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CASE REPORT
Other

ST-elevation myocardial infarction in a young patient with Duchenne's muscular dystrophy: a case report

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Background Duchenne's muscular dystrophy (DMD) is an X-linked muscular disease which is caused by the absence of dystrophin. This results in the death of muscle cells and cardiomyocytes and consequent substitution by fat and fibrous tissue. The clinical translation of this is muscle weakness and cardiomyopathy. We report on the case of a young patient with dilated cardiomyopathy on a background of DMD who developed ST-elevation myocardial infarction (STEMI).

Case summary A 19-year-old male patient with DMD, known dilated cardiomyopathy, and no risk factors for ischaemic heart disease presented with central crushing chest pain. His electrocardiogram revealed anterior ST elevation. His angiogram revealed distally occluded left anterior descending and second diagonal branch with no evidence of underlying coronary artery disease. He was treated with balloon angioplasty. An echocardiogram raised the suspicion of a left ventricular thrombus, and the mechanism of STEMI was felt to be embolism from the left ventricular thrombus on a background of dilated cardiomyopathy in the context of DMD. The patient was treated with anticoagulants (warfarin). On a repeat echocardiogram a few months later, the thrombus had resolved. At 3 years of follow-up, the patient did not present any more embolic events.

Discussion To our knowledge, this is the first case of STEMI secondary to thrombotic coronary occlusion that has been described in a patient with DMD. This case highlights an unusual complication of DMD. Based on this case, we discuss the dilemmas in the management and follow-up of this complex patient population.

Keywords Duchenne's muscular dystrophy • Dilated cardiomyopathy • ST-elevation myocardial infarction • Muscular dystrophy • Left ventricular thrombus • Coronary embolism • Case report

ESC Curriculum 3.2 Acute coronary syndrome • 6.5 Cardiomyopathy

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Learning points

- The cardiovascular complications of Duchenne's muscular dystrophy (DMD) are well recognized and consist mainly of dilated cardiomyopathy. However, unusual presentations such as an ST-elevation myocardial infarction (STEMI) can occasionally be observed.
- Although atherosclerotic plaque rupture is the main mechanism for the development of STEMI in the vast majority of patients, one must bear in mind other potential causes such as thromboembolism, especially in the case of young patients or patients with known dilated cardiomyopathy.
- Cardiovascular follow-up of patients with DMD in specialized clinics is crucial in order to ensure early detection of cardiovascular involvement such as heart failure and to co-ordinate appropriate treatment.

Introduction

Muscular dystrophies are a group of genetic disorders characterized by muscle degeneration and consequent substitution by fat and fibrous tissue. Duchenne's muscular dystrophy (DMD) is an X-linked disease, and it is caused by the absence of dystrophin due to defective synthesis.[1] The absence of dystrophin produces sarcolemmal fragility and muscle cell degeneration, as well as conformational changes in stretch-activated calcium channels, resulting in pathologic leakage of calcium in the muscle cytosol and cell death.[2]

Cardiac involvement is a well-recognized complication of muscular dystrophies. It is characterized by apoptosis of the cardiomyocytes and replacement by fibrotic tissue, which is often associated with compensatory hypertrophy of the surrounding muscle and can be a substrate for cardiac arrhythmias.[3] As myocardial fibrosis increases, the left ventricle progressively dilates, which eventually leads to the clinical picture of dilated cardiomyopathy.[3] Angiotensin-converting enzyme inhibitors or angiotensin receptor blockers and steroids are used to delay or protect against the development of DMD-related cardiomyopathy, although the evidence to support the use of the latter is controversial.[4,5] Once cardiomyopathy is established, traditional heart failure treatment is indicated.[5]

