Images in Cardiovascular Medicine

Giant Aortic Root Aneurysm

in a Patient with D-Transposition of the Great Arteries and Marfan Syndrome

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A 33-year-old man presented with 3 months of worsening dyspnea on exertion. His medical history included dextro-type transposition of the great arteries (D-TGA) by means of a Senning atrial switch at 12 months of age. He had been monitored by a pediatric cardiologist until age 18 years but was then lost to follow-up. Of note, his father had Marfan syndrome.

Cardiac magnetic resonance images showed an aneurysmal aortic root (diameter, 6.9 cm). The patient's systemic right ventricle was markedly dilated and hypertrophic; the ejection fraction was 0.34 (Fig. 1). Mutation analysis revealed a likely pathogenic mutation in the *FBN1* gene (p.Glu768Lys; c.2302 G>A in exon 20).

Aortic root replacement with use of a 28-mm Dacron graft and a 25-mm bioprosthetic Carpentier-Edwards Perimount Magna Ease Aortic Heart Valve (Edwards Lifesciences Corporation) was discussed. The team's consensus, however, was to re-evaluate right ventricular systolic function after a trial of medical therapy. Afterload reduction and β-blocker therapy led to notable symptomatic improvement. Ten months after initial presentation, the patient underwent aortic valve and root replacement (Fig. 2). Microscopic examination of his aorta revealed cystic myxoid medial degeneration (Fig. 3).

Comment

To our knowledge, comorbid D-TGA and Marfan syndrome have not been reported. The genetic patterns of D-TGA are incompletely understood, because familial clustering is rare.[1] Marfan syndrome is caused by mutations in the *FBN1* gene,[2] which encodes fibrillin-1. Defects in fibrillin-1 can lead to progressive aortic dilation caused by cystic medial degeneration.[3]

Histopathologic studies of ascending aortas from neonates born with D-TGA have revealed elastic fiber fragmentation, increased collagen deposition, and loss of smooth-

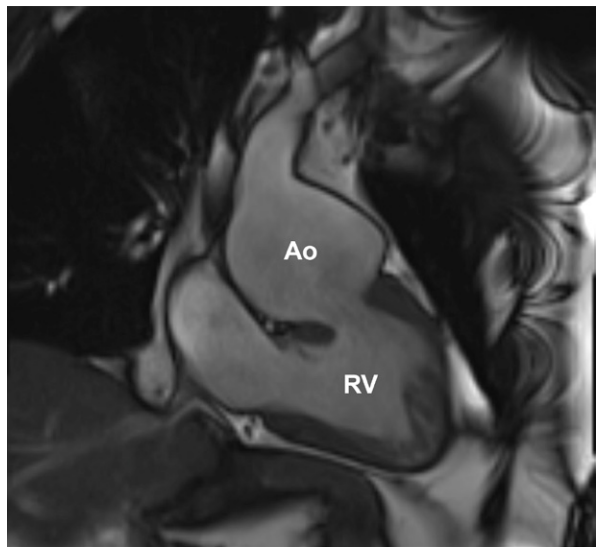

Fig. 1 *Cardiac magnetic resonance image shows a ventriculoarterial connection between the patient's systemic right ventricle (RV) and aneurysmal aorta (Ao). Note RV dilation and hypertrophy.*

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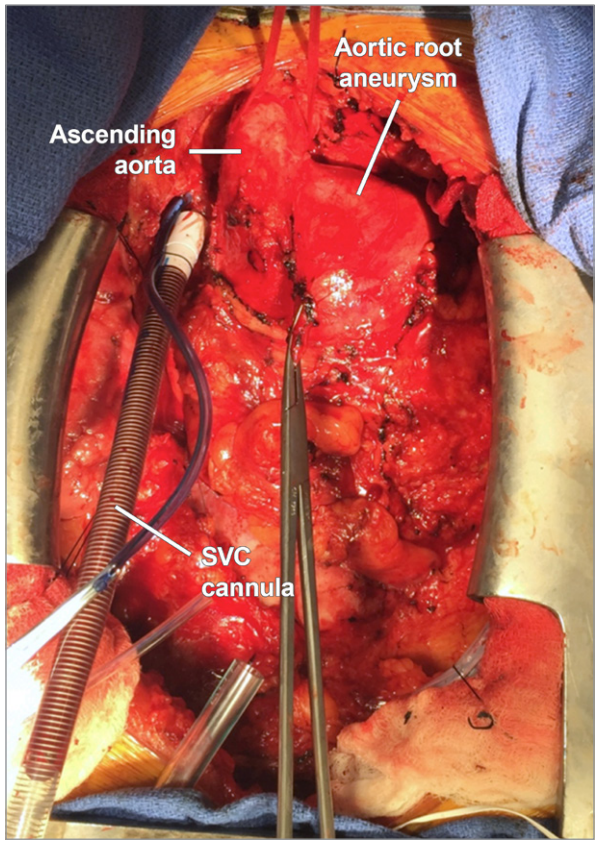

Fig. 2 *Intraoperative photograph shows the aneurysmal native aortic root with coronary buttons removed.*

SVC = superior vena cava

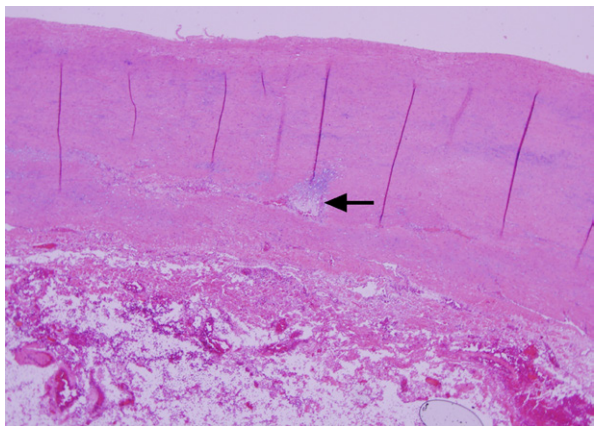

Fig. 3 *Photomicrograph of the aorta shows cyst-like degeneration in the triangular basophilic region (arrow) and more typical myxoid degeneration in the basophilic layers in the media (H & E, orig. ×40).*

muscle cells in the tunica media.[4] These findings correlate with data showing that approximately 50% of D-TGA patients who underwent a Jatene arterial switch had aortic root dilation 10 years later.[5] It therefore seems plausible that both D-TGA and Marfan syndrome contributed to the development of our patient's giant aortic root aneurysm.

References

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