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Rehabilitation Strategies in a Paediatric Case of Duchenne Muscular Dystrophy Presenting with Toe Walking and Seizures: a case report

Upasna Parashar[1], Dimple Choudhry[2*], Meetu Nagpal[2], Anamika Singh[1]

[1]MPT Scholar, College of Physiotherapy, Pt BDS PGIMS Rohtak, Haryana,

[2]Associate Professor, College of Physiotherapy, Pt BDS PGIMS Rohtak, Haryana.

***Corresponding Author**

Dimple Choudhry

Associate Professor, College of Physiotherapy, Pt BDS PGIMS Rohtak, Haryana.

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Abstract: -

Background:

Duchenne Muscular Dystrophy (DMD) is a progressive X-linked recessive neuromuscular disorder characterized by the absence of functional dystrophin protein, leading to degeneration of skeletal and cardiac muscle fibers. Early clinical signs such as toe walking, delayed motor milestones, and frequent falls are often subtle and may be mistaken for benign developmental variations, contributing to delayed diagnosis.

Case Presentation:

We report the case of a 9-year-old boy referred to the physiotherapy department with persistent gait abnormalities and a history of seizures starting at 1.5 years of age. Early motor development was globally delayed, with sustained toe walking noted throughout early childhood. Muscle biopsy revealed a significant reduction in delta-sarcoglycan expression, raising suspicion for dystrophinopathy. Genetic testing confirmed the diagnosis of DMD. Further evaluation revealed subtle white matter changes on brain MRI, minor cardiac involvement on echocardiography, and early respiratory compromise.

Discussion:

This case highlights an atypical presentation of DMD, complicated by early-onset seizures, and underscores the diagnostic challenges in such presentations. The physiotherapist played a pivotal role in early recognition of red flags during gait assessment, which facilitated timely referral for multidisciplinary evaluation.

Conclusion:

Early identification and intervention are critical in DMD to optimize functional outcomes and initiate supportive management. This case reinforces the importance of a multidisciplinary approach—including physiotherapy, neurology, cardiology, pulmonology, and genetics—in managing complex neuromuscular disorders and improving quality of life.

Keywords: *seizures, milestones, dystropin, delta- sarcoglycan, physiotherapy.*

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Introduction

Duchenne Muscular Dystrophy (DMD) is among the most common and severe forms of muscular dystrophy in childhood, with an estimated incidence of 1 in 3,500 to 5,000 live male births worldwide[1]. It is caused by mutations in the *DMD* gene located on the X chromosome, which encodes the dystrophin protein—essential for maintaining the structural integrity of muscle cell membranes[2]. The absence or severe deficiency of functional dystrophin leads to progressive degeneration of skeletal, cardiac, and respiratory muscle fibers, resulting in a relentless decline in motor and vital functions[1].

Clinical signs of DMD typically emerge between 2 and 5 years of age and may include delayed motor milestones, proximal muscle weakness, frequent falls, Gower's sign, and gait abnormalities such as toe walking[3]. These early symptoms can often be subtle, nonspecific, and misattributed to common developmental variations, leading to a delayed diagnosis and late initiation of multidisciplinary care[4]. Although seizures are not a classical feature of DMD, they have been reported in a small subset of patients and may reflect early central nervous system involvement[5].

Without effective disease-modifying therapy, children with DMD become wheelchair-dependent by the early teens, and most patients succumb to cardiopulmonary complications by their late twenties[6]. In contrast, Becker Muscular Dystrophy (BMD)—a milder allelic variant—presents with later onset, slower progression, and greater life expectancy[7]. In low- and middle-income countries, limited access to early diagnosis and intervention is often associated with higher morbidity and mortality, with many individuals not surviving beyond their early twenties[8].

This case report describes a 9-year-old boy presenting with early-onset toe walking and seizures, later diagnosed with DMD. The report highlights the diagnostic challenges posed by atypical features and emphasizes the critical role of early recognition and a multidisciplinary approach—including physiotherapy, neurology, cardiology, and genetics—in the comprehensive management of neuromuscular disorders.

Case Presentation

A 9-year-old boy was referred to the physiotherapy department with a confirmed diagnosis of Duchenne Muscular Dystrophy (DMD). The child was adopted from relatives and had a birth weight of 2.25 kg. He was born at full term and cried immediately after birth. Family history was unremarkable, with an elder brother showing no signs of neuromuscular disorders.

