The Heart Surgery Forum #2011-1133
15 (3), 2012 [Epub June 2012]
doi: 10.1532/HSF98.20111133

Online address: http://cardenjennings.metapress.com

Successful Surgical Treatment of Intramural Aortoatrial Fistula, Severe Aortic Regurgitation, Mitral Prolapse, and Tricuspid Insufficiency in a Patient with Ehlers-Danlos Syndrome Type IV

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ABSTRACT

Patients with Ehlers-Danlos syndrome (EDS) type IV, an inherited connective tissue disorder, are predisposed to vascular and digestive ruptures, and arterial ruptures account for the majority of deaths. A 31-year-old man with EDS presented with an intramural aortoatrial fistula, severe aortic regurgitation, mitral valve prolapse, and severe tricuspid valve insufficiency combined with a severely dilated left ventricle. Determining the best surgical option for the patient was not easy, especially regarding the course of action for the aortic root with a tear in the sinus of Valsalva. The fistula tract was closed at the aorta with suture and with a patch in the right atrium, the mitral valve was repaired with edge-to-edge suture and then annuloplasty with a Cosgrove ring, the aortic valve was replaced with a mechanical prosthesis, and a modified De Vega technique was used for the tricuspid valvuloplasty. The postoperative course was uncomplicated, and the patient was discharged 2 weeks later. The considerations made to arrive at the chosen surgical course of action in this complex case are reviewed.

INTRODUCTION

Ehlers-Danlos syndrome (EDS) type IV, also known as the vascular type of EDS, is a type of inherited connective tissue disorder. Patients with EDS type IV are predisposed to vascular and digestive ruptures, and arterial ruptures account for the majority of deaths. Vascular EDS, which is caused by abnormal synthesis of collagen type III and mutation in the COL3A1 gene, which encodes type III collagen, is considered the genetic etiology [Pepin 2000].

Received September 13, 2011; received in revised form December 5, 2011; accepted December 7, 2011.

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CLINICAL SUMMARY

A 31-year-old man with EDS type IV presented with progressive dyspnea. An intramural fistula tract (5.2 3.2 cm) of the right atrial wall originating from a defect of the right sinus of Valsalva (Figure 1) combined with severe aortic regurgitation (Figure 2) was demonstrated by echocardiography. Moderate mitral regurgitation secondary to anterior mitral valve prolapse and severe tricuspid insufficiency was confirmed. The left ventricle was dilated (end-diastolic diameter, 86 mm) with an ejection fraction of 38%. The aortoatrial fistula and the defect of the right sinus of Valsalva were verified by computed tomography (Figure 1).

The operation was performed with standard cardiopulmonary bypass. The aorta was clamped when the patient's temperature reached 28C. We found the defect of 0.6 cm in the right sinus of Valsalva adjacent to the ostium of the right coronary artery, which produced the intramural fistula of the right atrial wall. The fistula tract, which was approximately 6 3 2 cm, led to the right atrium. Because the surrounding aorta looked healthy, we closed the defect directly with pledgeted Prolene suture in the right sinus of Valsalva and concomitantly sutured a patch in the right atrium to repair the intramural fistula. The mitral annulus was dilated and a chorda supplying A3 of the anterior leaflet was torn. We made the repair with edge-to-edge suture and followed by annuloplasty with a 30-mm Cosgrove ring (Edwards Lifesciences, Irvine, CA, USA). The floppy and prolapsed native aortic valve was replaced with a 25-mm mechanical prosthesis (St. Jude Medical, St. Paul, MN, USA). The tricuspid valvuloplasty was performed with a modified De Vega technique. The postoperative course was uncomplicated, and the patient was discharged 2 weeks later.

DISCUSSION

The diagnosis of EDS IV type is mainly made clinically, and the major diagnostic criteria include arterial, digestive, or uterine fragility or rupture; thin, translucent skin; extensive bruising; and a characteristic facial appearance. All of these characteristics were found in our patient. Vascular EDS is caused by a deficit of type III fibrillar collagen. Although we lack laboratory evidence, the diagnosis of our patient was confirmed by the histologic observation in a biopsy sample of disorganization of collagen bundles in the dermis [Proske 2006].

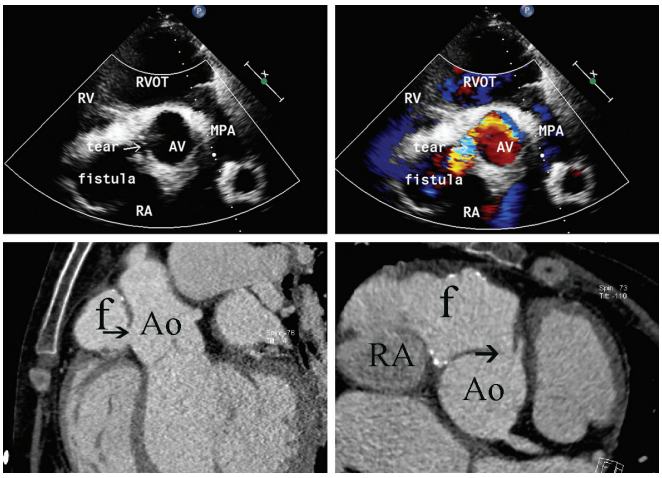

Figure 1. Echocardiography and computed tomography evaluations showed an intramural fistula tract (f) of the right atrial wall coming from the right sinus of Valsalva tear (arrow). RVOT indicates right ventricular outflow tract; RV, right ventricle; AV, aortic valve; MPA, main pulmonary artery; RA, right atrium; Ao, aorta.

