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An Unusual Presentation of Aortic Dissection and Aneurysmatic Disease in Marfan Syndrome: A Case Report

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Abstract

Marfan's syndrome (MFS) is an autosomal dominant disorder characterized by aortic root dilation, mitral valve prolapse and aortic regurgitation. The typical aneurysm location involves the thoracic aorta (60%). Thoracoabdominal aortic dissections are quite infrequent (10%). A type A aortic dissection can have fatal complications, such as aortic rupture and cardiac tamponade, and a prompt diagnosis is crucial. The present case involves a 46-year-old patient with Marfan's syndrome who was admitted to the emergency department for dyspnea. The patient presented congestive signs and manifestations suggesting acute heart failure. An initial transthoracic echocardiogram (TTE) showed ventricular hypertrophy and diastolic dysfunction. Additionally, TTE showed plurivalvular regurgitation, pulmonary hypertension (42 mmHg), aortic dilatation, and a dissection flap. Coronary computed tomography angiography (CCTA) was performed, confirming the suspicion of ascending aortic dissection. The patient underwent a three-stage intervention for the repair of all the defects, with adequate recovery. This was a rare presentation of aortic dissection in MFS, which was a diagnostic challenge for the heart team.

Keywords

Marfan-Syndrome, Aortic-Dissection, Heart-Failure

1. Introduction

Marfan's syndrome (MFS) is an autosomal dominant disease with an estimated

prevalence of 1 in 10,000 to 20,000 individuals [1]. Most fatal events associated with untreated MFS occur in early adulthood; the average age of death is estimated at 32 years [2].

The main clinical manifestations of MFS are long bone overgrowth, dislocation of the lens eye, and aortic root aneurysm; the aortic root dilation may be present in 60%, but the thoracoabdominal aorta is involved only in 10%, and aortic Stanford aortic dissection type A remains a frequent event [3] [4].

In approximately 80% of patients, sudden-onset chest pain that can radiate to the neck, jaw, and left extremity is the main manifestation, also can be accompanied by diminished or absent peripheral pulses, as well as a systolic murmur in the aortic valve. Only 6.4% of patients have an asymptomatic presentation and even fewer mimic heart failure [5] [6].

The treatment of aortic disease in MFS may include prophylactic β-blockers, calcium channel blockers, angiotensin II receptor inhibitors, and open surgical reconstruction [7]. Current guidelines call for prophylactic repair of the ascending aorta once the maximum diameter exceeds 5.0 cm in adults, or if a rapid rate of growth (>1.0 cm/year) or significant aortic regurgitation is observed [2]. The prognosis is determined by progressive aortic dilatation, dissection, and/or rupture [6].

The present case concerns a rare presentation of aortic dissection in MFS, which represented a diagnostic challenge, and the objective is to highlight the importance of multi-image assessment and adequate treatment to reduce the morbi-mortality and provide better outcomes in this pathology.

2. Observation

A 46-year-old male with a recent diagnosis of MFS and a 2-year history of ankle swelling and paroxysmal nocturnal dyspnea was admitted to the emergency department due to 3-days progressive dyspnea until it became at rest (NYHA functional class IV), without associated chest pain and medical treatment. Vital signs were within normal limits. On physical examination, jugular venous distention, and bilateral sub-crepitant rales in pulmonary fields were detected. Precordial auscultation revealed an aortic thrill and a grade IV Austin Flint murmur. Positive Quincke's, Landolfi's and Lian's signs were present.

Laboratory studies showed increased values of CK (330 ng/mL), CKMB (9.21 ng/mL), creatinine (1.7 mg/dL), TnI (63 ng/mL), NT-ProBNP (29,560 ng/mL), and CRP (2.3 mg/dL). The 12-lead electrocardiogram showed sinus rhythm, heart rate of 82 bpm, left ventricular (LV) hypertrophy, and biatrial enlargement (**Figure 1**). The chest X-ray revealed cardiomegaly with a cardiothoracic index of 0.62 and dilatation of the ascending and descending aorta.

