Case Report

A Silent and Catastrophic Cardiac Complication in a Marfan Syndrome Patient With Blurred Vision: A Case Report

Mehrdad Jafari Fesharaki[1], MD; Shideh Anvari[1*], MD; Hadi Allahverdi Nazhand[2], MD

ABSTRACT

Marfan syndrome is an autosomal dominant genetic disorder owing to insufficient fibrillin-1 that involves connective tissue, with an incidence rate of approximately 2–3/10000 in most populations. Almost 25% of cases are caused by new mutations and are sporadic. Marfan syndrome leads to not only skeletal and ocular complications (eg, ectopia lentis and retinal detachment) but also cardiac complications, which are the most significant. Critical cardiovascular complications that can occur include mitral valve prolapse, mitral regurgitation, aortic regurgitation, aortic aneurysms, dilation of the sinus of Valsalva, aortic dissection, and rupture. The diagnosis of Marfan syndrome is based on the Ghent nosology. Since cardiac manifestations are life-threatening, they need to be diagnosed and treated promptly. The treatment of cardiac complications comprises surgical and pharmacological therapy. Here, we introduce an asymptomatic case of Marfan syndrome with blurred vision and severe cardiac manifestations discovered during cardiac assessments before eye surgery. ***(Iranian Heart Journal 2024; 25(1): 112-117)***

KEYWORDS: Marfan syndrome, Aortic dissection, Aortic aneurysm, Aortic valve regurgitation

[1] Department of Cardiology, School of Medicine Shahid Beheshti University of Medical Sciences, Tehran, IR Iran.
[2] School of Medicine, Shahid Beheshti University of Medical Sciences, Tehran, IR Iran.

*** Corresponding Author:** Shideh Anvari, MD; Department of Cardiology, School of Medicine Shahid Beheshti University of Medical Sciences, Tehran, IR Iran.
Email: drsh.anvari@sbmu.ac.ir **Tel:** +982123601

Received: April 23, 2023 **Accepted:** August 12, 2023

Marfan syndrome is an autosomal dominant genetic disorder owing to insufficient fibrillin-1 that involves connective tissue. [1] In most populations, the incidence of Marfan syndrome is about 2–3/10000, and almost a quarter of cases are sporadic. [2] Critical cardiovascular complications can occur, the most notable of which are mitral valve prolapse, mitral regurgitation, aortic regurgitation, dilation of the sinus of Valsalva, aortic aneurysms, aortic dissection, and aortic rupture. Ocular complications (eg, ectopia lentis and retinal detachment) and skeletal complications may also be observed in this disorder. [3,4] Since cardiac manifestations are life-threatening, they need to be diagnosed and treated promptly to improve patients' outcomes. Here, we describe an asymptomatic case of Marfan syndrome with severe cardiac manifestations discovered during cardiac assessments.

CASE PRESENTATION

A 35-year-old man, a known case of Marfan syndrome, came to our hospital with a sudden onset of blurred vision caused by

retinal detachment and ectopia lentis. The patient had been referred to us for preoperative cardiac assessments before surgical treatment for ocular problems. He had no history of cardiac symptoms. On cardiac examination, a prominent early decrescendo diastolic murmur, indicating severe aortic regurgitation, was heard. Echocardiographic studies demonstrated severe aortic regurgitation (Movie 1), mild-to-moderate mitral regurgitation, and an aneurysm in the ascending aorta (the maximum diameter at the tubular part ≈ 7 cm) (Movie 2). There was evidence of an aortic dissection flap immediately after the aneurysmal sinus of Valsalva (the sinus of Valsalva diameter = 6.8 cm and the aortic valve annulus = 2.7 cm) (Fig. 1 & Movie 2).

Computed tomography angiography of the thoracic and abdominal aorta revealed an aneurysm at the sinus of Valsalva (75 mm) that extended to the proximal ascending aorta (52 mm) with a dissection flap in the right sinus of Valsalva. The abdominal aorta (19 mm) and the aortic arch (26 mm) were normal (Fig. 2 & 3).

The patient underwent surgery, which involved excision of the wall of the ascending aorta and the David operation with a No. 32 tube graft. During the operation, the aortic valve was successfully repaired, and cardiopulmonary bypass was performed. In the postoperative examination, signs of cardiac complications were not observed. Additionally, echocardiography and computed tomography angiography imaging showed successful results (Fig. 4).

