Isolated noncompaction of the ventricular myocardium

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Noncompaction of the ventricular myocardium is a rare congenital disorder characterized by the presence of numerous prominent trabeculations and deep intertrabecular recessess which communicate with the left ventricular cavity. The disease uniformly affects the left ventricle, and sometimes also affects the right ventricle. Echocardiographic findings are important clues for the diagnosis. Clinical symptoms include signs of left ventricular systolic dysfunction even to the point of heart failure, ventricular arrhythmias, and embolic events. We describe an illustrative case of isolated noncompaction of the left ventricular myocardium in a two-year-old child with the typical clinical and echocardiographic features of the disease. The literature on the topic is reviewed.

Key words: noncompaction, myocardium.

Noncompaction of ventricular myocardium (NCVM) is a rare disorder of endomyocardial morphogenesis[1]. NCVM refers to the arrest of compaction of loosely interwoven meshwork of myocardial fiber during embryogenesis[2]. CVM is normally more complete in the left ventricle than in the right ventricular myocardium[3]. This congenital disease is a distinct entity and should be classified as noncompaction among the unclassified cardiomyopathies[2]. Clinical manifestations include depressed left ventricular function, ventricular arrhythmias, and systemic embolization. We describe herein a child with NCVM and review the literature.

Case Report

A two-year-old infant was admitted to our hospital with heart failure. The medical and family histories were unremarkable. On physcial examination, his weight was 12,600 g and his height was 85 cm. He had normal facial features. Heart rate was 110 beat/min and blood pressure was 110/70 mmHg. The cardiac examination revealed a slightly increased apical impulse. There were soft holosystolic murmurs at tricuspid and mitral areas. The electrocardiogram (ECG) demonstrated biatrial enlargement and left ventricular hypertrophy. Chest roentgenography showed cardiomegaly and prominent ventricular markings. An echocardiographic examination revealed massive dilatation of the left atrium with normal left ventricular size and function. There was prominent trabeculation of the left ventricular wall with deep intertrabecular spaces and mild concentric left ventricular hypertrophy. There was second degree mitral regurgitation. The right ventricle was hypertrophic and there was second degree tricuspid regurgitation (Fig. 1). In Doppler echocardiographic evaluation, we found short mitral deceleration time, pulmonary vein atrial reversal consistently lasting longer than the mitral. A wave, and increased velocity and duration of pulmonary vein atrial reversal. The ventricular filling patterns were consistent with restriction, and the diagnosis of a restrictive cardiomyopathy of unknown etiology was made. Cardiac catheterization disclosed increased left ventricular end-diastolic pressure and pulmonary hypertension. Left ventriculography demonstrated the sponge-like appearance of the noncompacted ventricular wall during the diastolic phase and masked retention of the contrast medium in the trabecular recesses during the systolic phase (Fig. 2).

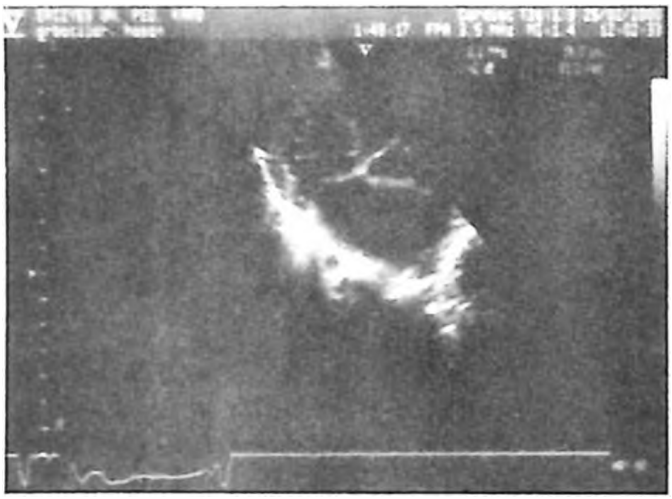

Fig. 1. Apical long axis view of the left ventricle (transthoracic echocardiography). Spongy myocardial appearance resulting from abundant myocardial trabeculations and intertrabecular recesses.

