Myocardial noncompaction: report of three cases

Fatma Çakmak, Pelin Hacıömeroğlu, Nazlıhan Günal, Kemal Baysal
Department of Pediatrics, Ondokuz Mayıs University Faculty of Medicine, Samsun, Turkey

SUMMARY: Çakmak F, Hacıömeroğlu P, Günal N, Baysal K. Myocardial noncompaction: report of three cases. Turk J Pediatr 2007; 49: 85-88.

Myocardial noncompaction is a rare type of cardiomyopathy which can be an isolated entity or in association with other congenital heart diseases.

We present three children with myocardial noncompaction: one male with isolated left ventricular noncompaction, another with right ventricular noncompaction and dysplastic tricuspid valve, and the last with left ventricular noncompaction, ventricular septal defect and coarctation of aorta, to stress especially the different clinical forms of the disorder and the importance of early diagnosis, as it may result in a fatal outcome.

Key words: myocardial noncompaction, cardiomyopathy.

Ventricular noncompaction is a rare type of cardiomyopathy resulting from arrested myocardial development during embryogenesis[1]. The prominent myocardial trabeculations and deep intertrabecular spaces can be demonstrated by echocardiography. Although the usual site of involvement is the left ventricle, the right ventricle can rarely be affected[2]. Patients can present with heart failure, arrhythmia or embolism. Myocardial noncompaction can be diagnosed as an isolated entity or in association with other congenital heart diseases[2-4]. We describe here three illustrative cases of myocardial noncompaction: a 10-month-old male with isolated left ventricular noncompaction, a 16-day-old male with right ventricular noncompaction and dysplastic tricuspid valve, and a 12-day-old male patient with left ventricular noncompaction, ventricular septal defect (VSD), and coarctation of aorta.

Case Reports

Case 1

A 10-month-old male patient was admitted to the emergency ward with the symptoms of heart failure. The physical examination revealed tachypnea, tachycardia and a systolic murmur at the apex. Liver was palpable 4 cm at the right subcostal margin. Chest X-ray revealed cardiomegaly and bilateral parenchymal infiltration. Electrocardiogram revealed sinus rhythm with intraventricular conduction delay. On echocardiographic study, severe left ventricular dysfunction with ejection fraction (EF) of 25% and a shortening fraction (SF) of 12% were detected. Color Doppler examination showed prominent left ventricular (LV) trabeculations with perfused intertrabecular spaces. The patient was hospitalized and digoxin, diuretic treatment, acetyl salicylate (3 mg/kg) and antibiotic regimen for the bronchopneumonia were started. He was discharged after 15 days of hospitalization with recovery of infection and heart failure. Echocardiographic study on discharge demonstrated EF 42% and SF 21% but ongoing LV trabeculations and perfused intertrabecular spaces (Fig. 1).

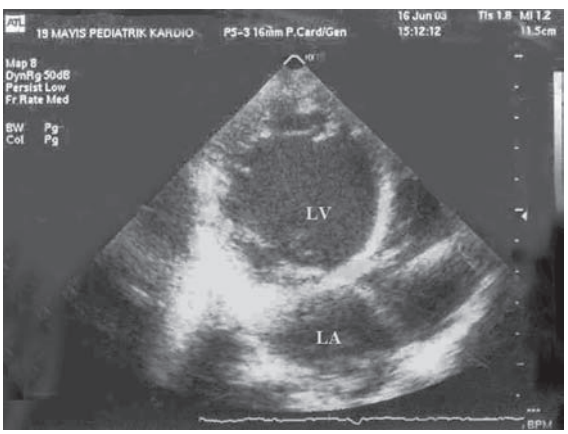

Fig. 1. Left ventricle of Case 1. Prominent left ventricular trabeculations.

Case 2

A 16-day-old male patient was evaluated in our hospital with perioral cyanosis from birth especially appearing during crying. He had tachypnea, tachycardia, intercostal retractions and a liver 3 cm palpable below the right costal margin. A 2/6 pansystolic murmur was heard along the left sternal border. Cardiomegaly was detected on the chest X-ray. Echocardiographic study demonstrated a large right ventricle with decreased systolic contractions (EF 38%, SF 26%), a muscular VSD 6 mm in diameter, and dysplastic, thick tricuspid valve leaflets. Color Doppler examination showed prominent right ventricular trabeculations with perfused intertrabecular spaces and severe tricuspid valve regurgitation. He was hospitalized and treated with digoxin and diuretic treatment. On the fifth day of hospitalization he died with a sudden cardiac arrest (Fig. 2).

