Case Report

Isolated Left Ventricular Noncompaction in Children: Two Cases with Different Manifestations

Çocuklarda İzole Sol Ventriküler Nonkompakşın: Değişik Belirtileri Olan İki Vaka

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Abstract

Isolated noncompaction of the ventricular myocardium is an uncommon disorder characterized by an excessively prominent trabecular meshwork seen in the early period of embryogenesis. The clinical presentation of noncompaction includes a high prevalence of heart failure, thromboembolic events and arrhythmias. Echocardiography is the procedure of choice for the diagnosis of noncompaction. We report two demonstrative patients with isolated noncompaction of the left ventricular myocardium and review the available related literature.

Keywords: Cardiomyopathy, Left ventricle, Thrombi, Wolf-Parkinson-White syndrome

Özet

Ventrikül miyokardının izole nonkompakşını nadir bir hastalıktır ve embriyogenezin erken döneminde görülen aşırı trabekülasyonların doğum sonrası sebat etmesi ile karakterizedir. Hastalık sıklıkla kalp yetersizliği, tromboembolik olaylar ve aritmilerle ortaya çıkar. Tanıda ekokardiyografi kullanılır. Bu çalışmada izole sol ventriküler nonkompakşın tanısı alan iki çocuk vaka sunulacak ve mevcut literature gözden geçirilecektir.

Anahtar Kelimeler: *Kardiyomiyopati, Sol ventrikül, Trombüs, Wolf-Parkinson-White sendromu*

Introduction

Isolated noncompaction of the left ventricular myocardium (INVM) is a rare, congenital abnormality resulting from an arrest in normal endomyocardial embryogenesis. Although INVM is a congenital myocardial disorder, the onset of symptoms is frequently delayed until adulthood. Although the most frequent presentation of the disease is depressed left ventricular systolic function, primary diagnosis of INVM is missed in most cases because of the infrequent occurrence of the disease. Patients with INVM may be asymptomatic or may present with heart failure, embolism or arrhythmia [1]. Here, we report two illustrative cases of left ventricular noncompaction with different manifestations.

Case Reports

Case 1:

A two-year-old boy was admitted with complaints of dizziness, breathlessness and swelling of the lower extremities. He had been healthy two months prior to admission. His family history included the sudden death of an uncle at 16 years of age due to an unknown heart disease.

On admission, the patient had signs of congestive heart failure (CHF). No obvious cardiac murmur was heard. Chest X-ray revealed marked cardiomegaly and hilar congestion. Electrocardiography (ECG) showed a heart rate of 130 beats per minute, with a PR interval of 140 ms, a QRS duration of 80 ms and a QRS axis 180 degrees. In addition to low voltage in extremity derivations, right atrial dilation, ST segment elevation in leads V4 and V5 and T wave inversion in lead V6 were present.

Two-dimensional ECG showed markedly enlarged cardiac chambers and global left ventricular hypokinesis. Ejection fraction (EF) and fractional shortening (FS) of the left ventricle were 31% and 11%, respectively. No associated congenital heart di-sease was present. The patient was diagnosed with dilated cardiomyopathy and CHF. Anti-congestive treatment including digoxin, furosemide and enalapril was started. On the second day of admission, follow-up echo cardiography revealed two thrombi in the left ventricle, with dimensions of 12.3x8.5 and 6.8x6.1 mm (Fig. 1A). A continuous heparin infusion was started. On the second day of heparin treatment, the patient developed left-side hemiplegia, and computerized tomography of the brain showed a hemorrhagic infarct area. During follow-up, no new embolic event occurred. Repeated echocardiographic studies were performed to assess the thrombi, and numerous trabeculations with deep recesses between them on the left ventricular lateral wall and apex were noted (Fig. 1B). The right ventricular myocardium was evaluated as normal. These findings revealed a diagnosis of INVM. Signs of CHF regressed during the first week of treatment. The thrombi reduced rapidly and disappeared completely by the 17th day of treatment. Heparin was stopped, and oral warfarin was started. Twenty-four hour Holter monitoring revealed infrequent, uniform ventricular extra beats. Rare ventricular couplets were identified, but no run of ventricular tachycardia was detected.

