

A different type of cardiomyopathy: ventricular noncompaction (evaluation of 8 cases)

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SUMMARY: Öztunç F, Babaoğlu K, Saltık L, Eroğlu AG, Ahunbay G. A different type of cardiomyopathy: ventricular noncompaction (evaluation of 8 cases). Turk J Pediatr 2006; 48: 209-212.

Ventricular noncompaction, characterized by numerous, prominent ventricular trabeculations and deep intratrabecular recesses, is thought to be due to an arrest of myocardial morphogenesis. We report eight patients with ventricular noncompaction diagnosed at our center in the previous one year. Two patients had associated congenital cardiac anomalies while the others were without coexisting cardiac abnormalities. Both ventricles were involved in one patient, only the right ventricle in one, and only the left ventricle in six patients. Seven patients had initially been diagnosed as having different types of cardiomyopathies. In conclusion, based on our limited experience, we propose that during the differential diagnosis of cardiomyopathies, ventricular noncompaction should be considered

Key words: noncompaction, cardiomyopathy.

Noncompaction of the ventricular myocardium is reported to represent an arrest in endomyocardial morphogenesis. It is characterized by numerous, excessively prominent trabeculations and deep intratrabecular recesses in the myocardium^{1,2}. The disorder has only recently been recognized as a distinct form of cardiomyopathy. We noticed that the primary diagnosis of this disease was missed or confused with other cardiomyopathies in our clinical practice.

In this report we present seven cases who were previously diagnosed as having different types of cardiomyopathies and one case previously diagnosed as complex cardiac anomaly.

Material and Methods

Patients were selected from the approximately 2,500 patients who underwent echocardiographic examination in our department during the last year. We detected eight cases (5 female, 3 male) of ventricular noncompaction and analyzed these patients. Although the age at presentation ranged from 20 days to 22 months, the age at diagnosis ranged from 8 months to 7 years.

Clinical assessments included clinical presentation and symptoms, primary diagnosis, and facial dysmorphism; personal and family history were recorded. Chest roentgenography, 12-lead electrocardiography and echocardiography were performed in all patients. Cardiac catheterization was undertaken in one patient for measurement of pulmonary vascular resistance.

Diagnosis of ventricular noncompaction was made according to the characteristic appearance of a two-layered structure of the myocardial wall consisting of a thin compacted epicardial layer and a thick noncompacted endocardial layer (ratio of noncompacted to compacted layers >2) with numerous, excessively prominent trabeculations and deep intertrabecular recesses that filled with blood from the ventricular cavity as visualized on color Doppler imaging on echocardiography as previously described³ (Fig. 1).

Results

The clinical and echocardiographic findings are presented in Table I. None of our patients' relatives had died of sudden cardiac death,

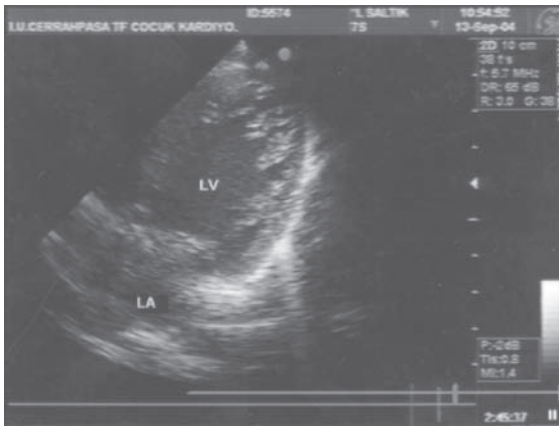


Fig. 1. Left ventricle view showing the characteristic picture of ventricular noncompaction with deep recesses and numerous trabeculations.

congestive heart failure, or stroke. There was no facial dysmorphism in any patient. All patients were admitted with signs or symptoms of heart failure at initial presentation.

The primary diagnosis of ventricular noncompaction had been missed in all of the patients. Incorrect diagnoses included dilated cardiomyopathy in five, and restrictive cardiomyopathy, hypertrophic cardiomyopathy with decreased ventricular systolic dysfunction, and atrioventricular septal defect in one case each. In the follow up, repeated echocardiographic examinations demonstrated ventricular noncompaction in four patients. The other four patients had been diagnosed as dilated cardiomyopathy at several centers and had been sent for further evaluation to our hospital later. We diagnosed ventricular noncompaction on echocardiographic examination in these patients also.

