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Familial Isolated Noncompaction of the Ventricular Myocardium in Asymptomatic Phase

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Isolated noncompaction of the ventricular myocardium (INVM) is a rare cardiomyopathy resulting from a failure of normal endomyocardial embryogenesis and it has been categorized as a form of unclassified cardiomyopathy. The disorder is characterized by an excessively prominent trabecular meshwork with deep intertrabecular recesses. Although the disorder is sporadic, familial incidence may occur. Clinical symptoms and prognosis of INVM may differ markedly, and range from an asymptomatic course to a severe cardiac disability. The diagnostic method of choice for IVNM is echocardiography, which reveals multiple prominent trabeculations with deep intertrabecular spaces communicating with the left ventricular cavity in the middle and apical segments of the left ventricle. The authors report a case of INVM in a family in which three adult members (a brother and two sisters) were found to be affected by this disorder. They were all asymptomatic. The diagnosis of the disorder was made first in the 36-year-old brother by transthoracic echocardiography (TTE) and multidetector CT (MD CT), during the process of preoperative evaluation for surgical treatment of low back intervertebral herniated disc. TTE and MD CT showed similar and peculiar findings of INVM. Echocardiographic screening in all first-degree relatives of this patient, in order to identify asymptomatic patients, demonstrated INVM in two elder sisters.

Key Words: INVM, familial occurrence, echocardiography, multidetector CT

INTRODUCTION

Isolated noncompaction of the ventricular myocardium (INVM) is a rare congenital form of cardiomyopathy.[1] The disorder is sporadic; however familial incidence may occur in some patients.[2-7] Early diagnosis and correct management of INVM are crucial as the clinical manifestation is characterized by important morbidity and mortality caused by early heart failure, life threatening ventricular arrhythmias, and systemic embolic events.[7-9] However the disease is not widely known and its diagnosis mostly missed because of lack of awareness and knowledge. We report a case of familial INVM in asymptomatic phase and present a review of the literature.

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CASE REPORT

A 36-year-old man presented with low back pain and severe bilateral radicular pain which was diagnosed as an intervertebral herniated disc at the L5-S1 level. He was referred to the department of cardiology due to an abnormal electrocardiogram (ECG), which was checked in the process of preoperative evaluation for surgical treatment. He had no past history of hypertension, diabetes or syncope. He had no significant family histories. On a review of the system, he didn't present with dyspnea, dizziness, palpitation or chest pain. At the time of reference, his blood pressure was 120/80 mmHg and other vital signs were stable. The physical examination was not remarkable. The chest X-ray and routine laboratory tests were normal, but ECG revealed first degree atrioventricular block, left axis deviation and left high voltage.

Transthoracic echocardiography (TTE) was performed. M-mode echocardiography showed nor-

mal right and left ventricular cavity dimensions. Left ventricular ejection fraction was normal (LVEF: 63%). Two-dimensional echocardiography revealed a thick left ventricular wall with an inhomogeneous appearance, multiple prominent muscular trabeculations present in the left ventricular apex and mid portion of the inferior and lateral wall, numerous separated bands inserting into the anterolateral wall near the apex, focal hypokinesia of noncompacted segments and deep recesses penetrating the myocardium. In addition, the left ventricular myocardial wall was observed on parasternal short axis view to consist of two layers of epicardial compacted zone and endocardial noncompacted zone. The end systolic ratio of noncompacted / compacted zone (N/C ratio) was 1.86 / 0.71=2.62. Color-flow imaging showed communication between intertrabecular spaces and the left ventricular cavity (Fig. 1).

These findings were compatible with the diagnosis of INVM. There were relaxation abnormalities of diastolic function. All cardiac valves appeared normal and there were no findings of any coexisting congenital lesion. Exercise testing and 24-h ECG monitoring were performed, since there is always the risk of serious arrhythmia in patients with INVM, and no significant supraventricular or ventricular arrhythmia was detected. Multidetector computed tomography (MD CT), performed to more closely investigate the endomyocardial morphology, showed wall thickening, multiple prominent muscular trabeculations, deep intertrabecular spaces without mural thrombi and numerous separated bands in the left ventricular apex and mid portion of the inferior and lateral wall of the left ventricle (Fig. 2). Left heart catheterization was performed to rule out concomitant coronary artery anomaly. Coronary arteriography

