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ASCENDING AORTIC DISEASE IN A PATIENT WITH MARFAN SYNDROME

Palabras clave: Aneurisma de la Aorta; Cirugía torácica; Aorta Torácica; Disección aórtica aguda, Marfan.

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ABSTRACT

Introduction. Acute thoracic aortic dissection is caused by a tear in the intimal lining of the aorta, and is a symptom of acute aortic syndrome. The dissection allows the blood to pass through the rupture and separates the tunica intima from the tunica media or the tunica adventitia, creating a false intravascular light. Early diagnosis directly affects the chances of survival, since it is a medical emergency that can lead to death, even with optimal treatment.

Case description. The following report presents the case of a 26-year-old man with a history of Marfan syndrome, retrosternal lancinating pain, nausea, vomiting, and medium effort dyspnea that evolved to orthopnea, perioral cyanosis, murmur of aortic insufficiency and mitral systolic murmur. Complementary studies (chest x-ray, electrocardiogram, angiography, tomography, and echocardiogram) were performed, obtaining a diagnosis of Stanford type A ascending aortic dissection. Surgical treatment was indicated to replace the aortic root using a composite prosthesis and Bentall and De Bono coronary reconstruction. During the procedure, right coronary button destructure occurred, so it was necessary to perform a venous bypass with a left internal saphenous venous hemoduct. Weaning extracorporeal circulation was achieved and then low expenditure of refractory character (despite vasopressor support at maximum dose), refractory ventricular fibrillation and asystole were observed. The patient did not recover and died as a consequence of acute transoperative myocardial infarction.

Conclusion. The treatment for ascending aortic dissection remains a therapeutic challenge. Timely diagnosis is directly related to life expectancy in patients who suffer from

this condition, hence the importance of proper diagnosis and management.

RESUMEN

Introducción. La incidencia de síndrome de Marfan que ha sido reportada a nivel mundial es de 1 en 5000 casos, de los cuales aproximadamente el 80% o más desarrollan complicaciones cardiovasculares. La disección aórtica torácica aguda requiere una rotura en la íntima de la aorta, que forma parte del síndrome aórtico agudo. En la disección, la sangre pasa a través de la rotura y separa la íntima de la media o la adventicia, lo que crea una falsa luz intravascular. Un diagnóstico temprano incide directamente en las posibilidades de supervivencia, pues se trata de una emergencia médica que puede llevar a la muerte, incluso con un tratamiento óptimo.

Descripción del caso. Se presenta el caso de un hombre de 26 años con antecedente de síndrome de Marfan, dolor retro esternal lancinante, náusea, vómito, disnea de medianos esfuerzos que evolucionó a ortopnea, cianosis peribucal, soplo de insuficiencia aórtica y soplo sistólico mitral. Se realizaron estudios complementarios (radiografía de tórax, electrocardiograma, angiografía, tomografía, ecocardiograma) y se le diagnosticó disección de aorta ascendente tipo A Stanford, por lo que se decidió iniciar tratamiento quirúrgico mediante reemplazo de la raíz aórtica mediante prótesis compuesta y reconstrucción coronaria tipo Bentall y de Bono. En el procedimiento se presentó desestructuración de botón coronario derecho, por lo que fue necesario realizar puente venoso con hemoducto venoso de safena interno izquierdo. Se logró destete de circulación extra corpórea y luego se observó bajo gasto de carácter refractario (pese a soporte vasopresor

a dosis máxima), fibrilación ventricular refractaria y asistolia; el paciente no mostró recuperación y falleció como consecuencia de un infarto agudo de miocardio transoperatorio.

Conclusión. El tratamiento de la disección aórtica de aorta ascendente sigue siendo un desafío terapéutico. El diagnóstico oportuno tiene relación directa con la esperanza de vida de quienes lo padecen, de ahí su importancia diagnóstica y su manejo.

INTRODUCTION

Marfan syndrome is a rare connective tissue disorder that equally affects men and women (autosomal dominant), with an approximate incidence rate of 1 in 5 000 people. It is characterized by an alteration in the production of fibrillin due to a mutation in the FBN1 gene of chromosome 15, which weakens the connective tissue of the body, whether tendinous, ligamentous, cartilaginous, vascular or heart valves. (1). It is worth noting that not all patients with this syndrome develop the same symptoms or severity (allelic heterogeneity) (2). Since the aorta wall is weak, a dilatation of the aorta occurs leading to an aortic aneurysm. In addition, if there is a rupture in the vascular wall, a double lumen may be formed, with the consequent passage of blood, which is called aortic dissection.

