Left ventricular noncompaction associated with Ebstein's anomaly

Ebstein anomalisine eşlik eden sol ventrikülde süngerimsi miyokart (noncompaction)

Levent Saltık, M.D.,[1] Elif Eroğlu, M.D., Fatih Bayrak, M.D., Muzaffer Değertekin, M.D.

[1]Department of Pediatric Cardiology, Cerrahpaşa Medicine Faculty of Istanbul University, İstanbul;
Department of Cardiology, Medicine Faculty of Yeditepe University, İstanbul

Left ventricular (LV) noncompaction is a congenital dysfunction of endomyocardial morphogenesis characterized by excessively prominent trabeculations and deep intratrabecular recesses of the myocardium. Its association with Ebstein's anomaly is very rare. A 13-year-old boy presented to the outpatient clinic for a cardiac evaluation before enrolling in a sports activity. On physical examination, there was a grade 1/6 systolic murmur at the tricuspid valve location. His electrocardiogram was normal. Transthoracic echocardiography revealed numerous large trabeculations and deep intratrabecular recesses at the apex, lateral wall, and the apical part of the interventricular septum. Color flow Doppler examination confirmed the presence of blood flow within the trabeculae. The size and ejection fraction of the LV was normal (65%). The attached margins of the septal and inferior leaflets of the tricuspid valve were apically displaced and there was mild tricuspid regurgitation. The distance between the mitral and tricuspid annuli was 20 mm. The right ventricle showed no dilatation and had normal systolic function, but the right atrium was moderately enlarged. As the patient was asymptomatic and the LV systolic function was preserved, he was scheduled for regular follow-up without medication except for low-dose aspirin (100 mg/day). After five years of follow-up, he was still asymptomatic with normal size and ejection fraction of both ventricles.

Key words: Child; Ebstein anomaly; echocardiography; heart defects, congenital; heart ventricles; myocardium/pathology.

Sol ventrikülde süngerimsi miyokart (noncompaction) endomiyokardiyal morfogenez sırasındaki bir duraklamadan kaynaklanan, belirgin trabeküller ve bu trabeküllerin arasında kalan derin girintilerle karakterize, doğumsal bir kalp hastalığıdır. Ebstein anomalisi ile birlikteliği çok nadirdir. On üç yaşında bir erkek çocuk, sportif bir etkinliğe katılım öncesinde kardiyak açıdan değerlendirildi. Fizik muayenede, triküspit kapaktan gelen 1/6 derecesinde sistolik üfürüm saptandı. Elektrokardiyogramı normaldi. Transtorasik ekokardiyografide, interventriküler septumun apeksinde, yan duvarında ve apikal kısmında çok sayıda büyük trabekül ve derin intratrabeküler girinti görüldü. Renkli akım Doppler incelemesinde trabeküller içinde kan akımı görüldü. Sol ventrikül boyutları ve ejeksiyon fraksiyonu (%65) normaldi. Triküspit kapağın septal ve inferior yaprakçıklarının birleştiği kenarda apikal yer değişikliği izlendi ve hafif triküspit yetersizliği vardı. Mitral ve triküspit halkalar arasındaki mesafe 20 mm ölçüldü. Sağ ventrikülde genişleme yoktu ve sistolik fonksiyonu normal idi; sağ atriyumda ise orta derece genişleme vardı. Hasta asemptomatik olduğundan ve sol ventrikül sistolik fonksiyonu korunmuş olduğundan, düzenli takip programına alındı ve düşük doz aspirin (100 mgr/gün) alması önerildi. Beş yıllık izlemi sonunda hala asemptomatik olan hastanın her iki ventrikülü normal boyutlarda ve ejeksiyon fraksiyonu normal değerler içindeydi.

Anahtar sözcükler: Çocuk; Ebstein anomalisi; ekokardiyografi; kalp defekti, doğuştan; kalp ventrikülü; miyokart/patoloji.

