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Diffuse Active Myocarditis in a Teenager with Duchenne Muscular Dystrophy - a case report

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Abstract

Introduction Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder caused by mutations in the dystrophin gene. These mutations result in the absence or reduced levels of dystrophin protein, leading to progressive skeletal and cardiac muscle deterioration. Cardiomyopathy is almost universally present in patients with DMD who survive into their third decade of life. The clinical onset of cardiac dysfunction is often asymptomatic and typically identified during routine evaluations. Acute deterioration of cardiac function in DMD patients is rarely reported in the medical literature.

Case presentation We report the case of a 13-year-old boy with DMD and preserved left ventricular (LV) function who presented with severe retrosternal chest pain and electrocardiographic (ECG) abnormalities. Laboratory testing revealed significantly elevated troponin levels, though coronary CT angiography (CTA) was normal. Cardiac magnetic resonance imaging (CMR) confirmed diffuse active myocarditis, demonstrating inflammation and fibrosis involving both the septal and lateral walls—a pattern of fibrosis atypical for Duchenne cardiomyopathy.

Conclusion Diffuse active myocardial injury with the pattern of active myocarditis is one of the causes of rapid deterioration of LV dysfunction in Duchenne cardiomyopathy and can be detected by CMR.

Keywords Duchenne, Myocarditis, Cardiomyopathy, CMR

Introduction

Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder caused by mutations in the dystrophin gene, leading to absent or reduced dystrophin protein levels. This deficiency results in progressive deterioration of skeletal and cardiac muscle function. Cardiomyopathy is nearly universal in patients with Duchenne who survive into their third decade. However, the initial clinical signs of cardiac deterioration are typically asymptomatic and often identified through routine evaluations. Acute deterioration of cardiac function in DMD patients is rarely reported in the medical literature. Here, we present the case of a 13-year-old boy with DMD who developed

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severe substernal chest pain accompanied by acute left ventricular (LV) dysfunction.

Case presentation

A thirteen-year-old boy diagnosed with DMD, diagnosed based on genetic testing six years ago, presented to the emergency department with new onset severe retrosternal and non-positional chest pain.

He had no significant family history of similar heart problems or related symptoms on either side of his family, and he hadn't experienced any previous episodes like this. He also mentioned that he hadn't had any recent flu-like symptoms. His last heart echocardiogram showed a left ventricular ejection fraction (LVEF) of around 50%. At the time, he was taking deflazacort twice daily, along with calcium supplements, coenzyme Q10, and L-carnitine, though he admitted he had missed his deflazacort doses for four days prior to this event. He was under the surveillance of a pediatric cardiologist regularly.

When examined, he appeared stable: his temperature was normal at 36.5 °C, blood pressure was 110/70 mmHg, heart rate was 90 beats per minute, breathing comfortably at 20 breaths per minute, and his oxygen level was good at 96% without supplemental oxygen. His lungs sounded clear, his heart examination didn't reveal any abnormalities, and he showed normal neck vein pressure (JVP) and good muscle strength in his legs.

The first electrocardiography (ECG) showed ST elevation in lateral leads.(Fig. 1).

The lab data showed: Cardiac Troponin I (cTnI): 50,000 µg/L, Normal Range: < 0.04 µg/L, N-terminal pro b-type natriuretic peptide (Nt-ProBNP): 3,270 pg/mL, Normal Range: < 450 pg/mL (age < 50); C-Reactive Protein (CRP): 12 mg/L, Normal Range: < 5 mg/L, Creatine Phosphokinase (CPK): 5,800 IU/L, Normal Range: 60–400 IU/L, Creatine Kinase-MB isoenzyme (CK-MB): 850 IU/L, Normal Range: < 25 IU/L, White Blood Cell Count (WBC): 8,100 cells/µL, Normal Range: 4,500–13,500 cells/µL, Haemoglobin (Hb): 12.9 g/dL, Normal Range; boys: 11.5–15.5 g/dL, Platelet Count (Plt): 260,000 cells/mm^3, Normal Range: 150,000–400,000 cells/mm^3, Aspartate Aminotransferase (AST): 580 IU/L, Normal Range: 10–40 IU/L, Alanine Aminotransferase (ALT): 280 IU/L, Normal Range: 10–35 IU/L, Hepatitis viruses and Human Immunodeficiency Virus Antibody (HIV-Abs) were negative and thyroid function test was normal.

