

Isolated Non-compaction of Ventricular Myocardium: A Report of Three Cases

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Abstract

Introduction: Isolated non-compaction of ventricular myocardium (INVM) is a rare disorder of myocardial morphogenesis in the absence of other cardiac anomalies. Both sporadic and sex-linked recessive forms have been described. It can be identified with two-dimensional echocardiography. **Clinical Picture:** We report a series of 3 cases of adult males with INVM in June 2000. Clinical manifestations were congestive cardiac failure with severely depressed left ventricle systolic function and stroke. One patient had biventricular INVM. **Conclusion:** Early diagnosis of INVM is important as it is associated with a high incidence of cardiac failure, ventricular arrhythmia and remobilisation. Anticoagulation and screening of first-degree relatives is advisable.

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Key words: Anticoagulation, Non-compaction, Screening, Sex-linked recessive

Introduction

Non-compaction of ventricular myocardium is a rare morphogenic arrest of compaction of the ventricular myocardial fibres during intrauterine life. Usually, the left ventricle is more compact and less trabeculated than the right ventricle. Isolated non-compaction of the ventricular myocardium (INVM) refers to non-compaction of the ventricular myocardium in the absence of any congenital cardiac anomalies such as pulmonary or aortic atresia. It can be sporadic or familial. Sex-linked recessive inheritance has been demonstrated and the gene is localised to the Xq28 region.

Case Reports

All the 3 patients presented in June 2000 (Table I). All were male adults. Their ages were 33, 61 and 42 years. A complete echocardiogram was done in all patients.

Diagnoses of INVM were based on:

1. numerous and excessive prominent ventricular trabeculations; and
2. deep intertrabecular spaces communicating with ventricular cavity, as visualised on colour Doppler.

On 2D echocardiogram, the main localisation of the disease process involved the right ventricle in 1 patient and

the left ventricular apical and mid anterolateral walls in all patients. Screening echocardiogram was offered to first-degree relatives but was rejected by all.

Patients 1 and 2 with severely impaired left ventricular systolic functions presented with congestive cardiac failure. One of them had restrictive diastolic dysfunction as well. Patient 3 had antiphospholipid syndrome secondary to systemic lupus erythematosus and presented with a large stroke in the territory of the left middle cerebral artery. No intracardiac thrombus was seen in any of the 3 patients.

None of the patients had any dysmorphic facial features. No specific ECG patterns were noted.

Discussion

Non-compaction of ventricular myocardium is characterised by prominent and excessive trabeculations with deep intertrabecular spaces of the ventricles. It usually affects the left ventricle but can be biventricular.¹⁻³ It is believed to be due to an arrest in the endomyocardial morphogenesis, leading to failure of myocardial compaction. In infants and children, it has occurred in association with pulmonary or aortic atresia in the presence of an intact ventricular septum.⁴⁻⁸ In this group of patients, it was postulated that the high intraventricular pressure as a result of the congenital cardiac malformation leads to the

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TABLE I: PATIENTS' CHARACTERISTICS

Pt.	Age	Sex	LVEF	Clinical presentation	ECG	Outcome
1	33	M	13%	CCF	Poor R progression, LAE	Survive
2	61	M	22%	CCF	Sinus bradycardia, LAD, complete RBBB	Survive
3	42	M	40%	Stroke	Normal	Survive

CCF: congestive cardiac failure; LVEF: left ventricular ejection fraction; LAE: left atrial enlargement; LAD: left axis deviation; Pt: patient; R: R wave; RBBB: right bundle branch

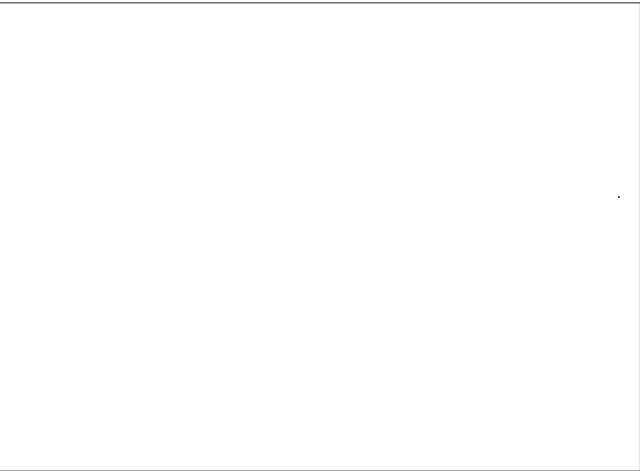


Fig. 1. Isolated noncompaction of ventricular myocardium. 2D ECG showing excessive trabeculations in the apex and anterolateral wall of the left ventricle.

