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Isolated ventricular non-compaction presenting with ventricular tachycardia

出現心室過速的單純性心肌致密化不全

Isolated ventricular non-compaction is a rare congenital cardiomyopathy, manifested morphologically as prominent myocardial trabeculations and deep inter-trabecular recesses that communicate with the ventricular cavity. Heart failure is the most common presenting condition. Other manifestations include arrhythmia and cardioembolic events. This report is illustrative of isolated ventricular non-compaction in a 78-year-old woman. The diagnosis was made when she presented with ventricular tachycardia many years after a stroke. She subsequently underwent implantation of a cardioverter-defibrillator. This report documents an uncommon presentation of this disease entity in the oldest patient at presentation as yet reported in the literature.

單純性心肌致密化不全是一種罕見的先天性心肌症，形態學上顯示心室內粗亂未致密化的肌小梁及深陷的小梁隱窩。心臟衰竭是最常見的徵狀。其他病徵包括心律失常和心臟栓塞。本報告報導了一名78歲婦女的單純性心肌致密化不全。患者中風多年後出現心動過速，由此診斷為患上此病。她隨後植入心臟復律及纖顫器。本報告記錄現有文獻中患上此病的最年老患者及其罕有病徵。

Introduction

Isolated ventricular non-compaction (IVNC) is a rare congenital cardiomyopathy, manifested morphologically as prominent myocardial trabeculations, and deep inter-trabecular recesses that communicate with the ventricular cavity. The condition is thought to arise from abnormal morphogenesis involving intrauterine arrest of normal compaction of the myocardium. Clinical manifestations include congestive heart failure (CHF), arrhythmia, and cardioembolic events. This report is of IVNC diagnosed in a 78-year-old woman presenting with ventricular tachycardia (VT). She subsequently underwent implantation of an implantable cardioverter-defibrillator (ICD).

Case report

A 78-year-old woman presented to the Pamela Youde Nethersole Eastern Hospital in December 2000 with sudden onset of palpitations and dizziness, associated with vague chest discomfort. She had a history of atrial fibrillation (AF), hypertension, coronary artery disease, diabetes mellitus, and a stroke 20 years previously. There was no known family history of cardiac disease or sudden death. At presentation, VT of 180 beats per minute was recorded, and blood pressure was 100/70 mm Hg. Intravenous lignocaine was ineffective. Cardioversion was then performed and resulted in AF. Electrocardiography (ECG) after cardioversion showed non-specific T inversion in the lateral leads, with no pathological Q-wave evident. Cardiac enzymes were not elevated.

To determine the underlying cause of the VT, echocardiography was performed. This showed prominent myocardial trabeculations over the lateral and apical regions of the left ventricle (LV). Deep inter-trabecular recesses that communicated with the cavity of the LV, and blood flow into the recesses was demonstrated on colour Doppler examination (Fig 1). No other congenital abnormalities were evident. Findings were consistent with IVNC. The inferior

