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Acute Testicular Torsion in an Adolescent with Duchenne Muscular Dystrophy: A Rare Case Report

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Abstract

Background: Testicular torsion is a urological emergency requiring urgent diagnosis and surgical intervention to preserve testicular viability. Although commonly encountered in adolescents, its occurrence in patients with neuromuscular disorders such as Duchenne Muscular Dystrophy (DMD) is exceedingly rare. DMD, an X-linked recessive progressive neuromuscular disease, presents unique diagnostic and perioperative challenges including limited mobility, altered pain perception, cardiomyopathy, and respiratory muscle weakness — all of which may delay diagnosis and complicate surgical management.

Case Presentation: We report a rare case of acute right testicular torsion in a 15-year-old adolescent with DMD who presented with sudden-onset right scrotal pain. Clinical assessment was challenging due to limited mobility and atypical pain expression. Colour Doppler ultrasonography demonstrated complete absence of intratesticular blood flow in the right testis. Emergency scrotal exploration revealed a 720° torsion of the right spermatic cord with a non-viable testis, necessitating right orchiectomy. Prophylactic contralateral orchidopexy was performed simultaneously. Anaesthetic management required meticulous multidisciplinary planning due to underlying cardiomyopathy and respiratory muscle weakness, employing total intravenous anaesthesia (TIVA) with avoidance of succinylcholine and volatile agents.

Conclusion: To the best of our knowledge, this represents the first reported case of acute testicular torsion in an adolescent with Duchenne Muscular Dystrophy. This case underscores the critical importance of maintaining a high index of clinical suspicion for acute scrotal emergencies in patients with neuromuscular disorders. Early imaging, prompt surgical exploration, and comprehensive multidisciplinary perioperative management are indispensable for optimising outcomes in this uniquely vulnerable patient population.

Keywords: Testicular torsion; Duchenne Muscular Dystrophy; Acute scrotum; Orchiectomy; Orchidopexy; Paediatric urology; neuromuscular disease; Perioperative anaesthesia; TIVA; Spermatic cord torsion

Introduction

Testicular torsion is a time-sensitive surgical emergency caused by twisting of the spermatic cord, resulting in compromised blood flow to the testis. Delay in diagnosis and intervention can lead to irreversible ischaemic damage and testicular loss, with salvage rates declining precipitously beyond six hours of symptom onset [1,2]. In males under 18 years of age, the annual incidence of testicular torsion is reported at approximately 3.8 per 100,000, with a well-recognised bimodal peak in the perinatal period and during adolescence [7]. While torsion most commonly affects otherwise healthy adolescents with a bell-clapper deformity of the tunica vaginalis, its presentation may be profoundly atypical in patients with physical disabilities or neuromuscular disorders [3].

Duchenne Muscular Dystrophy (DMD) is the most common and severe X-linked recessive muscular dystrophy, affecting approximately 1 in 3,500 male live births worldwide [4]. It is caused by pathogenic variants in the dystrophin gene, resulting in absent or severely reduced dystrophin protein, progressive skeletal and cardiac muscle degeneration, cardiomyopathy, and respiratory insufficiency [4,5]. With advances in multidisciplinary care including non-invasive ventilation and cardiac surveillance, median survival has improved significantly; however, patients continue to face substantial perioperative risk due to cardiomyopathy, respiratory muscle weakness, and heightened sensitivity to certain anaesthetic agents [6,7]. The diagnostic challenge posed by DMD in the context of acute surgical emergencies is considerable. Limited mobility, communication difficulties, altered pain perception, and restricted physical examination preclude the reliable use of classical clinical signs such as the cremasteric reflex and Prehn's sign. These factors collectively risk delayed presentation and missed diagnosis, potentiating testicular non-viability [3]. To date, testicular torsion has not been reported in adolescent patients with DMD, representing a unique lacuna in the paediatric urology and neuromuscular disease literature.

