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CASE REPORT

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Throwing thrombi: noncompaction cardiomyopathy causing renal infarct and catastrophic stroke - a case report

Madeeha Javed[1], Shivani Desai[1], Nathan DeRon Jr.[1*] and Miguel Villamil[1]

Abstract

Background Left ventricular noncompaction (LVNC) is a distinct cardiac phenotype characterized by prominent left ventricular trabeculae and deep intertrabecular recesses. It results in thickened myocardium with two layers consisting of non-compacted myocardium and a thin, compacted layer of myocardium. LVNC is a genetic condition associated with various cardiomyopathies, congenital heart disease, and environmental factors.

Case presentation A 60-year-old Afroamerican male with a past medical history of hypertension and chronic kidney disease stage 3a presented to the emergency department (ED) with sudden-onset abdominal pain and associated symptoms of nausea, vomiting, and diarrhea. The patient was provided antiemetics, antihypertensives, and pain control in the ED. An abdominal x-ray showed the small bowel with multiple fluid levels concerning for obstruction. Contrast-enhanced computed tomography of the abdomen showed a wedge-shaped attenuation in the lower pole of the right kidney concerning for infarction but negative for obstruction. There was also a nonocclusive thrombus in the superior mesenteric artery. A transthoracic echocardiogram (TTE) showed a newly reduced left ventricular ejection fraction of 20–25%, moderate dilatation of the left ventricle, and severe global hypokinesis, but did not reveal any thrombus. Cardiology was consulted and recommended a transesophageal echocardiogram (TEE) along with lifelong anticoagulation with apixaban. The TEE revealed a new finding of LVNC without thrombus. The patient underwent a left cardiac catheterization which showed no significant obstructive coronary artery disease. He was discharged on guideline-directed medical therapy (GDMT). Unfortunately, the patient was noncompliant with his GDMT and anticoagulation regimen. He presented approximately six weeks later with right hemiparesis. A repeat TTE showed a large thrombus in the left ventricle. The patient remained aphasic with right hemiparesis without significant recovery before discharge.

Conclusion This case highlights a rare cause of heart failure and catastrophic thromboembolism: noncompaction cardiomyopathy. This case is a prime example and reminder of the potential impact of LVNC on patient morbidity and should encourage medical providers to be conscious of this anomaly and its potential for severe clinical consequences.

Keywords Noncompaction cardiomyopathy, Cardiac noncompaction, Heart failure with reduced ejection fraction, Hypertension, Superior mesenteric artery thrombus

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Introduction

Left ventricular noncompaction (LVNC) is a distinct cardiac phenotype characterized by prominent left ventricular trabeculae and deep intertrabecular recesses. It results in thickened myocardium with two layers consisting of non-compacted myocardium and a thin compacted layer of myocardium [1]. LVNC is a genetic condition associated with various cardiomyopathies, congenital heart disease, and environmental factors. Its prevalence is estimated to be 0.014–1.3% [2]. Among patients with heart failure, the prevalence is 3–4%, and it is more frequent among Afroamerican patients [3]. LVNC is putatively due to intrauterine arrest of compaction of the meshwork that makes up the fetal myocardial primordium. The pronounced trabeculation may be due to altered cell proliferation, differentiation, and maturation during ventricular wall formation [1]. Each individual with LVNC may have unique trabeculations, and prominent trabeculations may also develop during adult life. This may explain LVNC phenotypes seen in athletes, patients with uncontrolled hypertension, pregnant patients, or patients with heart failure or hematologic disorders.

Table 1 Summary of inpatient laboratory findings

Lab	*Value*
Sodium	143 mmol/L
Potassium	2.9 mmol/L
Chloride	106 mmol/L
Carbon Dioxide	33 mmol/L
BUN	29 mg/dL
Procalcitonin	0.18 ng/mL
Urine Marijuana	Positive
Urine Cocaine	Positive
Urine Opiates	None Detected
Urine Amphetamines	None Detected
Urine Benzodiazepines	None Detected
Urine Barbiturates	None Detected
Urine beta-hCG	Negative
WBC	7.6×10^3/µl
Hemoglobin	12.4 g/dL
Platelet	378×10^3/µl
MCV	85.2 FL
RDW-CV	14.6%
INR	1.3
BNP	7190 pg/mL
Total CK	68 U/L
Troponin	0.098 ng/mL
COVID-19 by RT-PCR	Not Detected
Hemoglobin A1c	6.3%
Respiratory Viral Panel	Negative

