Biventricular noncompaction cardiomyopathy with severe systolic and diastolic dysfunction in a systemic sclerosis patient

Sistemik sklerozlu hastada ciddi sistolik ve diyastolik fonksiyon bozukluğuna neden olan biventriküler nonkompaksiyon kardiyomiyopati

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Summary– Non-compaction cardiomyopathy (NCM) is a rare congenital cardiomyopathy characterized by deep increased trabeculation in one or more segments of the ventricle. The apical segment of the left ventricle is most commonly affected, but left ventricular basal segment, biventricular involvement or right ventricle predominance have also been described. While some neuromuscular anomalies and myopathies had been described in systemic sclerosis patients, coexistence of chronic inflammatory disorders and NCM is unclear. This paper presents a case of biventricular NCM with severe systolic and diastolic dysfunction in a 40-year-old female diffuse cutaneous systemic sclerosis patient.

Özet– Nonkompaksiyon kardiyomiyopati ventrikülde artmış derin trabekülasyonlarla karakterize nadir görülen doğumsal bir kardiyomiyopatidir. Sol ventrikülün apeksi en sık etkilenen segmenttir ancak sol ventrikül bazal segmenti, biventriküler tutulum ve sağ ventrikülün baskın olarak tutulduğu olgularda tanımlanmıştır. Sistemik sklerozda bazı nöromüsküler anomaliler ve miyopatiler tanımlanmıştır. Ancak kronik enflamatuvar hastalıklarla nonkompaksiyon kardiyomyopati birlikteliği net değildir. Biz bu yazıda 40 yaşında diffüz kutanöz sistemik sklerozu olan bir kadında ciddi sistolik ve diyastolik fonksiyon bozukluğuna neden olan biventriküler nonkompaksiyon kardiyomiyopatili olguyu sunduk.

Non-compaction cardiomyopathy (NCM) is a rare congenital cardiomyopathy. It is characterized by deep increased trabeculation in one or more segments of the ventricle.[1] The left ventricle apical segment is the most commonly affected area, while left ventricular basal segment, biventricular involvement or right ventricle (RV) predominance have also been described.[2] While NCM has been described in association with some muscular dystrophies, glycogen storage disease and neuromuscular disease, the coexistence of chronic inflammatory disorders and NCM is unclear.

Abbreviations:

NCM	*Non-compaction cardiomyopathy*
RV	*Right ventricle*
SS	*Systemic sclerosis*

In this report is presented a case of biventricular NCM with severe systolic and diastolic dysfunction in a diffuse cutaneous systemic sclerosis patient.

CASE REPORT

A 40-year-old woman was admitted to the rheumatology outpatient clinic with dyspnea and finger cyanosis. She had sclerodactyly and Raynaud's phenomenon. She was anti-Scl 70 positive and anticentromere negative. C reactive protein was measured as 8 mg/L. Diffuse cutaneous systemic sclerosis (SSc) was diagnosed based on the results of her examinations and tests. Cardiomegaly was discovered during routine examinations in telecardiography. She was referred to our clinic for cardiological evaluation.

There was no history of sudden cardiac death or heart failure in her family. She presented with a worsening dyspnea and decreased exercise tolerance of three months duration. Electrocardiography (ECG) showed biventricular hypertrophy criteria, sinus

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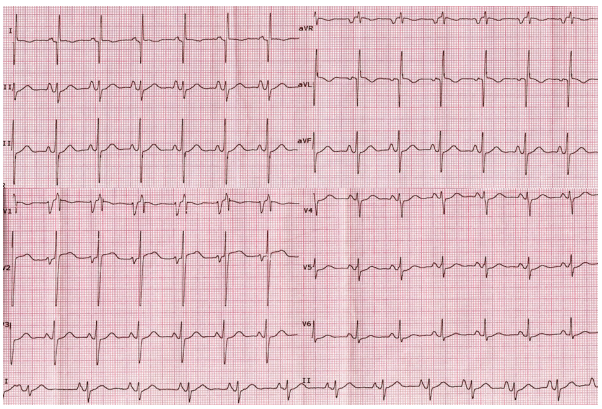

Figure 1. Electrocardiography showing biventricular hypertrophy criteria, sinus rhythm and biatrial enlargement.

rhythm and biatrial enlargement (Figure 1). Anterior posterior chest radiography revealed cardiomegaly and increased vascular. Arterial blood pressure was 120/60 mmHg, heart rate was 82 bpm and regular. A 2/6 grade systolic murmur was heard over the apical region. She also had mild ankle edema.

