Dilated Cardiomyopathy in a Patient with Marfan Syndrome Accompanied by Chronic Type A Aortic Dissection and Right Atrial Thrombus

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Abstract

Marfan’s syndrome (MFS) is an autosomal dominant disorder of connective tissue involving musculoskeletal, cardiovascular and ocular systems. Aortic disease is the leading cause of mortality in MFS. Among all, dilated cardiomyopathy in the absence of severe valvular dysfunction is a very rare cardiovascular feature of MFS. We report a case of biventricular heart failure in a patient with MFS, complicated by chronic type A aortic dissection and right atrial thrombus. This report clearly highlights the importance of close cardiovascular follow-up in patients with MFS.

Key words: Marfan syndrome, dilated cardiomyopathy, aortic dissection


Introduction

Marfan’s syndrome (MFS) is an autosomal dominant disorder of connective tissue caused by mutations in the protein fibrillin 1 (FBN1). Cardiovascular involvement is the leading cause of mortality in MFS. Cardiovascular features of this syndrome include proximal ascending aortic dilatation, dilatation of the main pulmonary artery, thickening and prolapse of either mitral or tricuspid or both atrioventricular valves, calcification of the mitral annulus, and dilated cardiomyopathy in the absence of severe valvular dysfunction (1, 2). Among all, aortic involvement is the most life-threatening feature. Therefore, understanding the pathophysiology and natural history of cardiovascular disease in MFS and awareness of atypical presentations earlier in life are very critical for all clinicians.

Case Report

A 27-year-old woman with Marfan’s syndrome (MFS) presented to the emergency unit with severe dyspnea and orthopnea. On physical examination, the patient was awake and alert. Vital signs at presentation were blood pressure 150/90 mmHg, heart rate 100 bpm with regular rhythm and respiratory rate 35/min. Peripheral edema, abdominal ascites and jugular venous distention were rather evident. On auscultation, inspiratory crackles were heard at both lung bases and the middle zones and expirium was prolonged. Electrocardiogram demonstrated sinus rhythm with no other abnormalities. Chest X-ray showed widening of the upper mediastinum with an increased cardiothoracic ratio. Her laboratory test results were normal. With intravenous diuretics, vasodilators and nasal oxygen, symptomatic relief was achieved. Transthoracic echocardiography (TTE) revealed biventricular dilation. Left ventricular ejection fraction was 20 percent with left ventricular end diastolic (LVEDD) and end systolic (LVESD) dimensions of 6.2 cm and 5.6 cm, respectively (Fig. 1A). The right ventricle was severely dilated and globally hypokinetic. Tricuspid annular plane systolic excursion (TAPSE) was severely impaired (9 mm). Tricuspid regurgitation was moderate and systolic pulmonary arterial pressure was measured as 50 mmHg. The mitral valve leaflets were mildly thickened, but there was no prolapse and the degree of mitral regurgitation was mild. Aortic root and the ascending aorta were dilated and an intimal flap was extending
from the aortic valve to the descending aorta (Fig. 1B-1D). There was only a mild aortic regurgitation. Modified parasternal short and long axis views showed a right atrial mass with diameters of 2.5×2 cm consistent with a thrombus. There was also a severe degree of spontaneous echo contrast in the right atrium (Fig. 1C, 1D). However, there was no filling defect in the pulmonary artery suggestive of an embolus on echocardiography. Following the surgical consultation, we decided to manage the patient conservatively, as the surgery imposed a significant risk with high peri-operative mortality. During inhospital stay, her clinical condition was stabilized. She was listed for heart transplantation.

**Discussion**

Dilated cardiomyopathy in the absence of severe valvular dysfunction is a rare feature of MFS (2). Although the criteria for dilated cardiomyopathy may not be always fulfilled, left ventricular dilation in the absence of severe valvular dysfunction seems to be present in a small group of MFS patients (3). It has been hypothesized that it may be due to either a fibrillin defect in the myocardium or increased aortic wall stiffness leading to increased LV afterload and associated LV dilation (4, 5). In our patient, the valvular insufficiencies were not severe enough to cause such a severe heart failure. Besides, there was no finding indicating an either a static or a dynamic coronary artery obstruction secondary to the dissection flap. Other significant clinical disorders known to affect the myocardial function, such as diabetes mellitus, anemia, thyroid disorders, renal or hepatic impairment were all evaluated and excluded. So we attributed this left ventricular dilation primarily to the MFS. It has also been mentioned that not only the left, but also the right ventricular (RV) systolic function may be primarily impaired in MFS. Using conventional echocardiography and tissue Dop-
Figure 2. Electrocardiogram demonstrating sinus rhythm with heart rate of 101 bpm.

Figure 3. Chest X-ray showing an increased cardiothoracic ratio with widening of the upper mediastinum.

Doppler imaging (TDI), Kiotsekoglou et al showed RV systolic dysfunction in MFS patients with a reduced dp/dt, tricuspid annular motion and peak TDI systolic velocities at the basal lateral wall (6). Both the left and the right ventricular systolic functions were rather impaired in the present patient and the thrombus with intense spontaneous echo contrast in the right atrium was probably related to the right ventricular dysfunction. Although transportation of the patient for a CT scan to completely rule out pulmonary embolism (PE) was not feasible due to the hemodynamic instability of the patient, the presence of inspiratory crackles on auscultation and the absence of a filling defect in the pulmonary artery suggestive of an embolus on echocardiography made the diagnosis of PE unlikely. We can not claim that the definite etiology of the biventricular heart failure in our patient was primarily MFS. The presence of aortic dissection restricted us from performing cardiac catheterization and endomyocardial biopsy to exclude secondary causes of heart failure such as infiltrative cardiomyopathies. In addition, a further genetic analysis was not possible. Although chronic type A dissections are not surgical emergencies in the absence of related symptoms, the majority of patients require a corrective surgery. The timing of this operation is more critical in MFS patients. Also the severe biventricular failure was rather increasing the risk of perioperative mortality in our patient. Aortic regurgitation was mild in severity and there was no symptom related to the dissection. So as an initial approach, we preferred to manage conservatively. Having stabilized her clinical condition, transplantation was considered as a choice of treatment. Clinical data on early and long-term survival following heart transplantation of even non-Marfan patients with chronic type A aortic dissection is lacking in literature. The largest group of patients (10 patients) with MFS and severe heart failure who underwent heart transplantation was reported by Knosalla et al (7); type-A aortic dissection was present in three patients and ascending aortic aneurysm was present in seven patients. They noted that transplantation in patients with MFS results in good long-term survival, similar to that of patients without MFS (7).

Conclusion

The present case is rather illustrative for the extent of cardiovascular involvement in MFS. The progression of cardiovascular disease may sometimes be rapid. Moreover, the presentations may sometimes be very atypical as seen in painless acute aortic dissections presenting later in the chronic stage. All physicians should be aware of the atypical characteristics of the cardiovascular involvement in MFS. The patients with MFS may require a closer cardiovascular follow-up.

References

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