Case Presentation

A 15-year-old adolescent male with a confirmed diagnosis of Duchenne Muscular Dystrophy presented to the emergency department with a two-hour history of sudden-onset right scrotal pain. He was wheelchair-dependent and had known dilated cardiomyopathy managed medically. There was no history of trauma, fever, urinary symptoms, or prior scrotal pathology. His regular medications included ACE inhibitors and prophylactic low-dose steroids. Assessment was significantly limited by restricted mobility, generalised limb weakness, and considerable difficulty in localising the pain precisely. On physical examination, the right hemiscrotum was tender with mild swelling and erythema. Classical signs of torsion — including the cremasteric reflex, high-riding testis, and Prehn's sign — were difficult to reliably assess due to patient discomfort and neuromuscular limitations. Given the acute presentation and diagnostic uncertainty, urgent colour Doppler ultrasonography was performed as the primary investigative modality. Ultrasound demonstrated complete absence of intratesticular blood flow in the right testis, with preserved normal vascularity in the contralateral left testis, consistent with acute right testicular torsion [8] (Figure 1). Prompt counselling was provided to the patient's legal guardian regarding the surgical emergency, and the patient was prepared for immediate operative intervention. The patient was taken for emergency scrotal exploration following a thorough and expedited multidisciplinary anaesthetic assessment. Intraoperatively, a 720° torsion of the right spermatic cord was identified. The right testis appeared dark, congested, and grossly non-viable, with no improvement in colour or turgor following detorsion and application of warm saline-soaked gauze over a ten-minute observation period. On this basis, a right orchiectomy was performed. Concurrently, prophylactic contralateral orchidopexy was carried out using a three-point fixation technique to prevent future contralateral torsion (Figure 2). The operative time was appropriate and the patient was transferred to a high-dependency unit postoperatively for close cardiorespiratory monitoring.

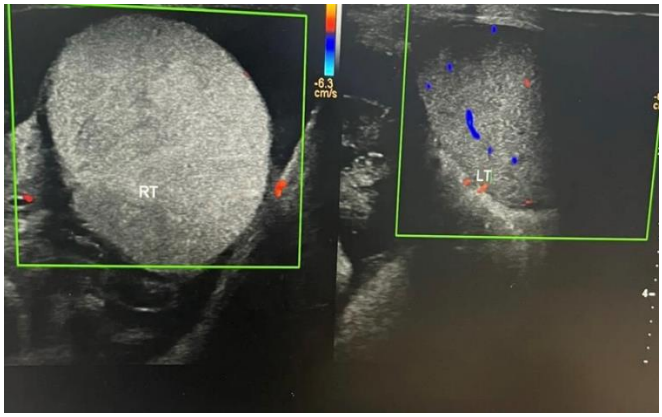

Figure 1: *Colour Doppler ultrasonography of the scrotum demonstrating complete absence of intratesticular blood flow in the right testis (left panel), consistent with acute testicular torsion. Normal vascularity is preserved in the contralateral left testis (right panel). Absence of Doppler signal in the affected testis is a highly sensitive and specific indicator of testicular torsion in the paediatric population [8].*

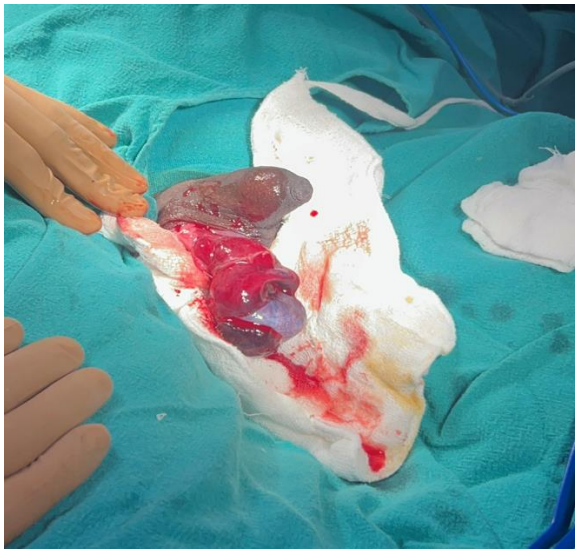

Figure 2: *Intraoperative photograph demonstrating the non-viable right testis following detorsion. The testis remains dark, congested, and non-responsive to warm saline application, confirming irreversible ischaemia and necessitating right orchiectomy. Prophylactic three-point fixation orchidopexy of the contralateral testis was performed in the same operative setting.*

