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A Reversible Cause of Acute Right Ventricular Failure After Heart Transplant



Una causa reversible de fracaso ventricular derecho agudo tras el trasplante cardíaco

To the Editor,

We report the case of a 41-year-old man, who was an active smoker, with no prior history of cardiac conditions or other history

of interest, and who was admitted to hospital for anterior ST-elevation myocardial infarction in Killip class IV. During admission, he experienced cardiac arrest unresponsive to advanced cardiopulmonary resuscitation. Peripheral venoarterial extracorporeal membrane oxygenation was used initially during the arrest and was subsequently switched to central extracorporeal membrane oxygenation. Intra-aortic balloon counterpulsation was also required. Coronary artery disease was detected in 3 vessels with chronic occlusion of the circumflex artery and right coronary artery and acute occlusion of the proximal left anterior descending

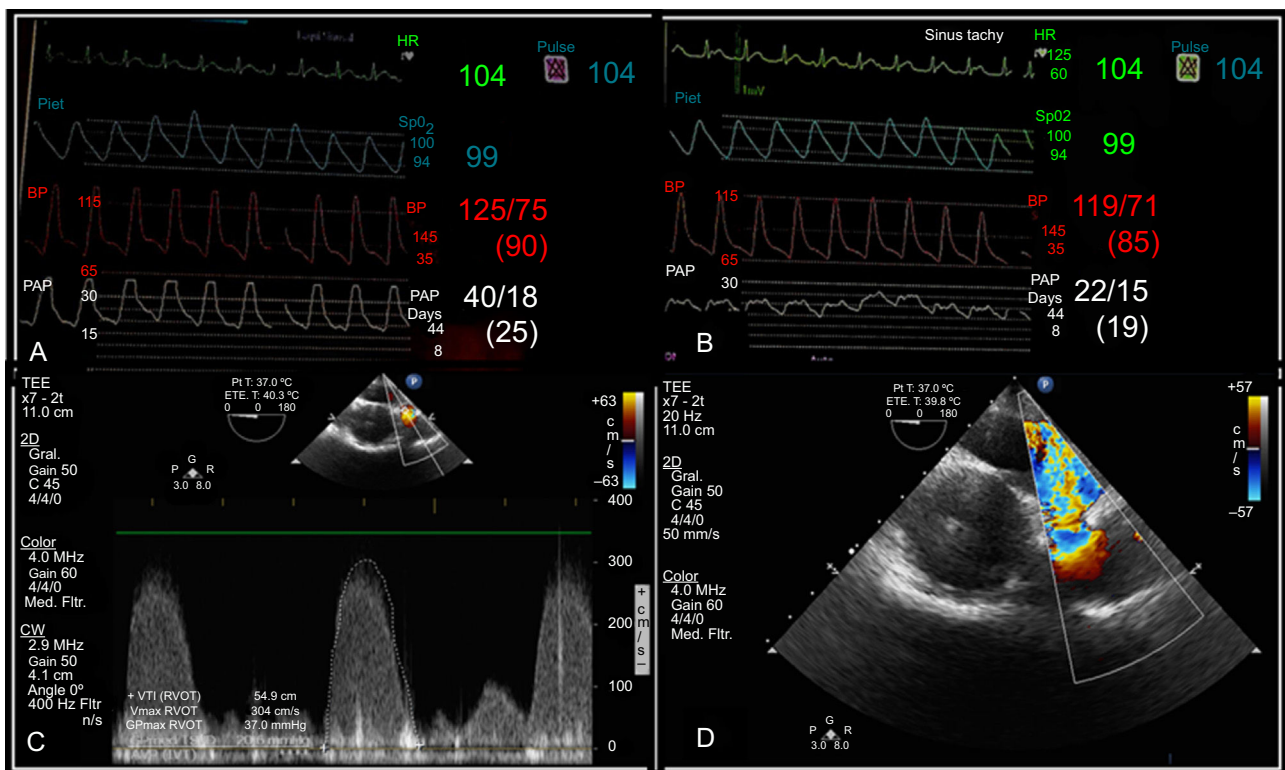
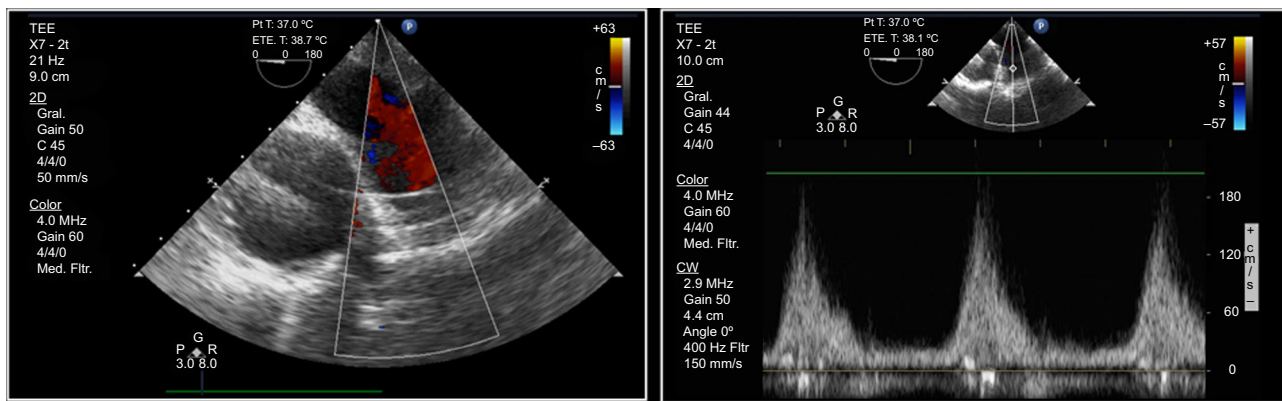