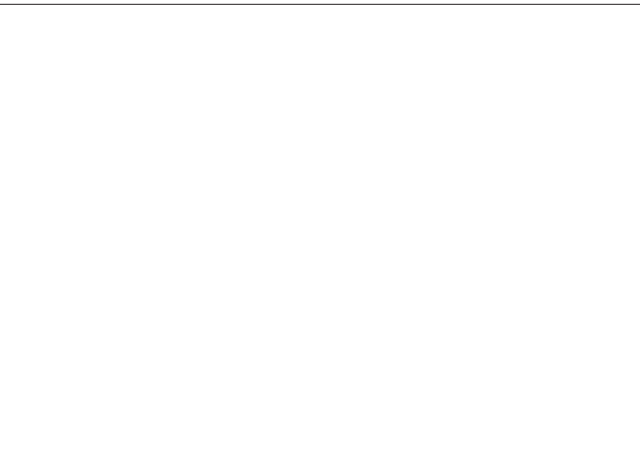


Fig. 2. M-mode tracing of left ventricle in patient 1 consistent with impaired systolic function.

arrest in myocardial compaction. However, the cause of isolated non-compaction of the ventricular myocardium, where there is no associated cardiac anomaly, remains unclear.

INVM can be sporadic or inherited by sex-linked recessive transmission. Genetic linkage has localised INVM to the Xq28 region, where other myopathies such as Barth syndromes, Becker’s muscular dystrophy and myotubular myopathy, have been located.⁹ This suggests that INVM may occur as part of the systemic myopathic process.

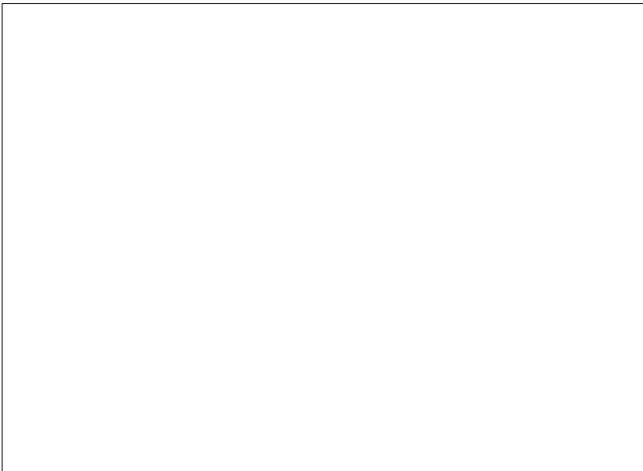


Fig. 3. Extensive trabeculations at the apices of both ventricles.

Although all the 3 patients were male, they have no features of myopathies. Unfortunately, their families refused screening.

The first large series of 8 cases of INVM, which included patients as young as 11 months, was published in 1990 by Chin et al.¹⁰ Subsequently, in 1997, Ritter et al¹ published a series of 17 adult patients collected over 10 years, demonstrating the rarity of this condition. We had 3 patients in 1 month and from anecdotal accounts, INVM may not be that rare in the Asians.

As illustrated by our patients, the clinical manifestations were similar to the above series, namely, cardiac failure with depressed systolic function and systemic embolisation. Arrhythmias, ranging from atrial fibrillation to ventricular tachycardia, and sudden cardiac death, have also been described. Arrhythmias could be secondary to impaired left ventricular function or scarring and fibrosis of the trabeculations seen on histology.

Postmortem studies have shown local thrombi within the deep intertrabecular recesses, not seen on echocardiography. INVM predisposes patients to systemic embolisation. It was speculative in patient 3 whether the stroke was cardioembolic or secondary to thrombosis from the antiphospholipid syndrome. However, it has been proposed that all patients with INVM, confirmed on echocardiogram, should be anticoagulated regardless of the presence of intracardiac thrombus.

In view of the possibility of familial occurrence, first-degree relatives of patients should be screened with an echocardiogram. 2D echocardiography and colour Doppler imaging are diagnostic in INVM. Early diagnosis of this condition is important in view of its associated high incidence of cardiac failure, arrhythmias and embolisation. Serial echocardiograms should be done to assess left ventricular function and for intracardiac thrombus.¹¹

Dysmorphic facial features such as prominent forehead, strabismus, low-set ears and micrognathia have been observed in some children in INVM.⁴⁻⁸ These were, however, not present in any of our adult patients.

Conclusion

INVM is rare. However, early recognition and follow-up are important because of its associated risk of cardiac failure, arrhythmia and embolisation. Anticoagulation and echocardiographic screening of first-degree relatives are advisable.

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