There was cardiomegaly on the chest roentgenography in all patients. Three patients showed abnormalities on baseline electrocardiography. Inverted T waves in the left precordial leads were detected in four patients (Cases 1, 4, 7 and 8) and right bundle branch block in one patient (Case 3).

Cardiac catheterization was performed in Case 3: right ventricle and pulmonary artery pressures were 90/5-15 and 90/40 (mean 50) mmHg, respectively. Pulmonary vascular resistance was 30 Wood's unit while systemic vascular resistance was 15 Wood's unit. Right ventriculography demonstrated the sponge-like

Table I. Patient Characteristics

No.	Sex	Age at presentation	Age at diagnosis	Clinical state	CTI	Echocardiography	FS (%)	Initial diagnosis	Follow-up (months)	Clinical course
1	F	16 months	17 months	Heart failure	0.60	VNC of LV, dilatation of LV	17	Dilated CMP	6	Exitus
2	M	3 months	4 years	Heart failure	0.59	VNC of LV and RV, dilatation of all cardiac spaces	25	Restrictive CMP	22	Doing well
3	M	5 years	7 years	Heart failure	0.78	VNC of RV, dilatation of RA and RV, complete atrioventricular septal defect	30	Severe PH, Complete atrioventricular septal defect	8	Exitus
4	F	20 days	6 years	Heart failure	0.54	VNC of LV, spontaneously closed VSD	20	Hypertrophic CMP, VSD	14	Doing well
5	F	4 months	8 months	Heart failure	0.61	VNC of LV, dilatation of LV	17	Dilated CMP	16	Doing well
6	F	22 months	2 years	Heart failure	0.72	VNC of LV, dilatation of LV	14	Dilated CMP	14	Doing well
7	M	20 months	2.5 years	Heart failure	0.68	VNC of LV, dilatation of LV	16	Dilated CMP	10	Doing well
8	F	9 months	3 years	Heart failure	0.54	VNC of LV, dilatation of LV	20	Dilated CMP	7	Doing well

CTI: Cardiothoracic index. VNC: Ventricular noncompaction. CMP: Cardiomyopathy. RA: Right atrium. RV: Right ventricle. LV: Left ventricle. RA: Right atrium. RV: Right ventricle. PH: Pulmonary hypertension. FS: Fractional shortening. VSD: Ventricular septal defect.

appearance of the noncompacted ventricular wall during the diastolic phase and marked retention of the contrast material in the trabecular recesses during the systolic phase (Fig. 2).

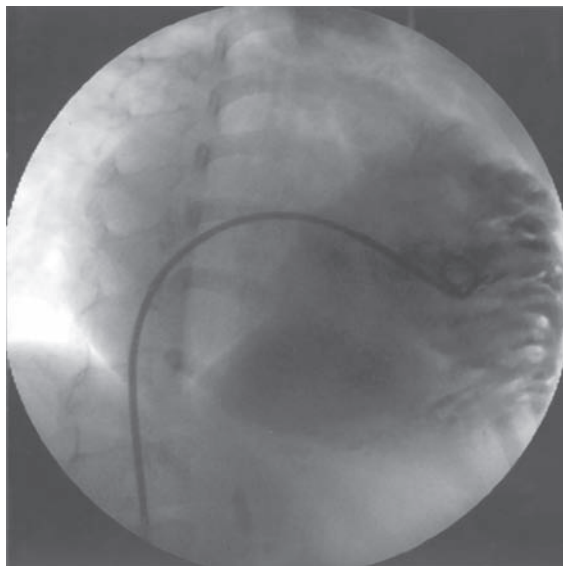


Fig. 2. Right ventriculography showing the sponge-like appearance of the noncompacted ventricular wall and marked retention of the contrast material in the trabecular recesses.

All patients were treated with anticongestive therapy such as digoxin, diuretics, angiotensin converting enzyme inhibitors and salicylate. Mean duration of follow-up was 12 months (range: 6-22 months). During follow-up, two of the patients died because of cardiac failure. The others are doing well with anticongestive therapy.