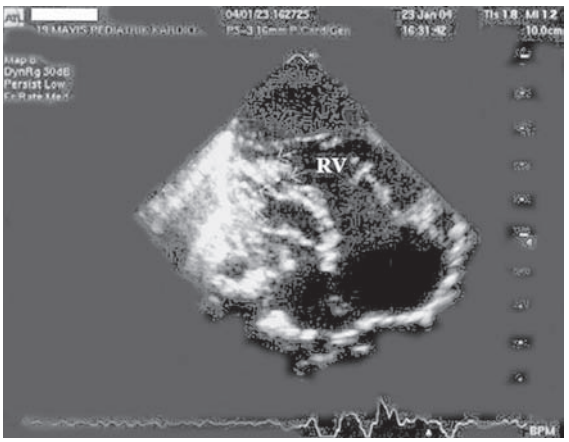

Fig. 2. Right ventricle of Case 2. Prominent right ventricular trabeculations (arrows).

Case 3

A 12-day-old male patient was admitted to the hospital with refusal of feeding and convulsions. The physical examination revealed perioral cyanosis, tachypnea, tachycardia, a liver 3 cm palpable below the right costal margin and weak femoral pulses bilaterally. A 2/6 systolic murmur was heard along the left sternal border. Cardiomegaly was detected on the chest X-ray. Blood pressures of upper and lower extremities were as follows: right arm 85/55, right leg 75/45, left arm 105/40, and left leg 65/30 mmHg. Meningitis was detected by lumbar puncture. The echocardiographic study demonstrated a large VSD, patent ductus arteriosus, aortic coarctation with segmentary narrowing of the lumen distal to the subclavian artery and pulmonary hypertension. The EF and SF were 62% and 30%, respectively. Color Doppler examination showed myocardial noncompaction (MN) with prominent LV trabeculations and perfused intertrabecular spaces. After the hemodynamic study, surgical correction for coarctation prior to VSD closure was planned. The patient was operated, ductus arteriosus was closed and end to end anastomosis was performed for coarctation. Two days after the operation, the patient died with sudden cardiac arrest in the intensive care unit.

Discussion

Myocardial noncompaction is a rare congenital disorder. The etiology is not clearly defined[5]. In the first month in utero before the development of coronary circulation, ventricles have prominent trabeculas and perfused intertrabecular spaces that nourish the fetal myocardium. Normally between the fetal 5th-8th weeks, intertrabecular spaces are obliterated and ventricular compaction occurs from the base towards the apex and from epicardium to endocardium, and an arrest in this progression of ventricular compaction results in MN[1,6,7].

The LV is the usual site of involvement, but involvement of both ventricles, and rarely isolated right ventricular (RV) noncompaction, can be seen. In our first and third cases, the LV was involved, whereas in our second case, a rare entity, the isolated involvement of the RV was demonstrated.

There are reported familial noncompaction cases in the literature. Familial cases were shown to have different mutations[8-11]. In a family with six MN cases, mutation on Xq28G4.5 was detected[8]. In another family with a member having LV noncompaction, mutation on Xq28G4.5 was shown and the mother was found to be a carrier[9]. 5q deletion was demonstrated in another family[10]. Mutation analysis was planned in our only living case. Echocardiographic screening should be planned for the first-degree relatives in familial cases[2,9]. In all of our cases, the echocardiographic study of first-degree relatives was normal.

Noncompaction can be isolated or associated with congenital heart diseases like VSD, atrial septal defect (ASD), dextrocardia, RV or LV

obstructive lesions, and cyanotic congenital heart diseases[3,4,12]. Our first case was an isolated MN, whereas the second was associated with VSD and tricuspid valve dysplasia and the third with coarctation of the aorta. To our knowledge, there is only one reported case having MN with tricuspid valve dysplasia in the literature, so our 16-day-old male patient is only the second such case.

Although MN can be demonstrated from birth, the symptoms may appear during childhood, adolescence or adulthood[2,13,14]. Patients with MN are usually admitted to the hospital with symptoms of heart failure, arrhythmias or embolism[13,14,15]. All of our cases were admitted with heart failure.

Transthoracic echocardiography (TTE) is the best diagnostic tool to demonstrate the ventricular trabeculations and perfused intertrabecular recesses. Transesophageal echocardiography, ventriculography, computed tomography, and magnetic resonance imaging are also used for diagnosis[16,17,18]. Noncompaction was diagnosed by TTE in our cases.

Prognosis may vary from a long asymptomatic course to progressive heart failure[1,15]. Patients may be asymptomatic for a long time or may deteriorate quickly, as in our second case in whom tricuspid valve dysplasia and regurgitation were thought to contribute to the progression of heart failure. If medical treatment fails, permanent pacemaker or even heart transplantation might be considered. In a retrospective study of 36 children with left ventricular MN (median age: 90 days at presentation) who were followed for 3.2 years, the authors reported that a significant number of patients had a transient recovery of cardiac functions followed by later deterioration, which might have accounted for many patients presenting as adults[19].