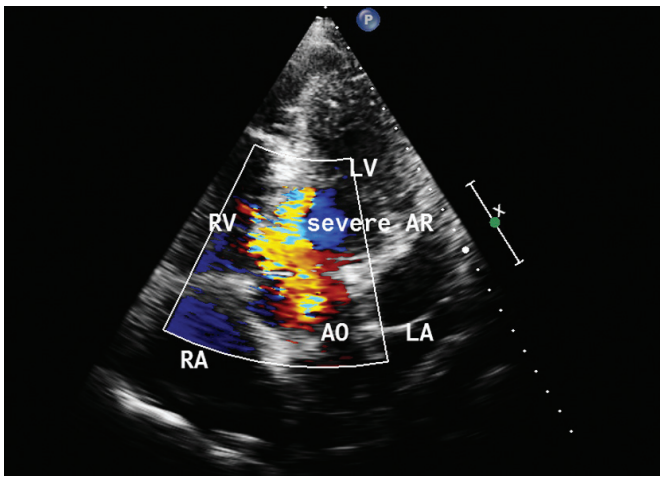

Figure 2. Echocardiography scan showing severe aortic regurgitation (AR). LV indicates left ventricle; RV, right ventricle; AO, aorta; RA, right atrium; LA, left atrium.

Arterial tears are considered the hallmark of EDS type IV [Pepin 2000]. Rupture of any artery into a free space is life-threatening and requires immediate intervention, even though the tissues are friable and repair is often difficult [Sèveve 2005]. Fortunately for our patient, the tear of the right sinus of Valsalva had led to the intramural wall of the right atrium and gradually produced the aortoatrial fistula, which gave us the opportunity for repair.

Mitral valve prolapse has been described to be a prevalent pathologic feature in EDS [Proske 2006]. Only a very limited literature exists with regard to successful surgical treatment of mitral regurgitation [Sèveve 2005], and the best surgical procedure has not yet been defined [Sauer 2010]. We undertook mitral valve repair to achieve the best hemodynamic benefits, although concerns had been raised regarding the long-term durability of mitral valve repair in patients with connective tissue disorders, owing to the tissue weakness that exists in the retained leaflets.

Little information is available regarding the successful treatment of severe aortic regurgitation in EDS type IV, although aortic regurgitation might be caused by dilation of the aortic root, which is commonly encountered in EDS patients [Oka 2001]. In our patient, the aortic root was not dilated significantly (38 mm in diameter), and the aortic leaflets were prolapsed and floppy. After further consideration of the hemodynamic benefits, we undertook the aortic valve replacement.

To the best of our knowledge, this report is the first to describe this combination of pathologies in EDS type IV: intramural aortoatrial fistula, severe aortic regurgitation, mitral valve prolapse, and severe tricuspid valve insufficiency, combined with a severely dilated left ventricle. Determining the best surgical option for our patient was not easy, especially regarding the course of action for the aortic root that included the tear of the sinus of Valsalva. Fortunately, the patient experienced no complications and recovered uneventfully. The echocardiographic result at the time of discharge was excellent, although close follow-up for the intermediate and long terms is warranted to determine if the operation chosen was correct.

REFERENCES

Oka N, Aomi S, Tomioka H, Endo M, Koyanagi H. 2001. Surgical treatment of multiple aneurysms in a patient with Ehlers-Danlos syndrome. J Thorac Cardiovasc Surg 121:1210-1.

Pepin M, Schwarze U, Superti-Furga A, Byers PH. 2000. Clinical and genetic features of Ehlers-Danlos syndrome type IV, the vascular type. N Engl J Med 342:673-80.

Proske S, Hartschuh W, Enk A. 2006. Ehlers-Danlos syndrome—20 years experience with diagnosis and classification at the university skin clinic of Heidelberg [in German]. J Dtsch Dermatol Ges 4:308-18.

Sauer M, Borger MA, Seeburger J, Mohr FW. 2010. Successful surgical treatment of atrial fibrillation, mitral regurgitation, and aortic root aneurysm in a patient with classical type Ehlers-Danlos syndrome. Ann Thorac Surg 89:273-5.

Sève P, Dubreuil O, Farhat F, Plauchu H, Touboul P, Broussolle C. 2005. Acute mitral regurgitation caused by papillary muscle rupture in the immediate postpartum period revealing Ehlers-Danlos syndrome type IV. J Thorac Cardiovasc Surg 129:680-1.