Case presentation

A 60-year-old Afroamerican male with a past medical history of hypertension and chronic kidney disease stage 3a presented with sudden onset abdominal pain, nausea, vomiting, and diarrhea. The patient reported smoking 1 to 2 cigars per day for many years, drinking multiple 24 ounce cans of beer several times per week, and occasional marijuana use. Patient was estranged from his family and unsure about any familial history of cardiac disease. Initial laboratory data showed hypokalemia, metabolic alkalosis, and positive urine screening for marijuana and cocaine (Table 1). Cardiac exam showed a regular heart rate and rhythm without any murmurs, rubs, or gallops. Abdominal exam revealed mild tenderness to palpation in the periumbilical and suprapubic region, and the lower extremities exhibited 2 + pitting edema bilaterally.

Electrocardiography revealed normal sinus rhythm with voltage criteria for left ventricular hypertrophy and nonspecific ST changes (Fig. 1). Chest radiography showed cardiomegaly with findings of pulmonary edema. Contrast-enhanced computed tomography (CT) of the abdomen revealed a wedge-shaped area of hypoattenuation in the lower pole of the left kidney compatible with infarction (Fig. 2). A filling defect of the distal superior mesenteric artery was also observed (Fig. 3). Transthoracic echocardiography revealed an ejection fraction of 25–30% with no clear thrombus noted. To further investigate, the patient underwent transesophageal echocardiography, which demonstrated left ventricular noncompaction (Fig. 4).

Given these findings, cardiology was consulted and recommended lifelong anticoagulation. The patient also underwent left heart catheterization to explore the etiology of the reduced ejection fraction; no significant coronary artery disease was observed. The patient was initiated on guideline-directed medical therapy (GDMT). The medications were carefully titrated as the patient's blood pressure would allow to avoid hypotension. He was counseled on the importance of continuing his medications, including anticoagulation, upon discharge.

However, approximately two months later, the patient presented again to the hospital with severe right-sided weakness and aphasia. Magnetic resonance imaging (MRI) revealed a large left middle cerebral artery distribution infarction involving the left temporal and parietal lobes (Fig. 5). The cerebral infarction was secondary to anticoagulation noncompliance and left ventricular thrombus embolization. The patient briefly required mechanical ventilation due to respiratory failure and did not recover meaningful function after extubation. The patient remained aphasic and eventually transitioned home with 24-hour supervision.

