



CLINICAL FEATURES, DIAGNOSIS, AND MANAGEMENT OF DUCHENNE MUSCULAR DYSTROPHY

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Abstract: Duchenne Muscular Dystrophy (DMD) is the most common and severe form of inherited muscular dystrophy, characterized by progressive muscle degeneration and weakness resulting from mutations in the dystrophin gene. The disease primarily affects boys and leads to significant physical disability, cardiopulmonary complications, and reduced life expectancy. Recent advances in genetic testing, early diagnostic techniques, and novel therapeutic approaches have improved disease management and patient outcomes. This review highlights the clinical manifestations, diagnostic methods, and current treatment strategies for Duchenne Muscular Dystrophy. Particular attention is given to emerging genetic therapies and multidisciplinary care approaches that aim to enhance quality of life and prolong survival.



Understanding the latest developments in DMD is essential for improving early diagnosis and optimizing long-term patient care.

Keywords: Duchenne Muscular Dystrophy, dystrophin gene, muscle weakness, genetic diagnosis, muscular dystrophy, exon skipping therapy, corticosteroids, rehabilitation, neuromuscular disorders.

Introduction

Duchenne Muscular Dystrophy (DMD) is an X-linked recessive neuromuscular disorder caused by mutations in the dystrophin gene located on chromosome Xp21. The absence or severe deficiency of dystrophin, a critical structural protein in muscle fibers, results in progressive muscle damage, degeneration, and replacement of muscle tissue by fat and fibrous connective tissue. DMD affects approximately one in every 3,500–5,000 live male births worldwide and represents the most prevalent form of childhood muscular dystrophy. Clinical symptoms usually appear during early childhood, often between the ages of two and five years. Children commonly present with delayed motor development, difficulty running, climbing stairs, and frequent falls. As the disease progresses, muscle weakness becomes more pronounced, eventually leading to loss of ambulation during adolescence. In addition to skeletal muscle involvement, cardiac and respiratory complications significantly contribute to morbidity and mortality in affected individuals. Advances in molecular genetics have revolutionized the diagnosis of DMD. Genetic testing now enables early and accurate identification of disease-causing mutations, facilitating timely intervention and genetic counseling. Furthermore, improvements in supportive care, corticosteroid therapy, cardiac management, respiratory support, and emerging gene-based treatments have significantly increased life expectancy compared to previous decades. Despite substantial progress, Duchenne Muscular Dystrophy remains a serious and life-limiting condition. Continuous research is focused on developing innovative therapeutic strategies aimed at restoring dystrophin expression and slowing disease progression. Therefore, a comprehensive understanding of the clinical features, diagnostic approaches, and management options is essential for healthcare professionals involved in the care of patients with DMD. The aim of this article is to review the clinical manifestations, diagnostic methods, and current management strategies of Duchenne Muscular Dystrophy, with particular emphasis on recent advances in treatment and future therapeutic perspectives.

Materials and Methods

This review was conducted to evaluate current evidence regarding the clinical features, diagnosis, and management of Duchenne Muscular Dystrophy (DMD). A comprehensive literature search was performed using major scientific databases, including PubMed, Scopus,



Web of Science, and Google Scholar. The search strategy included the keywords “Duchenne Muscular Dystrophy,” “dystrophin gene,” “clinical manifestations,” “genetic diagnosis,” “corticosteroid therapy,” “gene therapy,” “exon skipping,” and “multidisciplinary management.” Scientific articles published between 2015 and 2025 were selected to ensure the inclusion of recent advances in the field. Inclusion criteria comprised original research articles, systematic reviews, meta-analyses, clinical trials, and international clinical guidelines focusing on DMD. Studies with insufficient methodological quality, duplicate publications, and articles unrelated to the objectives of the review were excluded. The selected literature was analyzed according to major themes, including epidemiology, pathophysiology, clinical presentation, diagnostic approaches, therapeutic interventions, and emerging treatment strategies. Information from the included studies was synthesized to provide a comprehensive overview of current developments in the diagnosis and management of Duchenne Muscular Dystrophy.