Early Developmental History:

The child began toe walking around 1.5 years of age and experienced multiple seizure episodes during early childhood, the most recent occurring in July 2023. Early developmental milestones were significantly delayed, with late attainment of sitting and crawling. He began walking independently at 2.5 years of age, though with an unsteady gait. Seizure episodes were managed with medical treatment.

Clinical Progression:

Over time, the child developed progressive muscle weakness, increasing difficulty in rising from the floor, frequent falls, and noticeable fatigue with physical activity. At 8 years of age, a diagnosis of DMD was confirmed following clinical evaluation, elevated creatine kinase levels, and muscle biopsy, which revealed reduced delta-sarcoglycan (SGCD) expression.

Current Physical Status:

Upon physiotherapy assessment, the child presented with a thin body habitus, marked proximal muscle weakness, lumbar lordosis, calf pseudohypertrophy, and a positive Gower's sign. He demonstrated significant difficulty standing and was unable to ambulate independently without rest. Compensatory trunk movements were observed during attempts to maintain standing posture. A right-sided equinovarus deformity was noted, suggestive of muscle contractures resulting from chronic imbalance, a common complication in later stages of DMD.

Motor and Functional Findings

The patient demonstrated classic motor deficits associated with Duchenne Muscular Dystrophy. Proximal muscle weakness was prominent, particularly in the lower limbs and spinal extensors, consistent with the typical progression pattern where proximal muscles deteriorate earlier than distal ones. A positive Gower's sign and difficulty rising to a standing position were observed—hallmark features of DMD that typically emerge between 2 and 5 years of age. The child exhibited tight Achilles tendons and persistent toe-walking, indicative of early contracture development due to chronic muscle imbalance. Additionally, signs of respiratory difficulty were noted, likely resulting from diaphragmatic and intercostal muscle involvement, a complication commonly seen in later stages of the disease. These findings collectively reflect the progressive neuromuscular decline and functional limitations characteristic of DMD.

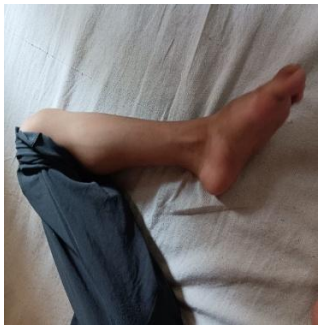


FIGURE 1 Calf pseudo hypertrophy

FIGURE 2 Part of Gower's sign

FIGURE 3 Toe standing

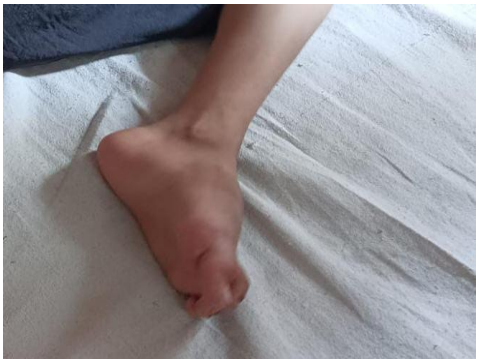


FIGURE 4 Equinovarus deformity

Neurological and Cognitive Findings

The patient currently experiences no active seizures, and cranial nerve examination revealed normal findings, supporting a neuromuscular etiology rather than a central neurological cause. There were no sensory deficits, which aligns with the typical presentation of DMD, a disorder that primarily affects motor function. Mild attentional difficulties were noted, although overall cognitive functioning appeared to be within average range. Such cognitive or behavioural challenges are occasionally observed in DMD and may be attributed to altered dystrophin expression in brain regions involved in attention and executive functioning.

Family and Social History

The child was adopted from extended family members. His biological elder brother is reportedly healthy with no known neuromuscular conditions. No similar disorders have been documented within the adoptive family. The lack of family history highlights the importance of genetic testing in confirming suspected neuromuscular conditions.