Noncompaction of the ventricle is a congenital dysfunction of ventricular morphogenesis characterized by excessively prominent trabeculations and deep intratrabecular recesses of the ventricular myocardium.[1] It is believed to result from premature arrest of muscle fiber compaction during early embryogenesis.[2] Left ventricular myocardium is most often affected, but biventricular noncompaction is also possible. It may occur as an isolated cardiac malformation, or in association with complex cyanotic heart disease,

Received: August 27, 2008 *Accepted:* November 12, 2008

Correspondence: Dr. Elif Eroğlu. Yeditepe Universitesi Hastanesi Kardiyoloji Bölümü, Devlet Yolu Ankara Cad., No: 102/104, 34752 Kozyatağı, İstanbul, Turkey. Tel: +90 216 - 578 47 49 e-mail: eroglu@yeditepe.edu.tr

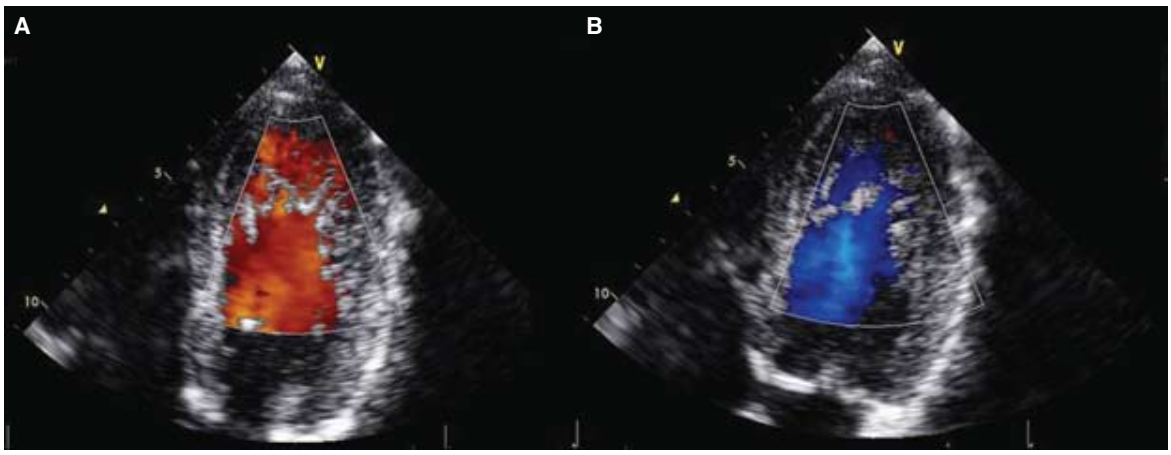

Figure 1. Color flow Doppler images of the left ventricle **(A)** at systole and **(B)** at diastole showing deep trabeculae and intratrabecular recesses with blood flow inside.

obstructive lesions of the right or left ventricle (LV), or congenital anomalies of the coronary arteries.[3] Ventricular noncompaction associated with Ebstein's malformation has rarely been reported.[4,5]

CASE REPORT

A 13-year-old boy presented to the outpatient clinic for a cardiac evaluation before enrolling in professional basketball training. His physical examination was normal except for a grade 1/6 systolic murmur at the tricuspid valve location. The 12-lead electrocardiogram was normal. Transthoracic echocardiography revealed numerous large trabeculations and deep intratrabecular recesses at the apex, lateral wall, and the apical part of the interventricular septum. Color flow Doppler examination confirmed the presence of blood flow within the trabeculae (Fig. 1). The size and ejection fraction (65%) of the LV was normal. The attached margins of the septal and inferior leaflets of the tricuspid valve were apically displaced and color flow Doppler showed mild tricuspid regurgitation. The distance between the mitral and tricuspid annuli was 20 mm (Fig. 2). The right ventricle showed no dilatation and had normal systolic function, but the right atrium was moderately enlarged. The 24-hour Holter monitoring showed normal sinus rhythm with rare isolated supraventricular premature beats. As the patient was asymptomatic and the LV systolic function was preserved, he was scheduled for regular follow-up without medication except for low-dose aspirin (100 mg/day). After five years of follow-up, he was still asymptomatic with normal size and ejection fraction of both ventricles.

