FACT SHEET
Duchenne Muscular Dystrophy

PURPOSE
This fact sheet is intended to provide an overview of Duchenne muscular dystrophy (DMD) including clinical presentation, diagnosis, prognosis, pharmacological management, and physical therapy management. The information provided will help the physical therapist and physical therapist assistant better understand the complex nature of this condition and plan appropriate examination, intervention, and care management, including durable medical equipment.

QUICK FACTS
- DMD is progressive neuromuscular disorder primarily affecting males.[1]
- DMD occurs in an estimated 15.9 per 100,000 U.S. male births.[2]
- Early signs include toe walking and difficulty rising from the floor (Gowers' sign), climbing stairs, and keeping up with peers.[1]
- Muscular weakness begins proximally in the hips and shoulder girdle and progresses distally, resulting in loss of function and ambulation.
- Although DMD currently has no cure, corticosteroids slow disease progression and prolong ambulation.[1,3,4]
- Updated Diagnosis and Management Care Guidelines were published in 2018.[1,5–7]

WHAT IS DUCHENNE MUSCULAR DYSTROPHY (DMD)?
DMD is an X-linked genetic neuromuscular disease caused by a mutation of the dystrophin protein gene.[1,8] Dystrophin is a cytoskeletal protein critical to muscle function and health.[8] Absence of dystrophin results in the breakdown of muscle tissue and replacement with fatty fibrous tissue,[8] leading to muscle atrophy and, particularly in the calf, pseudohypertrophy. The primary impairment in DMD is progressive muscle weakness with fatty infiltration, which leads to secondary complications of contractures, scoliosis, and progressive disability.[8] Standard of care medical intervention, including long term corticosteroids,[1,4] has extended the median age of ambulation loss from 10 years to over 13 years,[9] and the life expectancy for males with DMD into the fourth decade of life.[3]

DIAGNOSIS OF DMD
Blood tests for elevated serum creatine kinase can be used for screening when clinical presentation suggests DMD.[1,8] DMD diagnosis is confirmed by genetic testing to identify the specific dystrophin gene mutation, which informs eligibility for mutation specific treatments.[8] When genetic testing is non-confirmatory, muscle biopsy may be performed to test for the presence of dystrophin.[1] Diagnosis delays are common, especially in those without a family history.[10] The reported mean age of definitive diagnosis is 4.9 years, despite a mean symptom onset of 2.5 years.[10]

CLINICAL PRESENTATION OF DMD

Early signs of DMD include enlarged calf muscles (pseudohypertrophy), proximal muscle weakness, Gowers' sign when rising from the floor, difficulty walking or running, and an inability to keep up with peers.[1,10] Delays in gross motor, speech, and cognitive function are common.[11] Autism spectrum disorder (ASD) and attention-deficit/hyperactivity disorder (ADHD) are more prevalent in boys with DMD than in the general population.[11] Progression of muscle weakness and plantar flexor muscle contractures lead to a wide base of support and toe walking gait pattern, with increased lumbar lordosis, decreased walking endurance, and frequent falls. Median age at loss of ambulation is approximately 13 years,[9] after which further progression of lower extremity weakness and contractures, loss of upper extremity strength, and compromised respiratory function ensue.[7] In the late teens and early 20s, many individuals with DMD experience loss of independent sitting, difficulty with head control, loss of the ability to reach and bring hands to mouth, progression of scoliosis, and respiratory compromise. Onset of cardiac dysfunction is common between 10 and 20 years of age; progressive cardiovascular complications are a leading cause of mortality.[5,12]

More information about the progression of DMD and care considerations at each disease stage are available through the Parent Project Muscular Dystrophy (PPMD) website.[13]

PHYSICAL THERAPY MANAGEMENT OF DMD

Physical therapists play a critical role in supporting health related function in individuals with DMD through stretching, exercise/activity, and equipment recommendations.[1,7] Consistent administration of functional outcome measures at regular 6-month intervals is recommended to monitor disease progression and assist in rehabilitation management (see TABLES 1 and 2).[7]

TABLE 1: Priority Physical Therapy Tests and Measures[1,7]

