Case Report

Delayed Presentation of Duchenne Muscular Dystrophy: A Case Report

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Abstract

Duchenne Muscular Dystrophy (DMD) is the most prevalent genetic myopathic disease of childhood that usually occurs between the ages of 2 and 4 years and advances into progressive muscle impairment. Late introduction after early childhood is rare and can delay diagnosis and treatment. The current case of a 12-year-old male with DMD, who reported late with progressive muscle weakness, is significant in terms of awareness of clinicians in relation to non-traditional age of presentation. The prognosis is still guarded, regardless of the age at which one is diagnosed with the condition; therefore, early initiation of multidisciplinary treatment is beneficial.

Keywords: Duchenne muscular dystrophy, presentational delay, dystrophin, myopathy, pediatrics.

Received: 08 February 2026 **Revised: 27 January 2026** **Accepted: 13 February 2026** **Published: 27 February 2026**

Introduction

The most common and well-known early onset inherited progressive myopathic disorder is Duchenne Muscular Dystrophy (DMD). It is an X-linked recessive inherited disease with an incidence of about 1 in 3,500-5,000 living male births.[1] The disease is due to mutations in the dystrophin protein, which is the protein used to give the fibres their structure, and is located on the short arm of the X chromosome (Xp21).[4] The dystrophin gene is one of the largest genes in the human genome, with 79 exons, and about 30% of cases are due toClinically, DMD typically presents itself between the ages of 2 and 4 years of age with progressive proximal muscle weakness, delayed motor milestones, and loss of ambulation in early teenage years. Cardiorespiratory complications are common and contribute significantly to morbidity and mortality.[6] Beyond motor symptoms, neurodevelopmental comorbidities, including intellectual disability and autism spectrum disorders, affect a significant subset of patients due to a deficiency of brain dystrophin isoforms.[7]

Delayed presentation in late childhood or adolescence is uncommon and may lead to missed opportunities for early interventions. Diagnostic delays of over one year from symptom onset remain common, often due to a lack of awareness among frontline healthcare professionals.[8] We present a case of Duchenne Muscular Dystrophy with delayed clinical presentation to emphasize the need for awareness of such atypical cases.

A 12-year-old male child was brought to the pediatric outpatient department with complaints of progressive weakness of both upper and lower limbs for the past four years. The child had been unable to walk independently for the past two years. There was no history of difficulty in walking before the onset of symptoms. No significant perinatal history or family history of similar illness was reported. On physical examination, the patient was obese. The bulk of all four limbs appeared to be normal. Muscle tone was significantly decreased in all limbs. Contractures were present in the bilateral knee and ankle joints. Deep tendon reflexes, including knee and ankle jerks, were also absent. Pseudohypertrophy of the calf muscles was noted.

The cardiovascular examination findings were unremarkable. Two-dimensional echocardiography revealed no evidence of cardiomyopathy, with a normal ejection fraction.

Investigations

Laboratory evaluation revealed a markedly elevated serum creatine phosphokinase (CPK) level of 4047.4 U/L. A muscle biopsy was performed, and the results were consistent with muscular dystrophy. Whole exome sequencing confirmed a mutation in the DMD gene, establishing the diagnosis of Duchenne Muscular Dystrophy.

Management and Outcome

The patient was started on low-dose corticosteroid therapy. Supportive management, including respiratory and regular physiotherapies, was initiated. Parents were counseled in detail regarding the progressive nature of the disease, prognosis, and importance of regular follow-up with cardiopulmonary monitoring. Carrier testing was offered to the mother and at-risk

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DOI:
10.21276/amit.2026.v13.i1.388

How to cite this article: Goswami N, Bhattacharjee A, Parashar AK. Delayed Presentation of Duchenne Muscular Dystrophy: A Case Report. Acta Med Int. 2026;13(1):549-550.

female family members, along with appropriate genetic counseling.

At present, the child is under regular follow-up and wheelchair-dependent. Ongoing multidisciplinary care is being provided to optimize quality of life and manage complications.

Discussion

Duchenne Muscular Dystrophy classically presents in early childhood; however, delayed presentation, as seen in this case, though rare, has been reported. Ciafaloni et al. documented that diagnostic delays average approximately one year from the first evaluation to diagnosis, with incorrect referrals to therapists rather than neurologists contributing significantly to this delay. [8] Van Ruiten et al. similarly identified that delayed creatine kinase testing was the primary factor prolonging time to diagnosis.[9]

Variability in disease progression, partial dystrophin expression, or the absence of early detection could explain this abnormal presentation. Committed to a second diagnostic issue in the context of this case study, Donkervoort et al. reported the concept of the so-called double trouble, in which concomitant hereditary skeletal dysplasias are seen to confuse the typical presentation of DMD and delay the earlier diagnosis. In our case study, no such two-fold diagnosis existed; however, this report demonstrates that numerous factors can affect the diagnostic radiographic image of DMD.

According to a systematic review and Delphi consensus by Aartsma-Rus et al., the key factors contributing to diagnostic delay are a lack of awareness among healthcare professionals about DMD signs and symptoms and long waiting times to access specialists. The incomplete genetic analysis of the patient may show many such red flags, as in our patient, but he was diagnosed 4 years after the onset of symptoms.

The pattern of DMD gene mutations is well-defined: approximately 68% are deletions, 11% are duplications, and 20% are small mutations. Genetic testing is performed in stages: deletion/duplication testing using MLPA or CGH and sequencing if negative. A muscle biopsy is usually performed when DNA tests are negative or inconclusive.[12]

Comorbidities in DMD that relate to neurodevelopmental impairments have received growing interest. This is evidenced by Vaillend et al.'s observation that brain dystrophin deficiency is also a contributor to intellectual (~22%), autism (~6%), and attention-deficit disorders (~18%), and by the new genetic therapies that can restore brain dystrophin, which are promising in mice models.

DMD is progressive and has a poor prognosis, regardless of the age of onset. Nonetheless, when a timely diagnosis is achieved, it is still possible to provide supportive care, eliminate complications, and offer counseling to the family. This case highlights the need to suspect DMD in children and adolescents with progressive muscle weakness and increased CPK levels.

Conclusion

Even in children and teenagers who develop Duchenne Muscular Dystrophy later than expected, clinicians should be attentive to this disorder. A positive test for pseudohypertrophy, proximal weakness, hypoflexia, and significantly elevated creatine kinase levels in a male child must prompt immediate genetic testing, irrespective of the patient's age. The progressive nature of the disease and prognosis are not affected by delayed presentations. Early diagnosis and implementation of a multidisciplinary approach to treatment, involving physical training and continuous cardiorespiratory monitoring, are key to achieving successful outcomes in patient care.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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