve is still minimal, serious consideration should be given to whether or not surgical stabilization should be performed prophylactically before FVC declines to the point at which surgery is no longer a safe option. Partially successful conservative management beyond a point in time at which surgery remains an option should be guarded against, as later severe progression without surgical recourse should be avoided.

Compromised upper extremity function following surgical stabilization has been described in the individual who is dependent upon trunk leaning for hand function, but may be a small price compared to the benefits of stabilization. Functional losses, especially self feeding, are less demoralizing if anticipated pre-operatively, and if a plan is in place preoperatively to minimize or compensate for post-op losses. Surgical techniques (Duchenne instrumentation or DI) was described by Duport et al (1995) as providing improved stabilization with retained movement in some planes, thus preserving some useful truncal mobility.

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 keep up with his buddies ?”

***technology is the key to freedom in many situations

- motorized mobility
- power tilt, power recline, power elevating leg rests
- power seat elevation
- power standing
- powered lifts (including ceiling lifts, pivot lifts, stairclimbers, powered patient lifts)
- computers (including voice activated systems, adapted keyboards)
- internet
- environmental control units
- ramps and portable ramps
- modified sports equipment
- bathing and bathrooming equipment that fosters ease and independence
- power operated beds
- “PDA’s” (Palm Pilot)
- driving vehicles (vans)

GOALS AND OBJECTIVES OF PHYSICAL THERAPY INTERVENTION

Long term goals should include:

- to prevent deformity
- to maximize & maintain strength & endurance within limits of disease
- to maximize and maintain respiratory status at highest possible level
- to maintain ambulation as long as possible
- to maintain functional mobility throughout the course of the disease
- to maintain highest possible level of functional independence, using adaptive equipment and orthotic devices as needed
- to be an active team member in clinical decision making (including possible surgical intervention and adaptive equipment)
- to provide patient, family, and caregivers with comprehensive and timely information that allows understanding and active involvement in overall management of disability

Short term objectives should include:

- to increase/maintain range of motion as appropriate
- to increase/maintain strength and endurance as much as possible
- to promote optimal body alignment and symmetry
- to minimize the clinical impact of compensatory movement patterns and positions used for function
- to assist the team in preventing scoliosis
- to maintain functional ambulation as long as possible
- to maintain sitting ability
- to provide appropriate pre and post surgical intervention in coordination with the rest of the team
- to maintain functional, independent mobility throughout all phases of the disease progression
- to provide an active, ongoing respiratory program
- to maintain chest wall mobility
- to strengthen respiratory muscles and develop endurance
- to establish and maintain the most efficient breathing pattern
- to teach principles of pulmonary hygiene and assisted coughing
- to help preserve maximal hand function
- to initiate and monitor appropriate functional compensation for weakness/deformity as appropriate in all areas of self care, scholastic/vocational activities, and daily activities, with adaptive equipment used as needed, especially including motorized mobility, computer access, augmentative communication systems, and environmental control units.
- to consult with teachers and other community individuals regarding the individual's status and care as needed
- to assist the family in problem solving before and as problems arise
- to establish and monitor home programs
- to teach caregivers proper handling and transfer techniques
- to promote comfort
- to facilitate ease of nursing care when needed

helpful websites:

adaptive equipment / assistive technology / orthotic intervention:

orthotic intervention:
http://www.dafo.com/
http://www.eortho.com/anklesurestep.html
http://www.buyaircast.com/index.htm?srcad=air+casts

lifts, bathing, and bathrooming equipment:
http://www.arjo.com/nurse_default.asp?id_page=805&id_child=0&id_level=1&id_nav=819&id_market=13
http://www.image-management.com/
http://www.surehands.com/

respiratory care:
http://www.21stmedical.com/vol50volex.html
http://www.jhemerson.com/coughassist.htm
http://www.thevest.com/

oximeters:
http://www.portablenebs.com/nonin2120.htm

standers:
http://www.easystand.com/
http://www.mulhollandinc.com/Rocket.asp
http://www.permobilusa.com/templates/startpage.aspx?id=806

wheelchairs / mobility devices:
http://www.permobilusa.com/templates/startpage.aspx?id=806
http://www.pridemobility.com/
http://www.independencenow.com/ibot/index.html

cycling:
http://www.exnflex.com/

other:
http://www.portable-wheelchair-ramps.com/

DUCHENNE MUSCULAR DYSTROPHY

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Laura E. Case MS, PT, PCS

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