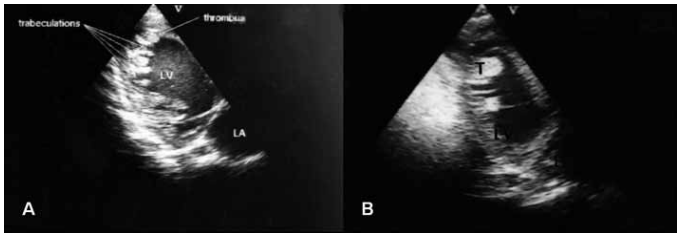

Fig. 1 — A: Two-dimensional recordings of the left ventricle show excessive trabeculations and deep recesses on the left ventricular apex in Case 1. B: Two-dimensional recordings of the left ventricle show multiple thrombi (T) between the trabeculations in Case 1. LV: left ventricle, LA: left atrium.

As of the 11th month, the patient still had signs of CHF and left hemiparesis. No new thrombus was detected. Warfarin was stopped, and aspirin was administered. Holter monitoring revealed infrequent ventricular premature beats and one ventricular triplet. Echocardiographic screenings of all members of the family were normal. Because the patient did not continue follow-up, we cannot report on his present clinical status.

Case 2:

A previously healthy 12-year-old girl was admitted to the emergency department with complaints of heart palpitation, chest pain and decrease in exercise capacity. These symptoms had started one month earlier and increased over time. In the last 10 days before admission, dyspnea and night sweating had occurred. Physical examination revealed weakness, tachycardia and weak femoral pulses bilaterally. A 2/6 systolic murmur was heard on the apex and along the left sternal border. The cardiothoracic index was 71%. The patient's electrocardiogram demonstrated characteristics of Wolf-Parkinson-White (WPW) syndrome (Fig. 2). Dilated cardiomyopathy (EF=31%, FS=12%) was detected by echocardiographic study. Color Doppler examination showed prominent left ventricular trabeculations with deep intertrabecular recesses that filled with blood from the ventricular cavity. The patient was hospitalized, and diuretic therapy (furosemide), acetyl salicylate (3 mg/kg) and captopril were administered. Holter monitoring revealed spells of supraventricular tachycardia. Amiodarone treatment was started.

The patient was discharged after seven days of hospitalization at the request of her parents. She did not continue follow-up and died at home four months after diagnosis.

Discussion

Isolated noncompaction of the left ventricular myocardium is an exceedingly rare cardiomyopathy. It is characterized by prominent and excessive trabeculation in a ventricular wall segment, with deep intertrabecular spaces perfused with blood from the ventricular cavity. Although it is a well-known pathology in adults, there are only a few case reports and series regarding children in

the literature [1].

The inheritance of INVM may be sporadic or familial. In familial cases, autosomal dominant inheritance is more common than X-linked inheritance [2]. The frequency of familial noncompaction was found to be 25% and 33% in two of the largest series [3,4]. The TAZ (G4.5) gene is located on the Xq28 chromosomal region and, thus far, is the only confirmed disease-causing locus [5]. In all of our cases, echocardiographic studies of first-degree relatives were normal. Genetic analysis was not available for our patients.

Patients with INVM may be asymptomatic or may have life-threatening ventricular arrhythmias, systemic embolization or heart failure from ventricular dysfunction. Asymptomatic patients with INVM are mostly diagnosed during school screenings or family screening of index cases. The only available data regarding long-term prognosis of these patients indicate a longer clinical course with gradually depressed left ventricular function. Some reports have suggested that asymptomatic children with conduction abnormalities and T wave changes on their ECGs must be evaluated carefully for INVM [1].