The echocardiogram at admission showed increased LV size and severely reduced LVEF of 30%. Regarding ST elevation and significant troponin rise, coronary CT angiography (CCTA) was performed, which revealed normal coronary arteries.

Cardiac magnetic resonance (CMR) using MAGNETOM Sola 1.5 T. Ventricular ejection fractions and volumes were assessed by steady-state free precession (SSFP) sequences. Inflammation was evaluated using parametric mapping and short tau inversion recovery (STIR-T2) images. An intravenous bolus of 0.15 mmol/kg Gadoterate meglumine (gadolinium-DOTA, Dotarem,

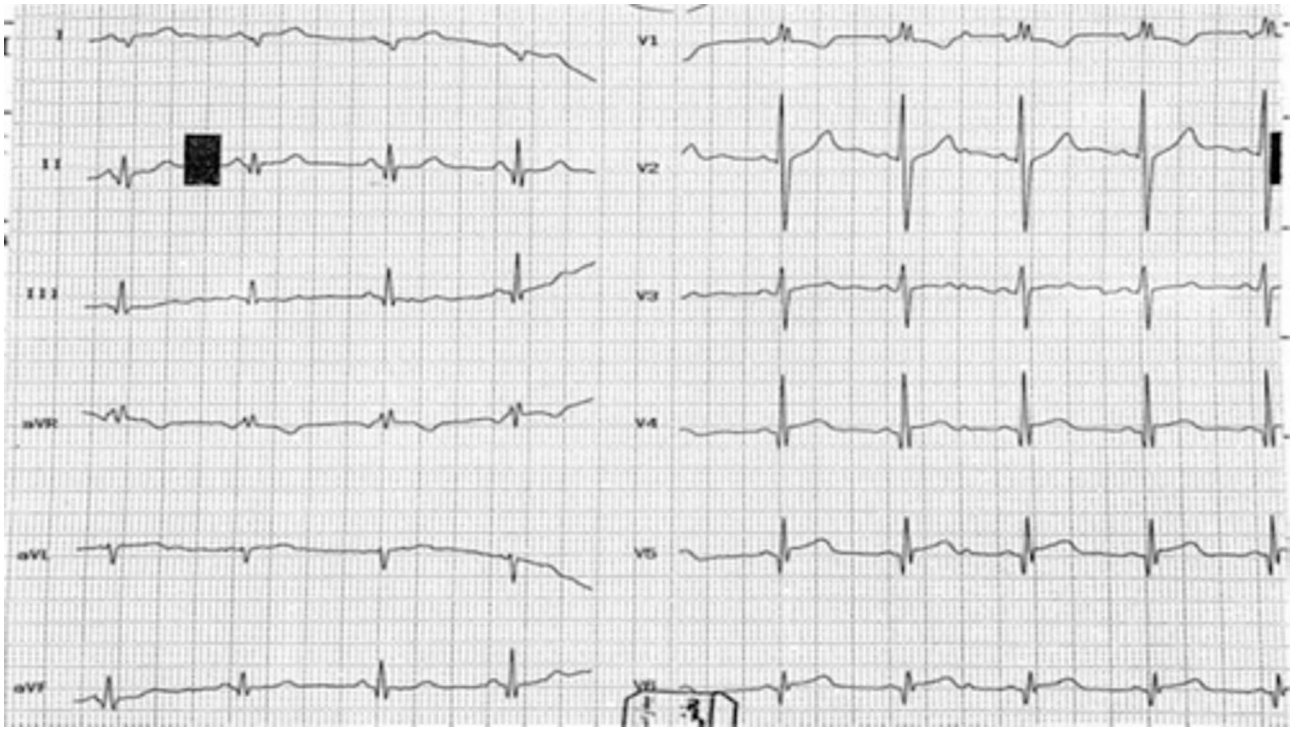

Fig. 1 12-lead ECG at admission shows lateral ST-segment elevation

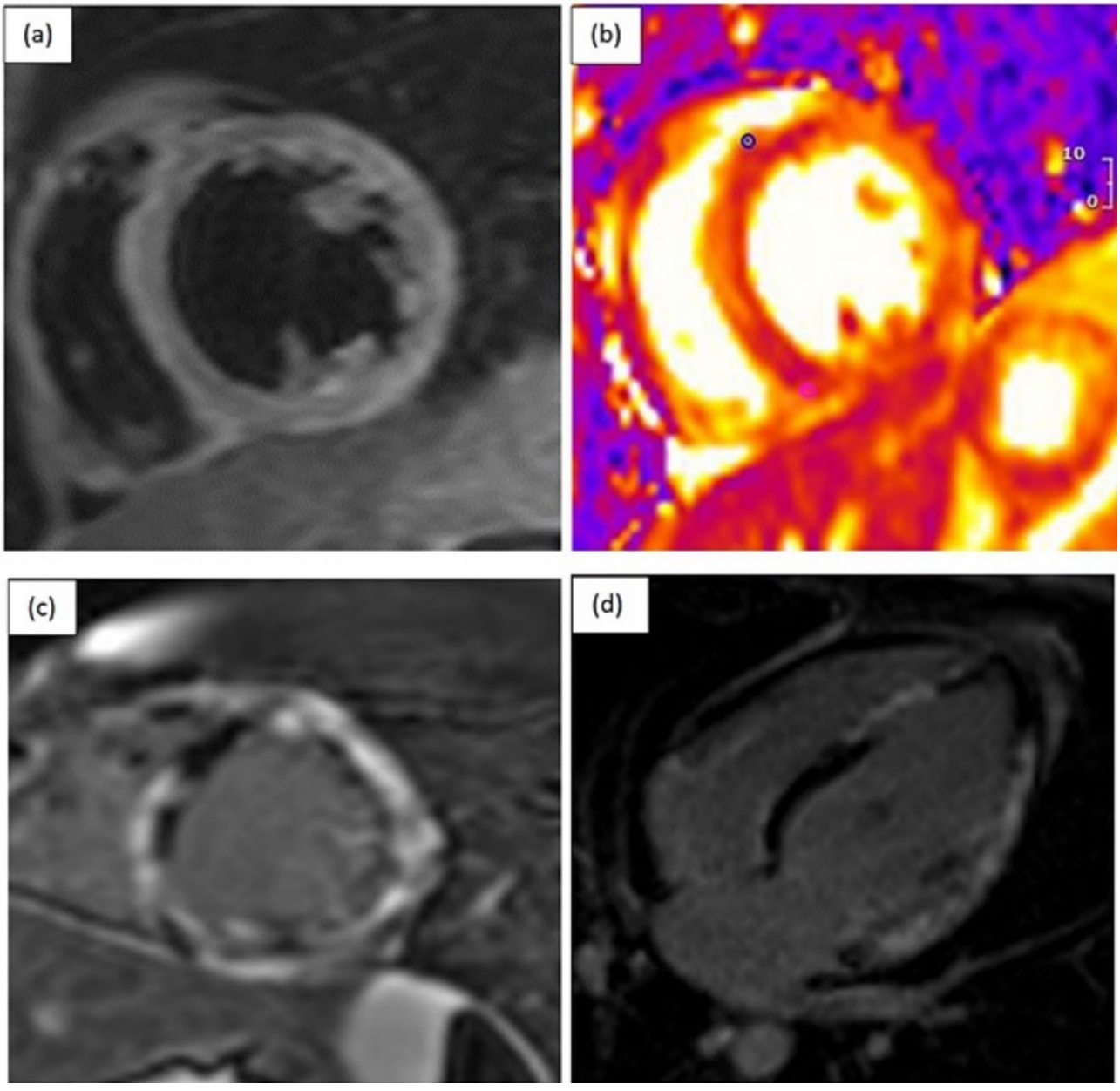

Fig. 2 **a**-**b**) STIR shows increased myocardial signal intensity at the lateral wall. T2 mapping shows diffuse myocardial inflammation is most remarkable at lateral segments **c**-**d**) LGE images show circumferential subepicardial near transmural enhancement in the short axis and 4 chamber

Guerbet S.A., Paris, France) was injected to evaluate fibrosis. The magnitude and phase-sensitive inversion recovery reconstructions of early and late gadolinium images were taken. The findings were as follows; Mildly increased LV size with LV end-diastolic volume index EDVI: 93ml/m^2, end-systolic volume index ESVI: 61ml/m^2, and LVEF 34%. Increased signal intensity in the STIR images in the basal to apical septal wall diffuse myocardial oedema, especially in the lateral wall with elevated T2 values (68 milliseconds) (Fig. 2a-b), and globally increased T1 value. In the late gadolinium images (Fig. 2c-d), there was circumferential subepicardial nearly transmural late gadolinium enhancement (LGE) in the basal to apical lateral, inferior, mid to apical anterior, and apical septal wall. All these findings were consistent with active, extensive myocarditis.