Transthoracic echocardiography revealed a severely dilated left ventricle with an ejection fraction of 25% (Figure 2a, Video 1*). Parasternal short-axis and apical four- chamber view showed deep increased trabeculation in the left and right ventricle (Figure 2b, c and Video 2, 3*) and blood flow in these recesses was revealed by color Doppler (Figure 2d). The ratio of non-compacted to compacted part was >2.0 measured at the end systole. Diastolic dysfunction was seen on the tissue Doppler image with septal E' velocity (Figure 3a). In addition, there was right atrial and ventricular enlargement, moderate tricuspid regurgitation and pulmonary hypertension (peak systolic pulmonary artery pressure 35+10 mmHg) (Figure 3b). Tricuspid annular plane systolic excursion was measured as 10 mm. We recommended hospitalization for optimizing of medication and assessment for intracardiac defibrillation device therapy.

DISCUSSION

First described in 1984, NCM is a cardiac pathology.[3] It occurs more frequently in males. Familial or sporadic cases of NCM and genetic association with a mutation in the G 4.5 gene and with chromosome Xq28 have been described.[4,5] Echocardiographic findings include a double layered myocardium with

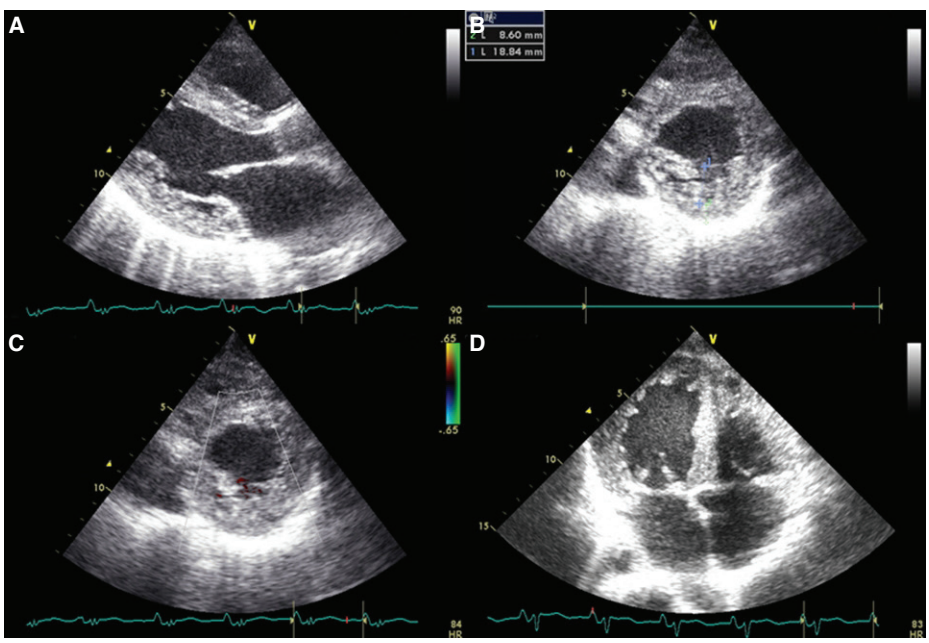

Figure 2. **(A)** Transthoracic echocardiography parasternal long-axis view showing severely dilated left ventricle and increased trabeculation in the left ventricle basal region. **(B)** Parasternal short axis-view of deep increased trabeculation in the left ventricle. **(C)** Apical four-chamber view showing right ventricle involvement. **(D)** Parasternal short-axis showing intertrabecular recesses and blood flow in these recesses on color Doppler.