Figure 1. A and B, Swan-Ganz catheter measurements. A: pulmonary hypertension according to pulmonary artery pressure prior to stenosis of the pulmonary artery anastomosis. B: normal pulmonary artery pressures after the stenosis of the pulmonary artery anastomosis. C: stenosis of the pulmonary artery anastomosis: flow > 3 m/s, maximum gradient 38 mmHg. D: stenosis of the pulmonary artery along the short axis of the aortic arch at the level of the esophagus.



**Figure 2.** Transesophageal echocardiography after reconstruction of the pulmonary artery anastomosis. Decrease in flow to 1.6 m/s (right).

artery, which was revascularized with conventional stenting. After a favorable assessment and confirmation of the absence of serious neurological damage, the patient was placed on the heart transplant waiting list with top priority. Orthotopic heart transplant was finally performed with the Shumway technique (male 25-year-old donor weighing 80 kg and 176 cm tall for a recipient weighing 80 kg and 178 cm tall). After surgery, mild right ventricular dysfunction was observed (on rapid discontinuation of extracorporeal circulation), with progressive hemodynamic worsening at 24 hours and a growing need for inotropic and vasopressor support. At 36 hours, his liver enzymes were significantly elevated, and transesophageal echocardiography showed progression to severe dysfunction and dilatation of the right ventricle. Elevation of pulmonary pressures (measured with Swan-Ganz catheter before the anastomosis of the pulmonary artery and decline in the cardiac index to 1.9 L/min/m<sup>2</sup>) were observed with stenosis of the pulmonary artery anastomosis with a flow rate of > 3 m/s and maximum gradient of 38 mmHg. In addition, severe tricuspid valve failure and extensive dilatation of the inferior vena cava with systolic flow reversal in the suprahepatic veins were also observed (Figure 1). Immediate surgery was performed (the chest was open due to hemorrhage after the intervention) to reconstruct the pulmonary artery anastomosis. Transesophageal echocardiography showed an immediate decrease in velocity across the anastomosis to 1.6 m/s and a notable improvement in right ventricular dysfunction (Figure 2). Vasoactive support could therefore be withdrawn and the patient was extubated.

The main surgical technique in heart transplant is the Shumway-Lower technique,<sup>1</sup> with end-to-end suture of the great vessels of the recipient and donor organs. It is important that the size mismatch between the donor organ and the recipient is not too large.<sup>2</sup> Acute right ventricular failure accounts for 50% of postoperative cardiac complications and 20% of early mortality after heart transplant.<sup>3</sup> Although the main causes of right heart failure are pressure or volume overload, myocardial ischemia, or arrhythmias, there have been 2 reports of stenosis of the pulmonary artery suture,<sup>4</sup> an uncommon but reversible cause of right heart failure that occurs more frequently in the lung transplantation setting than the heart transplantation setting.<sup>5</sup> In this complication, an etiological diagnosis and early treatment are essential to guarantee resolution. The complication is caused by mismatches in the size of the anastomosed vessels, suture constriction, intimal hyperplasia, or a combination of these. The consequences depend on the extent and site of the stenosis and the size of the affected vessel.<sup>4–6</sup> Initial diagnosis can be made by

echocardiography, preferably transesophageal; the great vessel view and the section of the aortic arch at the level of the upper esophagus provide the highest diagnostic yield and best quantification.<sup>3</sup> One case has been reported with a satisfactory outcome after percutaneous treatment,<sup>4</sup> as well as after surgery with anastomotic reconstruction. The second option was preferred in our patient because less than 36 hours had elapsed since the first intervention and the chest was still open, thus facilitating rapid access to the anastomosis.

This case represents an uncommon complication that is not widely reported in the literature but which should nevertheless be considered in the differential diagnosis of acute right ventricular failure in heart transplantation.<sup>3</sup> Likewise, it also reflects the importance of early diagnosis and treatment to avoid irreversibility. Anastomosis of the pulmonary artery should be assessed after withdrawal of extracorporeal circulation with transesophageal echocardiography, with the preferred views being the great vessel view and the aortic arch section at the level of the upper esophagus. Measurement of the gradient at the anastomosis with a Swan-Ganz catheter may also be useful.