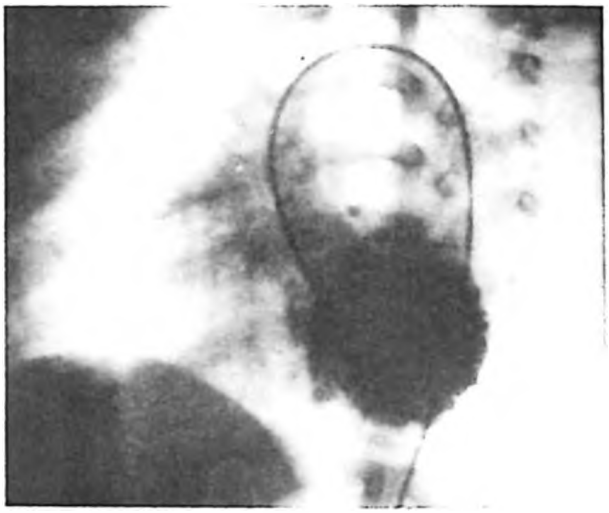

Fig. 2. Left ventriculography showing the sponge-like appearance of the noncompacted ventricular wall during diastolic phase.

Discussion

Isolated NCVM is a rare entity of unknown etiology that is characterized by numerous trabeculations within the ventricular walls. It has been previously described as involving either both ventricles or the left ventricle alone but never just the right ventricle[3,4]. Relatively rare in any case, ventricular compaction has almost invariably been associated with other congenital cardiac malformations, including anomalous origin of the left coronary from the pulmonary trunk and obstruction to right or left ventricular trunk[3]. Our patient had isolated noncompaction of the left ventricular myocardium, not associated with other congenital heart diseases.

The ECGs of the previously reported patients showed abnormal right or left frontal QRS axis deviations, first degree atrioventricular A-V block, abnormal p waves, intraventricular conduction defects of the left ventricle, right bundle branch block, and various arrhythmias. Ventricular conduction abnormality may develop later in life and could lead to progressive endocardial fibrosis in NCVM[3-5]. There were abnormal p waves in our patient.

Familial recurrence has been reported to occur more often in the pediatric population than in adults. A large family with six patients with isolated NCVM was reported by Bley et al.[6]. All reported cases were male, strongly suggesting x-linked recessive inheritance of this disorder[6,7]. In our patient, familial recurrence was not present.

Although the echocardiographic characteristics of numerous trabeculations and deep intertrabecular recesses have been well described and confirmed by necropsy, comparative hemodynamic properties assessed by cardiac catheterization have been reported in children[3]. Hook et al.[8] reported an exceptional case of NCVM presenting as restrictive cardiomyopathy, showing similarities to our data. Patients who are symptomatic at presentation and who follow a rapidly progressive clinical course may show hemodynamic properties similar to dilated cardiomyopathy, whereas asymptomatic patients may follow a slowly progressive course of the restrictive hemodynamic physiology, as our case demonstrated[3,4,8].

The differential diagnosis of NCVM includes the following: (a) Prominent normal myocardial trabeculation, commonly observed by echocardiography as a normal variant; (b) Hypertrophic cardiomyopathy in which ventricular hypertrophy may resemble the trabeculated myocardium of the noncompaction, as in our patient. The deep intertrabecular recesses characteristic of NCVM are, however, typically absent in hypertrophic cardiomyopathy; (c) Dilated cardiomyopathy, which may be accompanied by prominent myocardial trabeculations but to a lesser degree than in NCVM[3-5,8].

The treatment of patients with NCVM does not differ specifically from that of patients with other cardiomyopathies and is directed by the patient's symptoms and specific complications[3,4].

The prognosis of NCVM may range from a prolonged asymptomatic course to a fulminant course of progressive heart failure, leading to heart transplantation or death. Prognosis is worse in patients with symptoms than in patients without symptoms[3,4].

We conclude that isolated NCVM is a rare if not unique disorder with characteristic morphological features that can be identified by two-dimensional echocardiography. Proper recognization of this pathological entity is mandatory for adequae diagnosis and appropriate management and follow-up of this infrequently recognized cardiomyopathy.

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