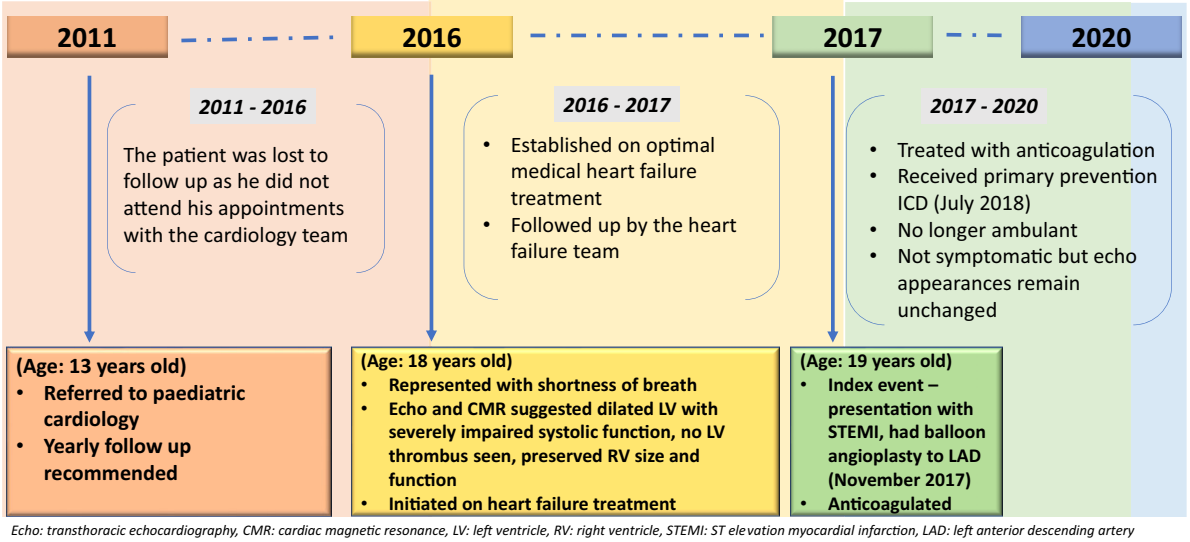
Case summary

We present the case of a 19-year-old Caucasian male patient with DMD who presented with ST-elevation myocardial infarction (STEMI) due to coronary embolism (CE).

This patient was diagnosed with DMD during the first years of his life (deletion of exons 51–54 of the DMD gene). He was on long-term steroids, and he was started on enalapril[5] at the age of 6. Unfortunately, the patient was lost to follow-up from the Paediatric Cardiology service until he presented with shortness of breath at the age of 18. At that point, the echocardiogram revealed dilated left ventricle (LV) with severely impaired systolic function. This was confirmed on cardiac magnetic resonance (CMR) imaging, which showed severely dilated LV with an LV ejection fraction of 16%. There was extensive patchy subepicardial late gadolinium enhancement of the inferior, inferolateral, anterolateral, anteroseptal, and apical anterior LV walls (*Figure 1*). The CMR findings were felt to be in keeping with DMD cardiomyopathy. Of note, there was no evidence of LV thrombus on this scan. The patient was established on heart failure treatment, and he was followed up at regular intervals. He remained in sinus rhythm with no evidence of atrial arrhythmias.

Timeline

TIMELINE - ST elevation myocardial infarction in a young patient with Duchenne's muscular dystrophy: a case report



Echo: transthoracic echocardiography, CMR: cardiac magnetic resonance, LV: left ventricle, RV: right ventricle, STEMI: ST elevation myocardial infarction, LAD: left anterior descending artery

