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DUCCHENE MUSCULAR DYSTROPHY - A CASE REPORT

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ABSTRACT Duchenne Muscular Dystrophy (DMD) is the most common and severe form of childhood muscular dystrophy, characterized by progressive muscle degeneration and weakness due to mutations in the dystrophin gene located on chromosome Xp21. The absence or near absence of dystrophin, an essential structural protein that stabilizes muscle cell membranes, leads to repeated cycles of muscle fiber damage and replacement by fat and fibrous tissue. This case report discusses a 12-year-old male presenting with progressive difficulty in getting up from sitting position and raising his arms above shoulder since early childhood. Clinical examination revealed proximal muscle weakness, positive Gowers sign, calf hypertrophy, and foot drop. The differential diagnosis includes Duchenne muscular dystrophy (DMD) and other neuromuscular disorders. Further investigations confirmed the diagnosis and initiate appropriate management.

KEYWORDS :

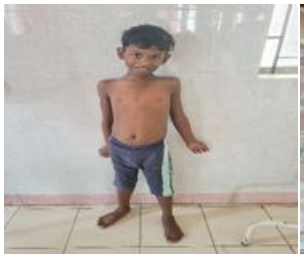
INTRODUCTION

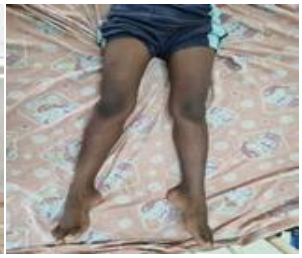
Duchenne muscular dystrophy (DMD) is a severe X-linked recessive disorder characterized by progressive muscle weakness and wasting. It typically manifests in early childhood with delayed motor milestones and difficulty in activities requiring proximal muscle strength, such as rising from the floor and climbing stairs. The disease primarily affects males due to the genetic mutation in the DMD gene, leading to absence of dystrophin protein. Diagnosis is based on clinical suspicion supported by markedly elevated serum creatine kinase (CK) levels, electromyography, genetic testing confirming dystrophin gene mutations, and muscle biopsy when required. Advances in molecular diagnostics have improved early detection and genetic counseling.

Although there is no definitive cure, current management strategies including corticosteroid therapy, physiotherapy, respiratory support, cardiac surveillance, and emerging gene-targeted therapies have significantly improved survival and quality of life. Early diagnosis is crucial for timely intervention and management to improve quality of life and delay disease progression.

CASE REPORT

12-year-old male, presented with difficulty in rising from sitting position since age 4 and difficulty raising his arms above shoulder level since age 8. These symptoms were insidious in onset and progressively worsened. He also experienced frequent falls and noticed wasting of thigh and biceps muscles. Neurological examination revealed positive Gowers sign, calf hypertrophy, foot drop, and fixed flexion contractures in bilateral knees. There were no sensory deficits, pain, or systemic symptoms.





Past Medical History is Unremarkable, with normal birth and developmental milestones. No relevant family history also. General examination showed a thin-built, poorly nourished adolescent with no systemic abnormalities except for musculoskeletal findings as described. Further investigations including creatine kinase levels, electromyography (EMG), muscle biopsy, and genetic testing for DMD mutation were recommended and shows positive findings. The clinical findings of progressive proximal muscle weakness, positive Gowers sign, calf hypertrophy, and foot drop along with investigation findings strongly suggest a diagnosis of Duchenne muscular dystrophy (DMD).

DISCUSSION

DMD is characterized by a mutation in the DMD gene, leading to absence of dystrophin protein essential for muscle integrity. Early symptoms typically appear in early childhood and progress relentlessly, causing significant disability by adolescence. The prevalence of DMD is less than 10 cases per 100,000 males. DMD in females is very rare (<1 per million) and is limited to case reports of individuals with Turner syndrome10–12, a translocation involving DMD or those with bi-allelic DMD mutations

The disease is caused by a mutation in the dystrophin gene that results in an absence or a decrease in dystrophin within muscle fibers. DMD is a progressive neuromuscular disease characterized by muscle weakness, associated motor delays, loss of ambulation, respiratory impairment and cardiomyopathy .Muscle weakness begins in the lower limbs and affects proximal more than distal muscles. The disease typically progresses through 5 stages: pre-symptomatic, early ambulatory, late ambulatory, early non-ambulatory, and late non-ambulatory. Diagnosis typically occurs during the early ambulatory stage around age 5 when the initial symptoms are seen including frequent falls, Gower's sign, and trouble with running and climbing stairs. Loss of ambulation occurs around age 12 and is associated with a decrease in health-related quality of life (HRQOL) and increase in economic burden.

A standard diagnostic work-up of patients with suspected DMD are enzymes (CK levels, AST &ALT), muscle biopsy (to assess whether dystrophin is properly localized or absent/reduced or has an altered size)and genetic testing(the presence and abundance of the 79 DMD exons is evaluated)

High-quality multidisciplinary care can slow disease progression, prolong functional independence and prolong life expectancy. Early detection of the disease, better clinical practice guidelines and increased ventilator use along with early intervention has improved the life expectancy of these patients Management of DMD involves multidisciplinary approach including physiotherapy, orthopedic interventions, respiratory support, and genetic counseling. The role of genetic testing is crucial for confirming the diagnosis and providing prognostic information. Newer therapies such as exon skipping and gene therapy hold promise in altering disease course, although access and affordability remain significant challenges.

CONCLUSION

This case underscores the importance of recognizing early signs and symptoms of Duchenne muscular dystrophy for timely diagnosis and intervention. The progressive nature of the disease necessitates comprehensive management strategies aimed at improving quality of life and delaying disability. Further research and advancements in therapeutic options are essential to mitigate the devastating impact of

DMD on affected individuals and their families.

This case report highlights clinical presentation, examination findings, and the diagnostic pathway towards a suspected diagnosis of Duchenne muscular dystrophy, emphasizing the need for heightened clinical suspicion and early intervention in similar cases.

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