Anaesthetic Considerations

Anaesthetic management in patients with Duchenne Muscular Dystrophy demands meticulous pre-operative evaluation and a tailored, non-triggering approach. In this case, several DMD-specific risks required careful consideration and multidisciplinary coordination involving the urology, anaesthesia, cardiology, and respiratory medicine teams. The primary anaesthetic concerns in DMD include the risk of life-threatening rhabdomyolysis and hyperkalaemia with succinylcholine and volatile halogenated agents [9,10]. Succinylcholine, a depolarising neuromuscular blocking agent, is absolutely contraindicated in DMD due to the risk of massive potassium release from fragile, dystrophin-deficient myocytes, which can precipitate fatal cardiac arrhythmias [11]. Similarly, inhalational volatile agents — including sevoflurane, isoflurane, and desflurane — have been associated with acute rhabdomyolysis and hyperthermia in DMD patients, in a syndrome clinically resembling but pathophysiologically distinct from malignant hyperthermia [12,13]. Accordingly, a total intravenous anaesthesia (TIVA) technique was employed in this case, utilising propofol and fentanyl for induction and maintenance, with rocuronium (a non-depolarising neuromuscular blocking agent) used judiciously for intubation under train-of-four (TOF) monitoring, and sugammadex available for reversal. The anaesthetic machine was prepared as a 'clean machine' free of volatile agent reservoirs. Continuous invasive arterial blood pressure monitoring, cardiac rhythm monitoring, and capnography were maintained throughout the perioperative period. Pre-operative echocardiography confirmed dilated cardiomyopathy with mildly reduced ejection fraction. Pre-operative pulmonary function assessment was limited by patient cooperation but clinical history was consistent with restrictive respiratory disease. Postoperatively, the patient was monitored in a high-dependency unit with non-invasive ventilatory support, analgesia titration, and early physiotherapy involvement. Serum creatine kinase (CK) and myoglobin were monitored post-operatively to screen for rhabdomyolysis; both remained within acceptable limits. The patient was discharged on day three without perioperative complications.

Discussion

This case, to the best of our knowledge, represents the first published report of acute testicular torsion in an adolescent with Duchenne Muscular Dystrophy, expanding the spectrum of acute surgical emergencies that may be encountered in this vulnerable population. It raises important clinical lessons at the intersection of paediatric urology, neuromuscular disease, and perioperative medicine. Testicular torsion is a time-critical emergency in which the duration of ischaemia is the principal determinant of testicular salvage. Published salvage rates of 90–97% are achievable within six hours of symptom onset, falling to 10% or less beyond 24 hours [1,2]. Recent large-scale registry data from Germany confirm that delayed presentation remains a major contributor to orchiectomy rates, underscoring the ongoing need for heightened clinical awareness [10]. In patients with DMD, this time-sensitivity is further compounded by the diagnostic challenges inherent to the condition. The clinical diagnosis of testicular torsion relies upon a constellation of findings including sudden-onset scrotal pain, absent cremasteric reflex, high-riding testis, and horizontal testicular lie [3,7]. In the present case, the patient's wheelchair dependence, generalised hypotonia, and difficulty localising pain rendered these classical signs unreliable or unobtainable. This diagnostic uncertainty exemplifies the broader challenge of evaluating acute surgical emergencies in patients with neuromuscular disorders, where altered pain perception, communication barriers, and limited mobility may obscure or delay presentation [3]. Colour Doppler ultrasonography (CDUS) proved indispensable in this case, enabling definitive non-invasive diagnosis by demonstrating the complete absence of intratesticular blood flow. CDUS has a reported sensitivity of 69–97% and specificity of 77–100% for testicular torsion in the paediatric population and represents the imaging investigation of choice in equivocal cases [8]. Importantly, its role should be complementary to, rather than a substitute for, clinical judgement; when clinical suspicion is high, immediate surgical exploration is warranted regardless of Doppler findings, as false negatives may occur in partial or intermittent torsion [7].

The degree of torsion identified intraoperatively — 720° — is among the more severe reported in the literature and is consistent with the absence of blood flow on Doppler and the non-viability of the testis at exploration. Prophylactic contralateral orchidopexy was performed in the same operative setting, in keeping with current evidence and guidelines, given the recognised risk of metachronous contralateral torsion associated with the underlying bell-clapper deformity, which is typically bilateral [1,5]. The perioperative management of DMD patients undergoing emergency surgery presents a distinct and high-stakes challenge. The combined burden of cardiomyopathy, respiratory compromise, and anaesthetic drug sensitivity necessitates rapid yet thorough preoperative optimisation and close post-operative monitoring. Radeka in a systematic review of anaesthesia in rare neuromuscular diseases affirmed that total intravenous anaesthesia (TIVA) with avoidance of both succinylcholine and volatile agents is the safest and most evidence-consistent approach in DMD [14,15]. The risk of succinylcholine-induced hyperkalaemia and fatal arrhythmia in DMD has been well-documented, with case reports of cardiac arrest and death following its administration [11,12]. Our case further supports the imperative for pre-operative DMD-specific anaesthetic protocols in any institution that may encounter this patient group, whether

on an elective or emergency basis. The absence of prior reports of testicular torsion in DMD patients likely reflects the rarity of the combination rather than a true biological immunity. DMD patients are increasingly surviving into adulthood due to improved multidisciplinary care, and urologists and paediatricians should be aware that acute urological emergencies may occur in this population. The principles of high clinical suspicion, early ultrasonographic assessment, and emergency surgical exploration remain unchanged, but must be adapted to the unique physiological and anaesthetic constraints of DMD.

