

Rehabilitation Standards of Care for Duchenne Muscular Dystrophy



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Objectives

- Review 2018 Duchenne muscular dystrophy (DMD) care considerations for rehabilitation
- Review overlapping components of care related to orthopedic management
- Review local resources that support rehabilitation care considerations and support individualized goals

THE LANCET Neurology



Articles

Pimavanserin for patients with
Alzheimer's disease psychosis
See page 213

Review

Diagnosis and management of
Duchenne muscular dystrophy
See page 251

Review

Clinical applications and
contraindications for lumbar puncture
See page 268



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Pediatric Rehabilitation Medicine

- Pediatric Rehabilitation Medicine (PRM)
 - utilizes an interdisciplinary approach
 - address the prevention, diagnosis, treatment, and management of congenital and childhood-onset physical impairments including related or secondary medical, physical, functional, psychosocial, cognitive, and vocational limitations or conditions
 - an understanding of the life course of disability



Duchenne Muscular Dystrophy

DMD results in:

- progressive muscle degeneration and weakness
- postural compensations
- risk of progressive contracture and deformity
- functional losses



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Duchenne Muscular Dystrophy

Improved DMD management has resulted in:

- prolongation of ambulation
- decreased prevalence of severe contracture and deformity, including scoliosis
- prolonged function and participation in all areas of life



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Duchenne Muscular Dystrophy

	Stage 1: At diagnosis	Stage 2: Early ambulatory	Stage 3: Late ambulatory	Stage 4: Early non-ambulatory	Stage 5: Late non-ambulatory
Rehabilitation management	Provide comprehensive multidisciplinary assessments, including standardised assessments, at least every 6 months				
	Provide direct treatment by physical and occupational therapists, and speech-language pathologists, based on assessments and individualised to the patient				
	Assist in prevention of contracture or deformity, overexertion, and falls; promote energy conservation and appropriate exercise or activity; provide orthoses, equipment, and learning support		Continue all previous measures; provide mobility devices, seating, supported standing devices, and assistive technology; assist in pain and fracture prevention or management; advocate for funding, access, participation, and self-actualisation into adulthood		

Birnkrant, David J (03/2018). "Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management.". *Lancet neurology* (1474-4422), 17(3), p. 251.



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Duchenne Muscular Dystrophy

Rehabilitation team members include:

- Physicians
- Physical therapists
- Occupational therapists
- Speech-language pathologists
- Orthotists
- Durable medical equipment providers
- Others



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Duchenne Muscular Dystrophy

The rehabilitation team focuses on:

- Individual's goals and lifestyle to optimize quality of life across the lifespan
- Assessment and anticipatory management

Stage 1:
At diagnosis

Stage 2:
Early ambulatory

Stage 3:
Late ambulatory

Stage 4:
Early non-ambulatory

Stage 5:
Late non-ambulatory

With the goal of minimizing :

- contractures
- deformity
- loss of function
- compromised skin integrity
- pain
- compromised cardiorespiratory status



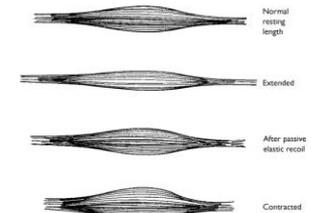
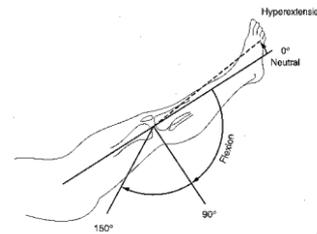
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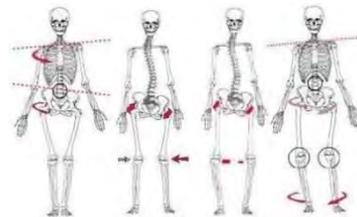
Assessment

Multidisciplinary rehabilitation assessment includes:

- measures of passive ranges of motion
- muscle extensibility
- posture and alignment
- strength
- function
- quality of life
- participation in all normal activities of everyday life



Source: Hall SJ: Basic Biomechanics, 9th Edition.
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Assessment

Specialized functional assessment may include:

- Analysis of patterns of movement
ex. 6-minute walk test (6MWT)
- Standardized assessments specific to DMD
ex. North Star Ambulatory Assessment



Goal:

- Predictive capabilities regarding functional motor changes that are important in monitoring clinical progression
- Assessing new and emerging therapies



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Assessment

Limitations and considerations:

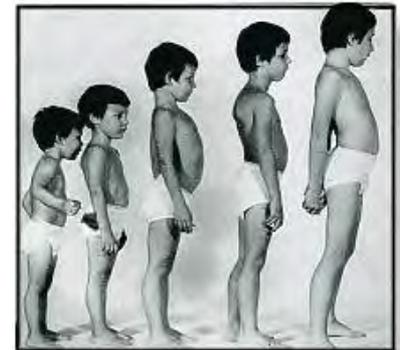
- Prediction of functional change in clinical settings should be made in the context of a patient's capabilities
- Awareness of limitations in effort-based assessments
- Potential interactions with musculoskeletal impairments such as contracture, and genetics
- Tests that predict potential upcoming changes can be used to guide proactive care, such as impairment-level interventions and future equipment needs



Assessment

Example:

- Before age 7 years, gains might occur in the 6-min walk test and timed function tests.
- After age 7 years,
 - a 6-min walk test result of less than 325 m,
 - time to stand more than 30 sec,
 - time to climb four stairs more than 8 s,
 - 10-min walk or run time more than 10–12 s,
 - Mean linearized NSAA 34 or less
- Have all been associated with greater functional decline in ambulation over the subsequent 12 months.



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Assessment

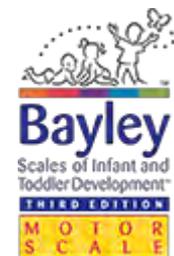
-Functional assessment includes the assessment of activities of daily living and the need for adaptive equipment or assistive technology.



-Various tools can be used to assess quality of life.



-Increasing use of standardized testing in infants and young children with DMD is timely
-potential for early diagnosis with newborn screening
-emergence of therapies that might work best if used in early childhood.



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Assessment

- Consistent use of the same functional measures by individual clinics is recommended for tracking change over time
- Assessment by rehabilitation specialists is recommended at least every 4–6 months throughout life
- More frequent assessment triggered by a clinical concern, a change in status, or specific needs.



Intervention

-Physical, occupational, and speech and language therapy

- Outpatient
- School
- Hospital admissions
- Home



-Continue throughout adulthood



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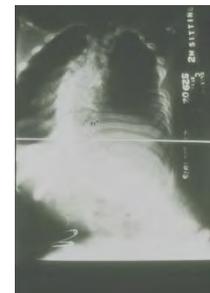
Targets of Intervention

-The goal of muscle extensibility and joint mobility management is to prevent or minimize contracture and deformity caused by:

- inability to move a joint through its full range of motion
- chronic static positioning
- muscle imbalance about a joint
- fibrotic changes in muscles



-Restricted patterns of breathing and fibrosis of intercostal muscles decrease chest wall mobility.



Intervention

-Maintenance of passive ranges of movement, muscle extensibility, chest wall mobility, and symmetry can:

- optimize movement and functional positioning
- maintain ambulation
- prevent fixed contractures and deformities
- optimize respiratory function
- maintain skin integrity

-Musculoskeletal management requires a team approach with input from:

- neuromuscular specialists
- physical therapists
- occupational therapists
- rehabilitation physicians
- orthotists
- orthopedic surgeons



Intervention

Prevention of contracture and deformity requires:

- daily passive stretching of joints, muscles, and soft tissues at risk of tightness.
- support of movement by decreasing the effects of gravity and optimizing biomechanics to allow more active movement.
- manual therapy techniques and prolonged elongation of soft tissues.
- optimal positioning, including individualized use of splinting, orthotic interventions, standing devices, serial casting, and custom seating and power positioning components in mobility devices.



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Intervention

- A daily preventive home stretching program should begin before the loss of passive ranges of motion under the guidance of physical and occupational therapists.
- Stretching is recommended for areas known to be at risk of contracture or deformity.
- Regular stretching of ankle, knee, and hip should begin soon after diagnosis and continue into adulthood.
- Stretching of the upper extremities is especially important after loss of ambulation.