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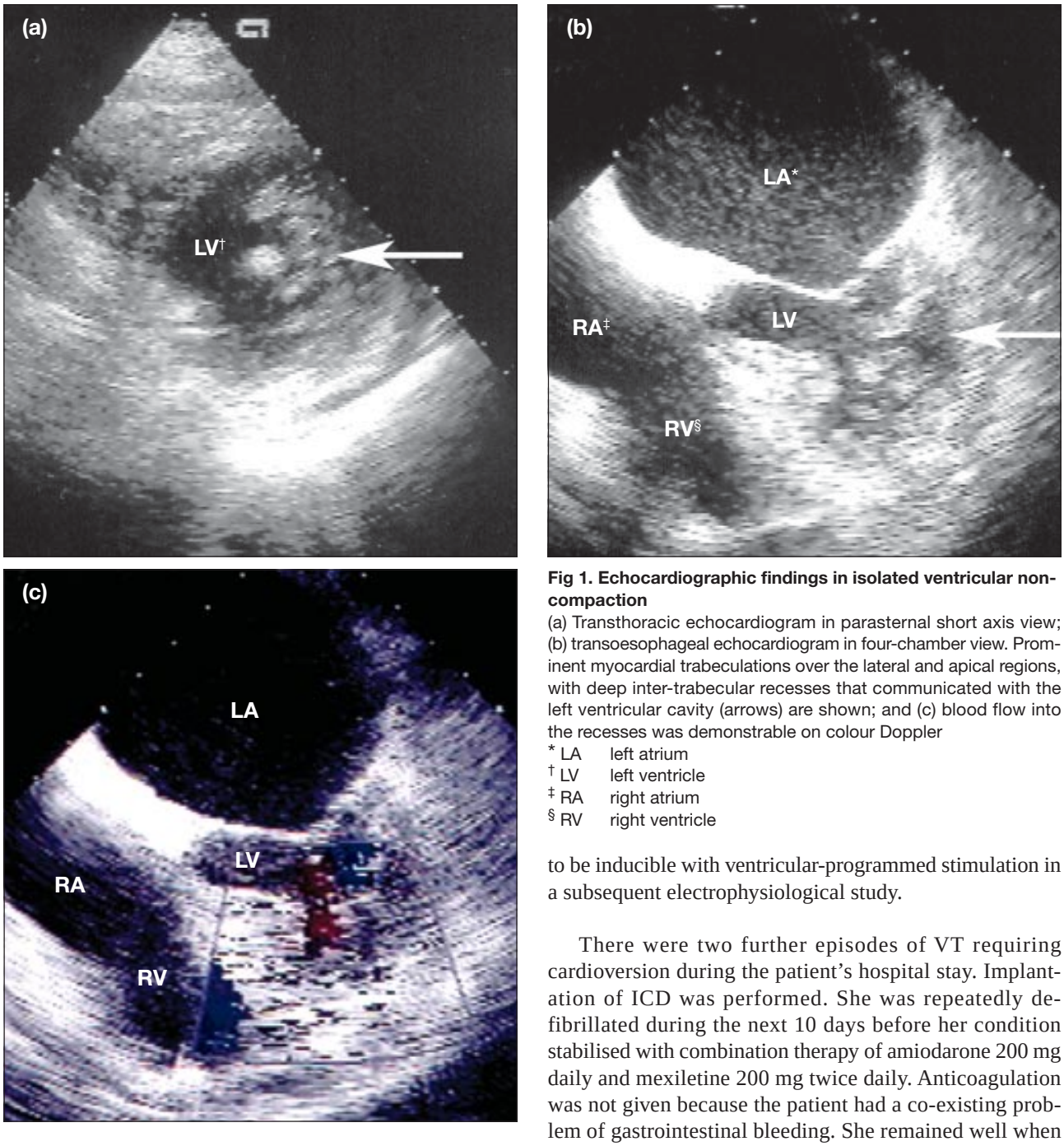

Fig 1. Echocardiographic findings in isolated ventricular non-compaction

(a) Transthoracic echocardiogram in parasternal short axis view; (b) transoesophageal echocardiogram in four-chamber view. Prominent myocardial trabeculations over the lateral and apical regions, with deep inter-trabecular recesses that communicated with the left ventricular cavity (arrows) are shown; and (c) blood flow into the recesses was demonstrable on colour Doppler

* LA left atrium
† LV left ventricle
‡ RA right atrium
§ RV right ventricle

regions of the LV appeared hypokinetic. Overall, LV systolic function was normal, with an estimated LV ejection fraction (LVEF) of 65%. There was evidence of LV diastolic dysfunction ('pseudonormal filling pattern'). The right ventricle (RV) was structurally normal. Coronary angiography showed significant stenosis (70%) in the mid-left anterior descending (LAD) artery that was treated with percutaneous angioplasty and stenting.

The prominent trabeculations of IVNC were obvious on left ventriculography (Fig 2), which also showed inferior hypokinesia. Estimated LVEF from ventriculography was 59%. Left ventricular end-diastolic pressure was elevated (16 mm Hg), confirming the diastolic dysfunction evident at echocardiography. Ventricular tachycardia was found to be inducible with ventricular-programmed stimulation in a subsequent electrophysiological study.

There were two further episodes of VT requiring cardioversion during the patient's hospital stay. Implantation of ICD was performed. She was repeatedly defibrillated during the next 10 days before her condition stabilised with combination therapy of amiodarone 200 mg daily and mexiletine 200 mg twice daily. Anticoagulation was not given because the patient had a co-existing problem of gastrointestinal bleeding. She remained well when seen for follow-up 1 year post-discharge. Family screening was offered, but was not undertaken as the patient did not have any first-degree relatives resident in Hong Kong.

