LETTER / *Cardiovascular imaging*

Incidental diagnosis of a familial left ventricular noncompaction on a chest CT angiography



Keywords: Cardiac imaging techniques; Magnetic resonance imaging; Echocardiography; Isolated noncompaction of the ventricular myocardium; Cardiomyopathies

Introduction

Left ventricular noncompaction (LVNC) is a rare genetic cardiomyopathy, which may progress to left ventricular (LV) dilatation and systolic dysfunction and often leads to cardioverter-defibrillator implantation and/or cardiac transplantation. Family investigation is essential. The diagnosis is based on three imaging examinations or autopsy. We report an incidental diagnosis of a familial form of LVNC, discovered on a chest computed tomography (CT) angiography performed in the index patient at the emergency department to rule out pulmonary embolism.

Case report

A 32-year-old man, practicing as a sports and aerobics teacher, was referred to the emergency department for chest pain, dyspnoea, and elevated D-dimer level. Contrast-enhanced chest computed tomography (CT) could eliminate a pulmonary embolism. It showed biventricular dilatation, prominent trabeculations of the LV (Fig. 1) and pulmonary oedema. During hospitalization, transthoracic echocardiography (TTE) confirmed a dilated cardiomyopathy, left ventricular ejection fraction (LVEF) was 20%.

A cardiac magnetic resonance was performed. The trabeculated LV mass exceeded 20% of the global LV mass according to Jacquier et al. method [1]. The ratio of non-compacted to compacted myocardium in end diastole was more than 2.3 (Petersen et al. method [2]). Trabeculations predominated in the mid-lateral and apical segments of the LV. LVEF was 15% and there was no delayed enhancement (Fig. 2). LVNC was diagnosed.

A contrast-enhanced TTE was then performed [3], confirming the diagnosis, by using the criteria of Jenni et al. [4] (no coexisting cardiac abnormalities, maximal end systolic ratio of non-compacted to compacted layers more than 2, mid-lateral predominant localization, color Doppler signal in intertrabecular recesses). TTE showed deep recesses filled up by the contrast agent and a maximal systolic compacted myocardium thickness less than 8 mm in the lateral apical segment [5] (Fig. 3).

Family investigation disclosed a history of cardiomyopathy. The index patient's father was treated by an implantable cardioverter-defibrillator. A history of cardiac transplantation was also found in the grand father's brother. A complete family investigation has been scheduled.

Cardiac CT and contrast-enhanced echocardiography were performed in the father (contraindicated to MRI), showing prominent trabeculations then fulfilling the diagnostic criteria of LVNC in mid-lateral and apical segments (Fig. 4).

Two weeks later, the index patient presented again to our hospital with acute abdominal pain. An enhanced thoracoabdominal CT showed left renal and splenic infarctions and mid-ventricular anterior and inferior thrombi. Anticoagulant therapy was prescribed.

Discussion

LVNC is classified as a genetic primary cardiomyopathy, characterized by a distinctive ("spongy") morphological appearance of the LV myocardium, resulting from an arrest in the normal embryogenesis [6]. Localization of non-compacted segments predominates in apical and mid-ventricular segments of both the inferior and lateral wall [7]. This disease may progress to LV dilatation and systolic dysfunction, heart failure, thromboembolism and arrhythmia. The diagnosis of LVNC is based on three imaging examinations: echocardiography (or contrast echocardiography), cardiac magnetic resonance and cardiac CT, using the above-mentioned criteria. In 2013, the definitive diagnosis remains based on autopsy.

Both familial and non-familial cases have been described [6] but LVNC is frequently familial, with at least 25% of asymptomatic relatives having a range of echocardiographic

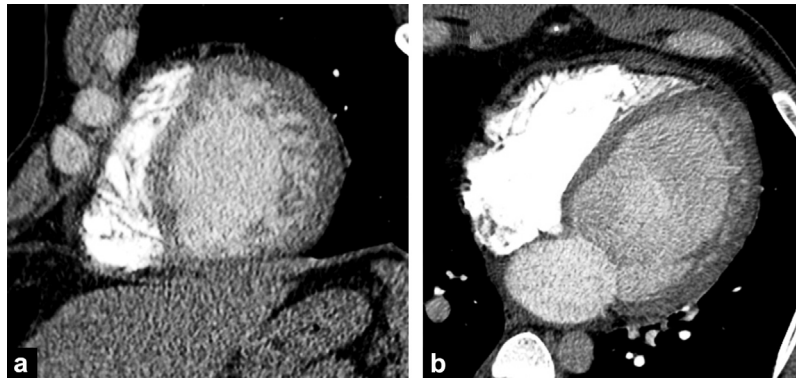


Figure 1. Chest CT angiography: multiplanar reconstruction. Mid-ventricular short-axis view (a) and long-axis four chambers view (b). Prominent trabeculations of the lateral wall and apex of the LV associated with biventricular dilatation. ED diameter of the LV is 77 mm. ED diameter of the right ventricle is 49 mm. CT: computed tomography; ED: end diastolic; LV: left ventricle.