Transthoracic echocardiography (TTE) (**Figure 2(A)**) demonstrated grade III left ventricular diastolic dysfunction, eccentric hypertrophy, normal left ventricular ejection fraction (LVEF 60%), biatrial enlargement, moderate mitral and tricuspid regurgitation, severe aortic regurgitation, and pulmonary hypertension

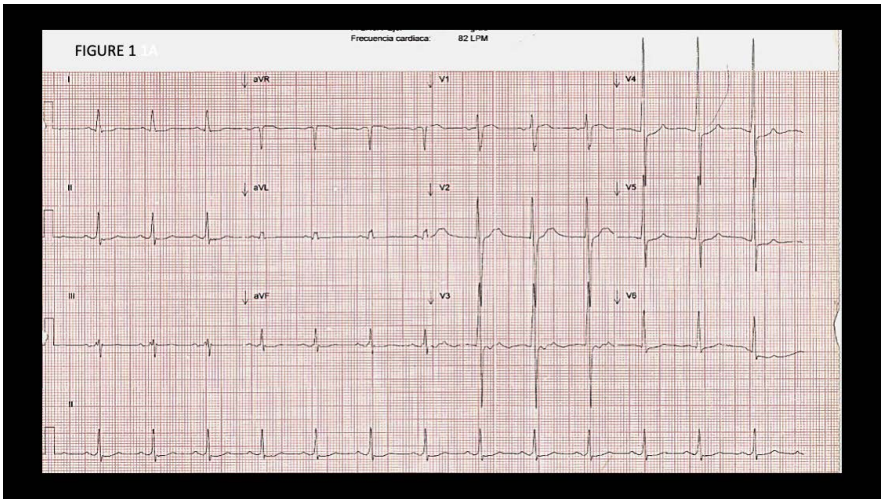

Figure 1. 12-leads electrocardiogram. Sinus rhythm, heart rate 82 bpm, left ventricular and biatrial dilatation.

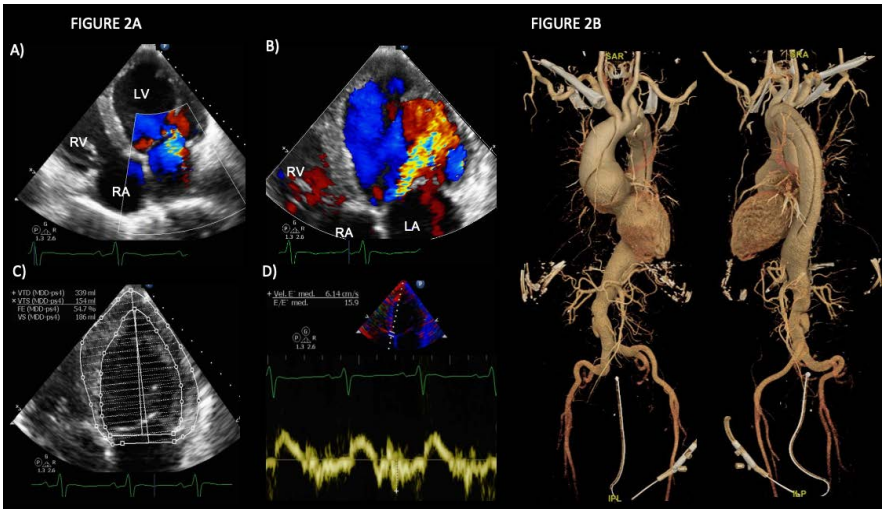

Figure 2. (A) Transthoracic echocardiography. A) Moderate mitral regurgitation (pressure half time 29.29 ms). B) Severe aortic regurgitation (pressure half time 163 ms, vena contracta 8.5 mm). C) LV eccentric hypertrophy (RWT-0.35, LVMI-290 gr/m^2) with an ejection fraction of 54.7%. D). left ventricular end-diastolic hypertension. (B) Coronary computed tomography angiography. 3D-CCTA shows a dissecting flap from the sino-tubular junction to T12, extending towards the proximal third of the brachiocephalic trunk; with a second dissection flap in the infrarenal aortic segment, with extension from L4 to S1; as well as an aneurysmal dilation of the aortic root (76 mm), sinuses of Valsalva (69 mm), sino-tubular junction (80 mm), ascending aorta (81 mm), and from the isthmus to the origin of the superior mesenteric artery (47 mm) at the level of T12 and inferior vena cava. Abbreviations: RV: right ventricle; LV: left ventricle; RA: right atrium; LA: left atrium; RWT: relative wall thickness; LVMI: left ventricular mass index.