Acute thoracic aortic dissection is the result of a tear in the wall of the aorta that allows blood flow between its layers separating them, which leads to the appearance of a double lumen within the same vessel, causing the blood flow through a false lumen to exert an occlusive effect on the lumen itself and condition the flow to the vessels that derive directly from the aorta. The importance of early diag-

nosis directly affects the chances of survival, since it is a medical emergency that can lead to death, even with optimal treatment.

The Stanford classification is used to better understand aortic dissection. It is divided into types A and B. Group A includes the dissections that involve the ascending aorta, which is why they are considered to pose higher risk because the dissection and false lumen that is generated can rapidly affect the coronary ostium and compromise the blood flow to the myocardium, leading to a massive and fulminating infarction, hence the importance of timely diagnosis and appropriate surgical treatment.

In many cases, if the aortic root is dilated, and depending on the degree of the dilation, the apparatus of the aortic valve is affected, which implies a backward flow of blood to the ventricles with the consequent decrease in the volume of ventricular ejection. To compensate this anomaly, the cavities of the heart increase their size, in other words, a cardiac dilation occurs.

The estimated mortality rate for type A aortic dissection is directly related to the time of evolution, being of 1% per hour within the first 24 hours, 29% at 48 hours, 44% after a week, and reaching 50% at two weeks. Therefore, emergency surgery is the best option.

Despite the advances for early diagnosis and the innovations in surgical management, type A aortic dissection of the ascending aorta has a high morbidity and mortality both in the short term and in the long term: at the hospital level, it ranges between 15% and 35%, with a 5-year survival rate in 65-75% of cases.

Type B dissections involve the aorta posterior to the left subclavian artery, with no compromise of the ascending aorta. Usually, its management is not urgent, although it is equally

important since relevant organs may also be affected, in which case other therapeutic options should be analyzed.

CASE PRESENTATION

26-year-old male, mestizo, single patient from Guayaquil (Ecuador), with a basic level of education, a history of Marfan syndrome and hypertension in treatment, who attended consultation reporting a clinical picture of 48 hours of evolution characterized by intense lancinating retrosternal pain and medium effort dyspnea that evolved to orthopnea. The vital signs on admission obtained after physical examination were: BP: 120/50 mmHg, HR: 108/min, RR: 22/min, oxygen saturation: 95%, temperature: 35.4°C, and height: 180 cm. He presented with disproportionate upper limbs in relation to thoracoabdominal structure, pale skin, arnodactyly, perioral cyanosis, and piriform thorax with sternal depression. Auscultation

revealed systolic and diastolic murmur and mitral systolic murmur.

Complementary diagnostic exams

1. Chest x-ray: cardiomegaly, mediastinal widening at the expense of ascending aorta (Figure 1a).
2. Electrocardiogram: sinus tachycardia, left ventricular hypertrophy with signs of diastolic overload.
3. Echocardiogram: significant aortic valve regurgitation with severe dilatation of the valve ring.
4. Computed tomography angiogram of the chest: aortic root of 90.2 mm, ascending aorta of 80.8 mm, double lumen observed at aortic root level correlating with aortic dissection (intimal flap) with visible leakage of contrast material towards false lumen. Normal mitral, tricuspid and pulmonary valves; insufficient tricuspid aortic valve and increased pulmonary systolic pressure (60 mmHg) (Figures 1b and 2).