Surgical treatment of the ophthalmologic complications was performed following the stabilization of the patient's heart condition.

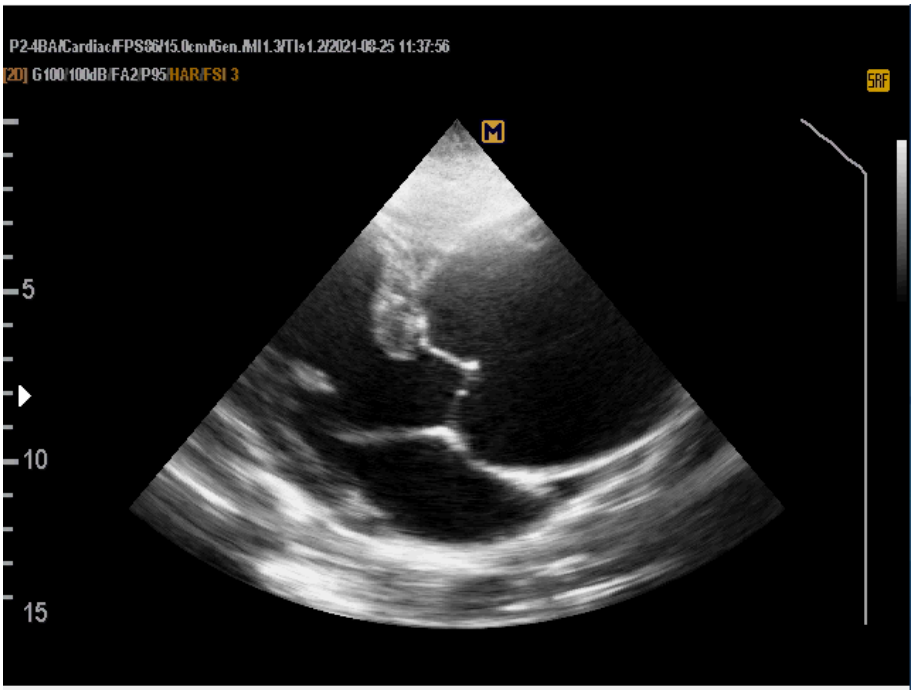

Figure 1: The image depicts an aneurysmal sinus of valsalva.

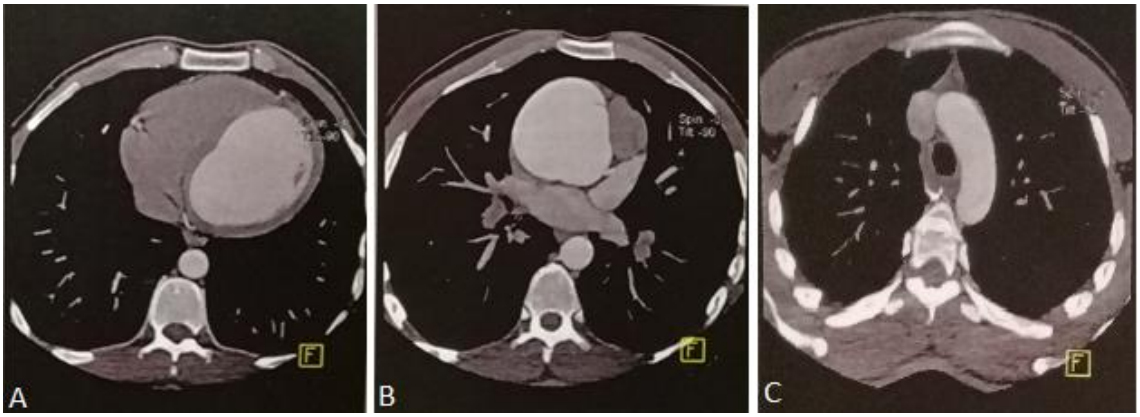

Figure 2: The images show A) aortic aneurysm and dissection at the sinus of Valsalva, B) extension of the aneurysm to the proximal ascending aorta, and C) a normal aortic arch.