with pulmonary systolic arterial pressure of 42 mmHg. Dilatation of the aortic annulus, Valsalva sinuses, sino-tubular junction, and ascending aorta was also documented.

CCTA (**Figure 2(B)**) showed two dissection flaps and an aneurysmal dilation

of the aortic root which were compatible with Stanford type A aortic dissection/ DeBakey I.

With these findings, the surgical team performed a Bentall and Bono procedure, including aortic and mitral valve replacement. The aortic root and aortic arch were also replaced. A woven Dacron tube was inserted for the brachiocephalic trunk, left carotid, and left subclavian arteries. The patient presented a type 5 myocardial infarction and atrial fibrillation during the surgical intervention. During the procedure, plateletpheresis, globular packages, fresh plasma, and cryoprecipitate transfusions were required. A chest X-ray and TTE were performed afterward, showing mild pericardial effusion, severe left ventricular systolic dysfunction with LVEF of 25%, and normally functioning mechanical aortic and mitral prosthetic valves (**Figure 3(A)** and **Figure 3(B)**). The pericardial effusion was treated with colchicine without any complications.

In the second and third surgical interventions, stents were placed in the ascending aorta, and resection of the femoral fusiform aneurysm was performed.

During hospitalization, the patient received medical treatment with digoxin, carvedilol, enalapril, acetylsalicylic acid, spironolactone, acenocoumarin, paracetamol, and amiodarone.

Fifty-three days after admission, the patient was hemodynamically stable with an INR of 2.49 and he was discharged.

After 24 days, the patient showed a clear improvement in his NYHA functional class. The dose of anticoagulant and carvedilol were adjusted, and amiodarone and digoxin were suspended.

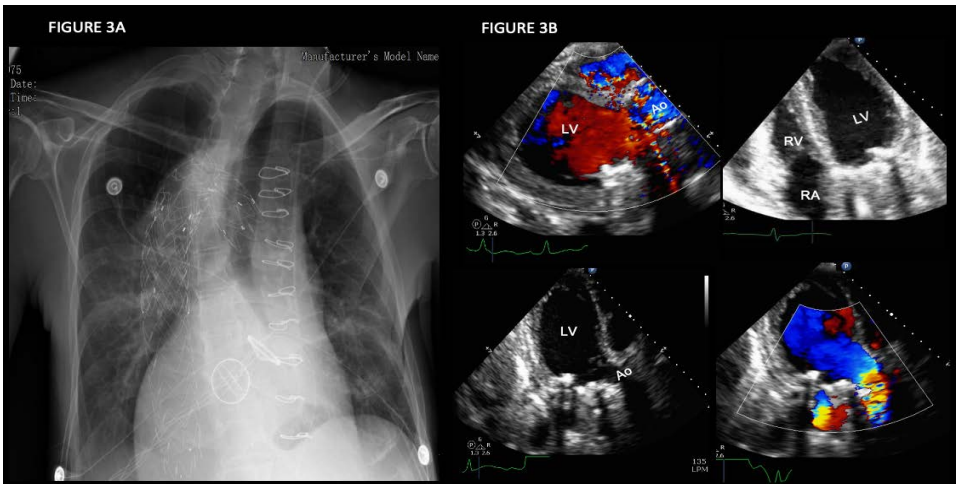

Figure 3. (A) Anteroposterior chest X-ray showing grade II cardiomegaly, endoprosthesis and coronary vascular stents in the ascending and descending thoracic aorta. Mechanical aortic and mitral prosthetic valves. (B) Transthoracic echocardiography. The mechanical mitral (mean gradient 4.9 mmHg, indexed EOA 1.46 cm^2/m^2) and aortic (mean gradient 3.9 mmHg, indexed EOA 1.46 cm^2/m^2) prosthetic valves are visualized. Also, left ventricular dilatation and hypertrophy are evident. Abbreviations: Ao: aorta, EOA: Effective orifice area. The other abbreviations as before.