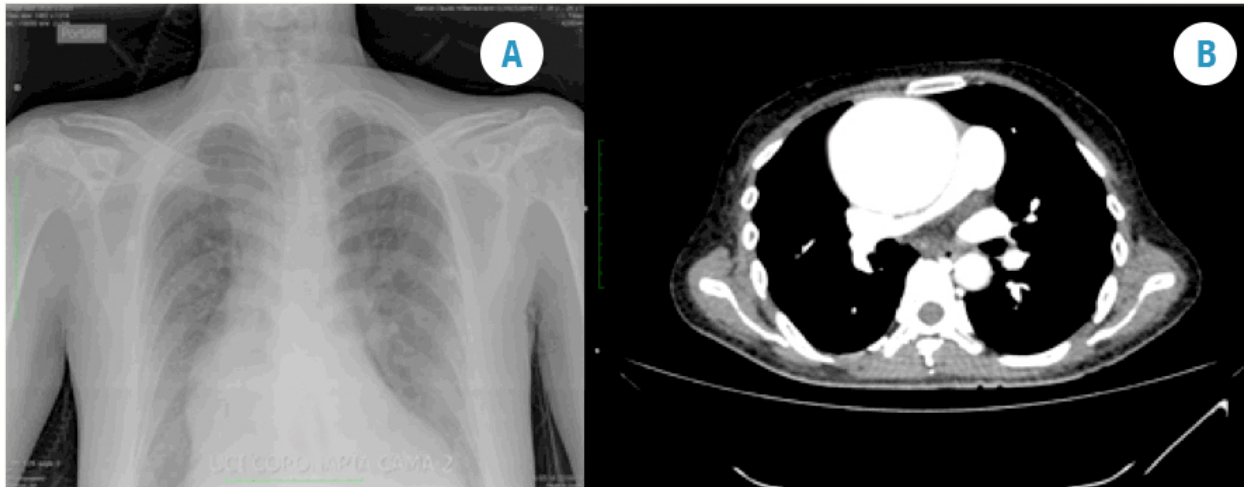


Fig 1A. Posteroanterior chest radiograph. Grade 4 cardiomegaly and mediastinal widening at the expense of ascending aorta.

Fig 1B. Computed tomography angiography: sagittal section. Aortic root of 90.2 mm, ascending aorta of 80.8 mm, insufficient tricuspid aortic valve and increased pulmonary systolic pressure (60 mmHg).

Source: Own elaboration based on the data obtained in the study.

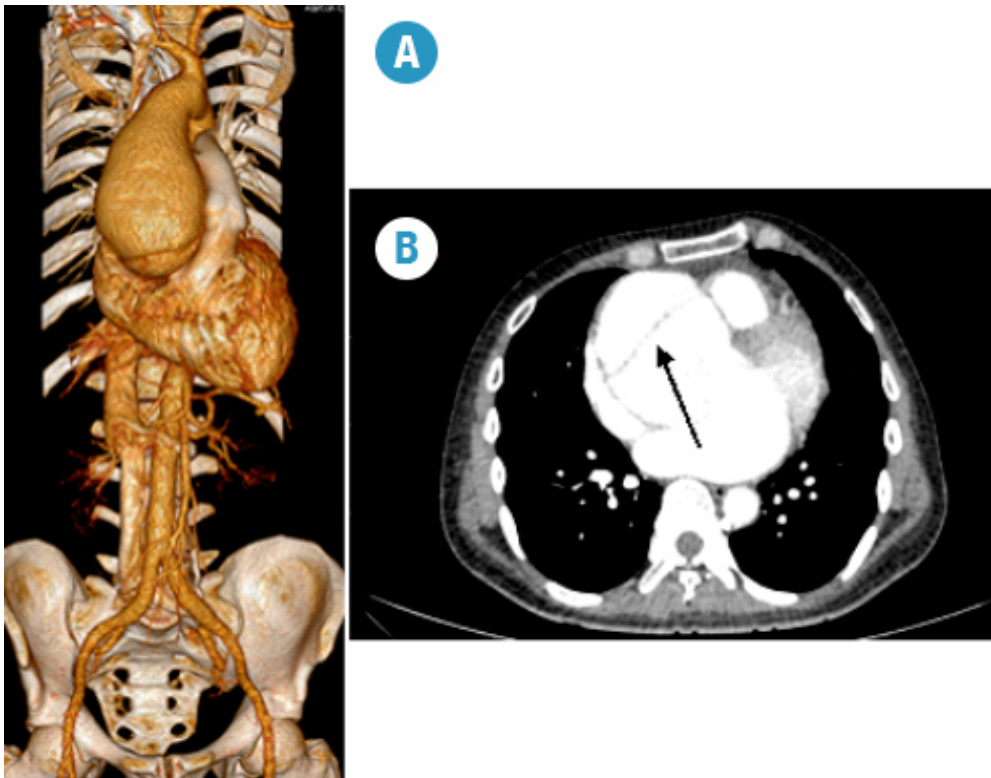


Fig 2A. 3D reconstruction of computed tomography angiography of the chest. Dilation of the ascending aorta. Aortic root 90.2 mm, anteroposterior diameter of ascending aorta of 80.8 mm.