Discussion

Pathogenesis

During a period of normal embryonic development, the myocardium exists as a loose meshwork of interwoven myocardial fibres that form trabeculae with deep inter-trabecular recesses. Through a process of compaction of the myocardium, the inter-trabecular recesses are transformed into the coronary circulation. Embryonic arrest of this process leads to the persistence of prominent myocardial

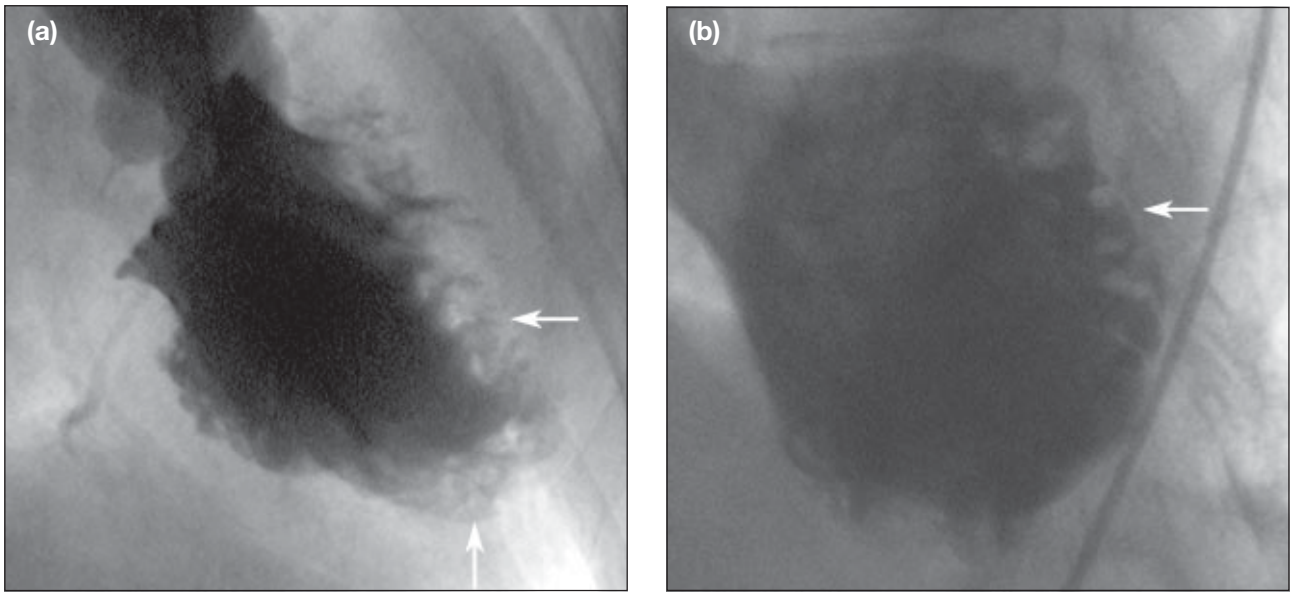

Fig 2. Left ventriculography showing the prominent trabeculations of isolated ventricular non-compaction
(a) Right anterior oblique projection; and (b) left anterior oblique projection. Prominent myocardial trabeculations can be seen over the lateral and apical regions of the left ventricle (arrows)

trabeculations, with deep inter-trabecular recesses that communicate with the ventricular cavity, a condition termed ventricular non-compaction.[1-3] In early reports, the condition was described in association with other congenital heart diseases such as obstructive lesions of the LV and the RV that cause intraventricular pressure overload.[1,4] In the absence of co-existing congenital heart abnormalities, the condition is termed IVNC. The cause of IVNC has not been elucidated, and both sporadic and familial cases have been described.[1,5,6] The genetic basis for IVNC has recently been identified.[7] Currently, the report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of Cardiomyopathies has categorised non-compacted myocardium as an unclassified cardiomyopathy.[8] With increasing recognition of this disease entity, its classification as a distinct cardiomyopathy has recently been suggested.[3,9]