3. Discussion

In MFS the most lethal presentations involve aortic root dilation and aortic dissection. The latter is found in 50% of young patients with MFS and is commonly associated with aortic regurgitation [3]. Mitral valve prolapse and dilation of the Sinuses of Valsalva have been identified as the most common cardiovascular features of MFS [8].

Typically, aneurysm formation of the ascending aorta begins in the aortic root and tapers down towards the aortic arch. Only 10% affect the thoracoabdominal aorta [1]. In the present case, multiple aneurysms were found in the sino-tubular junction, arising from the isthmus to the origin of the superior mesenteric artery, and mitral valve prolapse was not observed in them by echocardiography.

The most common clinical presentation is sudden-onset chest pain in approximately 80% of patients, followed by the discrepancy of blood pressures in the extremities, a pulse deficit, hypertension, or a diastolic murmur [3] [4]. However, 6.4% of patients are asymptomatic or have an atypical presentation, especially in type A dissections as occurred in the present case, and they are associated with higher mortality secondary to fatal complications. Therefore, physicians must have a high clinical suspicion that leads them to perform adequate paraclinical studies, especially in asymptomatic MFS [9].

Patients with high probability of aortic dissection, such as individuals with MFS, require cardiovascular imaging to identify and classify the location of the dissection and detect any valvular involvement. In aortic syndromes, CCTA is the best modality [3]. Aortic dissection in MFS usually arises from intimal tear in the proximal ascending aorta with the dissection involving the sino-tubular junction and aortic sinuses, resulting in prolapse of one or more commissures [8].

Concerning our case, intimal dissection flap, double lumen, and aortic dilation were compatible with acute aortic dissection.

The risk of rupture in cases of untreated ascending aortic dissection is estimated at 90%, with 75% of these causing hemorrhages into the pericardium, pleural space, or mediastinum. Cases of untreated dissection involving exclusively the descending aorta have a better prognosis, with mortality of 11% at one month, 16% at one year and 20% at 5 years [10].

Treatment includes both medical and surgical approaches. Medical therapy relies on adequate analgesia and maintenance of systolic blood pressure between 100 - 120 mmHg. Surgical interventions continue to evolve and include excision of the intimal tear, obliteration of the false lumen, and reconstruction of the aorta with a vascular graft [1].

Our patient met criteria for surgical replacement of the aortic root and the corresponding valves, achieving successful results [3] [9]. The risk of paraplegia in descending aortic dissections should always be considered. The most complex dissections are those affecting the aortic arch, as in our case, in which the brachiocephalic trunk was involved [3] [9].

A Bentall-type reconstruction was chosen because the sinuses of Valsalva were compromised. Bentall-type surgery in patients with Marfan syndrome shows extremely favorable results in terms of prognosis, with survivals greater than 90% at 5 years [11].

Current literature reports mortality in untreated patients with ascending aortic dissection at approximately 1% - 2% in the first hour after symptom onset, 33% within 24 hours, and 50% within 48 hours. In this patient, the follow-up demonstrated the utility of the multimodal cardiovascular approach in the diagnosis and surgical plan [6].

4. Conclusions

We report the case of a patient diagnosed with MFS and an atypical presentation of aortic dissection.

In this case, the poor NYHA functional class was attributed to severe aortic regurgitation and subsequent cardiac overload, consistent with chronic cardiac remodeling characterized by left ventricular eccentric hypertrophy on TTE.

Due to the lack of previous studies, the discovery of the dissection could be classified as an acute finding, responsible for the exacerbation of the previous cardiac manifestations.

Multimodality imaging such as TTE and CCTA were the main methods that contributed to the real diagnosis and modified the patient's prognosis.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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