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Mini Review

A Mini-Review on The Rehabilitation of Duchenne Muscular Dystrophy

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Abstract

Duchenne Muscular Dystrophy (DMD) is a rare, severe, progressive genetic disorder causing disability and premature death. Mutations in the DMD gene encoding the dystrophin protein leads to the dystrophinopathies DMD. It affects approximately 1 in 3500 to 5000 male births worldwide. Rehabilitation plays a critical role to overall management for individuals with DMD. A series of effective rehabilitation management strategies have been developed in multidisciplinary groups lead to help maintain a better patient's Quality of Life (QOL), as well as maximize the patient's physical and psychosocial functions. In this mini-review, we discuss revised guidelines for DMD care (known as DMD Care Considerations).

Keywords: Duchenne muscular dystrophy; Rehabilitation; Quality of life

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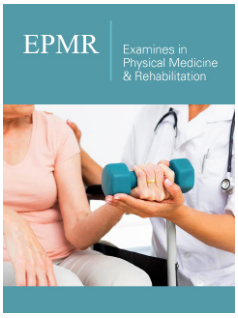

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Introduction

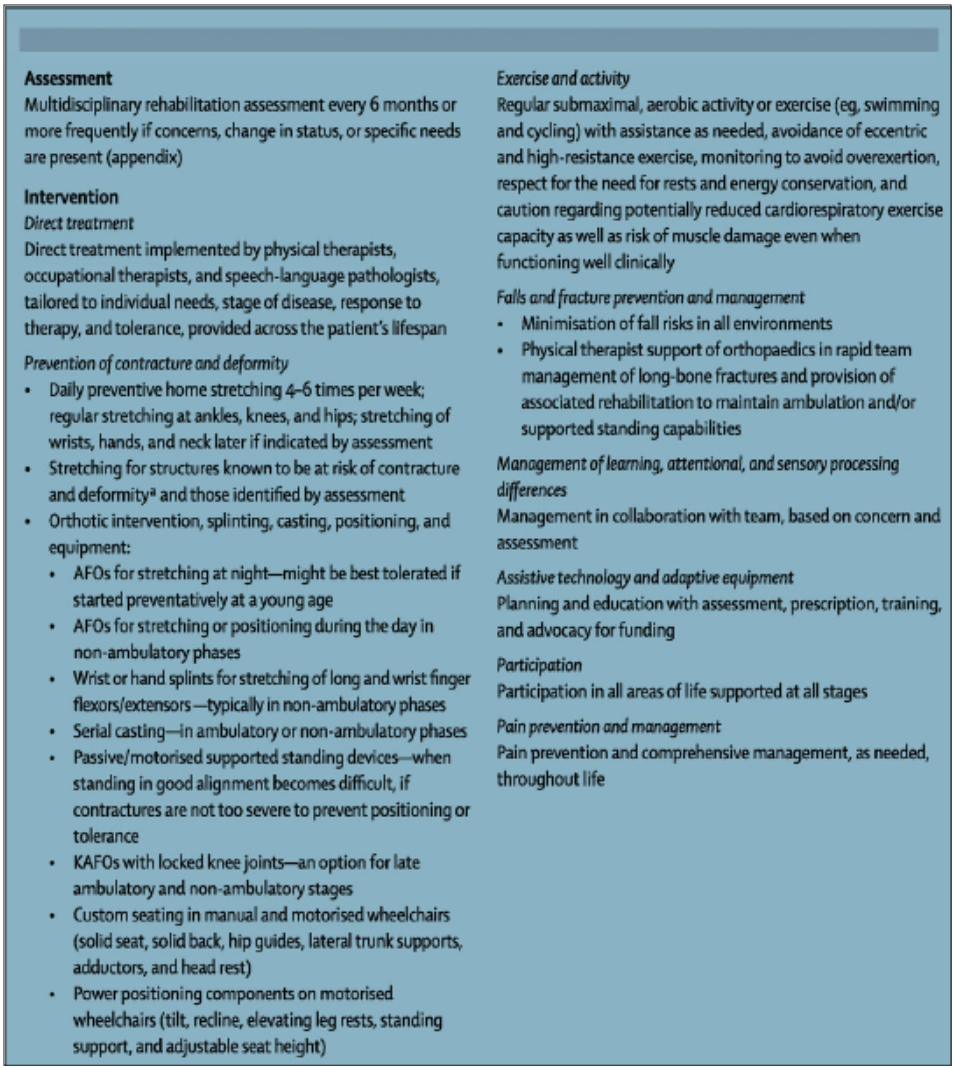
Duchenne muscular dystrophy, a rare X-linked disorder, is caused by a genetic mutation that prevents the body from producing dystrophin [1], a protein that enables muscles to work properly. It is one of the most common types of muscular dystrophy. DMD symptom onset is in early childhood, usually between ages 3 and 5 years. Over time, children with Duchenne will have difficulty walking and breathing, then lead to disability, dependence, and premature death. The disease primarily affects boys, but in rare cases, it also can affect girls. The prevalence of DMD is approximately 1 in 3500 to 5000 male births worldwide [2]. Muscle weakness is the principal symptom of DMD and worsens over time, first affecting the proximal muscles and later affecting the distal limb muscles. Patients with DMD progressively lose the ability to perform activities independently and often require a wheelchair by their early teens. As the disease progresses, life-threatening heart and respiratory conditions can occur [3]. In general, patients succumb to the disease in their 20s or 30s [3]; however, disease severity and life expectancy can vary.