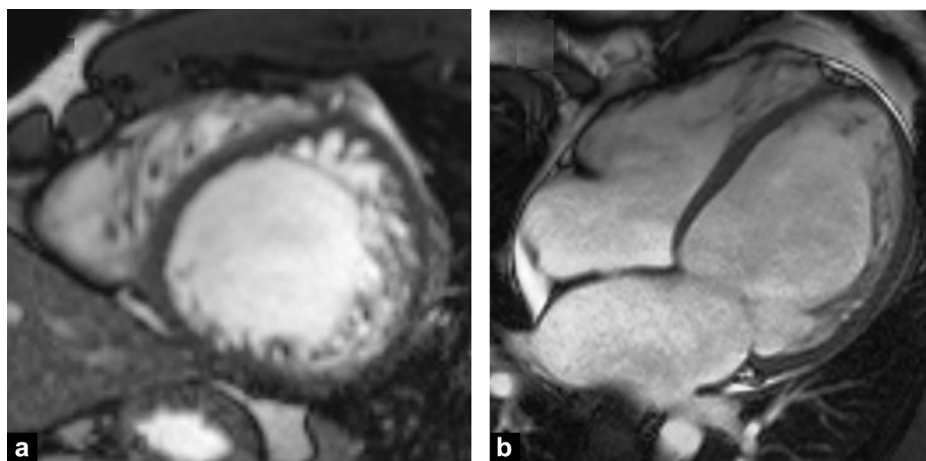


Figure 2. Cardiac magnetic resonance, cine steady-state free precession sequences in the short-axis mid-ventricular view (a) and long-axis four chambers view (b). Prominent trabeculations in mid-lateral and apical segments. Trabeculated left ventricular mass is 32 g/m^2 (23% of the global left ventricle mass). Ratio of non-compacted to compacted myocardium in diastole is 3.

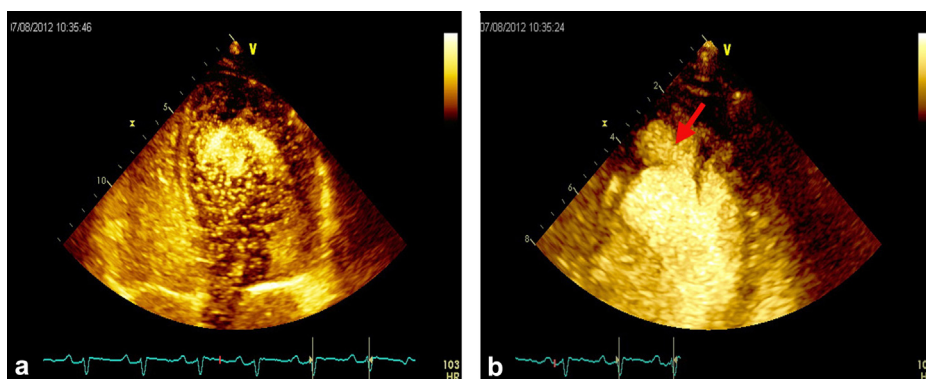


Figure 3. a: contrast-enhanced echocardiography: apical long-axis five chambers view. Apical and lateral prominent trabeculations, better delineated by contrast agent which seems to penetrate into the hypertrabeculated myocardium; b: focus on the apex showing contrast penetrating into the non-compacted myocardium.

abnormalities [8]. For this reason, family investigation is required.

We reported a case of LVNC with thromboembolic and heart failure complications, which proved to be a familial

form, firstly discovered on a chest CT angiography performed at the emergency department. This case illustrates the importance of a thorough analysis of cardiac structures on any chest CT.

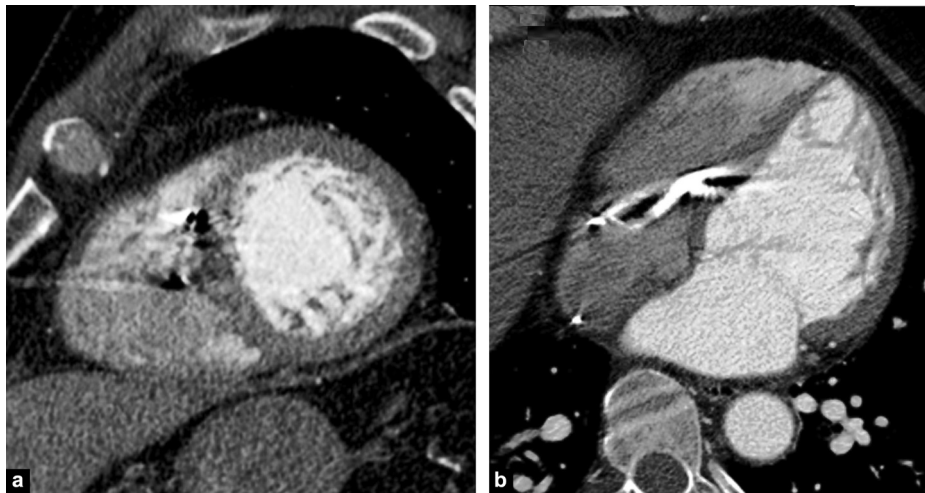


Figure 4. Cardiac CT of the index patient's father in apex short-axis view (a) and long-axis four chambers view (b) showing prominent trabeculations in mid-lateral and apical segments. Left ventricular ejection fraction is 45%. Note the implantable cardioverter-defibrillator leads artefacts in the right ventricle.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

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