Clinical manifestation

Isolated ventricular non-compaction may affect both sexes, although there is a male preponderance.[1,4] Early case series consisted largely of paediatric patients,[1,6] but there have been more recent reports of this condition in the adult population.[4,5,10] This patient represents the oldest patient at diagnosis currently reported. Patients may be asymptomatic but are identified when investigations are performed for family screening or for other reasons such as an abnormal ECG.[1,6] Clinical manifestations, if present, can include CHF, arrhythmia, and cardioembolic events.[1,4,5] Congestive heart failure is the most common presenting condition. In the largest case series to date, comprising 34 adult patients, heart failure was the reason for referral for 21 (62%) patients, with 12 (35%) patients in New York Heart Association classes III to IV.[4] Congestive heart failure can be a result of either systolic or diastolic ventricular dysfunction. Diastolic dysfunction is probably a result of the abnormal ventricular trabecular structure causing impaired ventricular relaxation and filling. The cause of systolic dysfunction is less clear. Chronic myocardial ischaemia due to coronary micro-circulatory dysfunction has been recently suggested as a possible mechanism.[11]

Arrhythmia, both tachyarrhythmia and conduction disorders, can occur in IVNC.[1,4,6] This patient had chronic AF, and presented with sustained VT. Oechslin et al[4] reported chronic AF in 26% of patients at presentation. Ventricular tachycardia, including sustained and non-sustained VT, was also observed in 41% of patients in that case series during follow-up. However, sustained VT leading to the diagnosis of IVNC, as seen in this patient, is rare.[1,4,12] Ventricular tachycardia usually occurs in association with poor LV systolic function,[5] and may contribute to sudden death. A single patient may have VT with different rates and morphologies.[12] This patient also had co-existing coronary artery disease (LAD artery disease) as a potential cause of VT. Clinical differentiation of the cause of VT is difficult. The absence of clinical features of acute coronary syndrome or previous myocardial infarction in the LAD territory, and the occurrence of inducible and spontaneous VT after revascularisation, suggest the underlying structural abnormalities as the likely cause, however. Cardioembolic events may complicate IVNC as a result of both AF and thrombus formation within the deep inter-trabecular recesses due to stagnant flow.[1,4] These may have been predisposing factors for this patient's past stroke.

Investigations and diagnosis

Echocardiographic findings are often diagnostic, but depend on clinician awareness of this rare disease entity. The affected myocardium shows a thick non-compacted endocardial layer, and a thinner compact epicardial layer.[1,3-6,13] A maximal end-systolic ratio of non-compacted layer to

compacted layer of two or more is considered diagnostic. The thick non-compacted layer consists of prominent trabeculations and deep inter-trabecular recesses, with direct blood flow from the ventricular cavity into the recesses demonstrable on colour Doppler imaging. The diagnosis of IVNC can be made in the presence of these echocardiographic features, in the absence of co-existing congenital structural abnormalities. The LV is primarily affected. The apical, lateral, and inferior segments are commonly involved. The RV may also be affected, but differentiation from normal RV trabeculations may be difficult. Left ventricle systolic and diastolic dysfunction is commonly present. Systolic dysfunction can occur in both non-compacted and normally compacted segments.[3,4] The morphological appearance of IVNC has also been described on angiographic,[6,10] computed tomographic,[14] and magnetic resonance imaging.[6]

Management and prognosis

The treatment of IVNC is directed at the patient's symptoms and complications. Patients with heart failure can be managed with usual medical therapy, and heart transplantation may be considered. Anti-arrhythmic medications are indicated for various arrhythmias. For patients presenting with sustained VT, as for this patient, a more aggressive approach is needed because of the risk of sudden death, and ICD implantation is indicated.[15] Anticoagulant therapy is required for patients with AF or a history of embolic events. Some researchers have also recommended its use even in the absence of such a history because of a high prevalence of thromboembolic events observed.[4,5] Because of possible familial occurrence, echocardiographic screening of first-degree relatives is also recommended.[1,5,6] With respect to prognosis, symptomatic patients carry a high risk of morbidity and mortality.[1,4-6] In one series, the event-free survival (in the absence of a combined end-point for death and heart transplantation) was 58% at 5 years.[4] For patients diagnosed at the asymptomatic stage, the short- to medium-term prognosis is more favourable, but progressive ventricular dysfunction is common.[6]

Conclusion

Isolated ventricular non-compaction is a rare congenital cardiomyopathy, with heart failure, arrhythmia, or cardioembolic events as the common presenting conditions. Distinct morphological features can be seen at echocardiography and are diagnostic of the disease. This case serves to alert clinicians to this rare disease entity.

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