DMD is caused by mutations in the DMD gene, which encodes the protein product called dystrophin. DMD gene is one of the largest of the identified human genes, spanning 2.4 Mb of a genomic sequence and corresponding to about 0.1% of the total human genome [4]. The gene consists of 79 exons encoding a 14,000 bp messenger RNA transcript that is translated into dystrophin [5]. The most common mutation responsible for DMD is a deletion spanning one or multiple exons. Such deletions account for 60-70% of all DMD cases. Point mutations are responsible for around 26% of DMD cases. Exonic duplications account for 10 to 15% of all DMD cases [6]. Mutations in the DMD gene disrupt the protein's reading frame causing premature stop codons, leaving little or no functional dystrophin protein produced in cells.

Despite major therapeutic advances over the past 30 years, there are currently no curative therapies for DMD. Gene therapy is a promising experimental method that uses genes (the fundamental units of heredity) to treat disorders that result from genetic mutations. Currently, several kinds of gene-based therapies including exon skipping are being developed to treat DMD. Rehabilitation plays a critical role in overall management for individuals with DMD. The rehabilitation team can include physicians, Physical Therapists (PTs), Occupational Therapists (OTs), Speech and Language Therapists (SALTs), dieticians, orthotists, and durable medical equipment providers. Rehabilitation management requires an understanding of DMD pathology, natural history, and disease progression; providers should consider each individual's goals and lifestyle to optimize quality of life across the lifespan [7-9]. Rehabilitation can be provided in outpatient and school settings and should continue

throughout the patient's life. The patient should be assessed by a rehabilitation specialist at least every 6 months or more frequently if concerns [10,11].

International guidelines for DMD care were initially published in 2010 [12,13], with recommendations for DMD management, assessment, and intervention. However, because of significant advances in the understanding and management of DMD since then, DMD care considerations were recently updated to address the evolving needs of DMD patients [7-9]. The new DMD care considerations represent a fundamental shift in the care of people with DMD. Although in the past the main goal of DMD care was the prolongation of survival, the focus has changed to optimization of the quality of life, psychosocial management, independence, and transition to adulthood. The guidelines continue to highlight the importance of rehabilitation for Duchenne patients. Recommendations include daily preventive home stretching programs and select orthotic interventions, splinting, casting, positioning, and equipment. Submaximal aerobic activity (e.g., swimming and cycling) and avoidance of eccentric and high-resistance exercise are also recommended [11-13]. Figure 1 includes an overall summary of rehabilitative care across all disease stages of DMD. Adapted from Laura E et al. [7-9,14].



Assessment

Multidisciplinary rehabilitation assessment every 6 months or more frequently if concerns, change in status, or specific needs are present (appendix)

Intervention

Direct treatment

Direct treatment implemented by physical therapists, occupational therapists, and speech-language pathologists, tailored to individual needs, stage of disease, response to therapy, and tolerance, provided across the patient's lifespan

Prevention of contracture and deformity

- Daily preventive home stretching 4–6 times per week; regular stretching at ankles, knees, and hips; stretching of wrists, hands, and neck later if indicated by assessment
- Stretching for structures known to be at risk of contracture and deformity[a] and those identified by assessment
- 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
  - Passive/motorised supported standing devices—when standing in good alignment becomes difficult, if contractures are not too severe to prevent positioning or tolerance
  - KAFOs with locked knee joints—an option for late ambulatory and non-ambulatory stages
  - Custom seating in manual and motorised wheelchairs (solid seat, solid back, hip guides, lateral trunk supports, adductors, and head rest)
  - Power positioning components on motorised wheelchairs (tilt, recline, elevating leg rests, standing support, and adjustable seat height)

Exercise and activity

Regular submaximal, aerobic activity or exercise (eg, swimming and cycling) with assistance as needed, avoidance of eccentric and high-resistance exercise, monitoring to avoid overexertion, respect for the need for rests and energy conservation, and caution regarding potentially reduced cardiorespiratory exercise capacity as well as risk of muscle damage even when functioning well clinically

Falls and fracture prevention and management

- Minimisation of fall risks in all environments
- Physical therapist support of orthopaedics in rapid team management of long-bone fractures and provision of associated rehabilitation to maintain ambulation and/or supported standing capabilities

Management of learning, attentional, and sensory processing differences

Management in collaboration with team, based on concern and assessment

Assistive technology and adaptive equipment

Planning and education with assessment, prescription, training, and advocacy for funding

Participation

Participation in all areas of life supported at all stages

Pain prevention and management

Pain prevention and comprehensive management, as needed, throughout life

Figure 1:

Conclusion

Rehabilitation plays a critical role to overall management for individuals with DMD. Lifelong rehabilitation is paramount in assisting people with DMD to reach their full potential.

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