The cause of impaired left ventricular function in patients with INVM is not clear. It has been postulated that intramural perfusion, particularly subendocardial perfusion, may be adversely affected by the prominent trabeculations and deep intertrabecular recesses characteristic of INVM [6]. Four of seven patients in Chin et al.'s study and 8 of 27 patients in Ichida et al.'s study had systolic left ventricular dysfunction on admission [1,7]. Most of them were reported to have severe CHF. Patients with INVM and depressed left ventricular function have poor prognoses. Our patients were admitted with severe CHF and severely depressed left ventricular systolic functions, and the initial diagnosis was idiopathic dilated cardiomyopathy in Case 1. In fact, primary diagnosis of INVM is missed in most cases, probably due to unfamiliarity with INVM [1].

Different types of electrocardiographic abnormalities, arrhythmias and conduction disturbances have been reported in children with INVM. In two series of children with INVM, ECG abnormalities were present in 81% of cases [1]. The most frequent abnormality was ventricular arrhythmia, ranging from isolated uniform ventricular extra beats, as in Case 1, to sustained ventricular tachycardia and ventricular fibrillation, which may be fatal [1,6,7]. Another frequent association is the presence of WPW syndrome, as in Case 2 [7].

The endomyocardial morphology of left ventricular noncompaction lends itself to the development of mural thrombi within the deep intertrabecular recesses, and depressed left ventricular systolic function facilitates such thrombus development. Embolic events are the least frequent complication of INVM in children [1,7]. In the available literature, mural thrombi have been documented by echocardiography in only one patient before an embolic event [8]. Case 1 is a good example indicating that patients with INVM may develop giant intraventricular thrombi and significant systemic embolization, thus necessitating frequent follow-up with echocardiographic studies to track newly developed mural thrombi.

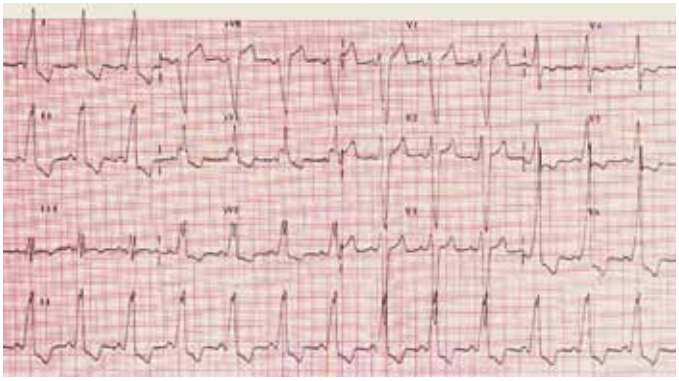

Fig. 2 — Electrocardiogram shows characteristics of Wolf-Parkinson-White syndrome in Case 2.

Echocardiography has been the diagnostic procedure of choice for INVM. Although computed tomography and magnetic resonance imaging are alternative means of diagnosing INVM, their superiority over echocardiography has not yet been definitively proven [9].

Management of the disease depends on associated comorbidities, including heart failure, thromboembolic events and arrhythmias. In cases with CHF, the treatment is similar to that of cases with other causes. Arrhythmias should be monitored at least annually by ambulatory electrocardiography. Although there are no clear suggestions for the use of internal cardiac defibrillators in the primary prevention of ventricular tachyarrhythmias, implantation of an automated defibrillator is indicated for secondary prevention [6]. Based on recent studies, anti-coagulation therapy is thought to be beneficial and necessary only in patients with significant systolic dysfunction, paroxysmal atrial arrhythmias and a history of embolic events [10]. Heart transplantation for patients with INVM and end-stage CHF has been suggested.

In conclusion, in children with signs of dilated cardiomyopathy, left ventricular noncompaction should be considered. We believe that the frequency of the disease among patients with dilated cardiomyopathy is higher than suggested, and in these patients, one must be alert for thrombus formation, embolic events and arrhythmias. Such patients should be evaluated with frequently repeated echocardiographic studies for thrombus formation and with Holter electrocardiography for arrhythmias.

Conflict interest statement The authors declare that they have no conflict of interest to the publication of this article.

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