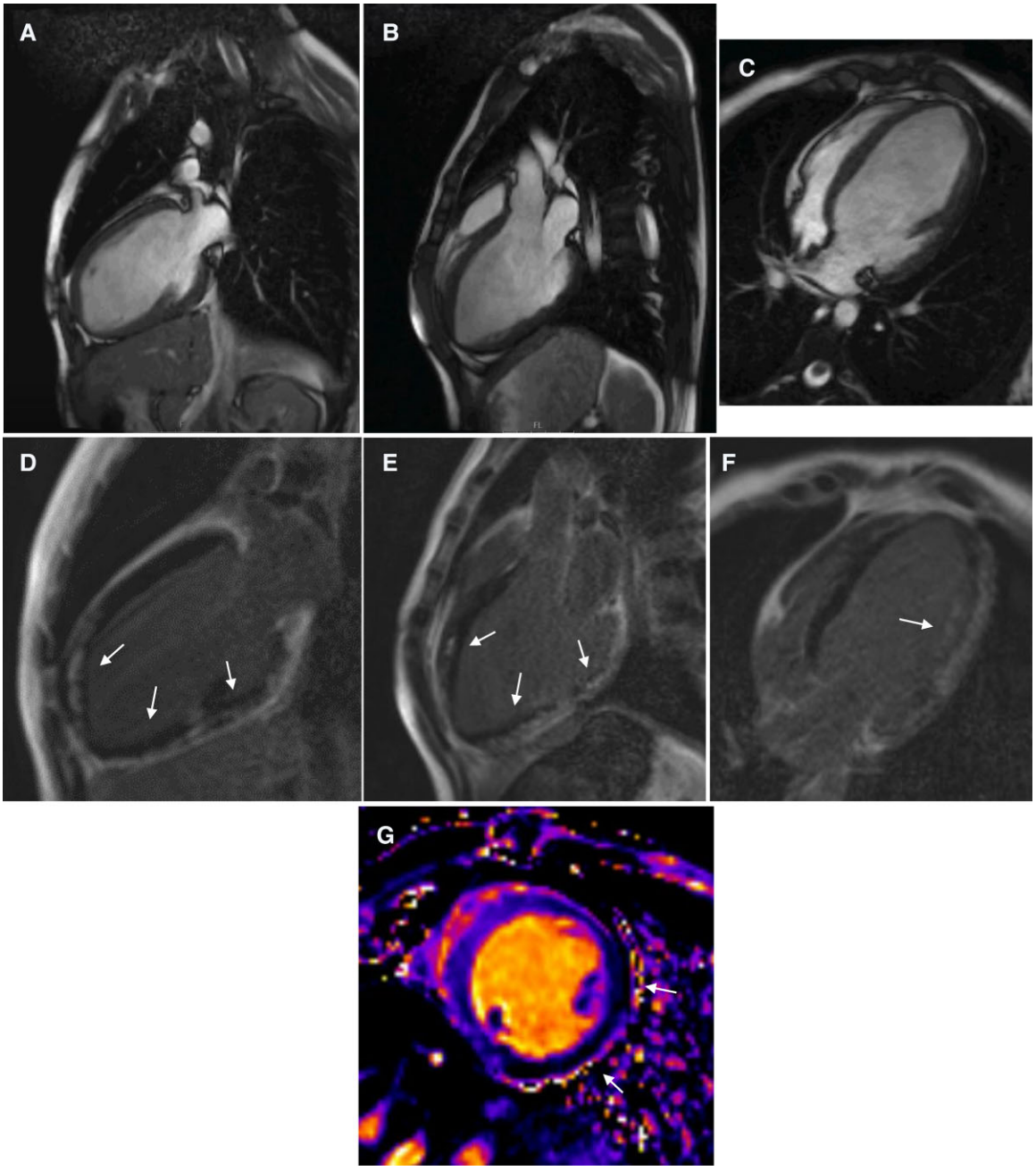


Figure 1 CMR at the time of diagnosis of dilated cardiomyopathy. (*A–C*) Cine views (*A*: 2-chamber view, *B*: 3-chamber view, and *C*: 4-chamber view). (*D–F*) Late gadolinium enhancement sequences in the same views. The extensive patchy subepicardial late gadolinium enhancement of the inferior, inferolateral, anterolateral, anteroseptal, and apical anterior left ventricular walls is demonstrated by the arrows. (*G*) T1 map was abnormal in the same left ventricular walls, as shown by the arrow.

At the age of 19, the patient presented with central crushing chest pain ongoing for one hour. He had no traditional risk factors for ischaemic heart disease. His electrocardiogram revealed anterior ST elevation (*Figure 2*). He was haemodynamically stable, and his clinical examination was unremarkable. He was taken to the catheter laboratory via the primary angioplasty pathway. His angiogram revealed occlusion of the distal left anterior descending (LAD) artery and second diagonal branch (*Figures 3A* and *4B*). There was no evidence of underlying coronary artery disease. He was treated with balloon angioplasty of the distal LAD and second diagonal branch with an excellent angiographic result (*Figure 3C*). Intravascular imaging was not performed as the occlusion was distal. The mechanism of STEMI was felt to be CE.