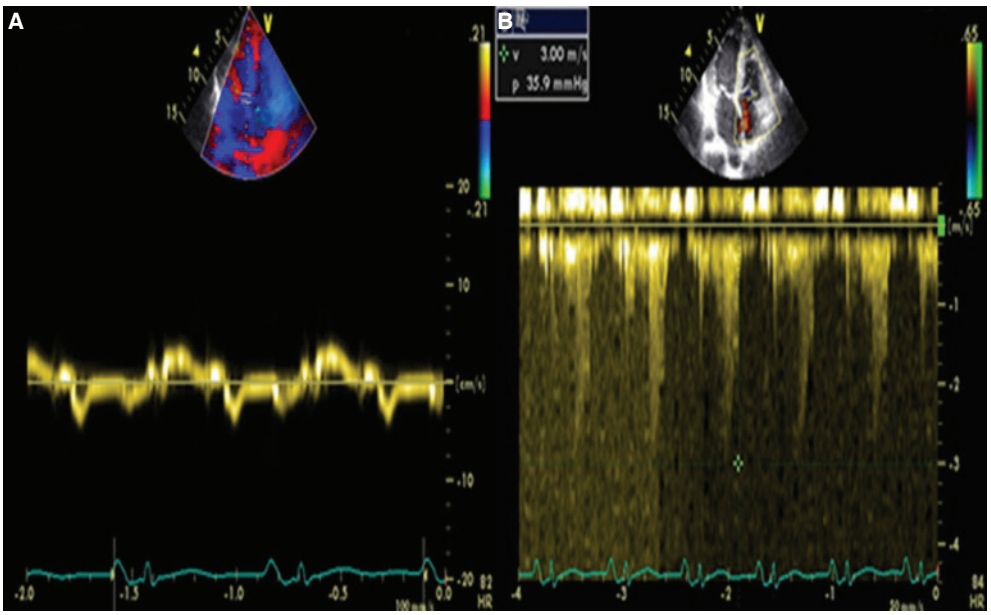

Figure 3. **(A)** Tissue Doppler image showing diastolic dysfunction. **(B)** Apical four-chamber revealing moderate tricuspid regurgitation and pulmonary hypertension by continuous wave Doppler.

a ratio of non-compacted to compacted myocardium >2 in the maximal thickened wall measured at the end systole, and multiple deep intertrabecular recesses filled with blood from the ventricular cavity.[6–8] Echocardiographic examination is usually sufficient for diagnosis. Besides contrast ventriculography, tomography and magnetic resonance imaging (MRI) are the alternative tools in diagnosis. The apical side of left ventricle is the most affected site of non-compaction, but RV involvement has been reported in some cases.[9] However, visualization of the RV is difficult because of its irregular shape.

Clinical manifestations are very heterogeneous and include heart failure, systemic embolic events and arrhythmias. Therapy options are adopted according to general clinical practice recommendations. Ventricular assist devices and heart transplantations are indicated for patients with symptoms refractory to conventional therapies.

NCM has been described in association with myotonic dystrophies, oculopharyngeal muscular dystrophy, tropomyosin-1 mutations, Danon disease, mitochondrial disorders, myoadenylate deaminase deficiency, Pompe's disease, glycogen storage disease-IV, fatty acid oxidation disorder, Charcot–Marie–Tooth neuropathy, hereditary cobolamine deficiency, beta-thalassemia, poliomyelitis and Friedreich ataxia.[10] However, coexistence of chronic inflammatory disorders and NCM has been described in only a few patients. Cutaneous sclerosis has been reported in a patient with NCM and in a neuromuscular disorder patient.[11] A case of NCM in a patient with systemic lupus erythematosus has also been reported.[12] The present case had left ventricular non-compaction, myopathy, polyneuropathy, pancytopenia and eosinophilia, but there was no severe ventricular dysfunction or right ventricle involvement.

Systemic sclerosis is an autoimmune disease of multifactorial etiology, triggered by a combination of genetic and environmental factors.[13] in SSc patients, scleroderma heart disease is very common and a major risk of death. Myocarditis is a common finding in SSc patients with recent-onset cardiac involvement that is revealed histologically.[14] Primary myocardial involvement is related to repeat focal ischemic injury causing subsequent irreversible myocardial fibrosis.[15] Echocardiographic examination is the main assessment for scleroderma heart disease. Cardiovascular magnetic resonance (CMR) also demonstrates cardiac lesions in patients with symptomatic connective tissue disease with normal echocardiography.[16] Early detection of scleroderma heart disease allows for initiation of immunosuppressive treatment, thus preventing progression of cardiac damage. Successful control of

scleroderma myocarditis using immunosuppressive treatment has been shown in most patients.[17]

This case shows that systemic sclerosis may be associated with NCM. In the presence of congestive symptoms in patients with systemic sclerosis, advanced cardiac examination should be kept in mind.

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****Supplementary video files associated with this article can be found in the online version of the journal.***

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