Fig 2B. Arrow. Intimal flap.

Source: Own elaboration based on the data obtained in the study.

Evolution

One of the most important aspects is the length of hospital stay, since this is directly related to morbidity and mortality. In this case, approximately 48 hours passed between the completion of the complementary examinations, the confirmation of Stanford type A ascending aortic dissection as definitive diagnosis, and the decision to start surgical treatment through Bentall and De Bono surgery, which represented an increase of about 29% in the mortality rate.

The clinical management of the patient was holistic, and one of the parameters actively treated was blood pressure through carvedilol 6.25 mg VOc/12h, which kept the values within normality ranges.

Surgical procedure

Once the patient was under general anesthesia and strict asepsis, proximal and distal control of the right axillary artery was performed by cannulation for extracorporeal circulation (ECC), followed by medial sternotomy, exposing the pericardial sac through an inverted T incision, which exposed the ascending aortic aneurysm from the aortic annulus, with a diameter greater than 100 mm that deformed the base of the heart and extended to the distal third of the ascending aorta. Cardiomegaly IV/IV was found in both right and left cavities, along with global dilation of the ventricles and intraoperative pulmonary artery pressure over 60 mmHg according to Swan-Ganz catheter measurements.

The extracorporeal circulation entrance was then isolated by aortic clamping and the dissected aortic wall was opened longitudinally, which allowed to identify false and true lumen. Then, through a partial resection of the dissection focus, the distal end of the aorta was verified and the false lumen was sealed with BioGlue 1. Resection of the tricuspid aortic valve was continued, isolating left and right coronary buttons and implanting a valved tube sj # 27. Once the normal function of the prosthetic discs was confirmed, the left and right coronary button reimplant was placed and its permeability was checked. Immediately, distal anastomosis was performed from the valved tube to the distal ascending aorta. Finally, a prosthetic neo-ostium venous bypass implant was placed since the right coronary button was unstruc-

tured with suture dehiscence, which caused an acute intraoperative myocardial infarction.

Upon removing ECC, acute right ventricular distension and sustained ventricular fibrillation were observed, which required defibrillation (20-30 joules) together with antiarrhythmic therapy. Finally, arrhythmia was controlled. The patient presented three episodes of ventricular fibrillation which responded to electrical cardioversion.

ECC removal was attempted on three occasions, but low expenditure was observed despite maximum inotropic support, then the patient suffered refractory ventricular fibrillation and later asystole despite an epicardial pacemaker and died. The aortic clamping time was 150min and ECC time, 305 min (Figures 3, 4 and 5).

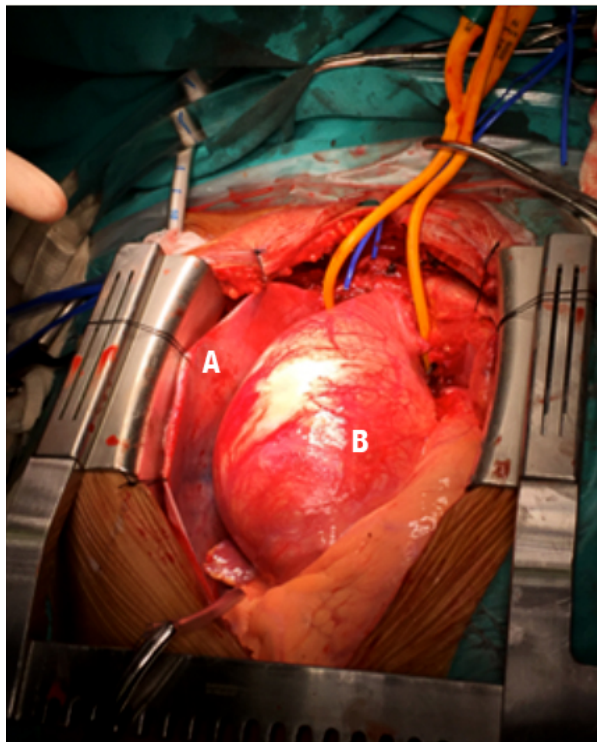


Fig 3. Surgery. Exposure by median sternotomy. A. Pericardial sac. B. Aneurysmal ascending aorta.

Source: Own elaboration based on the data obtained in the study.