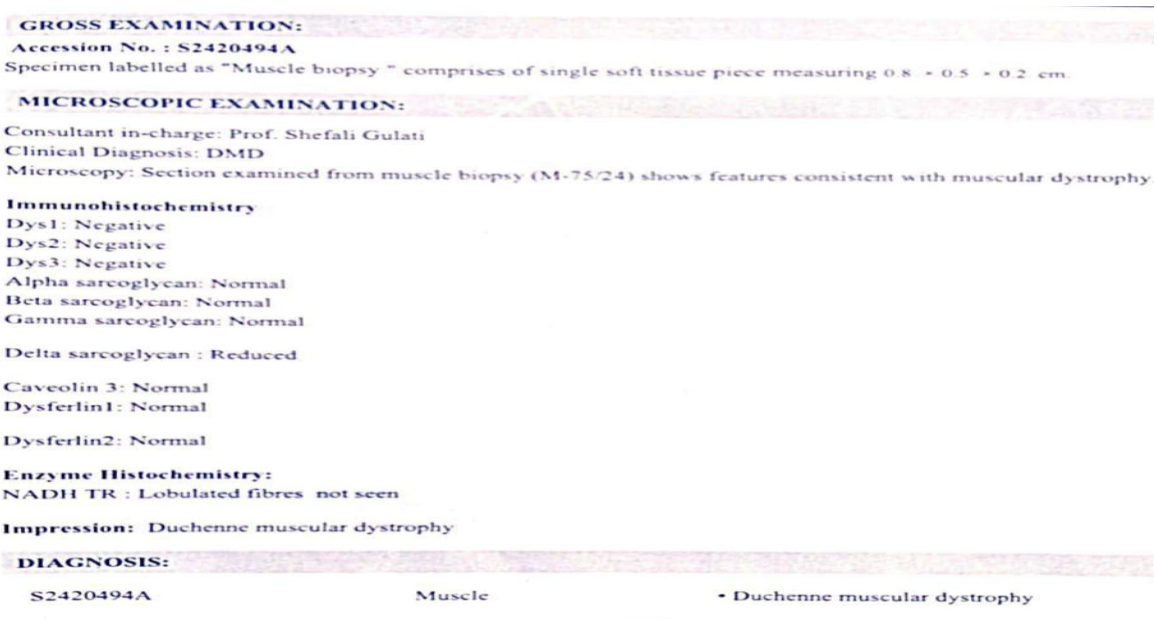
Investigations

- **Histopathology (09/05/2024):**

  A muscle biopsy of the quadriceps demonstrated a marked reduction in delta-sarcoglycan expression. Delta-sarcoglycan is an integral component of the dystrophin-glycoprotein complex, and its deficiency is commonly observed in dystrophinopathies such as DMD. These histopathological findings supported the clinical suspicion of a muscular dystrophy and provided direct tissue-level evidence of a dystrophin-associated disorder.

- **Genetic Testing (18/01/2024):**

  Molecular DNA analysis confirmed the diagnosis of Duchenne Muscular Dystrophy, identifying a pathogenic deletion consistent with the DMD phenotype. Genetic testing not only serves as the diagnostic gold standard for DMD but also enables essential genetic counseling for the family and assists in planning for future therapeutic interventions



GROSS EXAMINATION:

Accession No. : S2420494A

Specimen labelled as "Muscle biopsy " comprises of single soft tissue piece measuring 0.8 × 0.5 × 0.2 cm.

MICROSCOPIC EXAMINATION:

Consultant in-charge: Prof. Shefali Gulati
Clinical Diagnosis: DMD
Microscopy: Section examined from muscle biopsy (M-75/24) shows features consistent with muscular dystrophy.

Immunohistochemistry
Dys1: Negative
Dys2: Negative
Dys3: Negative
Alpha sarcoglycan: Normal
Beta sarcoglycan: Normal
Gamma sarcoglycan: Normal

Delta sarcoglycan : Reduced

Caveolin 3: Normal
Dysferlin1: Normal

Dysferlin2: Normal

Enzyme Histochemistry:
NADH TR : Lobulated fibres not seen

Impression: Duchenne muscular dystrophy

DIAGNOSIS:

S2420494A Muscle • Duchenne muscular dystrophy

FIGURE 5 Histopathology Report shows reduction of Delta sarcoglycan protein.