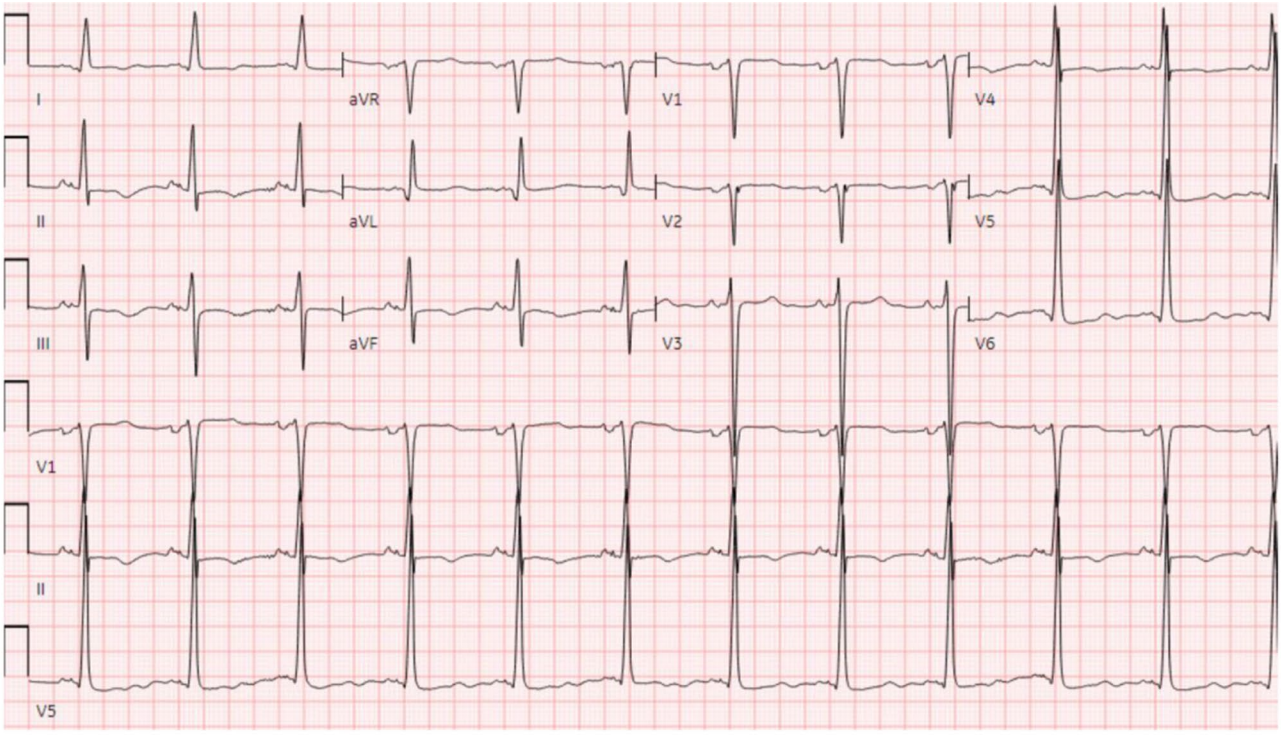

Fig. 1 ECG on admission showing normal sinus rhythm at a rate of 60 with left ventricular hypertrophy and nonspecific ST changes

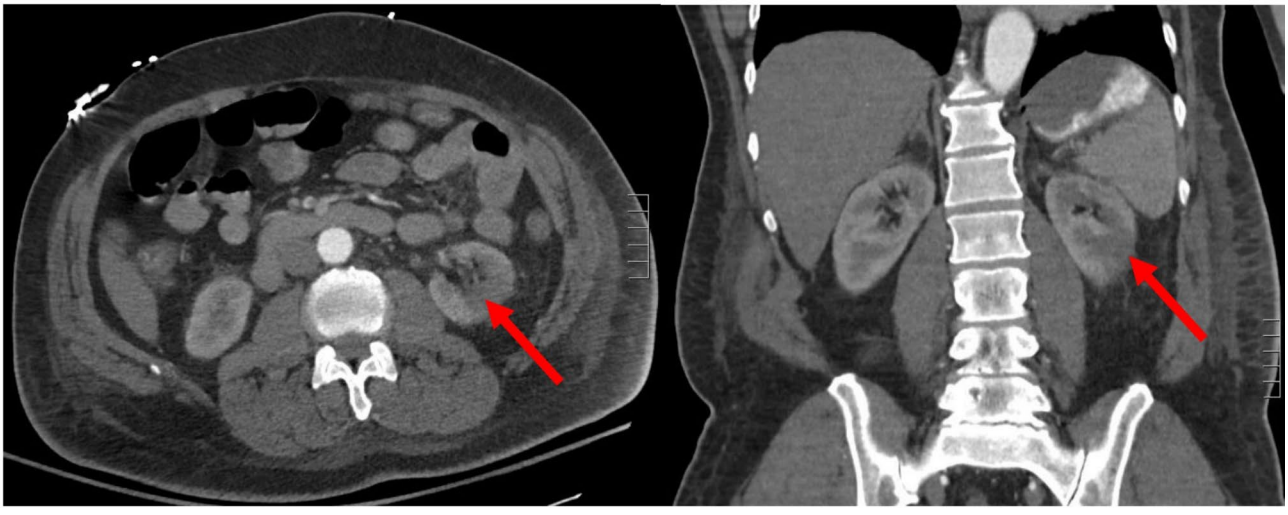

Fig. 2 Contrast-enhanced computed tomography images of the abdomen revealing a wedge-shaped infarct in the mid to lower pole of the left kidney (red arrows)