Priority Tests & Measures	Disease Phase	Rationale
Body Functions and Structures		
Range of Motion (lower extremities)	Ambulatory Non-ambulatory	*Prioritize ankle plantar flexors, iliotibial bands, hip/knee flexors to guide stretching and positioning.*
Range of Motion (upper extremities)	Ambulatory Non-ambulatory	*Prioritize forearm pronators, wrist and elbow flexors/extensors, and shoulder elevation to guide stretching and positioning.*
Posture	Ambulatory Non-ambulatory	*Evaluate sitting and standing posture to guide stretching, bracing, equipment, and positioning recommendations.*
Pain	Ambulatory Non-ambulatory	*Monitor for signs of overexertion and guide supported standing, seating, and wheelchair positioning recommendations.*
Rating of Perceived Exertion	Ambulatory Non-ambulatory	*Use to guide activity pacing and exercise prescription intensity recommendations.*
Activities, Participation, and Quality of Life		
North Star Ambulatory Assessment (NSAA)	Ambulatory boys >3 years of age	*Measure functional motor abilities in ambulatory children with DMD. Use to monitor disease progression and treatment effects, and to anticipate mobility equipment needs.*
Performance of Upper Limb Module (PUL)	Ambulatory Non-ambulatory	*Measure upper limb function. Use to monitor disease progression and treatment effects.*
Brooke Upper Extremity Scale	Ambulatory Non-ambulatory	*Classifies upper extremity function in ambulatory and non-ambulatory children with DMD.*

Egen Klassifikation Scale Version 2 (EK2)	Non-ambulatory	*Measures functional abilities and activities of daily living in non-ambulatory children with DMD.*
Six- or Two- Minute Walk Test	Ambulatory	*Measures aerobic capacity and walking endurance. Use to monitor disease progression, treatment effects, and anticipate mobility equipment needs.*
Pediatric Quality of Life Inventory (PedsQL)	Ambulatory Non-ambulatory	*Measures health-related quality of life.*
Pediatric Evaluation of Disability Inventory (PEDI)	Ambulatory Non-ambulatory	*Assesses key functional capabilities and performance.*

TABLE 2: DMD Presentation and Physical Therapy (PT) Management through the Lifespan[7,14]

At time of Diagnosis *(Infancy/Childhood)*		
Presentation	Children in this stage may present with mild symptoms and have gross motor delays when compared to norm-referenced development. Calf pseudohypertrophy and Gowers' maneuver may be observed. Cognitive and speech delays may be present.	
PT Intervention	Stretching:	Plantar flexors, hamstrings, hip flexors, and iliotibial bands to slow contracture progression.
	Exercise:	Facilitate acquisition of expected gross motor milestones, low impact age-appropriate play, cycling (low or no resistance), and swimming.
	Equipment:	Resting AFOs, stroller for community distances, as needed.
		Avoid eccentric exercise (i.e., bounce houses, trampolines). Avoid activities with collision or fall risk.
	Education:	Provide family with DMD resources and education. Coordinate care among early intervention/ school/ outpatient/ multidisciplinary care teams.
Early Ambulatory *(Childhood)*		
Presentation	Children in this stage may present with frequent tripping/falling, use of Gowers' maneuver to transition from floor to stand, difficulty with jumping/hopping, delayed speech, and difficulty lifting the head in supine. Pseudohypertrophy of the calves may be observed. Gait deviations include increased lumbar lordosis, wide base of support, toe walking, and "Ducheene" Jog" when running. Children in this stage may have difficulty keeping up with and require rest breaks.	
PT Intervention	Stretching:	Plantar flexors, hamstrings, hip flexors, and iliotibial bands to slow contracture progression.
	Exercise:	Low impact, sub-maximal age-appropriate aerobic activity, and play, cycling (low or no resistance), respiratory muscle training, and swimming to prevent disuse atrophy, functional training for ADLs.
	Equipment:	Resting AFOs, manual wheelchair/stroller/motorized scooter for community distances, depending on child's size, maturity, safety awareness, endurance, and mobility needs.
	Education:	Avoid eccentric exercise (i.e., repeated stair descent, squatting, or jumping) Avoid activities with collision or fall risk Discuss energy conservation strategies Encourage self-recognition of when rest breaks are needed Coordinate care among school/outpatient/multidisciplinary care teams