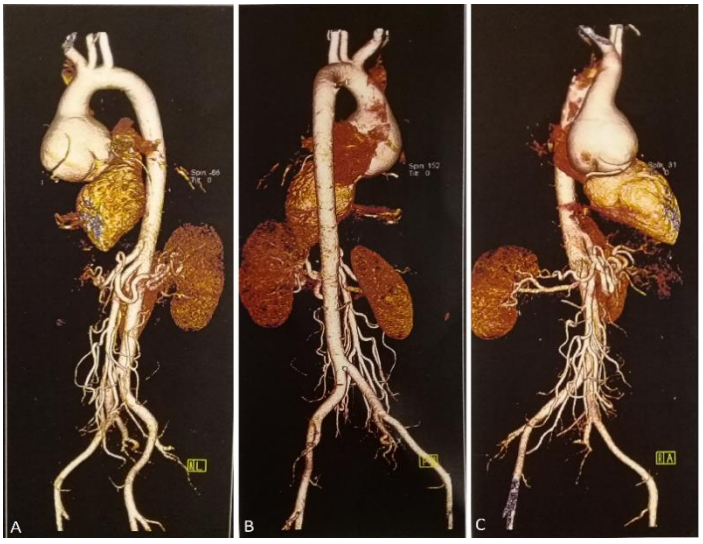

Figure 3: The images present velocity rendering technique (VRT) reconstruction: A) the left anterior view (spin = -66), B) the right posterior view (spin = 152), and C) the right anterior view (spin = 31).

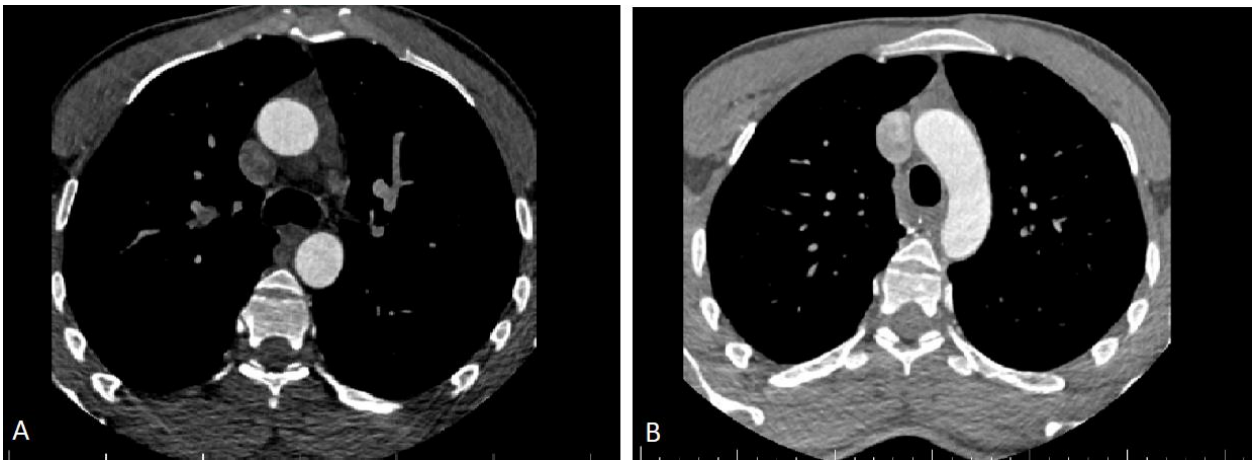

Figure 4: The postoperative images illustrate A) the ascending and descending aorta and B) the aortic arch.