Discussion

The pathogenesis of noncompaction cardiomyopathy starts during embryonic development. Initially, the myocardium is a loose network of fibers which is separated by deep recesses that connect the myocardium to the left ventricle. Usually, during weeks five to eight of embryonic development, the trabeculations halt proliferation and compact to form the ventricular myocardium creating the spongy appearance of the left ventricle. Noncompaction of the myocardium is thought to be caused by arrest of this normal embryological process [4, 5]. The direct mechanism is unknown, but one proposed hypothesis is that noncompaction occurs due to ventricular wall growth into the lumen in a trabecular fashion rather than incomplete formation of the myocardial meshwork [6]. Usually, only the left ventricle is affected, but biventricular noncompaction has been reported [7, 8].

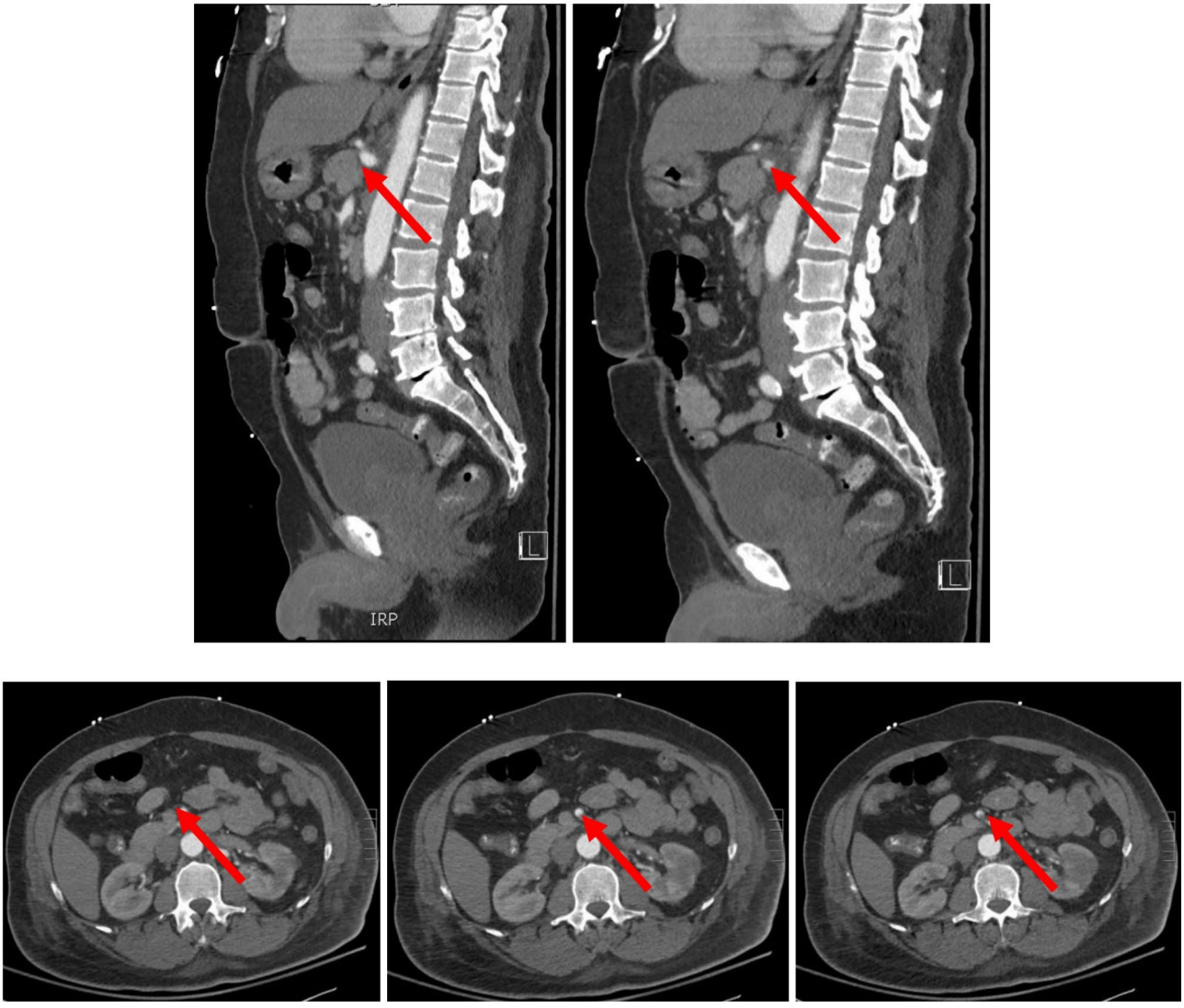

Fig. 3 Contrast-enhanced computed tomography images of the abdomen and pelvis demonstrating a filling defect in the superior mesenteric artery (red arrows)

Noncompaction cardiomyopathy encompasses both sporadic and familial forms. Several genetic factors have been reported. In a family of six affected children, a mutation in the G4.5 gene in the Xq28 chromosome region was identified illustrating an X-linked pattern [9]. Another report showed a link between noncompaction cardiomyopathy and mutations in the G4.5 and alpha-dystrobrevin genes [10].

The median age at noncompaction cardiomyopathy diagnosis is 7 years with ages ranging from 11 months to 22 years [4]. However, noncompaction has been diagnosed, although more rarely, in older adults [11]. A noncompaction cardiomyopathy diagnosis is made by two-dimensional and color Doppler echocardiography. Deep intertrabecular recesses are observed within multiple ventricular trabeculations. Color Doppler imaging shows blood flow that is continuous from the deep recesses to the ventricular cavity [4]. There have also been quantitative approaches to diagnosing noncompaction including using trabeculation peak to trough ratios [12]. Other studies have proposed a diagnosis as at least a 2:1 ratio of the maximum thickness of noncompacted to compacted layers measured at the end of systole in a parasternal short axis (echocardiographic view) [13]. This ratio provides a quantifiable method to distinguish LVNC from normal myocardial structure, which typically has a lower ratio. A higher ratio correlates with more severe forms of the disease and can help predict outcomes. It may suggest a higher likelihood of developing complications like heart failure or arrhythmias. Therefore, monitoring changes in this ratio over time can help assess disease progression and guide treatment