During hospitalisation, his routine medication was restarted in addition to prednisolone 15 mg daily. Commencing corticosteroids resulted in a remarkable improvement in clinical symptoms and laboratory findings. The discharge LVEF by echo was 45%.

Discussion

We report the case of a teenager with DMD who developed acute myocarditis following cessation of chronic steroid treatment. Given his acute presentation, ECG abnormalities, and significantly elevated cardiac biomarkers (particularly troponin), CCTA was performed to assess the coronary arteries, which revealed normal epicardial coronary arteries. CMR demonstrated characteristic features of diffuse acute myocarditis, including inflammation and fibrosis involving both the septal and lateral walls—a pattern not typically observed in Duchenne cardiomyopathy, which usually involves fibrosis in the lateral wall.

DMD, which affects 1 in 3,500 males, is an X-linked recessive condition brought on by mutations in the p21 band of the X chromosome that is responsible for Dystrophin, a protein located on the inner surface of the sarcolemma. The dystrophin protein is absent in the great majority of DMD patients. Dystrophin has an essential structural role in muscle by connecting the internal cytoskeleton to the extracellular matrix. Its absence causes sarcolemmal instability and muscle cell degeneration [1].

Cardiomyopathy is almost always present in DMD individuals who survive into their third decade of life. It has been proposed that mechanical stress placed on a metabolically and anatomically disordered heart causes myocardial injury in DMD [2]. Duchenne-related dilated cardiomyopathy (DCM) is characterized by thinning of the left ventricular (LV) wall and progressive LV enlargement, indicative of ongoing myocyte loss. Repeated mechanical stress initiates apoptosis and fibrotic replacement beginning at the epicardium and progressing toward the endocardium, commonly originating from areas near the posterior wall and mitral valve apparatus. Eventually, fibrosis extends toward the apex, leading to dilated cardiomyopathy [3]. Acute deterioration of cardiac function in DMD patients is rare in medical literature, with only a few cases documented that clinically mimic acute myocardial infarction [4].

Increasing evidence indicates that inflammation may trigger the development of cardiomyopathy in DMD patients. Acute myocarditis can result from various mechanisms, including viral infections that provoke an immune response and subsequent inflammation of the myocardium. Viruses such as coxsackievirus B have been linked to acute myocarditis in these individuals [5]. The absence or dysfunction of dystrophin may further increase susceptibility to viral myocarditis. Autoimmune processes, involving molecular mimicry between viral and self-antigens, may also provoke myocardial inflammation [6]. Dysregulated immune pathways and cytokine expression could also contribute to the development of myocarditis in specific individuals. Moreover, ongoing chronic inflammation is shown in mildly DMD cardiomyopathy [7].

Echocardiography remains an important initial imaging technique in assessing suspected myocarditis, although it has limited sensitivity in detecting myocarditis without evident LV dysfunction and lacks specificity in distinguishing between acute, chronic, ischemic, and non-ischemic cardiac conditions. In contrast, CMR is highly sensitive for tissue characterisation in cardiomyopathy related to dystrophinopathy, typically revealing subepicardial fibrosis in the inferolateral LV wall [1]. Moreover, CMR provides accurate and reproducible quantification of LV volumes and ejection fraction, making it especially useful for early detection of cardiac involvement and guiding decisions regarding initiation of cardioprotective therapies. In addition to tissue characterisation, CMR provides reproducible LV volume and ejection fraction quantification, making this modality highly suited to identify early cardiac involvement and assist clinicians in determining when to start cardioprotective medication [8].

In our patient, prior to the onset of chest pain, LVEF was 50%. During the acute presentation, CMR revealed diffuse elevation of myocardial T2 values and high signal intensity on T2-weighted STIR images involving the septal and lateral walls. Additionally, extensive fibrosis was noted in the same regions, consistent with diffuse active myocarditis [9]. This fibrosis pattern contrasts with the typical fibrosis seen in Duchenne cardiomyopathy, usually limited to the lateral wall [3].

The standard pharmacological management for dystrophin-associated cardiomyopathy includes ACE inhibitors, beta-blockers, and steroid therapy [10]. It is generally accepted that steroid therapy delays the onset of cardiomyopathy and may also reduce the likelihood of acute myocarditis events.

