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ROLE OF PHYSICAL THERAPY IN THE MANAGEMENT OF MUSCULAR DYSTROPHY

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ABSTRACT

Muscular dystrophy is a genetic disorder cause progressive muscle wasting of skeletal muscle that affect the children 3 to 5 years of age and have affect from childhood to adulthood. MD generally caused by gene mutations i.e. dystrophin gene which require by muscle for proper functioning. Among all the types of MD, Duchenne MDs is most prevalent in boys as compared to girls affecting 1in 3600 lives male birth, which leads to muscle degeneration and premature death whereas Myotonic Dystrophies (DM1 and DM2) both in combined are prevalence affecting 18.1 per 100000 individuals. Physical exercise like stretching minimize contractures and preventing the muscles pain. Exercise like stretching, swimming, bracing and serial casting boost the muscles efficiency and reduce tiredness. As MDs cannot be cure but some new therapy and medication is used for reducing muscle weakness.

KEYWORDS: Muscular Dystrophy, Aquatic exercise, Dystrophin, Physical exercise, Breathing-assistance.

INTRODUCTION

The muscular dystrophies are genetic disorder comprise a heterogenic group that cause progressive muscle weakness and wasting of skeletal muscle.[1] Muscular dystrophies (MDs) is a neuromuscular disorder which generally affect the children of age 3 to 5 years.[2] Different forms of muscular dystrophies MDs exhibit which involve not only skeletal muscles but also present in cardiomyopathy features. These disorders vary in age of onset, severity, life expectancy and may affect various muscle groups differently. MDs are generally

caused by gene mutations in over 50 distinct genes, i.e. change in DNA sequence that affect protein called dystrophin that muscle require to function normally and many of them are caused by different genetic mechanisms.[3,4] In the absence of dystrophin, muscle fibers are susceptible to progressive degradation and fibrosis. Dystrophin protein is expressed in various tissues, including the heart, skeletal muscles, smooth muscles, retina, and brain.[5] MDs cannot be cure, So physical therapy are performed for slowing the progressive muscle weakness**.** Different types of muscular dystrophies are Duchenne MDs, Becker MDs, Myotonic MDs, and Facioscapulohumeral MDs. Among these most prevalent types of muscular dystrophy are Duchenne MDs and Myotonic Dystrophies (DM1 and DM2 both in combined are prevalence).

Table 1: Characteristics of different types of muscular dystrophy.

Muscular dystrophies	Gene	Age of onset	Affected muscles	Symptoms	Prevalence
Duchenne MDs	Dystrophin	Between the age of 2-5 years.	Skeletal and smooth muscle i.e. legs, arm and spine.	Muscle weakness and may cause premature death.	1/3500 live male births.[6]
Becker MDs	Dystrophin	Between 5 to 20 years, up to 60.	Affects skeletal muscle and may cause heart problems.	Weakening muscle. Affects only males.	0.1-1.8 per 10000 male individuals.[7]
Myotonic Dystrophies (DM)	CTG, CCTG repeat expansion	Childhood to adulthood.	Skeletal and smooth muscle, can also affect CNS.	Muscle weakness, cardiac abnormalities, CNS dysfunction and sleep disorder.	0.5-18.1 per 100000 individuals.[8]
FSHD type 1 FSHD type 2	DUX4 (Repeat contraction). SMCHD1, DNMT3B and LRIF1.	Teenage and adults.	Affects face, shoulder, upper arm and abdominal muscles.	Muscle weakness cause difficulty in chewing, talking and cause considerable morbidity.	Total FSHD1: 5-12/100000 individuals. FSHD2: 5% of patients with FSHD1.[9]

Orthopaedic Management in Duchenne Muscular Dystrophy by S.C Ranade, 2025. https://doi.org/10.1016/j.jposna.2024.100154.

Physical therapy in muscular dystrophy

Physical therapy is an important aspect for the treatment of child with DMD. Physical exercise like stretching minimize contractures and help in managing compromised skin

integrity. Consistent exercises can make it easier for child and adult to use home modification.

1. Muscle Extensibility and Joint mobility training

People with muscular dystrophy have reduced muscle extensibility and joint contractures due to aberrant muscle function, which causes restricted joint movement and muscle imbalance. This kind of change also affects the chest wall's movement, which results in breathing problems. Stretching the ankle, knee, and hip on a regular basis should start as soon as a diagnosis is made and last into adulthood. Stretching program should be started under the guidance of physical and occupational therapists. Splinting, serial casting, and manual therapy techniques are necessary in addition to stretching techniques to prevent joint contractures.[10,11]

2. Muscle Strength and Conditioning

Physiotherapists may suggest moderate, low-impact aerobic workouts for individuals with muscular dystrophy who are in the early ambulatory to early non-ambulatory phases of the condition. Exercises enhance cardiovascular function, boost muscle efficiency, and reduce tiredness. Muscular dystrophy patients engaging in an exercise program should be able to explain the symptoms of myoglobinuria and exhaustion, which include feeling weaker rather than stronger 30 minutes after exercise, excessive muscle soreness 24 to 48 hours later, severe muscle cramping, heaviness in the limbs, and significant shortness of breath.[12]