DISCUSSION

Ventricular noncompaction is categorized as an unclassified cardiomyopathy by the World Health Organization report on definition and classification of cardiomyopathies.[6] In the literature, noncompaction has been described in isolation or in combination with several other congenital heart abnormalities. Although the clinical aspects of the disease are highly variable and have not completely been defined, classical clinical manifestations include heart failure, arrhythmias, and thromboembolic events.[7,8]

Echocardiography is the procedure of choice for the diagnosis of ventricular noncompaction. Typical findings include regional increase in wall thickness,

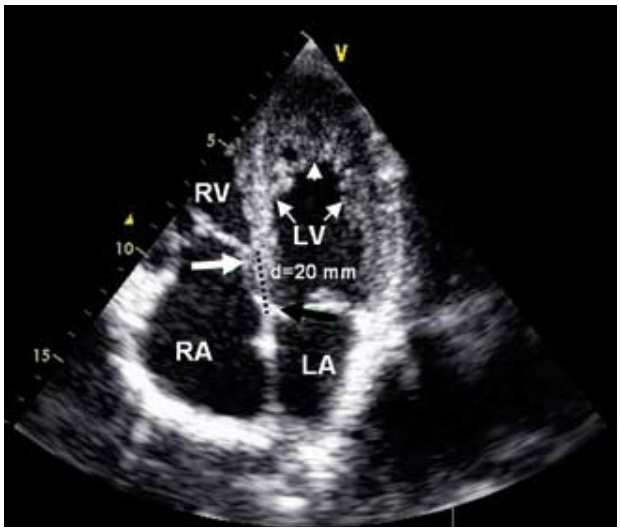

Figure 2. Left ventricular noncompaction associated with Ebstein's anomaly diagnosed by two-dimensional echocardiography. The small arrows indicate the prominent trabeculae and deep recesses at the left ventricular apex. The white and black arrows indicate the interventricular septal attachments of the tricuspid and mitral valve leaflets, respectively, and 'd' indicates the distance between these attachments. A normal distance should be less than 10 mm; however, in this case the distance was measured as 20 mm.

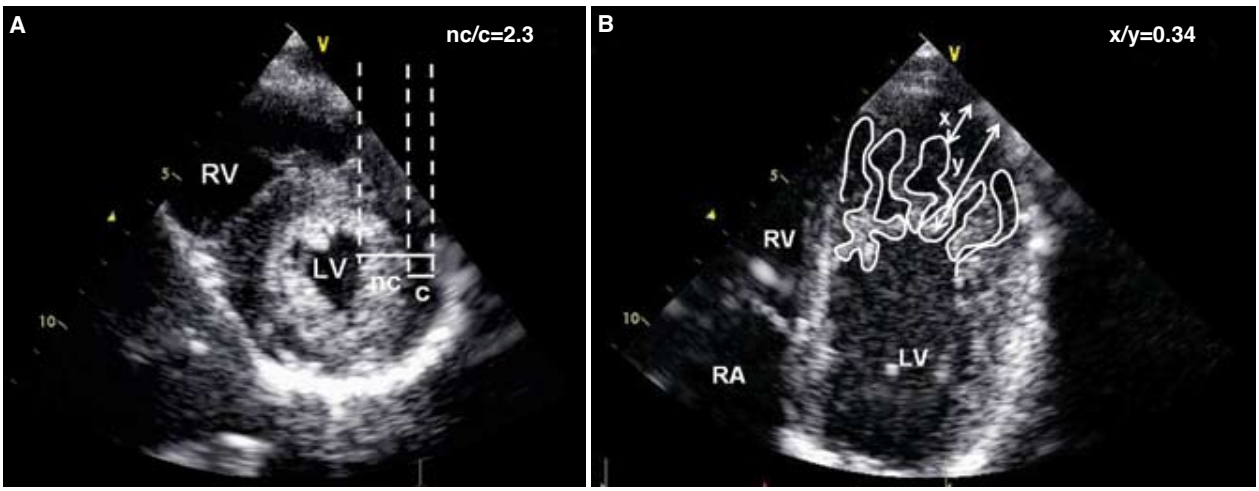

Figure 3. (A) Calculation of noncompaction ratio at end-systole. The ratio of the thicknesses of the noncompacted and compacted layers at the end of the systole is 2.3 (nc: noncompacted; c: Compacted). **(B)** Calculation of noncompaction ratio at end-diastole. X represents the distance between the epicardial surface and a trabecular recess and Y represents the distance between the epicardial surface and maximum trabeculation. The ratio is 0.34, which is diagnostic for noncompaction. nc: Noncompacted; c: Compacted LV; Left ventricle; RV: Right ventricle; LA: Left atrium; RA: Right atrium.