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Intervention

- Orthotic intervention, splinting, casting, positioning, and equipment:

- AFOs for stretching at night (might be best tolerated if started preventatively at a young age)

- AFOs for stretching or positioning during the day in non-ambulatory phases

- Wrist or hand splints for stretching of long and wrist finger flexors/extensors, typically in non-ambulatory phases

- Serial casting-in ambulatory or non-ambulatory phases



Intervention

-Orthotic intervention, splinting, casting, positioning, and equipment:

- Passive/motorized supported standing devices—when standing in good alignment becomes difficult, if contractures are not too severe to prevent positioning or toleran
- KAFOs with locked knee joints—an option for late ambulatory and non-ambulatory stages
- Custom seating in manual and motorized wheelchairs (solid seat, solid back, hip guides, lateral trunk supports, adductors, and head rest)
- Power positioning components on motorized wheelchairs (tilt, recline, elevating leg rests, standing support, and adjustable seat height)



Intervention

- Power stand-and-drive motorized wheelchairs are now frequently used in place of knee-ankle-foot orthoses to support standing mobility.
- KAFOs might still be an appropriate choice in some situations, but should be viewed as therapeutic rather than functional tools, supplementing rather than replacing motorized mobility.
- Technological innovations may be simple or advance
 - Simple devices
 - elevated lap trays and
 - adaptive straws) to more
 - Advanced technologies
 - robotics
 - bluetooth capabilities that permit remote activation of devices
 - infrared environmental controls
 - smart phones
 - tablets and computers
 - advanced access capabilities such as voice activation in the home



Intervention

- Possible adaptive equipment and home renovations include:
 - lifts for safe transfers
 - ramps
 - stair lifts
 - bathing and bathroom equipment or renovations
 - special beds and mattresses
 - vehicle modifications



- Personal care attendants can help to optimize independence and participation.



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Intervention

- Physical therapists prescribe, monitor, and guide exercise, which can **prevent an unnecessarily sedentary or immobile lifestyle** and the associated problems of social isolation and overweight.
- The effects of exercise on muscle degeneration in dystrophinopathies, though not fully understood, can include damage due to structural fragility of muscles, metabolic abnormalities, nitric oxide abnormalities contributing to ischemia during exercise, and reduced exercise capacity.
- Eccentric muscle activity or exercise and high-resistance exercise or strength training should be avoided.
- Submaximal aerobic exercise or activity has been recommended, especially early in the course of the disease - **avoiding overexertion and overwork, and allowing adequate rest.**



Intervention

- Swimming is highly recommended from the early ambulatory stage and can be frequently continued adulthood.



- Cycling has been recommended as a submaximal aerobic form of activity and assisted cycling and robotic-assisted movement can be used into adulthood.



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Intervention

- Safe physical activity can be supported by appropriate adaptive equipment and assistive technology.
- Pain must be assessed and addressed in individuals at all ages.
- Interventions require comprehensive team management, including physical therapy, postural correction, orthotic intervention and splinting, wheelchair and bed enhancements that allow independent weight shift, position change and pressure relief, and pharmacological approaches.
- Back pain, particularly in the context of glucocorticoid treatment, should prompt assessment for vertebral fractures.

Stage 1:
At diagnosis

Stage 2:
Early ambulatory

Stage 3:
Late ambulatory

Stage 4:
Early non-ambulatory

Stage 5:
Late non-ambulatory



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Orthopedic Intervention



- Children in the ambulatory stage might benefit most from surgery, but it is recommended less frequently than in the past.
- Though the 2010 care considerations included recommendations for multilevel surgeries, the consensus of the current panel is that surgery on the foot to improve the varus positioning and on the Achilles tendon to improve dorsiflexion range might be sufficient to improve gait in patients with clinically significant ankle contracture and good quadriceps and hip extensor strength.
- Interventions related to the hips and knees are not recommended.



Orthopedic Surveillance

- Assessment for scoliosis should be done at least annually, although onset is unusual in the ambulatory stage.
- Visual assessment is appropriate, with radiographic assessment only if a curve is observed on examination or if visual inspection alone is inadequate, such as in children with obesity.
- Use of spinal orthoses is not generally recommended in the setting of a compression fracture.



Orthopedics - Fractures

- Anticipatory guidance during routine clinic visits is an important part of a fracture prevention program throughout all disease stages.
- Corticosteroids have been associated with osteoporosis and subsequent vertebral fractures in DMD.
- In a study of 143 boys with DMD, the long-bone fracture rate in those treated with corticosteroids was 2-6 times greater than in those who had never received steroids.
- A lower-limb fracture during the ambulatory stage might need aggressive management to maintain ambulation.
- Internal or external fixation allows for early mobilisation compared with casting or splinting.