Neuroimaging and Cardiac Evaluation

- **MRI Brain (09/01/2023):**

  As part of the evaluation for early-onset seizures, magnetic resonance imaging (MRI) of the brain was performed. The scan revealed subtle bilateral posterior periventricular white matter FLAIR hyperintensities. While nonspecific, these findings may represent delayed myelination or mild leukodystrophy-like changes. Such alterations have been reported in some cases of Duchenne Muscular Dystrophy, particularly in patients with early neurological symptoms. These changes are thought to be associated with the absence or dysfunction of brain-specific dystrophin isoforms, suggesting a potential degree of central nervous system (CNS) involvement in DMD.

- **Echocardiography (05/11/2023):**

  Cardiac evaluation demonstrated trivial tricuspid regurgitation (TR) without significant valvular abnormality or chamber enlargement. Left ventricular function was preserved at the time of examination. Although no overt cardiomyopathy was present, the progressive nature of cardiac involvement in DMD warrants regular cardiac monitoring as part of long-term management.

Differential Diagnoses

When evaluating a child with progressive muscle weakness and delayed motor milestones, it is crucial to consider and rule out other neuromuscular disorders that may mimic Duchenne Muscular Dystrophy (DMD). Several conditions fall within the differential diagnosis spectrum:

- **Limb-Girdle Muscular Dystrophy (LGMD):**

  LGMD can present with clinical features similar to DMD, including proximal muscle weakness and difficulty with ambulation. However, it typically manifests later—often during the second decade of life. Cognitive function is usually preserved, calf pseudohypertrophy is less prominent, and serum transaminase levels are not markedly elevated compared to DMD. Genetic testing is essential for distinguishing between LGMD subtypes and dystrophinopathies.

- **Emery-Dreifuss Muscular Dystrophy (EDMD):**

  EDMD is another important differential diagnosis, characterized by early-onset joint contractures (particularly of the elbows, ankles, and neck), humeroperoneal muscle weakness and wasting, and cardiac abnormalities, including arrhythmias and dilated cardiomyopathy. Unlike DMD, EDMD has a more localized pattern of muscle involvement and a slower disease progression.

- **Spinal Muscular Atrophy (SMA):**

  SMA is a genetic motor neuron disorder marked by degeneration of the anterior horn cells, leading to lower motor neuron signs such as hypotonia, muscle atrophy, areflexia, and tongue fasciculations. SMA does not involve elevated creatine kinase levels and lacks the pseudohypertrophy and Gower's sign commonly seen in DMD. Additionally, cognitive development is typically normal.

Differentiating between these conditions requires a combination of clinical assessment, family history, enzyme studies (such as CK levels), neuroimaging, muscle biopsy, and confirmatory genetic testing.

Physiotherapy Intervention

In children diagnosed with Duchenne Muscular Dystrophy (DMD), physiotherapy plays a pivotal role in preserving functional independence, delaying disease progression, and enhancing overall quality of life. Early and proactive rehabilitation is critical, as muscle degeneration begins well before overt clinical signs emerge. The physiotherapy goals in this case focus on three key priorities:

1. **Maintaining Ambulation:**

   Preserving the ability to walk independently for as long as possible is associated with improved cardiovascular and respiratory outcomes, greater independence in daily activities, and a delay in the need for assistive mobility devices.

2. **Preventing Contractures:**

   Muscle imbalances and reduced activity levels increase the risk of joint contractures, particularly in the lower limbs. Preventing or minimizing soft tissue shortening is crucial to maintaining range of motion, postural alignment, and functional mobility.

3. **Enhancing Quality of Life:**

   Physiotherapy not only targets physical function but also supports emotional well-being, social participation, and engagement in age-appropriate activities, all of which are essential to the child's holistic development.

Rehabilitation Plan

The physiotherapy plan is individualized, goal-oriented, and designed to address the child's current capabilities while anticipating future needs. Key components of the intervention include:

- **Passive Range of Motion (PROM) – Lower Limbs:**

  Regular PROM exercises target joints most susceptible to contractures—ankles, knees, and hips. These exercises help preserve joint mobility, reduce stiffness, and support proper positioning, especially as the disease advances and active movement becomes limited.

- **Active-Assisted Range of Motion (AAROM) – Upper Limbs:**

  AAROM exercises for the arms and shoulders help sustain upper limb function, which is vital for self-care activities such as feeding, writing, and dressing. This approach encourages neuromuscular activation while compensating for progressive weakness.