Late Ambulatory *(Late Childhood/Adolescent/Young Adult)*	
Presentation	Children and adolescents in this stage present with increased lumbar lordosis and a wide base of support in standing, progressive lower extremity contractures, toe walking, limited walking endurance, fatigue with mobility, and more frequent falls as ambulation becomes difficult. Loss of stair climbing ability and marked increases in timed tests including the Six- and Two- Minute Walk Tests, Timed Floor to Stand, Time to Climb 4 Stairs, and Timed 10 Meter Walk are observed.
PT Intervention	Stretching: Iliotibial bands, tensor fascia latae, hip flexors, hamstrings, gastroc/soleus, posterior tibialis, plantar fascia to prevent progressive contracture. Exercise: Standing program to be initiated prior to loss of ambulation, low impact, sub-maximal aerobic activity, cycling (low or no resistance), respiratory muscle training and aquatic activity to prevent disuse atrophy, functional training (including compensatory strategies) for ADLs, transfers, and bed mobility. Equipment: Resting AFOs, stander, power wheelchair or motorized scooter, back-up manual wheelchair, cough assist, shower/commode chair. Education: Avoid eccentric exercise (i.e., repeated stair descent or squatting). Outline fracture risk and fall prevention. Discuss strategies for transfers and access at school, home, work, community. Coordinate care among school/outpatient/multidisciplinary care teams.
Early Non-Ambulatory *(Adolescent/young adult)*	
Presentation	Adolescents and young adults in this stage use a wheelchair or scooter for household and community mobility, and present with increasing leg and arm weakness, and contractures of the upper and lower extremities. Progressive loss of overhead and reaching activities is often observed.
PT Intervention	Stretching: Lower extremity muscle groups above, plus elbow flexors and pronators, wrist and finger flexors and/or extensors, lumbricals, and intrinsic hand muscles. Exercise: Daily standing program, aquatic activities, cycling (low or no resistance), respiratory muscle training, functional training (including compensatory strategies) for ADLs, transfers, and bed mobility. Equipment: Resting AFOs, stander, power wheelchair, back-up manual wheelchair or motorized scooter, cough assist, mechanical lift, shower/commode chair. Education: Discuss adaptations and equipment needs to preserve and optimize independent ADLs. Discuss strategies for transfers and access at school/work, home, and community. Coordinate care among school/outpatient/multidisciplinary care teams.
Late Non-Ambulatory *(Adult)*	
Presentation	Adults in this stage present with diffuse weakness, progressive contractures, scoliosis and cervical mobility limitations. Individuals may be dependent for bed mobility, at increased risk for pressure sores, and in need of trunk and head support to maintain sitting. Progressive contractures and weakness may result in decreased tolerance for supported standing programs. Impairments in respiratory function, speaking, and swallowing may be present.
PT Intervention	Stretching: Lower extremity muscle groups above, plus head/neck range of motion, elbow flexors and pronators, wrist/finger flexors and/or extensors, lumbricals, and intrinsic hand muscles. Exercise: Respiratory muscle training.

	Equipment:	Power wheelchair, adaptive devices for upper limb function, upper extremity resting orthoses, cough assist, mechanical lift, shower/commode chair.
	Education:	Teach compensatory strategies for ADLs, transfers, and bed mobility. Encourage frequent position changes in wheelchair for skin protection, respiratory function, back/hip pain management, edema control. Coordinate care among outpatient/multidisciplinary care teams.