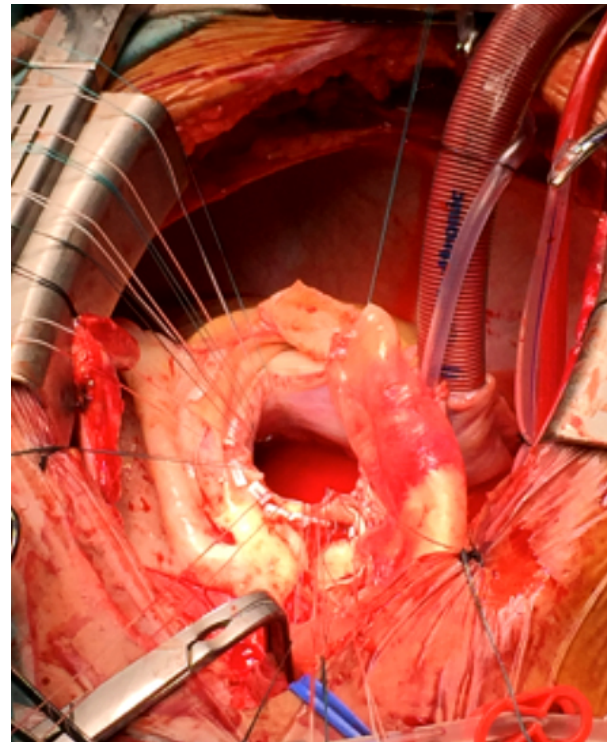


Fig 4. Exposure of the aortic ring with appropriate fixation points for subsequent placement of prosthetic material.

Source: Own elaboration based on the data obtained in the study.

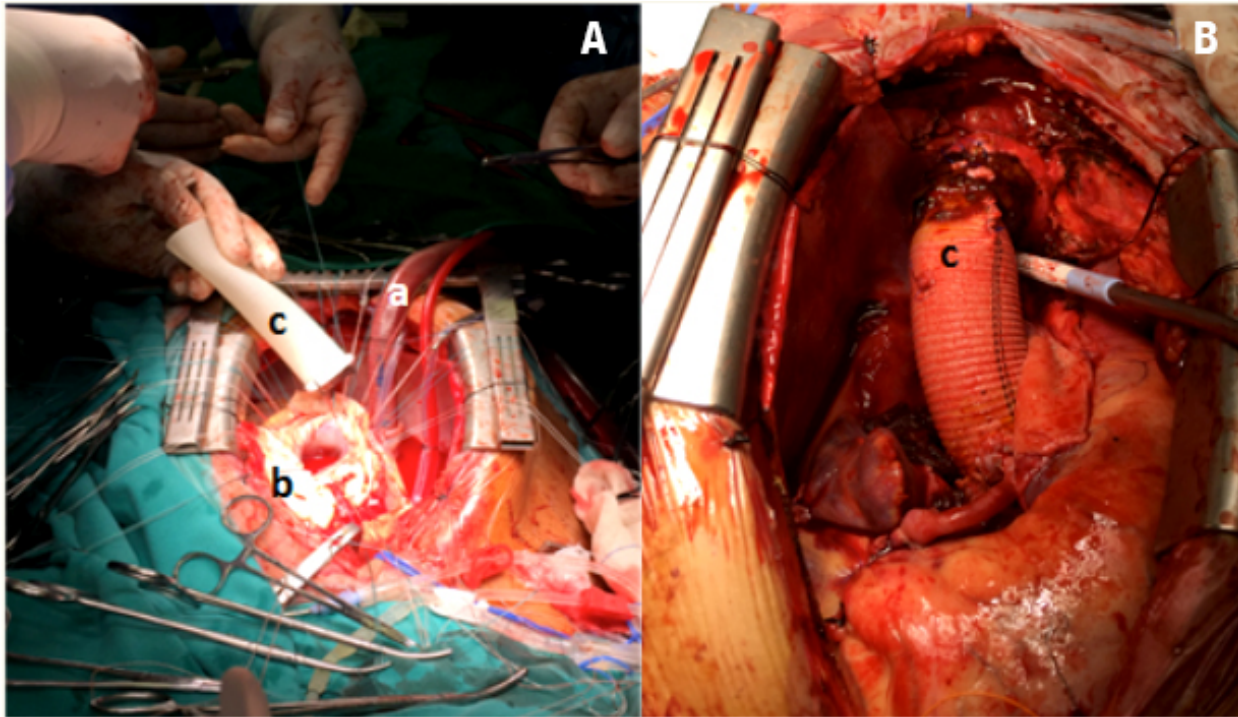


Fig 5A Exposure of the aortic ring with the appropriate fixation points after prosthesis placement.