His echocardiogram during this admission showed severely dilated LV (LV end-diastolic volume indexed for body surface area of 128 mL/m^2, indexed LV end-systolic volume of 120 mL/m^2) and suggested an LV thrombus (heterogenous mass measuring

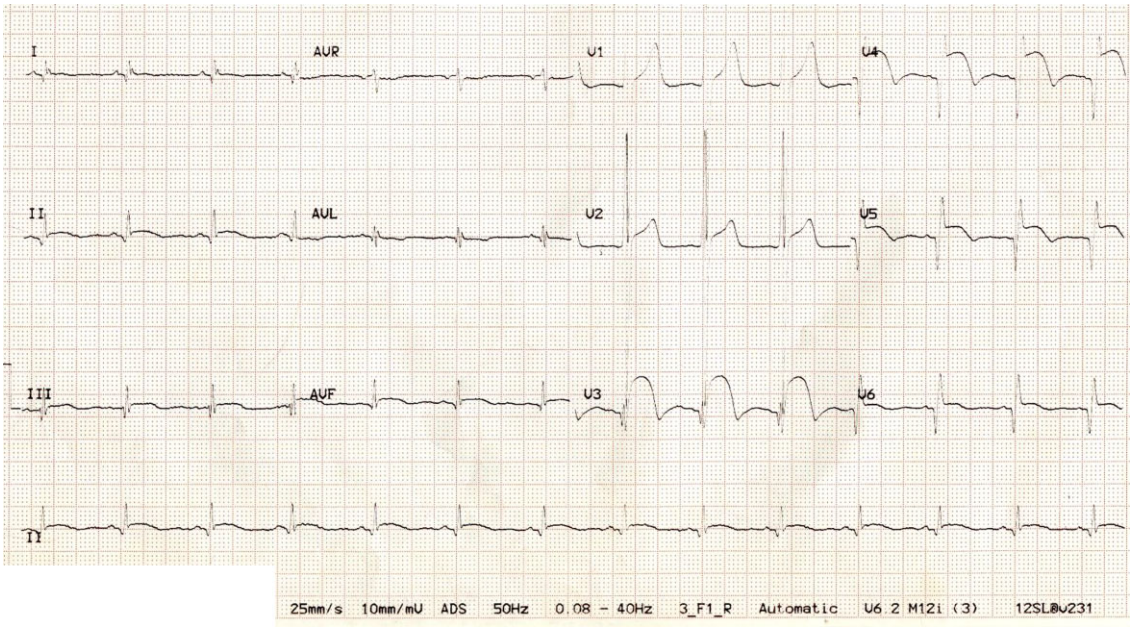


Figure 2 Electrocardiogram at the time of presentation with chest pain, showing anterior ST elevation.