Key Learning Points / Clinical Messages

- Testicular torsion, although rare in patients with neuromuscular disorders, is a potentially life-altering emergency that should be actively considered in any adolescent with DMD presenting with acute groin or scrotal pain, regardless of the atypicality of the clinical presentation.
- The classical clinical signs of testicular torsion — cremasteric reflex, high-riding testis, and Prehn's sign — may be unreliable or impossible to elicit in patients with Duchenne Muscular Dystrophy due to hypotonia, restricted mobility, and altered pain perception.
- Colour Doppler ultrasonography is the diagnostic investigation of choice in equivocal cases and should be performed urgently. However, a high index of clinical suspicion should prompt immediate surgical exploration without delay for imaging where clinical features are strongly suggestive.
- Succinylcholine and volatile inhalational anaesthetic agents are absolutely contraindicated in DMD due to the risk of life-threatening rhabdomyolysis, hyperkalaemia, and cardiac arrhythmia. Total intravenous anaesthesia (TIVA) with non-depolarising neuromuscular blockade under TOF monitoring is the recommended approach.
- Emergency surgery in DMD patients mandates a multidisciplinary approach involving urology, anaesthesia, cardiology, respiratory medicine, and intensive care — even when the operation itself is brief and technically straightforward.
- Prophylactic contralateral orchidopexy should always be performed at the time of emergency scrotal exploration given the bilateral nature of the predisposing anatomical anomaly (bell-clapper deformity).
- As survival in DMD improves with modern multidisciplinary management, urologists and emergency physicians should anticipate a broader range of acute urological emergencies in this population. Institutional protocols for DMD-specific perioperative management should be established proactively (Table 1).

Table 1: *Clinical and Anaesthetic Profile.*

Parameter	Detail
Age / Sex	15-year-old male
Underlying diagnosis	Duchenne Muscular Dystrophy (DMD) — confirmed, wheelchair-dependent
Presenting complaint	Sudden-onset right scrotal pain, 2-hour history
Key co-morbidities	Dilated cardiomyopathy, respiratory muscle weakness, generalised hypotonia
Clinical examination	Right hemiscrotum tender, mildly swollen; classical signs unreliable due to NMD
Imaging	Colour Doppler USG: absent intratesticular blood flow right testis; normal left
Intraoperative finding	720° torsion, right spermatic cord; non-viable testis
Surgical procedure	Right orchiectomy + prophylactic left orchidopexy (three-point fixation)
Anaesthetic technique	Total intravenous anaesthesia (TIVA): propofol + fentanyl; non-depolarising NMB (rocuronium); sugammadex reversal
Agents avoided	Succinylcholine (absolute CI), volatile halogenated agents (rhabdomyolysis risk)
Monitoring	Continuous invasive arterial BP, ECG, capnography, TOF, SpO_2, post-op CK & myoglobin
Post-operative care	High-dependency unit; non-invasive ventilatory support; physiotherapy
Outcome	Uneventful recovery; discharged day 3; no perioperative complications
Novelty	First reported case of testicular torsion in a DMD adolescent in English literature

Conclusion

Testicular torsion, though rare in patients with neuromuscular disorders, is a genuine and potentially testis-threatening emergency that must remain within the differential diagnosis of any adolescent with Duchenne Muscular Dystrophy presenting with acute groin or scrotal pain. The atypicality of clinical presentation in DMD — driven by neuromuscular limitations, altered pain perception, and restricted physical examination — heightens the risk of diagnostic delay and mandates a lower threshold for urgent ultrasonographic evaluation and surgical exploration. Prompt multidisciplinary management, with

particular emphasis on DMD-specific anaesthetic precautions including TIVA and absolute avoidance of succinylcholine and volatile agents, is essential to achieving safe and optimal outcomes. This case adds a novel and instructive entry to the surgical emergency literature in patients with rare neuromuscular diseases.

Patient Consent

Written informed consent was obtained from the patient's legal guardian for publication of this case report and all accompanying clinical images, in accordance with the Declaration of Helsinki. The patient's identity has been protected and no personally identifiable information has been disclosed.

Conflicts of Interest

The authors declare no conflicts of interest relevant to this publication. No financial or non-financial competing interests exist.

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