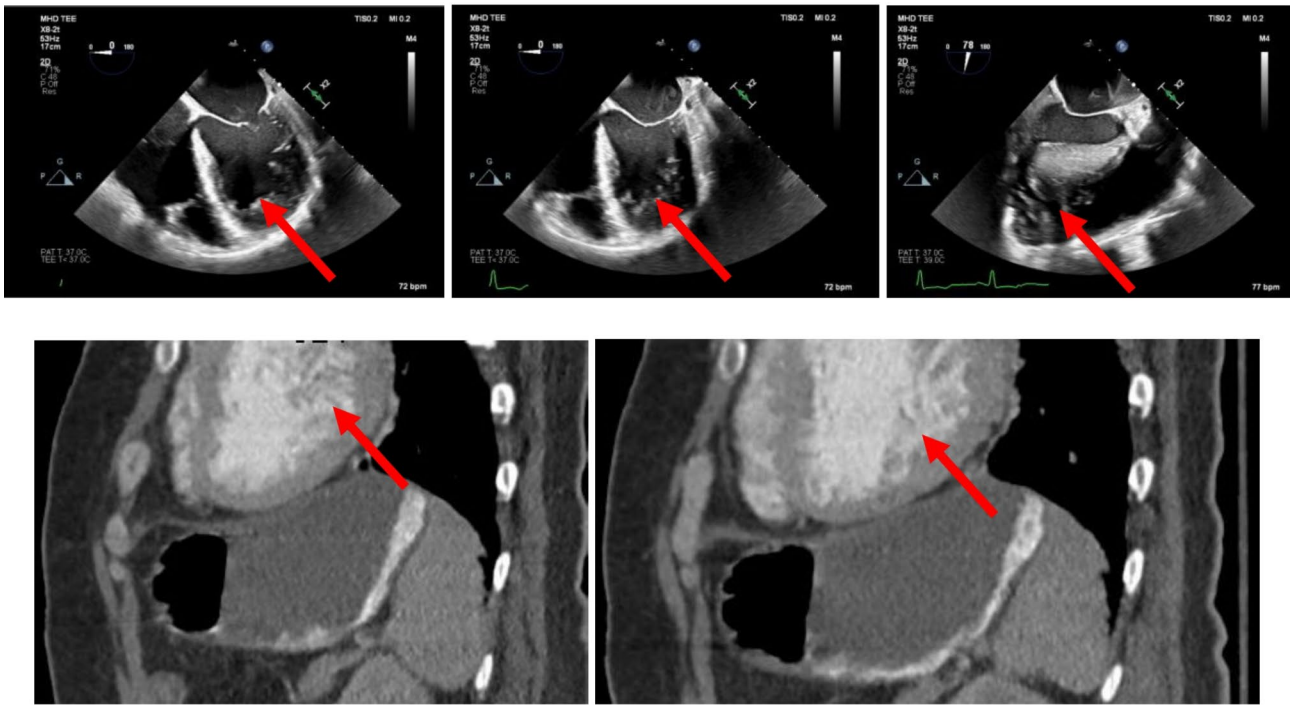

Fig. 4 Transesophageal echocardiography 4 chamber view (top row) and computed tomography of the chest (bottom row) images illustrating left ventricular noncompaction morphology (red arrows), severely dilated left ventricle without hypertrophy, and severely dilated left atrium

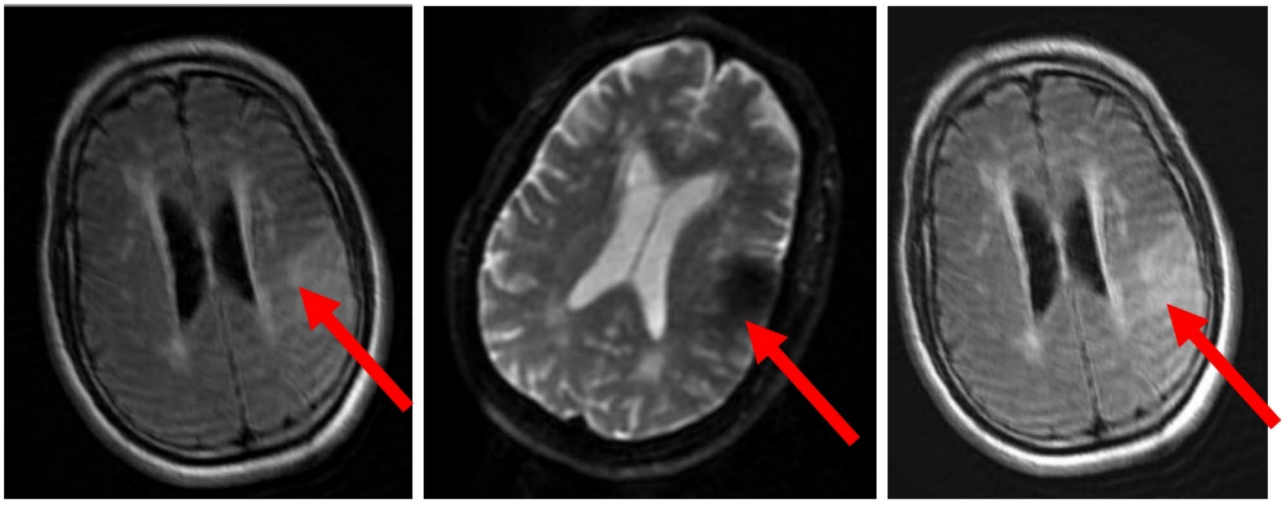

Fig. 5 Magnetic resonance imaging of the brain without contrast showing infarction in the distribution of the left middle cerebral artery (red arrows)

decisions. Although echocardiography is used to make the diagnosis of noncompaction, other modalities such as contrast ventriculography, CT, and MRI can be utilized. Other coronary artery anomalies and pathologies need to be excluded for a true diagnosis of noncompaction of ventricular myocardium.