Here, three different MN cases, one with isolated left ventricular involvement, another with right ventricular MN together with tricuspid valve involvement, which is an exceedingly rare condition, and a third case with left ventricular involvement along with VSD and coarctation of aorta were reported to emphasize the variance of the disorder and the importance of early diagnosis, as it may result in a fatal outcome.

REFERENCES

1 Çelik S, Görgülü S, Gürol T, Dağdeviren B, Eren M, Tezel T. Miyokardiyal noncompaction: İki olgu nedeniyle derleme. Anadolu Kardiyol Derg 2003; 3: 48-53.

2. Yüksel C, Necmi A, Bilgin T, et al. Noncompaction of the ventricular myocardium: report of two cases with bicuspid aortic valve demonstrating poor prognosis and with prominent right ventricular involvement. Echocardiography 2003; 20: 379.

3. Taylor GP. Cardiovascular system. In: Dimmick JE, Kalousek DK (eds). Developmental Pathology of the Embryo and Fetus. Philadelphia: J.B. Lippincott; 1992: 121-135.

4. Özkutlu S, Ayabakan C, Çeliker A, Elshershari H. Noncompaction of ventricular myocardium: a study of twelve patients. J Am Soc Echocardiogr 2002; 15: 1523-1528.

5. Chin TK, Perloff JK, Williams RG, Jue K, Morhmann R. Isolated noncompaction of left ventricular myocardium: a study of eight cases. Circulation 1990; 82: 507-513.

6. Agmon Y, Connolly HM, Olson U, Khanderia BK, Seward JB. Noncompaction of ventricular myocardium. J Am Soc Echocardiography 1999; 12: 859-863.

7. Elshershari H, Okutan V, Çeliker A. Isolated non-compaction of ventricular myocardium. Cardiol Young 2001; 11: 472-475.

8. Bleyl SB, Mumford BR, Brown-Harrison MC, et al. Xq28-linked noncompaction of the left ventricular myocardium: prenatal diagnosis and pathologic analysis of affected individuals. Am J Med Genet 1997; 72: 257-265.

9. Fukiko I, Shinichi T, Karla RB, et al. Novel gene mutation in patients with left ventricular noncompaction or Barth syndrome. Circulation 2001; 103: 1256.

10. Pauli RM, Scheib-Wixted S, Cripe L, Izumo S, Sekhon GS. Ventricular noncompaction and distal chromosome 5q deletion. Am J Med Genet 1999; 85: 419-423.

11. D'Adamo P, Fasone L, Gedeon A, et al. The X linked gene G4.5 is responsible for different infantile dilated cardiomyopathy. Am J Hum Genet 1997; 61: 862-867.

12. Lauer RM, Fink HP, Petry EL, Dunn MI, Diehl AM. Angiographic demonstration of intramyocardial sinusoids in pulmonary valve atresia with intact ventricular septum and hypoplastic right ventricle. N Engl J Med 1964; 271: 68-72.

13. Mukhopadhyay S, Yusuf J, Rangasetty UC, Trehan V. Noncompacted left ventricle in association with dysplastic tricuspid valve. Ind Heart J 2003; 55: 382-383.

14. Hussein A, Schmaltz AA, Trowitzsch E. [Isolated abnormality "noncompaction" of the myocardium in 3 children] (German). Klin Padiatr 1999; 211: 175-178.

15. Ichida F, Hamamichi Y, Miyawaki T, et al. Clinical feature of isolated noncompaction of the ventricular myocardium: long-term clinical course, hemodynamic properties, and genetic background. J Am Coll Cardiol 1999; 34: 233-240.

16. Boyd MT, Seward JB, Tajik AJ, Edwards WD. Frequency and location of prominent left ventricular trabeculations at autopsy in 474 normal human hearts: implications for evaluation of mural thrombi by two-dimensional echocardiography. J Am Coll Cardiol 1987; 9: 323-326.

17. Conces DJ Jr, Ryan T, Tarver RD. Noncompaction of left ventricular myocardium: CT appearance. Am J Roentgenol 1991; 156: 717-718.

18. Magnus B, Ingrid J, Michael K, Gunther S, Benno H, Michael B. Cardiac imaging in isolated left ventricular noncompaction. Circulation 2002; 108: 16-17.

19. Pignatelli RH, McMahon CJ, Dreyer WJ, et al. Clinical characterization of left ventricular noncompaction in children: a relatively common form of cardiomyopathy. Circulation 2003; 108: 2672-2678.