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### Large Family With Marfan Syndrome Demonstrating the Pathogenicity of a “Synonymous” Variant (p.Ile2118=) in the Fibrillin-1 Gene



#### Extensa familia con síndrome de Marfan en la que se demuestra la patogenicidad de una variante «sinónima» (p.Ile2118=) en el gen de la fibrilina 1

#### To the Editor,

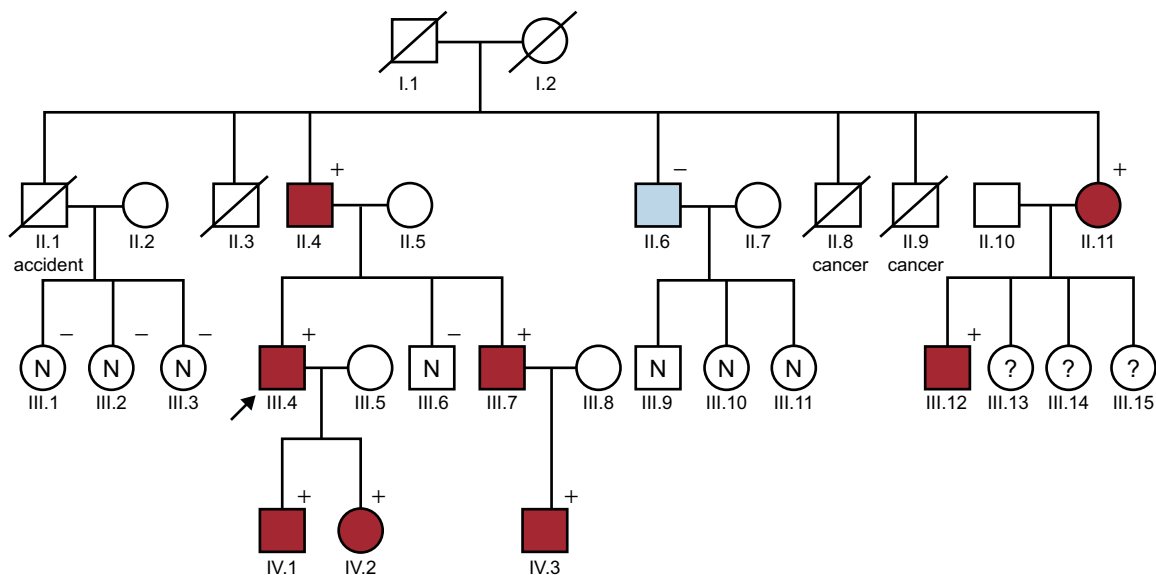
We present a family with 8 members from 3 generations with Marfan syndrome (MFS), in whom a complete familial genetic study confirmed the pathogenicity of a variant previously thought to be “synonymous”, identified on the fibrillin 1 gene (*FBN1*).

The proband is a 50-year-old Caucasian male who underwent emergency surgery for a type A aortic dissection. His 79-year-old father underwent surgery at age 56 years for dilatation of the ascending aorta with replacement of the aorta and aortic valve (Bentall procedure) and was operated on again at age 75 years for dilatation of the descending aorta. The proband had been diagnosed with ectopia lentis prior to his aortic dissection. In view of this and his father’s history, MFS was suspected. In 2008, he underwent a genetic study of the *FBN1* gene in another center,

using Sanger sequencing and multiplex ligation-dependent probe amplification (MLPA), but this was reported as negative. His 2 children were under follow-up as suspected MFS in a third center.

The familial study was centralized in our hospital. When the family tree was drawn (Figure), it identified other members with aortic dilatation or variable expression of features of MFS (Table).<sup>1</sup> The younger brother, who also had ectopia lentis, recently underwent surgery for a descending aortic aneurysm, and received a Bentall valved tube. This patient declined the aortic valve-sparing surgery offered (David procedure) due to the good outcomes in his father and brother, who had the Bentall procedure.

Given the obvious familial disease, the negative results of the previous genetic study were striking. In MFS, the percentage of patients with a positive genetic study has been reported as close to 90%.<sup>2</sup> Another genetic study was performed, this time using next generation sequencing with a panel that included 30 genes involved in aortic disease. This study identified only 1 “synonymous” variant (meaning that the change in nucleotide does not produce a change in the amino acid) in the *FBN1* gene (c.6354C>T, p.Ile2118=). The fact that this does not produce a change in the amino acid could indicate that this variant is nonpathogenic or of borderline pathogenicity. It is located in a coding region of the gene (at position 40 of exon 52, isoform



**Figure.** Family tree. Square, male; circle, female; arrow, proband; red, has MFS; blue, bicuspid aorta; diagonal line, deceased; N, assessed and healthy; ?, unknown; +, p.Ile2118= carrier; -, noncarrier.