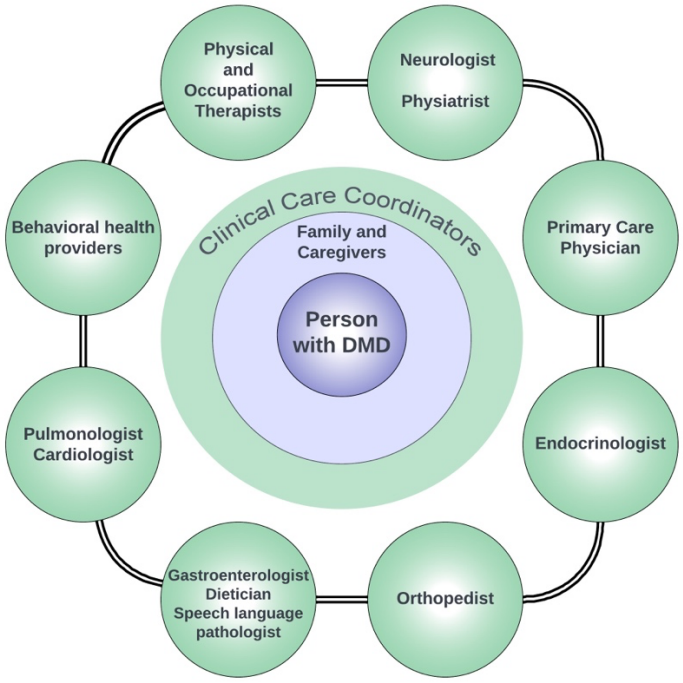
PHARMACOLOGICAL MANAGEMENT OF DMD

- Glucocorticoid treatment (e.g., Prednisone or Deflazacort) is a standard of care in DMD. Benefits of glucocorticoid therapy include ≥2-year delay in loss of mobility and functional milestones, including walking and upper limb function.[4]
- Glucocorticoid prescription typically begins in the Early Ambulatory Phase to preserve muscle strength, ambulation, respiratory function, and upper limb function.[1,4] The optimal age for treatment initiation and dosing regimens for steroids are still being evaluated.[1,4,15]
- Common side effects of glucocorticoid therapy include weight gain, osteopenia, risk of fractures, behavioral changes (e.g., increased aggression, attention difficulties), insulin resistance or diabetes, cushingoid features, and cataracts.[1,3] Balancing the benefits and side effects of glucocorticoids is essential; treatment should be closely monitored by a qualified medical professional.
- Several treatments targeting specific gene mutations have recently been approved and more promising treatments are under study.[8]

THE MULTIDISCIPLINARY CARE TEAM

Management of individuals with DMD involves patient- and family-centered care coordinated across a host of medical specialists (see FIGURE 1). Care considerations for each specialty are outlined in the 2018 DMD Care Guidelines.

FIGURE 1: The Multidisciplinary Care Team



DURABLE MEDICAL EQUIPMENT

The following equipment may be considered to support and maintain functional abilities of those with DMD. For more information on medical equipment visit the Cure Duchenne Durable Medical Equipment Guide

- Orthoses for contracture management[1,7]:
  - Resting custom Ankle Foot Orthoses (AFOs) to be worn 6-8 hours daily (often nighttime) *AFOs that restrict dorsiflexion or plantar flexion should not be worn when ambulating.*
  - Wrist hand orthoses to be worn nightly to slow progression of finger flexion contractures.
- Stander[7]: initiate standing program in the late ambulatory and early non-ambulatory phases, preferably prior to the loss of ambulation or development of contractures which may limit tolerance and functional benefit.[1,16]
- Powered wheeled mobility[7]: Proactive evaluation and planning in late ambulatory phase to allow time required for insurance authorization, home modifications, vehicle adaptation, and device assembly/delivery. Ensure wheelchair provides adequate postural support and alignment. Important functions include power tilt/recline, power seat elevation, power leg rests; consider power stand feature.
- Transfer devices[7]: slide board/sheets, mechanical lift.
- Bed[7]: high density foam overlay, hospital/motorized bed.
- Bathroom modifications[7]: grab bars, shower/commode chair.
- Home modifications: stair lift, ceiling lift, ramp, vehicle lift.

SUMMARY

- Physical therapists play a critical role in supporting health-related function in those affected by DMD through designing and implementing stretching programs to minimize contractures, promoting sub-maximal, non-fatiguing exercise/activity to maintain cardiovascular health and protect fragile muscles, guiding equipment recommendations and management to optimize function, and providing patient and family education tailored to each disease stage.[1,7]
- Children with DMD should avoid muscle overexertion and eccentric exercise.[7]
- Children with DMD, especially those treated with long-term corticosteroids, are at increased risk of fractures which can accelerate functional loss and result in health emergencies, including fatty embolisms.[5] Education to reduce fall risk and avoid collision activities are important priorities.[7]
- Updated Diagnosis and Management Care Guidelines published in 2018 inform best practice PT care.[1,5–7]

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ADDITIONAL RESOURCES

- Parent Project Muscular Dystrophy: https://www.parentprojectmd.org/
- Muscular Dystrophy Association: https://www.mda.org/
- Duchenne.com: https://www.duchenne.com/
- Cure Duchenne: https://www.cureduchenne.org/

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