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Case Report:

A Child with Duchenne Muscular Dystrophy: A Case Report of a Rare Diagnosis in Cumilla, Bangladesh

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Abstract

Background: *Muscular dystrophies encompass a diverse range of conditions that share the common feature of gradually worsening muscle weakness. Among these, Duchenne Muscular Dystrophy (DMD) stands out as the most prevalent X-linked form affecting children, usually manifesting in the early years of life. This condition primarily weakens muscles near the body's core and causes distinctive calf muscle enlargement in boys. Children with DMD typically show delayed motor milestones and progressive deterioration and ultimately bedbound. The condition often leads to early death due to cardiac or respiratory complications. Current interventions, including corticosteroid therapy and respiratory support, have enhanced daily functioning, mobility duration, overall well-being, and survival rates. However, researchers continue pursuing novel treatments in hopes of finding a definitive cure for this serious disease. This report presents the clinical case of a boy aged 10 years and 11 months diagnosed with DMD, focusing on his distinctive clinical presentation and other significant findings.*

Introduction: Duchenne muscular dystrophy (DMD) represents an early-onset, severe, and rapidly progressive neuromuscular condition classified within the dystrophinopathy spectrum, characterized primarily by muscle weakness as its predominant clinical feature [1,2.]

DMD constitutes a debilitating disorder of early onset resulting from functional dystrophin deficiency. Affected individuals typically require wheelchair assistance by age twelve and frequently experience mortality due to cardiac, respiratory, or combined organ failure during their mid-to-late twenties. Becker muscular dystrophy (BMD), a milder variant, can be differentiated from DMD through its later onset, delayed wheelchair dependency, and extended lifespan.

The etiology of DMD involves mutations in the X-linked recessive dystrophin gene[3]. The global incidence affects approximately 1 in 3,500 male births, with regional variations ranging from 0.95 per 100,000 in certain sub-Saharan African regions to 16.76 per 100,000 in some European countries [4-6].

In developing nations, DMD is associated with premature mortality, typically occurring during the late adolescent years or early third decade of life[7]. Accurate diagnosis of muscular dystrophies necessitates comprehensive medical assessment, including evaluation of weakness patterns, age at onset, familial history, and disease-specific characteristics. Throughout many Asian nations, the convergence of inadequate healthcare infrastructure, limited literacy, delayed diagnosis, and postponed therapeutic intervention may contribute to elevated morbidity and mortality rates[8].

Case presentation

An 10-year- 11 months old male patient presented to the medical clinic with a history of progressive generalized weakness spanning over six years, during which he had been evaluated in multiple hospitals without a definitive diagnosis and had received empirical treatments including multivitamins and antibiotics with no clinical benefit. His parents reported poor academic performance, frequent falls,

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inability to keep up with peers during sports due to excessive fatigue, difficulty climbing stairs, and progressive muscle weakness; there was no history of muscle pain, preceding trauma, or symptoms suggestive of infection. Notably, family history revealed that his twin brother, currently the same age, had experienced similar symptoms and had become bedridden. On examination, the child was fully conscious but had mild wasting and a Montreal Cognitive Assessment score of 20, indicating significant cognitive impairment. His temperature was normal (36.7°C). Neurological examination showed a waddling gait, toe walking, reduced power of proximal lower limb muscles, calf hypertrophy, flat feet (pes planus), and a positive Gowers' sign; cranial nerve examination was unremarkable and there were no upper or lower motor neuron signs. Muscle tone was mildly reduced, reflexes were preserved without spasticity, and cardiac and respiratory examinations were normal. Clinical history also included difficulty rising from the floor over the past six months, frequent falls, delayed motor milestones with walking achieved at 20 months, and calf enlargement noted by parents.

Laboratory investigations revealed serum creatine phosphokinase of 1983 U/L (reference: 24–195 U/L), LDH of 1209 U/L (135–225 U/L), vitamin D deficiency (19.1 ng/mL), low serum calcium (9.4 mg/dL), and low serum creatinine (0.45 mg/dL). Bone densitometry of the lumbar spine (L1–L4) showed a z-score of -2.8 and BMD of 0.447 g/cm². Electromyography demonstrated a myopathic pattern. Genetic testing by multiplex ligation-dependent probe amplification identified a deletion of exons 45–50 in the DMD gene. Although not routinely required when genetic results are conclusive, a muscle biopsy was performed, revealing extensive loss of skeletal muscle fibers replaced by fat and fibrosis, consistent with DMD. Baseline echocardiogram and electrocardiogram were normal. The patient was started on prednisolone at 0.75 mg/kg/day and daily physiotherapy, with baseline spirometry and cardiac evaluation showing normal function; genetic counseling was provided to the family, including referral for maternal carrier testing. A multidisciplinary long-term plan was arranged involving neurology, cardiology, pulmonology, physiotherapy, and psychosocial support, alongside regular monitoring for anticipated progressive loss of ambulation around age 12 and future need for mobility aids and ventilatory support. Based on the combination of clinical presentation, family history, laboratory results, genetic testing, and biopsy findings, a diagnosis of Duchenne Muscular Dystrophy was established. His twin brother was also given some treatment plan.

