CASE REPORTS

A DOUBLE DOSE OF AORTIC STENOSIS: AN UNUSUAL CASE

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Abstract

Supravalvular aortic stenosis is a rare congenital anomaly (less than 0.05% of all congenital heart defects). This aortic root anomaly consists in a narrow aortic lumen immediately above the aortic valve and represents the least common form of left ventricular outflow tract obstruction. Clinical presentation is usually in the first decades of life. In most cases, the aortic valve leaflets are morphologically normal. However, aortic insufficiency due the high systolic pressure proximal to the sinotubular junction is the most commonly abnormality described. There are very few cases described in the literature with concomitant valvular and supra-valvular aortic stenosis.

Keywords: *Supravalvular aortic stenosis; aortic valve stenosis; left ventricular outflow tract obstruction*

INTRODUCTION

Supravalvular aortic stenosis (SVAS) is a rare congenital anomaly reported in less than 0.05% of all congenital heart defects. This aortic root anomaly consists in narrowing aortic lumen immediately above the aortic valve and represents the least common form of left ventricular outflow tract obstruction (LVOTO). LVOTO lesions account for approximately 6% of congenital heart disease, and SVAS occurs in 5-10% of all patients with LVOTO.[1] Clinical presentation of SVAS is usually in the first decades of life.[2] SVAS is seen in syndromic as well as non-syndromic patients. It has classically been described in association with Williams-Beuren syndrome caused by a deletion on chromosome 7q11.23 which affects the elastin gene. The sporadic form is more common than the autosomal dominant form.[2] Severity of SVAS ranges from localized ringlike thickening to diffuse involvement including pulmonary arteries, aortic arch and its branches.[3] In most cases, the aortic valve leaflets are not fused and are morphologically normal. However, aortic insufficiency due the high systolic pressure proximal to the sinotubular junction is the most commonly abnormality described.[4] There are very few cases described in the literature with concomitant valvular and supravalvular aortic stenosis. We present the case of a 62-year-old man with a double doses of aortic stenosis.

CASE REPORT

A 62-year-old man presents with complaints of fatigue to moderate physical efforts, which progressively worse over the last months. At physical examination, a panfocal mid-systolic murmur was described, with no other abnormalities reported. He had a medical history of hypertension. An echocardiogram was performed, which

revealed a thickened calcified aortic valve, with reduced opening and mild regurgitation, a peak LV/Ao gradient of 97mmHg and a mean gradient of 59mmHg, an estimated area of 0.5 cm2, moderate hipertrophy of left ventricle and ventricular septum, good biventricular function and mild mitral regurgitation. (Figure 1 A) Cardiac catheterization showed a significant narrowing of aortic lumen in the aortic root which was not previously known. (Figure 1 C) CT angiogram confirmed a supravalvular aortic stenosis, originating 17mm above the aortic valve with a length of 25mm, and a maximum luminal diameter of 10 mm, while the aortic sinuses and post-stenosis ascending aorta were wider (33 mm and 23 mm, respectively). (Figure 1 D) Genetic studies for mutations involving elastin gene were negative and acquired causes were excluded. There was no familiar history of aortopathies. The patient was submitted to aortic valve replacement with a mechanical prosthesis by patient preference and a supravalvular aortic repair with pericardium patch enlargement. (Figure 2) There were no complications, and the patient was discharged on the 5th postoperative day.

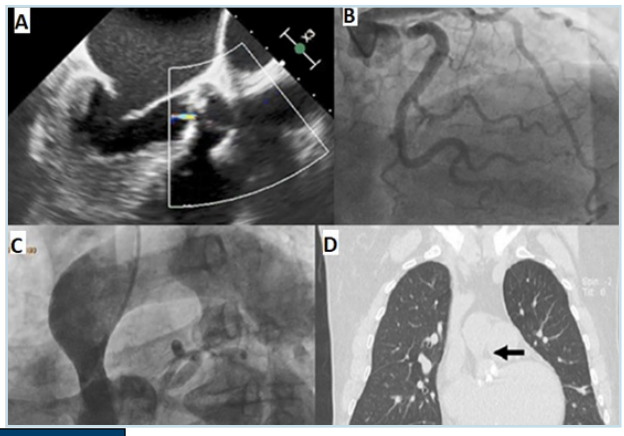

Figure 1

1- (A) Transthoracic echocardiogram shows thickened calcified aortic valve, with reduced opening and mild regurgitation, and a narrowed lumen of the aortic root. (B) Coronariography shows left coronary artery dilated and tortuous. (C) Aortography shows a significant narrowing of aortic lumen in the aortic root. (D) CT angiogram shows a supra-valvular aortic stenosis.

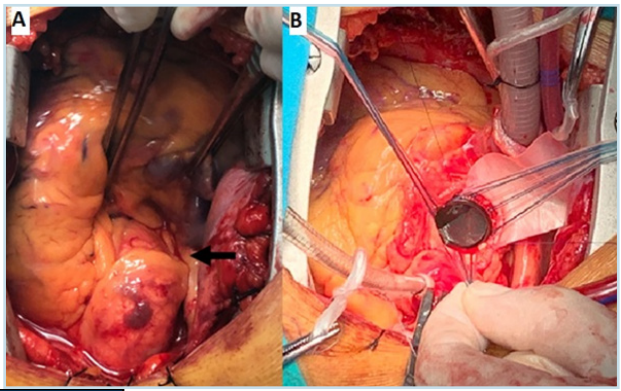

Figure 2

(A) Intraoperative view of supravalvular aortic stenosis. (B) Aortic valve replacement with a mechanical prosthesis and aortic repair with pericardium patch.

COMMENTS

Surgical intervention is associated with good long-term results in most patients. A few surgical options are available. Ross procedure, which consists of replacing the diseased aortic valve with the patient's own pulmonary valve, is usually reserved for small children. Aortic valve and aortic root replacement with coronary reimplantation (Bentall procedure) is an option for older patients.[5] A percutaneous approach with transcatheter aortic valve implantation (TAVI) with transcatheter stent placement could also be considered although it is a rarely utilised option. We opted for aortic valve replacement and aortic repair with pericardium patch. We found, intraoperatively, left ventricular hypertrophy, which is a common finding in this form of LVOTO due to pressure overload. The increased myocardial metabolic demand in this situation makes myocardial protection an even more important step during surgery. The coronary arteries are often markedly dilated and tortuous, secondary to their constant exposure to extremely high pressures proximal to the sinotubular junction (Figure 1 B). They also displayed accelerated coronary artery sclerosis. Sudden death is frequent if patients are left untreated due to the combination of left ventricular hypertrophy and premature coronary artery disease.[2] Supravalvular aortic stenosis is a rare congenital condition, and clinical presentation usually manifests in the first decades of life, not at older ages. The combination of valvular and supra-valvular aortic stenosis is infrequently reported in the literature, which makes this case unusual and particularly challenging from a diagnostic and therapeutic point of view.

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