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Case Report

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Clinical Insights and Multidisciplinary Tertiary Prevention in Duchenne Muscular Dystrophy: A Case Report

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ABSTRACT

Duchenne Muscular Dystrophy (DMD) is a severe X-linked recessive disorder characterized by progressive muscle atrophy, loss of mobility, and systemic complications, predominantly affecting males. This case report presents a 10-year-old boy with progressive walking difficulties, frequent falls, and hallmark features such as a positive Gower's sign, waddling gait, lumbar lordosis, calf hypertrophy, and proximal muscle weakness. Symptoms began at age 4, with a notable family history of similar symptoms in an older sibling. Elevated creatine kinase levels and clinical findings strongly suggest DMD, with genetic testing underway for confirmation. Management focused on tertiary prevention strategies, including corticosteroid therapy, physiotherapy, orthotic support, respiratory care, and nutritional counselling. A multidisciplinary approach emphasized the importance of early interventions, assistive devices, and adjunct therapies such as yoga to improve functional outcomes and quality of life. This case highlights the critical role of comprehensive, personalized care and multidisciplinary collaboration in addressing the complex challenges of DMD, ultimately aiming to enhance the well-being and independence of affected individuals.

Keywords: Duchenne muscular dystrophy, genetic neuromuscular disorder, multidisciplinary care, tertiary prevention, quality of life

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INTRODUCTION

Duchenne muscular dystrophy (DMD) is the most common genetic neuromuscular disorder, predominantly affecting males due to its X-linked recessive inheritance.[1] It impacts approximately one in 3600 male infants and is characterized by progressive muscle atrophy, leading to severe disability and early death. Initial symptoms, such as difficulty ascending stairs, a waddling gait, and frequent falls, appear between ages 2 and 3. By ages 10–12, most patients require wheelchairs, and ventilation support is needed around age 21.[2] Due to proximal muscle weakness, most affected persons cannot run and jump adequately, which also causes them to adopt the traditional Gowers manoeuvre while getting up from the floor. Affected individuals can also have somewhat delayed motor milestones. When a patient's physical ability sharply deviates from that of their peers at age five on average, they are diagnosed. Boys who go untreated lose muscle strength and need a wheelchair before they turn ten. Muscle function has already decreased by the time a parent becomes concerned about DMD, which typically takes 1.6 years to diagnose.[3] If nothing is done, heart, lungs, and joint difficulties occur,

and the average age at death is about 19 years. There may also be non-progressive cognitive impairment.[4] Early diagnosis and intervention can significantly improve outcomes, allowing patients to live into their fourth decade.

CASE REPORT

A 10-year-old boy presented with progressive difficulty walking and frequent falls over several years. Symptoms began at age 4 with difficulties in walking and rising from the floor. His 12-year-old brother exhibits similar symptoms. The patient struggles to keep up with peers, often complains of leg pain and fatigue, and has delayed developmental milestones. No family history of neuromuscular or genetic disorders was reported.

Initially met developmental milestones until age 4. At present, the patient requires assistance with daily activities such as dressing and using the restroom and experiences increased fatigue after physical activities. Notable difficulties include getting up from the floor, climbing stairs, frequent falls, and walking long distances without support. The patient had positive Gower's sign, waddling gait, lumbar lordosis, decreased muscle strength in proximal lower extremities, calf hypertrophy, hamstring rigidity, and poor oral hygiene. Weakness of the proximal muscles leads to foot drop and tight heel cord (contracture) leads to walking on tiptoe are shown Figure 1 and 2.

The patient had high creatine kinase levels, which are an indicator of muscle damage. Genetic testing is pending, with DMD suspected based on clinical presentation. Further assessments might include electromyography and nerve conduction studies, though muscle biopsy is typically avoided but could reveal DMD-specific histopathological changes.

DISCUSSION

DMD is characterized by muscle atrophy and degeneration.[5] Treatment guidelines focus on diet, exercise, and cardiovascular health to slow disease progression. Corticosteroids such as prednisolone and deflazacort are commonly used to improve muscle strength and function.[6] Tertiary prevention involves managing symptoms and enhancing life quality, including monitoring for scoliosis and using physiotherapy and orthotic support to prevent contractures and maintain posture.[7] Respiratory issues are man-

Figure 1. Foot drop due to proximal weak muscles.

Figure 2. Tight heel cord (contracture) leading to toe walking.

aged through ventilation support and therapies to maximize respiratory capacity.

Rehabilitation plays a crucial role in DMD management, aiming to optimize respiratory function, bone density, muscle strength, and coordination.[8] Standard therapies include physical therapy, stretching, orthopaedic surgery, ventilatory support, scoliosis management, and nutritional supplements. A multidisciplinary team approach is essential, involving various health-care specialists to address the multifaceted challenges of DMD.

Yoga has shown benefits in improving autonomic regulation and parasympathetic control in DMD patients, supporting its use as an adjunct therapy.[9] A multidisciplinary rehabilitation team, including physicians, therapists, and equipment providers, supports patients in various settings to enhance their functional mobility and quality of life.[10] Motorized stand-and-drive wheelchairs can aid in maintaining independence and reducing fall risks.

During the ambulatory stage, key strategies include preventing deformities and falls, promoting specific activities or exercises, and providing necessary equipment and orthoses.[11] In addition, supporting funding for social services, transportation access, and community involvement is vital for the well-being of individuals with DMD.

CONCLUSION

Tertiary prevention is critical in managing DMD, addressing various complications through a team-based approach. This includes cardiomyopathy treatment, respiratory support, mobility aids, and psychosocial support. Staying updated with new therapies and clinical research is essential. End-of-life care focuses on providing palliative care to maintain comfort, dignity, and quality of life. Overall, a comprehensive, personalized strategy aims to maximize the well-being of DMD patients and their families in the face of this challenging illness.

Disclosures

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