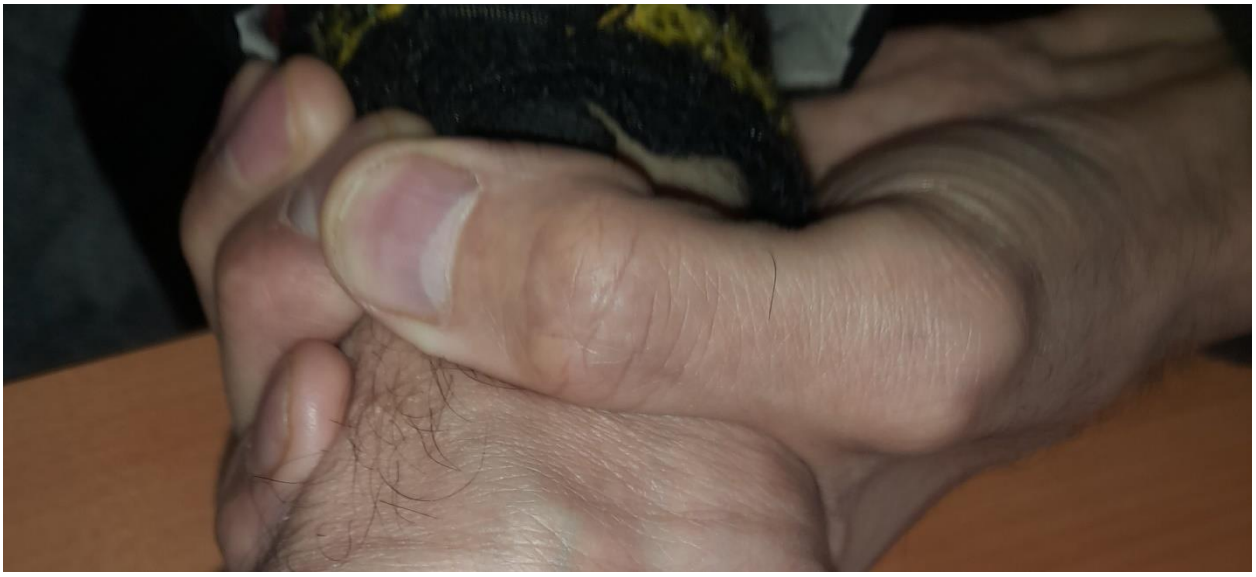

Figure 5: The image shows the patient's wrist sign.

DISCUSSION

One of the most common inherited connective tissue disorders is Marfan syndrome, which has an autosomal dominant pattern of inheritance. This disorder results in serious cardiac, ocular, and skeletal (eg, scoliosis and long limbs) complications. The diagnosis is based on the Ghent nosology (Table 1), including the criteria, systemic scores (Table 2), and gene sequencing tests. [5, 6]

The most frequent ocular involvement in Marfan syndrome is ectopia lentis, present in 50-80% of patients. Patients often present with symptoms of pain, blurred vision, and diplopia. [7] Retinal detachment is also a major ocular complication, observed in 5–11% of cases, especially in individuals with ectopia lentis. [8-10]

Cardiovascular complications are critical in these patients. Proximal aortic dilatation in patients with Marfan syndrome places them at risk of dissection. [11] Although Marfan syndrome most commonly affects the ascending aorta, the ascending and descending aorta has also been shown to be abnormal. [12-14] The diameter of the aortic root is crucial to predicting severe complications. [15, 16] Determining the size of the aorta at the level of the sinus of Valsalva is an integral component of the assessment of aortic dissection in patients with Marfan syndrome who have ascending aortic aneurysms. [17] In these individuals, in addition to the aortic diameter, the risk of dissection is associated with factors such as the spread of dilatation beyond the sinotubular junction, premature aortic dissection in the family history, high blood pressure, strenuous exercise, sleep apnea, and rapid increase of the aortic diameter. [18-23] Aortic dilatation is apparently no more common in men than in adolescent women, but the risk of dissection is greater in men, which could be due to the protective effect of the X chromosome. [24, 25] Mitral valve prolapse is also common in these patients, [26] and its prevalence increases with age. [27] Recently, heart failure and arrhythmias have been suggested as cardiovascular abnormalities in patients with Marfan syndrome. [28] Significant valvular insufficiency could lead to volume overload, left ventricular dysfunction, and heart failure. [29-31] In these patients, echocardiography plays a significant role in the diagnosis of cardiovascular involvement. [11]

Early diagnosis, regular follow-up, medical treatment, and preventive surgery dramatically reduce the risk of dissection. [11, 32]

Moreover, apart from transthoracic echocardiography, computed tomography angiography or transesophageal echocardiography could be used to diagnose aortic diseases.

Our patient had a positive family history of Marfan syndrome in his uncle and grandmother and revealed serious cardiac and ocular manifestations, in addition to a positive wrist sign (Fig. 5), indicative of this disorder. Since an aortic root diameter of at least 45 mm and high risk of aortic dissection are considered indications for surgical treatment, [3] our patient, with an aortic root diameter of 75 mm and aortic dissection, had to undergo surgery. We followed up the patient after surgery by physical examinations, echocardiography, and computed tomography angiography, which demonstrated the success of the operation. He was administered β-blockers and angiotensin-converting enzyme inhibitors as post-surgical pharmacological therapy.

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