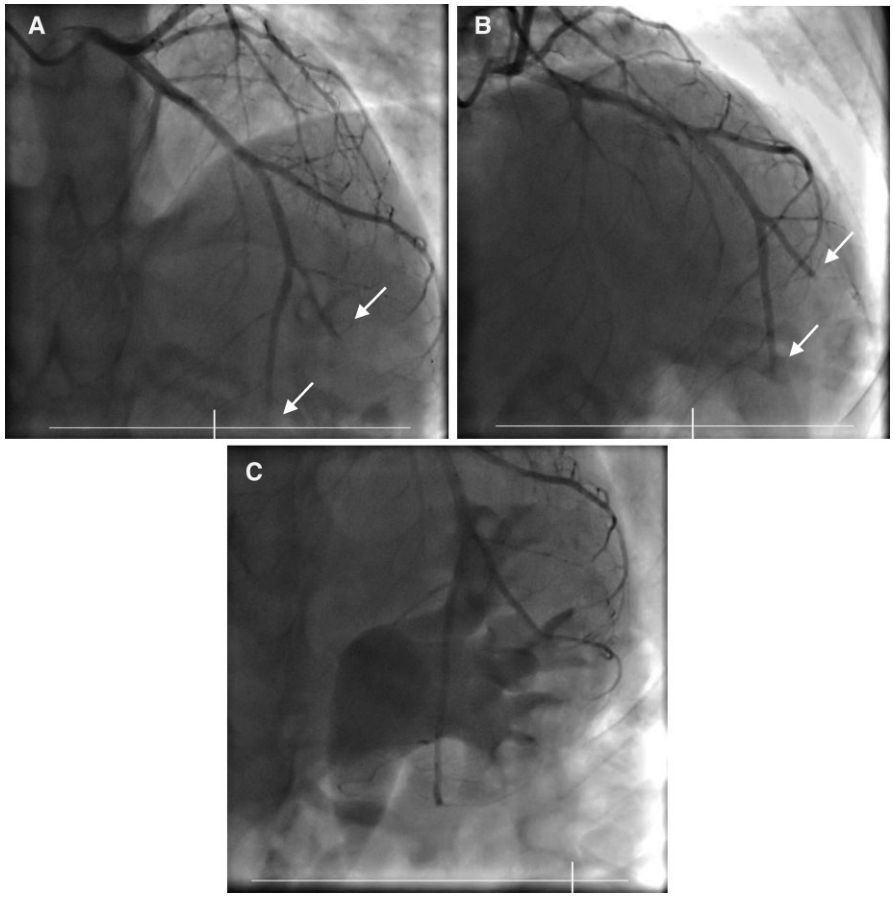


Figure 3 (*A*,*B*) Thrombotically occluded left anterior descending and second diagonal branch (arrows). (*C*) Results following balloon angioplasty of left anterior descending and second diagonal branch.

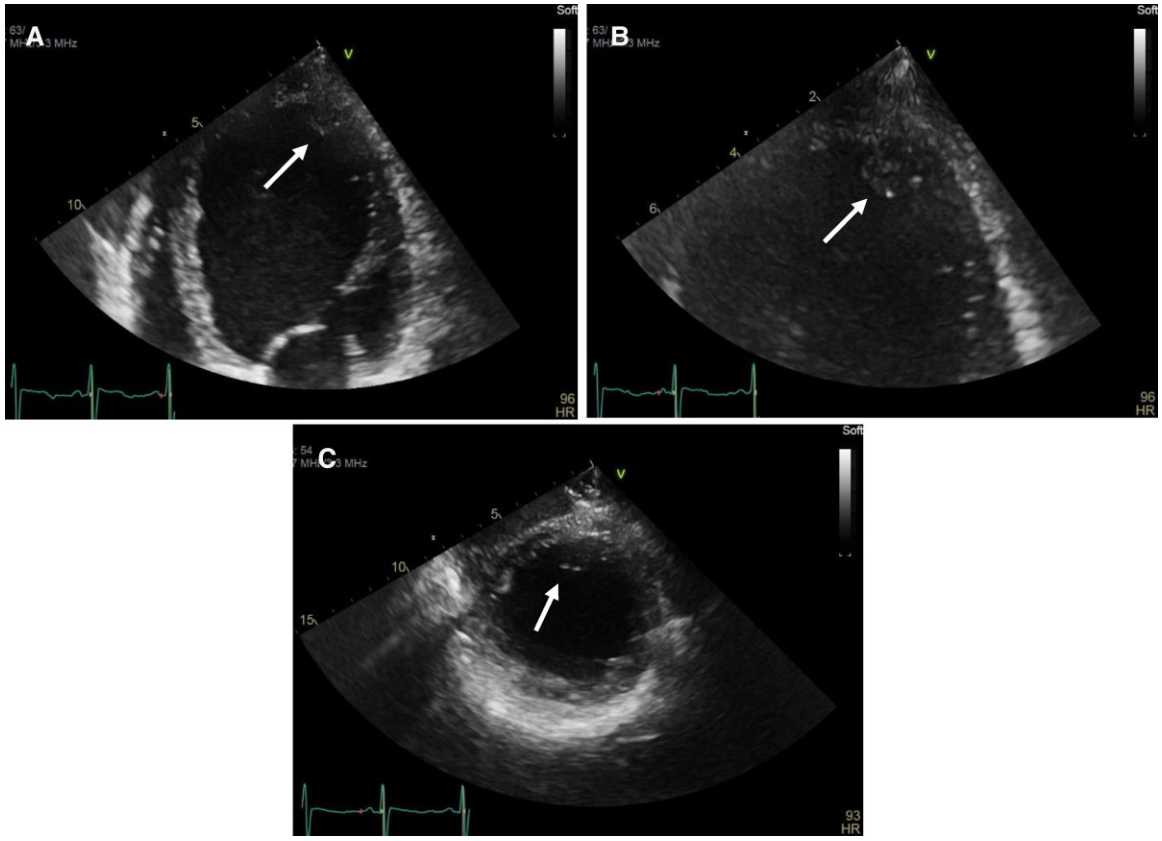


Figure 4 Transthoracic echocardiography images following the STEMI raised the suspicion of apical LV thrombus. (*A*,*B*) Apical 4-chamber view, (*C*) Parasternal short-axis view.

0.9 × 1.0 cm visualized in the LV apex—*Figure 4*). This was felt to be the most likely cause of the CE. His troponin I was elevated at 3.10 μg/L (0–0.04 μg/L), and his creatine kinase was 21 040 U/L (40–320 U/L).