Results

The reviewed literature demonstrated that Duchenne Muscular Dystrophy remains the most common inherited neuromuscular disorder of childhood. Clinical manifestations typically begin between two and five years of age and are characterized by progressive proximal muscle weakness, delayed motor milestones, frequent falls, difficulty climbing stairs, and the Gowers' sign. Calf muscle pseudohypertrophy was identified as one of the most characteristic physical findings. Recent advances in molecular diagnostics have significantly improved the early detection of DMD. Genetic testing, including multiplex ligation-dependent probe amplification (MLPA) and next-generation sequencing (NGS), has become the primary diagnostic approach for identifying dystrophin gene mutations. These techniques provide high diagnostic accuracy and facilitate early genetic counseling. The analysis also revealed substantial progress in disease management. Corticosteroids continue to be the standard pharmacological treatment and have been shown to delay disease progression and preserve motor function. In addition, multidisciplinary care involving neurologists, cardiologists, pulmonologists, physiotherapists, and rehabilitation specialists has contributed to improved quality of life and prolonged survival. Emerging therapies such as exon-skipping agents, stop-codon read-through therapies, and gene replacement strategies have demonstrated promising clinical outcomes. Several studies reported improved muscle function and slowed disease progression in selected patient populations receiving these novel treatments.

Discussion

The findings of this review highlight the significant advances achieved in the diagnosis and management of Duchenne Muscular Dystrophy during recent years. Early recognition of clinical symptoms combined with modern genetic testing has enabled more accurate and timely



diagnosis, allowing earlier therapeutic intervention and improved disease monitoring. The widespread use of molecular diagnostic techniques has transformed clinical practice by reducing diagnostic delays and increasing the identification of specific genetic mutations. This progress is particularly important because many emerging therapies are mutation-specific and require precise genetic characterization before treatment initiation. Although corticosteroids remain the cornerstone of treatment, their long-term use is associated with adverse effects, including weight gain, osteoporosis, growth retardation, and metabolic complications. Consequently, there is increasing interest in targeted therapies that directly address the underlying genetic defect. Exon-skipping therapies and gene-based approaches represent major milestones in the development of personalized medicine for DMD. Furthermore, multidisciplinary management has become an essential component of patient care. Regular cardiac and respiratory assessments, physiotherapy, nutritional support, and psychosocial interventions have significantly improved survival rates and quality of life. Current evidence suggests that comprehensive care can extend life expectancy well into adulthood. Despite these advances, Duchenne Muscular Dystrophy remains a progressive and life-limiting disorder. Challenges related to treatment accessibility, high costs of novel therapies, and long-term efficacy remain unresolved. Future research should focus on optimizing gene-editing technologies, improving therapeutic delivery systems, and developing curative strategies capable of restoring dystrophin expression throughout the body.

Conclusion

Duchenne Muscular Dystrophy is a severe X-linked genetic disorder characterized by progressive muscle degeneration and significant functional impairment. Advances in molecular genetics and diagnostic technologies have enabled earlier and more accurate identification of affected individuals, facilitating timely intervention and improved disease management. Current treatment strategies, including corticosteroid therapy, rehabilitation programs, respiratory support, and cardiac care, have significantly improved patient survival and quality of life. Furthermore, emerging genetic therapies such as exon-skipping technologies, gene replacement approaches, and genome-editing techniques represent promising developments that may alter the natural course of the disease. Despite considerable progress, Duchenne Muscular Dystrophy remains associated with substantial morbidity and mortality. Continued research is required to improve treatment accessibility, evaluate long-term therapeutic efficacy, and develop more effective disease-modifying interventions. A multidisciplinary and patient-centered approach remains essential for optimizing clinical outcomes and enhancing the quality of life of individuals living with Duchenne Muscular Dystrophy.



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