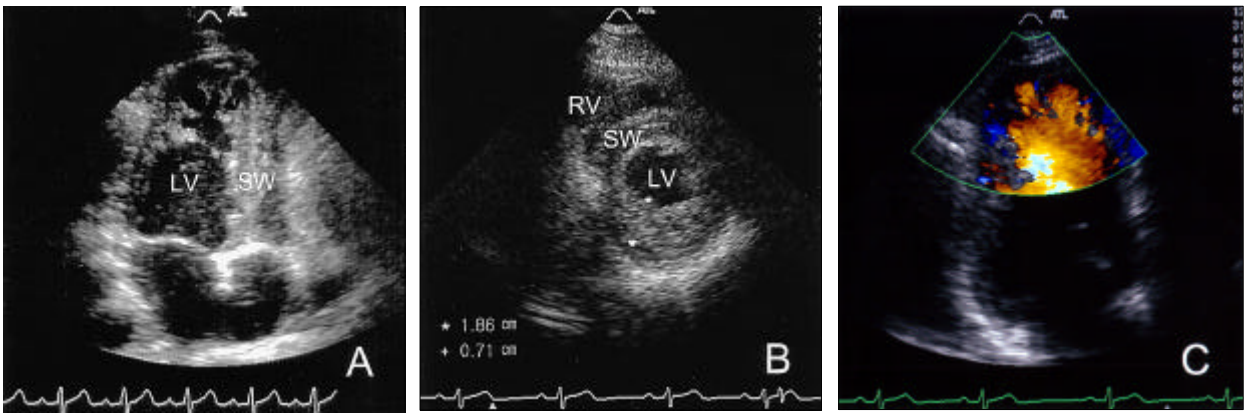

Fig. 1. Transthoracic echocardiogram (TTE) of the proband. (A) Apical 4-chamber view shows prominent left ventricular trabeculations with multiple deep intertrabecular recesses. (B) Parasternal short axis view shows a thin compacted epicardial layer and a thickened endocardial layer at end systole. (C) Color-flow imaging shows communication between intertrabecular spaces and the left ventricular cavity. LV, left ventricle; RV, right ventricle; SW, septum wall.

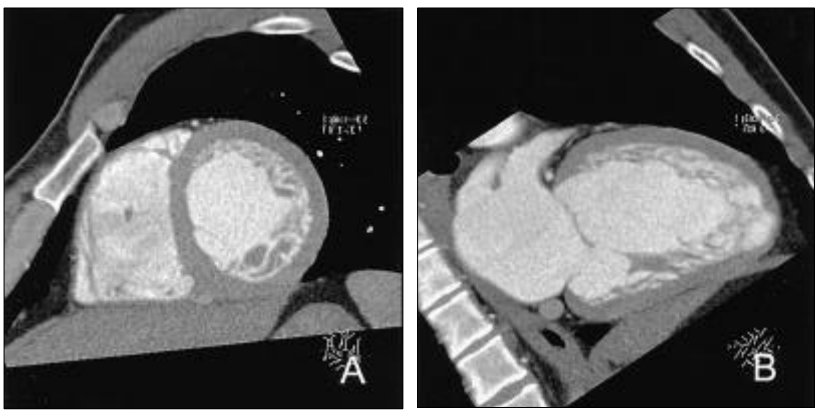

Fig. 2. Multidetector computed tomography (MD CT) of the proband. (A) Postcontrast coronal computed tomography (CT) section at the level of apex to mid-portion of the left ventricle shows thick left ventricular wall with an inhomogeneous appearance, multiple prominent muscular trabeculations and deep intertrabecular recesses. (B) Postcontrast midsagittal CT section shows a peculiar appearance characterized by the presence of numerous separated bands inserting in to the anterior wall near the apex.

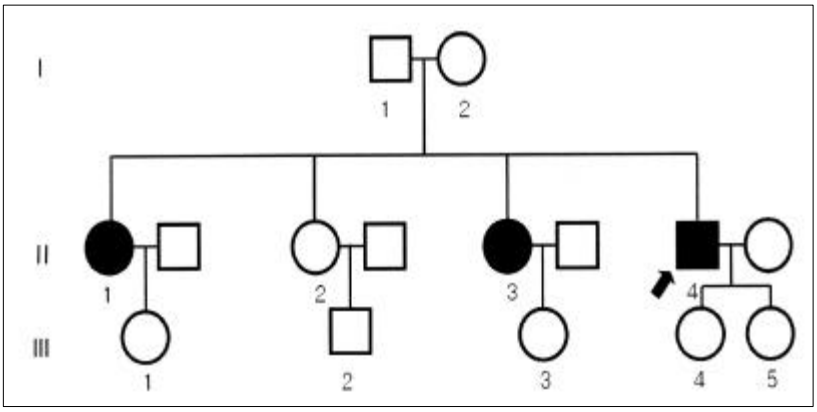

Fig. 3. Three-generation pedigree of the affected family. Generation number is listed along the left margin, and individuals are numbered underneath each symbol. Thus, the proband (indicated by an arrow) corresponds to Patient II-4. Solid-shaded symbols correspond to affected members.