Fracture Prevention

Assessment and education

Assessment and education by occupational or physical therapist

- Minimise fall risks in all environments, including consideration of walking surface, terrain, and obstacles
- Provide training for patients and families on wheelchair safety; raise awareness that falls out of wheelchair are a common cause of injury
- Provide training for families in safe lifting and transfers to and from wheelchair and various surfaces in all environments

Common considerations or possible modifications

Safety of home environment

- Remove obstacles such as rugs, toys, cords, and clutter

Avoidance of falls from wheelchair or mobility device

- Use seatbelt at all times
- Use anti-tippers on wheelchairs

Safety on uneven or slippery surfaces

- Take special care when outdoors because of uneven surfaces
- Wear pool shoes for protection against falls when walking on slippery surfaces around water
- Use non-slip treads on ankle-foot orthoses at night to decrease fall risk when walking to and from bathroom

Safe transfer in and out of wheelchair

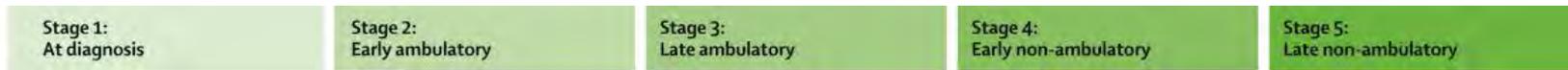
- Consider adaptive equipment and patient lift systems early for use in all settings to provide safe support and minimise risk of falls or injury during transfers, toileting, and bathing or showering

Potential home modifications

- Non-slip mats in shower or bathtub
- Grab bars for shower or bathtub
- Bath seat or other adaptive equipment for bathing
- Non-slip treads for bare-wood steps
- Handrails on both sides of stairways



Orthopedic



Early non-ambulatory stage

Foot and ankle surgery to improve equinovarus foot might help with foot positioning in the wheelchair or for shoe wear, but is typically done only if a patient requests the procedure.

After foot and ankle surgery, use of ankle-foot **orthoses will be needed** during the daytime to prevent a recurrence of the contractures.



Orthopedic

- Inspection of the spine should be part of every clinical examination.
- Experienced clinicians should be able to monitor the spine in non-ambulatory boys by visual inspection alone; however, less experienced clinicians should obtain a spine radiograph when a child first becomes non-ambulatory.
- A spine radiograph is also useful when inspection is unhelpful, such as in children with obesity.



Orthopedic

- Once a curve has been detected with radiography, further surveillance depends on the skeletal maturity:
 - skeletally immature individuals should undergo radiographs once every 6 months, and
 - skeletally mature individuals should undergo radiographs at least once a year.
- A curve of 20° or more should warrant involvement of an orthopedic surgeon.
- The use of spinal orthoses is not recommended.
- Those treated with corticosteroids have milder spinal curvatures and less frequent need for spinal surgeries.



Orthopedic

- Posterior spinal fusion in young men with DMD is recommended due to the positive effect on:
 - function,
 - sitting balance and tolerance,
 - pain, and
 - quality of life

Observed in non-randomized, prospective cohort studies.



Orthopedic

- Posterior spinal instrumentation and fusion are recommended in:
 - non-ambulatory individuals
 - with a spinal curve in the sitting position greater than 20-30°,
 - who have not yet reached puberty, and
 - who have not been treated with corticosteroids (because the curve is expected to progress).

- Although patients treated with corticosteroids can still develop scoliosis, the progression might be less predictable, so observation for clear evidence of progression is a reasonable approach before intervening.



Orthopedic

- Anticipatory fracture prevention guidance should continue through the non-ambulatory stages.
- A more conservative approach to management of lower-limb fractures is advised in non-ambulatory children because the goal is no longer to bear weight.
- Internal fixation might be necessary for an unstable fracture, but splinting might be sufficient for bone healing and pain control.



Orthopedic

- Surgical interventions to manage contractures involving the upper or lower extremities are not recommended during the late non-ambulatory stage of DMD unless there problems with:
 - pain
 - Positioning
 - skin integrity



Orthopedic

- Individuals with known scoliosis should have yearly anteroposterior upright spinal radiographs when there is any concern about progression.
- Posterior spinal fusion is recommended during the late non-ambulatory stage for those with a progressive curve.
- It is essential to consult with the patient's respiratory physician and cardiologist to ensure that lung and heart function are sufficient to proceed with this surgical intervention.
- Some studies indicate that spinal fusion slows the progression of respiratory decline, whereas others show no significant difference in the rate of decline postoperatively.



Partners

Teamwork is the ability to work as a group toward a common vision, even if that vision becomes extremely blurry.

-Unknown Author



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Partners



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Thank you!



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