mainly affecting the apex and mid-apical portions of the lateral and inferior walls of the LV, evidence for direct blood flow from the ventricular cavity into deep intratrabecular recesses by color Doppler echocardiography, and a two-layered structure of the LV wall, with a thin compacted epicardial, and much thicker noncompacted endocardial zones.[7,8] In order to obtain a more accurate diagnosis, calculation of the ratio of noncompacted to compacted zones at end-systole and end-diastole is recommended. The more commonly used method is to calculate the end systolic ratio of the thicknesses of the noncompacted and compacted layers, as defined by Oechslin et al.[9] A ratio of >2 is considered to be diagnostic for noncompaction. In addition, Chin et al.[10] introduced a method of quantifying the depth of penetration of the intratrabecular recesses, through calculating the X-to-Y ratio at the diastole, where X and Y represent the distances from the epicardial surface to the trabecular recess and to the peak of the trabeculation, respectively. We used both methods for the diagnosis of LV noncompaction in our patient and illustrated the findings in Fig. 3a and 3b.

Ventricular noncompaction associated with Ebstein's anomaly is a rarely reported pathology. Attenhofer Jost et al.[4] reported three cases of left ventricular, and Sinkovec et al.[5] reported two cases of biventricular myocardial noncompaction associated with Ebstein's malformation. All these cases had LV dilatation and reduced systolic function in varying degrees. The poor prognosis in ventricular noncompaction has been associated with poor functional capacity and systolic dysfunction of LV. In contrast, patients without heart failure can remain asymptomatic for a long period without experiencing any cardiovascular events. Our patient had a favorable long-term outcome during five years; he was asymptomatic and had normal LV ejection fraction.

REFERENCES

1. Agmon Y, Connolly HM, Olson LJ, Khandheria BK, Seward JB. Noncompaction of the ventricular myocardium. J Am Soc Echocardiogr 1999;12:859-63.
2. Jenni R, Goebel N, Tartini R, Schneider J, Arbenz U, Oelz O. Persisting myocardial sinusoids of both ventricles as an isolated anomaly: echocardiographic, angiographic, and pathologic anatomical findings. Cardiovasc Intervent Radiol 1986;9:127-31.
3. Stöllberger C, Finsterer J, Blazek G. Left ventricular hypertrabeculation/noncompaction and association with additional cardiac abnormalities and neuromuscular disorders. Am J Cardiol 2002;90:899-902.
4. Attenhofer Jost CH, Connolly HM, Warnes CA, O'leary P, Tajik AJ, Pellikka PA, et al. Noncompacted myocardium in Ebstein's anomaly: initial description in three patients. J Am Soc Echocardiogr 2004;17:677-80.
5. Sinkovec M, Kozelj M, Podnar T. Familial biventricular myocardial noncompaction associated with Ebstein's malformation. Int J Cardiol 2005;102:297-302.
6. Richardson P, McKenna W, Bristow M, Maisch B, Mautner B, O'Connell J, et al. Report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of cardiomyopathies. Circulation 1996; 93:841-2.
7. Jenni R, Oechslin E, Schneider J, Attenhofer Jost

C, Kaufmann PA. Echocardiographic and pathoanatomical characteristics of isolated left ventricular noncompaction: a step towards classification as a distinct cardiomyopathy. Heart 2001;86:666-71.
8. Chrissoheris MP, Ali R, Vivas Y, Marieb M, Protopapas Z. Isolated noncompaction of the ventricular myocardium: contemporary diagnosis and management. Clin Cardiol 2007;30:156-60.
9. Oechslin EN, Attenhofer Jost CH, Rojas JR, Kaufmann PA, Jenni R. Long-term follow-up of 34 adults with isolated left ventricular noncompaction: a distinct cardiomyopathy with poor prognosis. J Am Coll Cardiol 2000; 36:493-500.
10. Chin TK, Perloff JK, Williams RG, Jue K, Mohrmann R. Isolated noncompaction of left ventricular myocardium. A study of eight cases. Circulation 1990;82:507-13.