Discussion

DMD represents the most prevalent muscular dystrophy globally, including in Bangladesh, resulting from dystrophin gene mutations that impair the body's capacity to synthesize dystrophin protein essential for muscle contraction. Each muscle contraction cycle induces damage that undergoes repair; however, the deficient protein results in compromised muscle restoration. This perpetual cycle of damage, repair, and eventual replacement of muscle tissue with fibrofatty tissue underlies the characteristic clinical manifestations of progressive muscle wasting and degeneration, typically becoming apparent by 3-4 years of age [9-12].

DMD originates from mutations in the DMD gene, which encodes dystrophin protein. This protein localizes to the cytoplasmic surface of the sarcolemma in skeletal muscle, forming a component of the dystrophin-associated glycoprotein complex. Dystrophin comprises an N-terminal actin-binding

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domain, 24 spectrin-like repeat units separated by four hinge regions, a cysteine-rich domain, and a C-terminal domain. The cysteine-rich domain binds laminin-2 via alpha and beta-dystroglycan, establishing a mechanical connection between cytoskeletal actin and the extracellular matrix. The DMD gene encompasses 79 exons representing merely 0.6% of the gene, with the remainder consisting of extensive introns. This substantial gene size increases mutation susceptibility, resulting in dystrophin functional loss and producing a prematurely truncated, unstable dystrophin protein. Intragenic deletions constitute the majority of mutations, accounting for 65-72% of all DMD cases. The precise mechanism by which dystrophin deficiency leads to muscle fiber degeneration remains incompletely understood. Dystrophin absence at the plasma membrane causes delocalization of dystrophin-associated proteins, cytoskeletal disruption with consequent membrane instability, and increased mechanical stress susceptibility. Additionally, altered membrane permeability and abnormal calcium homeostasis contribute to pathogenesis, with elevated cytosolic calcium concentrations activating proteases such as calpains [11].

Affected males typically present with running difficulties, challenges rising from the ground, frequent falls, or toe-walking. Clinical findings include waddling gait, calf enlargement, and lumbar lordosis that resolves when seated. Proximal lower limb muscle weakness manifests as patients using their hands and arms to "walk" up their body from squatting positions due to hip and thigh muscle strength deficiency, indicating positive Gower's sign [13]. In the present case, the affected child demonstrated delayed motor development, walking and stair-climbing difficulties, positive Gower's sign, and muscle weakness.

Diagnosis confirmation requires elevated serum marker levels of CK, muscle biopsy, electromyography, and genetic analysis. Increased sarcolemma permeability from repeated contractions in DMD patients causes protein leakage, including CK, into plasma, resulting in characteristically elevated serum CK levels. Other enzymes including alanine transaminase, aspartate transaminase, aldolase, and lactate dehydrogenase are similarly elevated[10,14]. In this case, serum markers including CK, lactate dehydrogenase, and alanine transaminase levels were markedly increased, with muscle biopsy and electromyography yielding positive results.

Current DMD management encompasses physiotherapy and corticosteroid therapy, which delays ambulatory loss by 1-3 years without providing cure, as implemented in our case. However, corticosteroids are associated with significant adverse effects, including weight gain, decreased bone mineralization, Cushing syndrome, and behavioral disturbances. Alternative regimens have been investigated, though their efficacy compared to daily dosing remains incompletely studied [15,16]. Emerging evidence suggests particular benefit from treatment initiation before age 5 years, although supporting data remain limited[17]. Recent therapeutic modalities include gene therapy and stem cell therapy, which demonstrate promise and suggest beneficial effects from dystrophin-like protein upregulation. Prenatal counseling and genetic testing, including multiplex ligation-dependent probe amplification, are being utilized to provide hope for patients with this progressive and ultimately fatal muscular dystrophy, aiming to prolong and enhance quality of life [18].

Conclusion

Differential diagnosis considerations for conditions presenting similarly to DMD remain crucial. BMD is typically regarded as the milder phenotypic variant of the disease spectrum. Clinical manifestations generally appear later in the disease course, with cardiac failure during the second decade commonly representing the initial presentation. While DMD prevalence is perceived as low in Bangladesh, limited studies exist to substantiate this assumption. Conventional genetic testing demonstrates reduced sensitivity, while advanced testing methods remain cost-prohibitive and rarely accessible. Consequently, conducting observational studies, including case-control designs, is essential to identify unique genetic mutations prevalent in Bangladesh, which would enhance diagnostic precision and reduce biopsy dependency. Additionally, clinicians should maintain heightened suspicion for DMD and other dystrophinopathies in patients, particularly children and adolescents, presenting with early-onset progressive weakness accompanied by clinical features suggestive of dystrophinopathies, including waddling gait, toe walking, and positive Gowers' sign.

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Declaration of Patient Consent

The authors confirm obtaining all appropriate patient consent documentation. The patient consent forms indicate that patients have provided authorization for their images and clinical information to be published in the journal. Patients acknowledge that names and initials will not be disclosed, and appropriate measures will be implemented to protect their identity, though complete anonymity cannot be assured.

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Conflicts of Interest

The authors declare no conflicts of interest.

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