The patient was treated with standard post-myocardial infarction care, and he was initiated on anticoagulation with warfarin with target INR (international normalized ratio) between 2 and 3. Bridging therapy with intravenous heparin was administered until the INR was therapeutic. Antiplatelets were not felt to be appropriate in this case as there was no evidence of underlying coronary artery disease. The steroid therapy that he was already established on (prednisolone 20 mg once a day) was continued at the same dose. There was no clinical evidence of other systemic embolism.

Subsequent echocardiogram 6 months following anticoagulation treatment with well-controlled INR showed no evidence of LV thrombus (*Figure 5*). However, a decision was made to continue treatment with warfarin long term, taking into consideration the persistent substrate for thrombus formation (LV systolic impairment).

His LV function did not improve despite maximum tolerated medical heart failure treatment, and a decision was made to insert a primary prevention ICD (implantable cardioverter-defibrillator) 9 months after his presentation with STEMI. The patient was not a candidate for cardiac resynchronization therapy as he had a narrow QRS complex.

At the time of the last follow-up (3 years following the presentation with STEMI), his heart failure symptoms were well controlled, and he had not shown any evidence of further embolic events.

Discussion

We report on the case of a young patient with dilated cardiomyopathy on a background of DMD who developed STEMI secondary to CE, most likely secondary to LV thrombus.

To our knowledge, this is the first case of STEMI secondary to thrombotic coronary occlusion described in a patient with DMD. Cases of STEMI with normal coronaries in DMD patients have been reported in the literature.[6] These cases were felt to possibly represent acute myocardial necrosis, and pulse corticosteroid therapy was given to halt this process, although there is no evidence from randomized control trials to support this treatment approach. In our case, the coronary angiogram revealed thrombotically occluded coronary arteries. According to the criteria proposed by Shibata *et al.*[7] for the clinical diagnosis of CE, our case met the requirements for definite CE based on the presence of two major criteria (angiographic evidence of coronary artery embolism and thrombosis without atherosclerotic components, and concomitant embolization at multiple sites). All minor criteria were also met (<25% stenosis on