3. Swimming or aquatic exercise

Aquatic exercise is most common physical activities combines strengthening and aerobic exercises to maintain muscle structure and function. Exercises in the pool is safe and controlled, For children with altered muscle tone and severe contractures. Warm water allow children with DMD to perform target stretches , exercises and function-based and play activities that are progressively lost to them on dry skin,[13,14] When compared to strengthening and stretching activities performed on dry skin, the buoyancy and anti-gravity properties of the water relieved pressure on joints that result into reduction of pain and reduce joint morbidity.[2]

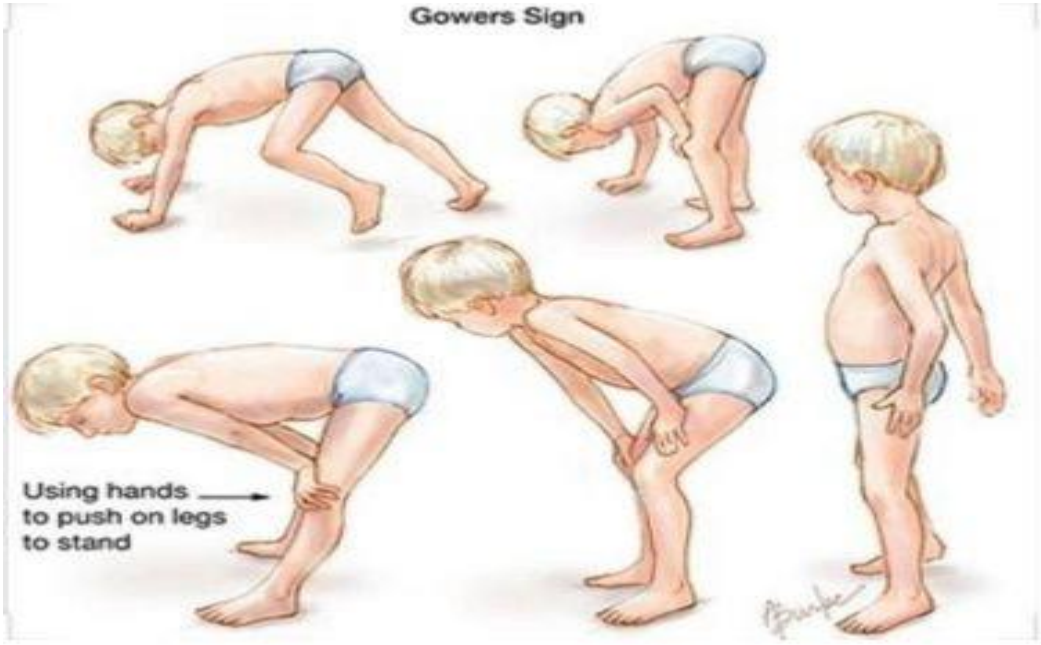
4. Respiratory rehabilitation

Respiratory disease in DMD is major cause of morbidity and mortality, there is inadequate awareness of its treatable nature. Most of the patients with DMD do not realize when they

have lost respiratory muscle strength that leads to a prolonged cough or to pneumonia.[15] However, Respiratory failure is the main causes of death in patients affected by DMD, So it is extremely important to measure lung and respiratory muscle function to monitor the progression of the disease.[16] The weakened respiratory muscles cannot fully expand the lungs up to the maximal capacity, leading to reduction of chest wall through shortening and stiffening of the unstretched tissue and fibrosis of Dystrophic Muscle.[17] Stretching and strengthening of inspiratory muscle can help to delay requirement for assisted ventilation in case of hypoventilation.[2]

5. Physiotherapy

It has a significant impact on the rehabilitation management of people with muscular dystrophy. The main goal of this is to prolong the function of unaffected muscle groups. Exercise can accelerate the breakdown of muscle fibers even if it helps preserve muscle function.[18] Muscular dystrophy progression can be simply divided into functional and unable to walk stages so availability of flexible gadgets is essential for promoting involvement and functionality. Therefore provision of a suitable wheelchair and seating, can aid to maximize freedom in daily activities, function, and engagement.[19]