- **Achilles Tendon Stretching:**

  Toe walking, often one of the earliest signs of DMD, is commonly linked to tightness in the gastrocnemius-soleus complex. Daily Achilles tendon stretching reduces the risk of fixed equinus deformity, supports safer ambulation, and helps maintain ankle flexibility.

- **Core Strengthening and Balance Training:**

  Weakness in proximal muscles impacts posture, gait, and transitional movements. Core exercises are introduced to improve trunk control and postural stability. Balance training is also essential for minimizing fall risk and enhancing the child's confidence during movement.

- **Postural Management – Prone-on-Elbow Positioning:**

  This positioning strategy encourages upper trunk activation and spinal extension, counteracting the development of lumbar lordosis due to weak back musculature. It also promotes better sitting posture and contributes to respiratory efficiency.

- **Respiratory Support – Deep Breathing Exercises:**

  Since respiratory muscle involvement is a known progression in DMD, deep breathing exercises are incorporated early to maintain chest wall mobility, optimize lung expansion, and delay the need for ventilatory assistance. These exercises also promote breathing awareness and coordination.

- **Family Education and Home Exercise Program:**

  Active caregiver involvement is a cornerstone of effective rehabilitation in DMD. Parents are educated about disease progression, therapy goals, and home-based interventions. A structured home exercise program ensures continuity, encourages adherence, and empowers the family to participate actively in the child's care.

- **Multidisciplinary Coordination:**

  Optimal care for a child with DMD requires a team-based approach. Collaboration among physiotherapists, pediatric neurologists, occupational therapists, respiratory therapists, orthopedic specialists, and social workers ensures comprehensive, individualized, and holistic management. Regular team communication allows for coordinated interventions and timely adjustments based on the child's evolving needs.

Discussion

This case report highlights a young male patient presenting with clinical features suggestive of a dystrophinopathy, confirmed by genetic and histopathological findings. Mutations in the DMD gene, which encodes the dystrophin protein, are well-established causes of Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD). Dystrophin plays a critical role as part of the dystrophin-associated glycoprotein complex (DGC) located in the sarcolemma of skeletal muscle fibers[9]. The DGC functions to stabilize the muscle cell membrane by linking the intracellular cytoskeleton to the extracellular matrix, thereby maintaining cellular integrity during muscle contraction[10].

In this patient, genetic analysis revealed missense mutations rather than the more common frameshift or nonsense mutations typically associated with classic DMD. Missense mutations result in the substitution of a single amino acid, potentially allowing for the production of a partially functional dystrophin protein. This partial expression was supported by the presence of dystrophin detected on muscle biopsy, correlating with a milder clinical phenotype in comparison to typical DMD cases where truncated or absent dystrophin leads to a more severe disease course. This distinction is consistent with the observed clinical presentation, including later onset of functional decline and relatively preserved muscle function[8,11].

Early clinical signs such as persistent toe walking and seizures complicated the diagnostic process in this patient. Although seizures are not a hallmark of DMD, emerging evidence suggests that central nervous system (CNS) involvement can occur, possibly related to the absence or dysfunction of dystrophin isoforms expressed in the brain. This neurological involvement may contribute to the variability in clinical manifestations and can delay diagnosis when overshadowed by early neurological symptoms[12].

The case underscores the critical role of early and sustained physiotherapy intervention in managing DMD. Physiotherapy aims to maintain mobility, prevent contractures, and delay the progression of functional decline, ultimately improving the child's quality of life. The importance of a multidisciplinary approach, including genetic counseling and regular cardiac and respiratory monitoring, is also emphasized.

Lastly, the presence of a healthy biological sibling supports the X-linked inheritance pattern of DMD, highlighting the genetic variability and the need for family screening and counselling.

Conclusion

This case highlights the complexity and variability in the presentation of Duchenne Muscular Dystrophy, especially when early signs such as toe walking and seizures are present. The identification of missense mutations leading to partially functional dystrophin correlates with a milder phenotype and emphasizes the spectrum of dystrophinopathies. Early diagnosis and timely physiotherapy intervention are essential to preserving mobility and improving quality of life. A multidisciplinary approach, including genetic counseling and regular cardiac and respiratory monitoring, is vital for comprehensive care. Awareness of atypical presentations can facilitate earlier recognition and management, ultimately enhancing patient outcomes

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