Conclusion

Diffuse myocarditis can be one of the potential causes of rapid cardiac dysfunction in patients with DMD. CMR is a valuable non-invasive imaging method that helps detect early or subtle cardiac involvement in DMD. It also helps in identifying the cause of new or acute cardiac symptoms. Additionally, CMR is safe and effective for long-term monitoring of heart function in DMD patients.

Abbreviations

DMD	Duchenne muscular dystrophy
LVEF	Left ventricular ejection fraction
ECG	Electrocardiography
JVP	Jugular venous pressure
ProBNP	Pro–B–type natriuretic peptide
CRP	C–Reactive protein
CPK	Creatine phosphokinase
CK	MB–creatine kinase–MB isoenzyme
WBC	White blood cell
Hb	Hemoglobin

Plt	Platelet
ALT	Alanine aminotransferase
AST	Aspartate aminotransferase
HIVAb	Human immunodeficiency virus antibody
CCTA	Coronary CT angiography
CMR	Cardiac magnetic resonance
SSFP	Steady–state free precession
STIR	Short tau inversion recovery
EDVI	End diastolic volume index
ESVI: End	systolic volume index
LGE	Late gadolinium enhancement
DCM	Dilated cardiomyopathy

Supplementary Information

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Supplementary Material 1

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Author contributions
GH, MM, and SM wrote the main manuscript, and GMH and HP prepared the figures. All authors reviewed the manuscript.

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Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate
Following institutional guidelines, IRB approval was not required for this case report. Written informed consent for the publication of clinical details and images was taken.

Consent for publication
Informed consent was obtained from the patient's parents for the publication of clinical details and/or clinical images.

Competing interests
The authors declare no competing interests.

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References

1. Puchalski MD, Williams RV, Askovich B, Sower CT, Hor K, Su JT, et al. Late gadolinium enhancement: precursor to cardiomyopathy in Duchenne muscular dystrophy? Int J Cardiovasc Imaging. 2009;25(1):57–63.
2. Verhaert D, Richards KE, Rafael-Fortney JA, Raman SV. Cardiac involvement in patients with muscular dystrophies. Circ Cardiovasc Imaging. 2011;4(1):67–76.
3. Adorisio R, Mencarelli E, Cantarutti N, Calvieri C, Amato L, Cicenia M, et al. Duchenne dilated cardiomyopathy: cardiac management from prevention to advanced cardiovascular therapies. J Clin Med. 2020;9(10):3186.
4. Abutaleb ARA, McNally EM, Khan SS, Anderson AS, Carr JC, Wilcox JE. Myocarditis in Duchenne muscular dystrophy after changing steroids. JAMA Cardiol. 2018;3(10):1006–10.
5. Lim BK, Peter AK, Xiong D, Narezkina A, Yung A, Dalton ND, et al. Inhibition of Coxsackievirus-Associated dystrophin cleavage prevents cardiomyopathy. J Clin Invest. 2013;123(12):5146–51.
6. Xiong D, Lee GH, Badorff C, Dorner A, Lee S, Wolf P, et al. Dystrophin deficiency markedly increases enterovirus-induced cardiomyopathy: A genetic predisposition to viral heart disease. Nat Med. 2002;8(8):872–7.
7. Mavrogeni S, Papavasiliou A, Spargias K, Constandoulakis P, Papadopoulos G, Karanasios E, et al. Myocardial inflammation in Duchenne muscular dystrophy as a precipitating factor for heart failure: a prospective study. BMC Neurol. 2010;10:33.
8. Silva MC, Magalhães TA, Meira ZMA, Rassi CHRE, Andrade AC, de Gutierrez S. Myocardial fibrosis progression in Duchenne and Becker muscular dystrophy: A randomized clinical trial. JAMA Cardiol. 2017;2(2):190–9.
9. Ferreira VM, Schulz -Menger, Jeanette, Holmvang G, Kramer CM, Carbone I, Sechtem U, et al. Cardiovascular magnetic resonance in nonischemic myocardial inflammation. J Am Coll Cardiol. 2018;72(24):3158–76.
10. Hansen MR, Hróbjartsson A, Videbæk L, Ennis ZN, Pareek M, Paulsen NH, et al. Postponement of death by Pharmacological heart failure treatment: A Meta-Analysis of randomized clinical trials. Am J Med. 2020;133(6):e280–9.

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