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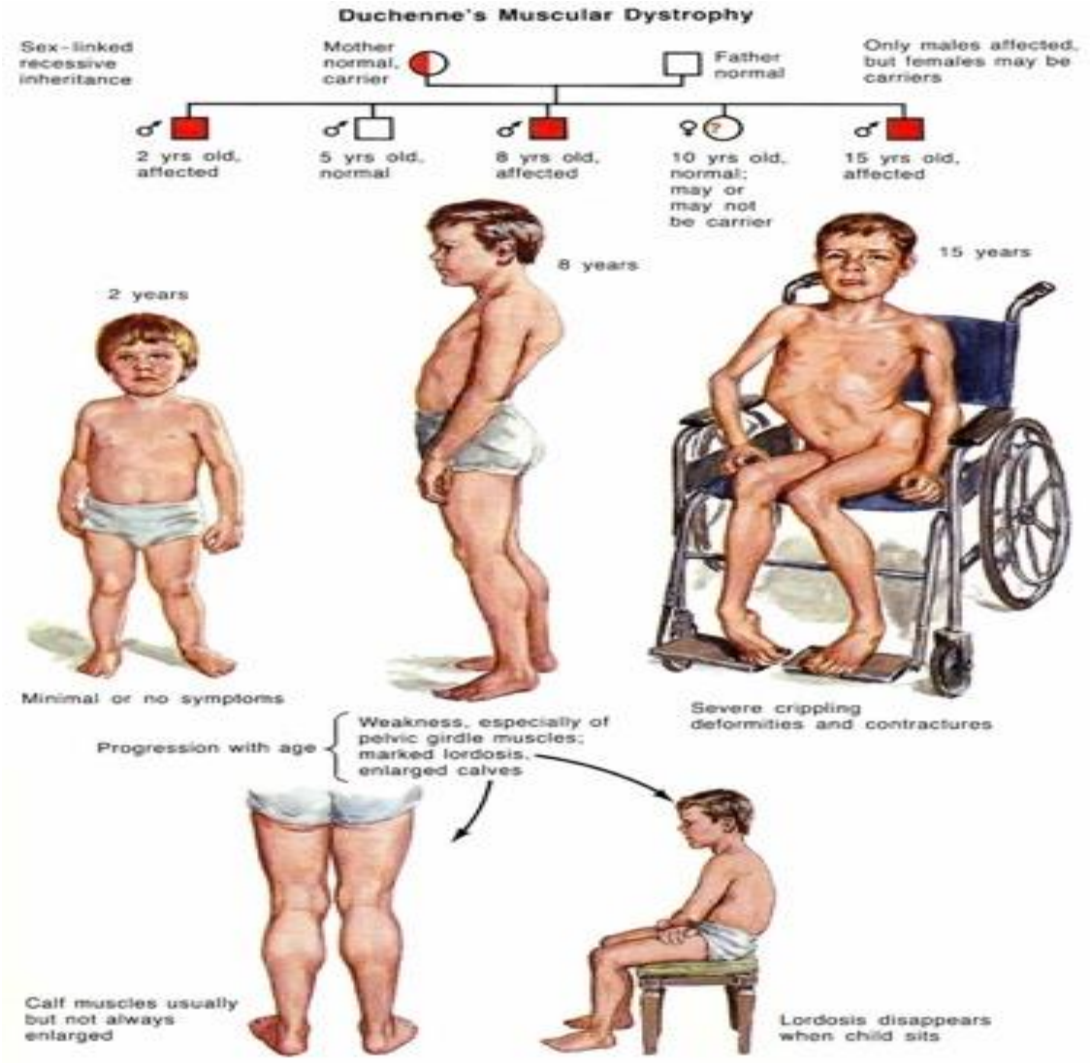

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New theraphy or medication used for muscular dystrophy

a. Duvyzat's drug

Duvyzat's (Givinostat) is first nonsteroidal drug approved medication for the treatment of patients with all genetic variants of Duchenne Muscular Dystrophy (DMD) in patient six years of age and older[22] Duvyzat's is a histone deacetylase (HDSC) inhibitor, targeting the deregulated activity of HDACs in dystrophic muscle tissue. By inhibiting HDAC pathological overactivity, Duvyzat's mitigates the sequence of events that lead to muscle damage, this potentially slowing the progression of muscle deterioration associated with the disease.[23]

b. K884 Medication

McGill university researcher have discovered an experimental compound called K884 which can boost the natural repair abilities of muscle stem cells.[24] K884 work by inhibiting two

enzymes i.e. PTPN1 and PTPN2, these enzyme activity disrupt the JAK/STAT signalling pathway which is essential for myogenic commitment. K884 restore proper STAT3 activation and this treatment significantly improved muscle repair outcomes.[25]

c. Stem cell therapy

Stem cell therapy is considered to be one of the most promising method for treating Muscular Dystrophies. Stem cells are responsible for the development and maintenance of tissue and organs.[26] Isolation of stem cell can be from embryonic, adult or donor cell. There are a lot of ongoing stem cell research for muscle weakening and wasting conditions, Several cell types with different characteristics and tissues of origin, including myogenic stem and progenitor cells, stromal cells, and pluripotent stem cells, have been investigated.[27]

CONCLUSION

A key component of treating muscular dystrophy (MD) is physical therapy, which helps patients with the condition live better lives overall, lessen pain, and delay the progression of muscle weakness. Even though MD cannot be cured, there are several advantages to therapeutic procedures such respiratory rehabilitation, water therapy, stretching, and strengthening exercises. Long-term preservation of muscle function and independence can be achieved with early and regular physical therapy programs that include joint mobility training, muscle conditioning, and the use of assistive devices. Furthermore, there are promising novel approaches to controlling MD and slowing its progression, such as stem cell therapies, Duvyzat, and K884. In the end, improving requires a comprehensive strategy comprising medical specialists, physical therapists, and new treatment approaches the muscular dystrophy.

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