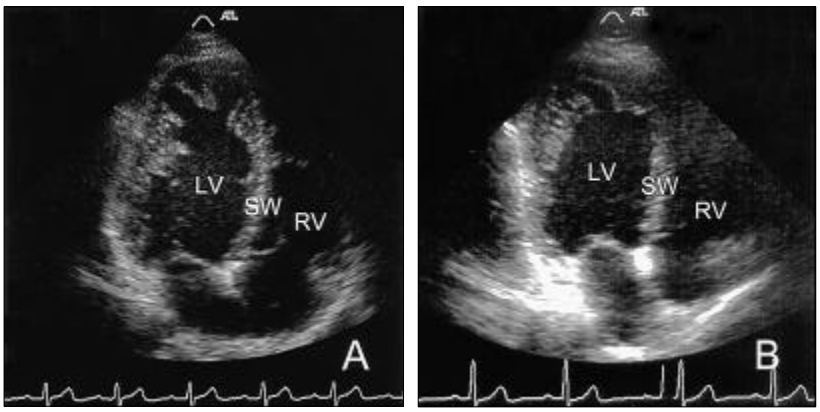

Fig. 4. Transthoracic echocardiograms (TTE) of the affected other family members. (A) Apical 3-chamber view of Patient II-1 and (B) apical 4-chamber view of Patient II-3 show the peculiar appearance of INVM, characterized by prominent trabeculations and deep recesses. LV, left ventricle; RV, right ventricle; SW, septum wall.

was normal and left ventriculography showed normal LV global systolic function (LVEF: 60%) with hypokinesia of the apex and irregular endocardial surface. Echocardiographic screening of the other members of the family over three-generations demonstrated INVM in two elder sisters (Fig. 3). The peculiar echocardiographic findings of INVM were demonstrated in them without any symptom (Fig. 4).

DISCUSSION

INVM is a rare congenital form of cardiomyopathy resulting from an intrauterine arrest in the normal process of trabecular compaction of the myocardium.[1] The disorder is sporadic; however in some patients it may be due to chromosomal abnormalities and familial incidence may occur.[2-7] The familial form has been observed in 40 - 50% of cases in the pediatric population[9] and in about 18% in the adult population[7] with INVM. In our case, INVM was demonstrated in three adult members incidentally.

INVM was previously named as persistent myocardial sinusoid or spongy myocardium, and was reported in the past in patients with the concomitant presence of congenital obstructive lesions of the left and right ventricular outflow tract.[10-13] INVM was grouped under unclassified cardiomyopathies by the World Health Organization in 1996.[14] The frequent clinical manifestations of INVM are heart failure, ventricular arrhythmias, cardioembolic events and sudden cardiac death.[7-9] The most frequent symptom of the disease is heart failure, both systolic and diastolic, mainly of the left ventricular origin. Clinical symptoms and the onset of the disorder may differ markedly, so the prognosis of patients with INVM ranges from a prolonged and completely asymptomatic course to a severe cardiac disability, leading to heart transplantation and death.

In the course of ten months of follow-up, our patient and his sisters remained asymptomatic.

Mortality in INVM is high after having entered the symptomatic phase and some patients die due to dangerous ventricular arrhythmias. Echocardiography is the method of choice to diagnose IVNM. The peculiar echocardiographic finding of IVNM is a two-layered structure of the ventricular myocardial wall consisting of a thin, compact, epicardial layer and a thick, noncompact, endocardial layer, with prominent trabeculations and deep intertrabecular recesses, which is lined with thick endocardium and communicating with the left ventricular cavity in the apical and mid-ventricular segments of both the inferior and lateral wall of the left ventricle, rather than with other segments, thereby sparing the left ventricular base in the absence of other congenital or acquired heart disease.[7] Excessive trabeculations have been reported in 68% of normal hearts and can also be observed in hypertrophic hearts secondary to valvular, hypertrophic or dilated cardiomyopathy (DCM).[15,16]

Oechslin et al.[7] and Jenni et al.[17] proposed the introduction of the end systolic ratio of the noncompacted to compacted layers of > 2 as being diagnostic for INVM as allowing distinct differentiation from hypertrophic cardiomyopathy and DCM. Multiplane transesophageal echocardiography (MTE) may be used when transthoracic studies cannot reliably exclude other processes such as apical hypertrophic or infiltrative cardiomyopathy and apical thrombus.[18] In addition, one report described the use of contrast echocardiography with sonicated albumin in a patient with INVM. Contrast echocardiography may be helpful when standard echocardiographic image quality is limited or the diagnosis is questionable.[19]

Although echocardiography has been the diagnostic test of choice for INVM, other modalities have been used for diagnosis, including contrast ventriculography,[20,21] computed tomography,[21-23] and magnetic resonance imaging (MRI).[24,25] The superior resolution of MD CT over MTE enabled the diagnosis of INVM to be confirmed and other pathologies of myocardium to be ruled out. Therefore, cardiac tomography should be considered the secondary diagnostic modality for evaluating patients with high clinical suspicion of INVM.

In conclusion, although INVM is an extremely rare disorder, its diagnosis is not difficult if physicians are aware of its possibility. Thus, careful examination using TTE is indicated to ascertain clinical diagnosis of INVM, and MTE and cardiac tomography, by MRI or MD CT, seem to be useful examinations as complementary tools when the diagnosis is not certain. When INVM is demonstrated, family screening of first-degree relatives of the patient is mandatory to identify asymptomatic patients, since the disorder is often familial. In addition, careful, long-term follow-up is obviously needed because mortality in INVM is high after having entered the symptomatic phase, even though a patient may have remained asymptomatic for a long time.

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