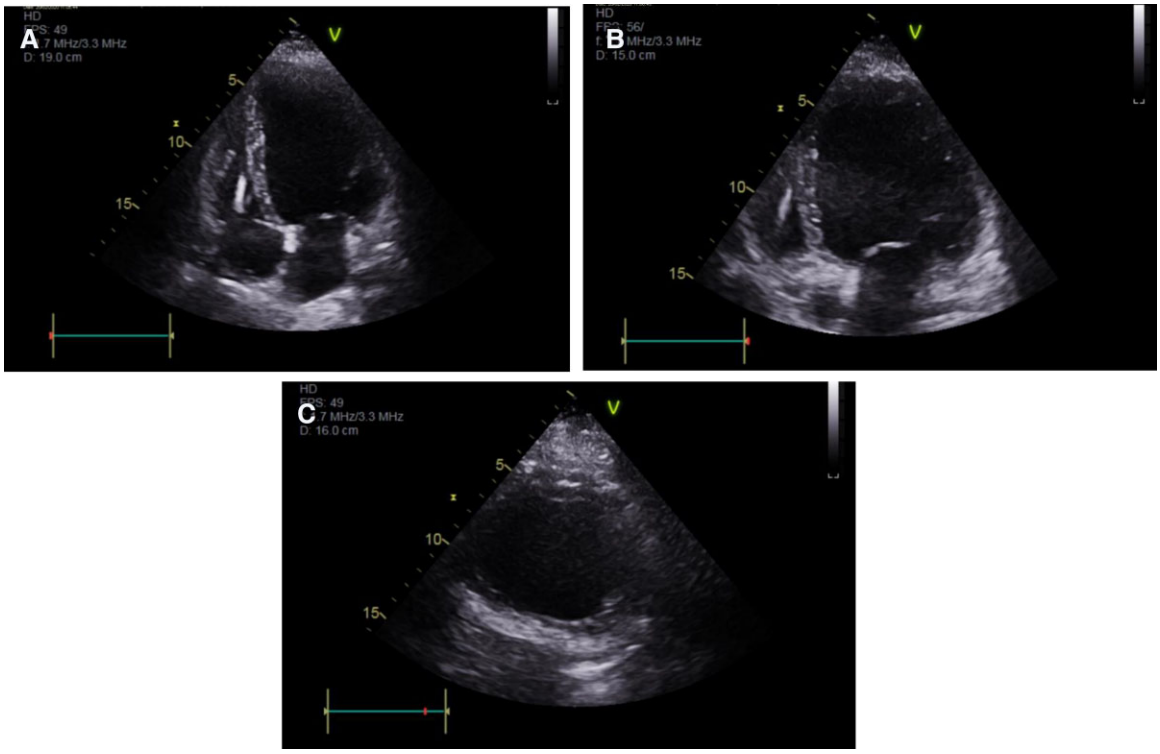


Figure 5 Transthoracic echocardiogram following treatment with anticoagulation showed no convincing evidence of apical left ventricular thrombus within the limitations of the study (limited endocardial definition). (*A*) Apical 4-chamber view, (*B*) Apical 5-chamber view, (*C*) Parasternal short-axis view. *Please note the study was performed without* electrocardiogram *monitoring as per departmental protocol during the first wave of the COVID-19 pandemic.*

coronary angiography except for the culprit lesion, evidence of embolic source detected on echocardiography, and presence of dilated cardiomyopathy, which is an embolic risk factor).

CE is a rare and potentially under-recognized cause of myocardial infarction, with significant mortality and morbidity.[8] Intracardiac sources of CE are well recognized.[9] To date, there are no treatment guidelines, and most of the published experience comes from case series or case reports.[8] Different treatment options that have been employed in the interventional treatment of CE include thrombectomy, thrombus aspiration, balloon angioplasty, and occasionally stent placement. The medical management following coronary intervention is also controversial. According to a recent meta-analysis of 147 cases from case reports and case series, only 33% of patients with CE were prescribed anticoagulation and/or antiplatelet agents following the index event.[8] Intracoronary imaging has been used to differentiate atherosclerotic aetiology of the myocardial infarction and to avoid unnecessary stenting.[10] In our case, we did not use intracoronary imaging as the vessel occlusion was distal.

Patients with severe LV systolic dysfunction are known to be at an increased risk of LV thrombus formation, which can cause cardioembolic events. Cases of cardioembolic stroke[11] or other sites of systemic embolization[12] as a result of LV thrombus have been described in DMD patients. Furthermore, cases of ischaemic stroke in the absence of LV thrombus or documented atrial arrhythmias despite extensive investigation have been reported,[13] indicating that this patient population is at risk of cardioembolic events even in the absence of traditional risk factors. It has been suggested that DMD patients with cardiac dysfunction may exhibit systemic subclinical coagulation and fibrinolysis activation,[14] although further evidence to support this is required. Currently, the use of anticoagulation or antiplatelet therapy in patients with heart failure who are in sinus rhythm is not recommended.[15] Equally, there is no clear guidance on screening asymptomatic heart failure patients for atrial arrhythmias or LV thrombus. These limitations apply to all heart failure patients, and they are particularly relevant in patients with DMD.

Conclusion

DMD is often associated with dilated cardiomyopathy, which can be complicated by the formation of intracardiac thrombus. The latter can cause embolization in the systemic circulation; this includes thrombus embolization in the coronary arteries, which can present as STEMI.

This case highlights the complexity of the management of DMD patients with severe LV systolic dysfunction. It also discusses the limited evidence and the dilemmas in the treatment of myocardial infarction secondary to CE.

Lead author biography

Dr Panagiota Mitropoulou is a Cardiology Registrar (ST6 level) in Wessex Deanery, UK. She was awarded her medical qualification with Honours (National and Kapodistrian University of Athens, Greece), and she moved to the UK for her postgraduate training. Her subspecialty interest is Adult Congenital Heart Disease (ACHD) and Heart Disease in Pregnancy, and she is currently in a 2-year ACHD Fellowship. She also has a strong interest in medical education, having completed a postgraduate certificate in medical education, and until recently she was the Wessex Deanery IMT (Internal Medicine Training) Implementation Fellow.

Supplementary material

Supplementary material is available at *European Heart Journal—Case Reports* online.

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Slide sets: A fully edited slide set detailing this case and suitable for local presentation is available online as Supplementary data.

Consent: The authors confirm that written consent for submission and publication of this case report, including image(s) and associated text has been obtained from the patient in line with COPE guidance.

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