Discussion

The noncompacted ventricular myocardium, characterized by excessively prominent trabecular meshwork and deep intertrabecular recesses within the ventricular walls, is a part of normal cardiac development in the early period of embryogenesis.

Similar myocardial patterns of 'persisting sinusoids' are associated with congenital obstructive lesions of the left or right ventricular outflow tract, such as pulmonary atresia with intact ventricular septum⁴. In these patients, regression of the embryogenic sinusoids is impaired during ontogenesis by ventricular pressure overload. In contrast, isolated ventricular noncompaction is characterized

by an altered structure of the myocardial wall as a result of intrauterine arrest of myocardial morphogenesis in the absence of any coexisting structural heart disease. However, ventricular noncompaction may also coexist with simple or complex congenital heart defects⁵.

In our patients, six patients had isolated noncompaction. One patient had spontaneous resolution of ventricular septal defect and one patient had complete atrioventricular septal defect. The disorder can be readily identified in cross-sectional echocardiography by its characteristic morphological features.

Although this myocardial disease has been considered rare, recent studies have proposed that the prevalence of this cardiomyopathy is not as rare as previously reported. Nugent et al.⁶ identified 314 new cases of primary cardiomyopathy over a 10-year period. They found that dilated cardiomyopathy accounted for 58.6% of cases, hypertrophic cardiomyopathy 25.5%, restrictive cardiomyopathy 2.5% and left ventricular noncompaction 9.2% of cases, and thus noted that ventricular noncompaction is more common than previously recognized. Similarly, Pignatelli et al.⁷ found that ventricular noncompaction was responsible for 9.5% of cardiomyopathies over a five-year period⁷.

Ventricular noncompaction typically involves the left ventricle, although involvement of the right ventricle has been rarely reported to date⁸. In our study, both ventricles were involved in one case, only the left ventricle in six cases, and only the right ventricle in one case. Noncompaction of the right ventricle without involvement of the left ventricle was defined in a case with atrioventricular septal defect. In this case, right ventricular myocardium had the characteristic appearance with noncompaction. This may be secondary to ventricular pressure overload as mentioned before. Tavlı et al.⁹ defined the typical echocardiographic appearance of noncompaction of the right ventricle following Senning operation with the diagnosis of dextraposition of the great arteries.

The primary diagnosis of ventricular noncompaction was missed in most cases due to similarities between ventricular noncompaction and other cardiomyopathies and to the examiner's unfamiliarity with its specific diagnostic pattern. Ichida et al.¹⁰ evaluated 27 subjects with the diagnosis of

isolated ventricular noncompaction; they found that most patients were falsely diagnosed as dilated cardiomyopathy, hypertrophic cardiomyopathy, restrictive cardiomyopathy or myocarditis. They suggested that several echocardiographic examinations were required to diagnose ventricular noncompaction in most cases. Hook et al.¹¹ also reported that one case presented as restrictive cardiomyopathy. Alehan¹² reported that the primary diagnosis of isolated ventricular noncompaction had been missed in four of nine patients. Similarly, in our group, five patients (1, 5, 6, 7 and 8) had been initially erroneously diagnosed as having dilated cardiomyopathy and myocarditis; the 2nd and 4th patients had been followed up in our center with diagnoses of restrictive and hypertrophic cardiomyopathy.

Although ventricular noncompaction should be present at birth in all patients, ventricular dysfunction might become clinically overt during infancy, childhood, adolescence or in adulthood. Major morbidity during long-term follow-up includes heart failure, arrhythmias and thromboembolic events. Heart failure was the main cause for hospital admission in all our patients.

Ritter et al.¹³ reported that prognosis in the asymptomatic patients was clearly better than in symptomatic patients; however, Ichida et al.¹⁰ reported that most patients with a follow-up period of longer than 10 years, whether symptomatic or asymptomatic, developed ventricular dysfunction. Pignatelli⁷ showed a significant number of patients have transient recovery of function followed by later deterioration, which may account for many patients presenting as adults, some manifesting an undulating phenotype.

In conclusion, based on our limited experience and the previous reported studies, we propose that during the differential diagnosis of cardiomyopathies, ventricular noncompaction should be considered. Pediatric cardiologists should keep in mind the diagnosis and should

recognize the echo pattern of ventricular noncompaction so that the true prevalence and long-term prognosis of this disease will be better elucidated and understood in the future.

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