Fig 5b. Replacement of the aortic root using composite prosthesis and Bentall coronary reconstruction. a: extracorporeal circulation system. b: aortic ring with corresponding fixation points. c: prosthetic material.

Source: Own elaboration based on the data obtained in the study.

DISCUSSION

The Marfan syndrome is an autosomal dominant pathology with a relatively low frequency, which tends to cardiovascular complications in most cases. The evolution of Marfan syndrome leads to an aneurysmal dilation of the aorta, which may be added to a rupture of its wall with the subsequent development of aortic dissection. In this case, a type A dissection was diagnosed based on the Stanford classification, the most frequently used nowadays, or DeBakey type II (3).

The ascending aortic aneurysms have a relatively low prevalence, as well as an important variability that can be accompanied by an aortic dissection or rupture of the aorta, increasing its mortality rate to almost 100%. The

complications observed in the patients who suffer from this condition are closely related to the diameter of the aorta, its growth rate and the time of evolution. In vessels with a diameter greater than 5 cm, the risk of rupture and dissection increases by up to 45% (4). The symptomatology is variable and depends on the type of aneurysm, as well as on its location and relationship with neighboring structures. An aortic aneurysm can cause fatigue, hoarse or strained voice and pain in the left shoulder. If it is accompanied by aortic dissection, it can cause intense lancinating pain in the anterior chest, which usually radiates towards the back in the interscapular region.

Aortic dissection is characterized by the creation of a false lumen in the middle layer of the aortic wall. It is classified according to

the presence and location of the first tears, as well as the retrograde or anterograde extension of the dissection (5).

If coronary vessels are compromised since birth, associated acute coronary syndrome symptoms are added, which constitutes an emergency and modifies both the management and the necessary times.

Opting for surgical treatment depends on several factors and choice is based on the anatomical conditions of the aorta, the underlying disease, the risk of anticoagulation, the age of the patient, the presence of an active infection, among others. Several surgical techniques have been developed that reflect the evolution in the management of the ascending aortic aneurysms; each has its own advantages, limitations and risks. The replacement of the aortic root and the ascending aorta with valved tubular grafts, known as the Bentall procedure, along with annuloaortic ectasia is the most appropriate choice of the treatment for this condition. (6).

The literature reports that the Bentall procedure has a low morbidity and a mortality of 13%, being septic shock and ventricular fibrillation the most common causes of death (7). These figures vary considerably depending on the moment when surgery is performed: if surgery is scheduled, mortality rates range between 1.7% and 17.1%, but if surgery is the consequence of an emergency, the values increase to between 23% and 50% (7). The survival rate at 5 years ranges between 73 and 92%, while survival at 10 years is between 60 and 73% (8). However, surgical mortality can vary greatly depending on the hospital center, the experience of the medical team, the available resources and the heterogeneity of the patients (9). It is worth noting that dissection distal to the operated segment persists in

60% to 75% of the patients who undergo a dissection of the ascending aorta (10).

Due to its comorbidity rates and its degree of evolution, Bentall procedure is the most favorable choice for this type of patients. In this case, the procedure consisted in performing a prosthetic replacement that compensated aortic valve deficiency as a result of the large dilatation of the aortic annulus and subsequent replacement of the damaged aortic wall. Finally, the aortic buttons were reimplanted to the prosthesis.

However, there are other surgical options. For example, David's technique, developed by the Canadian surgeon Tirone David in 1992, achieves a greater stabilization of all the components of the root of the aorta, which requires the proper function of the aortic valve (6). In this case, the patient had a large dilatation of the aortic annulus that made this procedure difficult to use.

It is important to consider the need for developing new therapeutic options for this type of patients in order to improve their survival rates and quality of life.

CONCLUSION

The case reported here demonstrates that the treatment of patients with Marfan syndrome should be done from a multidisciplinary and joint approach, where timely diagnosis together with follow-up by specialized services and a qualified team allow a timely action and, thus, avoid that the patients with this condition look for care when it is already at a late stage.

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INFORMED CONSENT

The authors met the regulations on of anonymity of the case study.

CONFLICT OF INTEREST

None stated by the authors.

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