Clinical features of LVNC include heart failure, embolic events, and arrhythmias. Some patients present asymptomatically, and others present with severe debilitating congestive heart failure. The patient in this case initially presented with abdominal pain, and CT of the abdomen revealed a left kidney infarct and filling defect of the distal superior mesenteric artery. According to Chin et al., approximately 38% of patients present with systemic embolism. Patients can present with both systolic and diastolic dysfunction in heart failure. The cause of systolic dysfunction is unclear, but developing research indicates that it may be due to subendocardial hypoperfusion and microcirculatory dysfunction [14]. The diastolic dysfunction is thought to be due to the abnormal relaxation

and restrictive filling due to the extensive, prominent trabeculae. Arrhythmias are common, with atrial fibrillation presenting in approximately 25% of reported cases of adults with noncompaction. Ventricular tachyarrhythmias are common in patients with LVNC [15]. Sudden cardiac death, paroxysmal supraventricular tachycardia, and complete heart block have also been reported in patients with isolated noncompaction of the ventricular myocardium [1]. Although LVNC leading to myocardial fibrosis has not been well defined in the literature, recent studies have reported a significant prevalence of mitochondrial myopathy and genetic mutations in patients with LVNC [20]. These mutations and myopathies may be the etiology of fibrosis and eventually lead to severe LV dysfunction, a process that takes several years and may explain why heart failure typically presents in the fourth decade of life in patients with LVNC.

Management of LVNC focuses on treating the three major clinical manifestations: heart failure, systemic embolic events, and arrhythmias. GDMT is imperative for treatment of heart failure. Ambulatory ECG monitoring should be performed annually to assess for atrial and ventricular arrhythmias [16]. Cardiac transplant can be considered in patients with LVNC and refractory heart failure. More data is needed to associate noncompaction and benefit from an implantable cardioverter-defibrillator in the setting of sudden cardiac death. Prevention of embolic events is another component of management, and is recommended by several authors that long-term prophylactic anticoagulation is warranted [17]. While it is imperative that patients with underlying atrial fibrillation or previous thromboembolic events be placed on anticoagulation, it seems reasonable to consider the same strategy for patients with LVNC given the elevated risk of thromboembolism. For this reason, formulation of a better clinical stratification and pathophysiologic understanding are necessary. Screening of first-degree relatives by echocardiography is also recommended due to a possible genetic component of LVNC [18].

Previous case reports for non-compaction demonstrated cryptogenic or overtly embolic stroke or myocardial infarction [19], but data on embolism to specific areas such as the kidney or mesentery has not yet been reported in the literature.

Conclusion

Noncompaction cardiomyopathy is an uncommon cause of heart disease often present in younger populations. Usual presenting symptoms include dyspnea, NYHA class III or IV heart failure, palpitations, chest pain, and syncope. Here, we present the case of newly diagnosed reduced ejection heart failure in a 60-year-old male who was initially found to have a nonocclusive thrombus of superior mesenteric artery and an embolic wedge-shaped infarction of the kidney. This case highlights the rare instance of noncompaction cardiomyopathy as the underlying cause of heart failure as well as the increased propensity to develop thrombi. Clinicians should be aware of this diagnosis and include it on the differential especially when evaluating younger patients presenting with signs and symptoms of heart failure or older adults with new ischemic events. Additionally, this case highlights the importance of counseling patients with noncompaction cardiomyopathy on lifelong compliance with anticoagulation.

Supplementary Information

The online version contains supplementary material available at https://doi.org/10.1186/s12872-024-04439-0.

Supplementary Material 1

Author contributions

Dr. Madeeha Javed performed the literature review and wrote and edited the manuscript. Dr. Shivani Desai performed the literature review and wrote and edited the manuscript. Dr. Nathan DeRon, Jr. wrote and edited the manuscript and generated the table and figures. Dr. Miguel Villamil oversaw the project and reviewed and edited the final manuscript.

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Data availability

All data and materials are available upon reasonable request.

Declarations

Ethics approval and consent to participate

Ethics approval was not applicable to this type of manuscript. Informed consent to participate was obtained from the patient.

Consent for publication

Written and signed consent was obtained from the patient to publish this information prior to publication.

Competing interests

The authors declare no competing interests.

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