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Congenital aortic insufficiency with ascending aortic aneurysm: Dealing with a challenging case

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ABSTRACT

Congenital aortic insufficiency is a challenging condition; without appropriate management from birth, it can lead to left ventricle dilation and heart failure. Aortic insufficiency due to leaflet agenesia is rarely described in literature. We report here a case of severe congenital aortic regurgitation that required surgical treatment until the boy reached five-year-old thanks to prenatal diagnosis and adequate medical care since birth. The patient underwent a mechanical Bentall procedure for associated dystrophic pulmonary valve with mild stenosis, ascending aortic aneurysm of 34 mm and anomalous position of the left coronary ostium. We safely performed a rare paediatric procedure with encouraging short-term results.

1. Introduction

Aortic regurgitation in children is a challenging condition; without appropriate management, it can lead to left ventricle dilation and heart failure. It can be associated with connective disorder and ascending aortic aneurysm. Depending on the patient's age, different surgical options can be considered.

We report here a case of severe congenital aortic regurgitation associated with aortic aneurysm that, despite adequate medical management, required urgent surgical treatment in a five-year-old boy.

2. Case Report

Systematic antenatal follow-up showed echographic signs of left ventricular dysfunction secondary to massive aortic valve leakage. Birth was given at 40 weeks of gestational age in a tertiary hospital. The child was admitted to the cardiac intensive care unit for inotropic support (milrinone).

Congenital heart disease was associated with bilateral radial hypoplasia with thenar hypotrophy and transient neutropenia. The patient's sister, younger by seventeen months, displayed normal results at musculoskeletal and echocardiographic screenings. Genetic testing did not find any disorder: karyotype 46, XY, normal array-CGH (comparative genomic hybridization), normal genetic sequencing of TBX5, SALL4 (Holt Oram syndrome) and G4.5 (Barth syndrome). His parents refused further genetic testing.

First transthoracic echocardiography, at four months of age, showed a massive aortic regurgitation reaching the apex and believed to be due to a bicuspid aortic valve [Fig. 1], a dystrophic pulmonary valve with mild stenosis (maximum peak gradient: 35 mmHg), a hypokinetic and dilated left ventricle (left ventricular end-diastolic diameter: 24.4 mm), and a dilated ascending aorta (diameter: 16.1 mm).

The boy was treated with conversion enzyme inhibitors and regularly followed with echocardiography. He reached 5 years old without symptoms but transthoracic echocardiography showed a severe dilation of the left ventricle (left ventricular end-diastolic diameter: 65 mm), of the aortic annulus (measured at 19 mm), and of the ascending aorta (at 34 mm, versus 18 mm for the descending aorta). Massive aortic regurgitation and severe left ventricular systolic dysfunction (ejection fraction of 15%) indicated urgent surgical intervention.

Surgery was conducted via median sternotomy. Massive aneurysm of the ascending aorta involving the root was found. Cardiopulmonary bypass was established with distal aortic, superior and inferior vena cava cannulation. Under moderate hypothermia (32 °C), with vent into the right superior pulmonary vein, aorta was cross-clamped and opened. Direct ostial crystalloid cardioplegia was administered and repeated every 30 min.

We were surprised by the absence of the left coronary cusp of the aortic valve and the presence of two other thickened leaflets, the right and the non-coronary cusps. The left coronary ostium had a very low origin, only a few millimetres above the annulus. The left anterior descending coronary artery, the circumflex artery, and a third branch emerged very early in a ‘gun barrel’ shape that made cardioplegia tricky and dissection risky. The option of a mechanical Bentall was chosen

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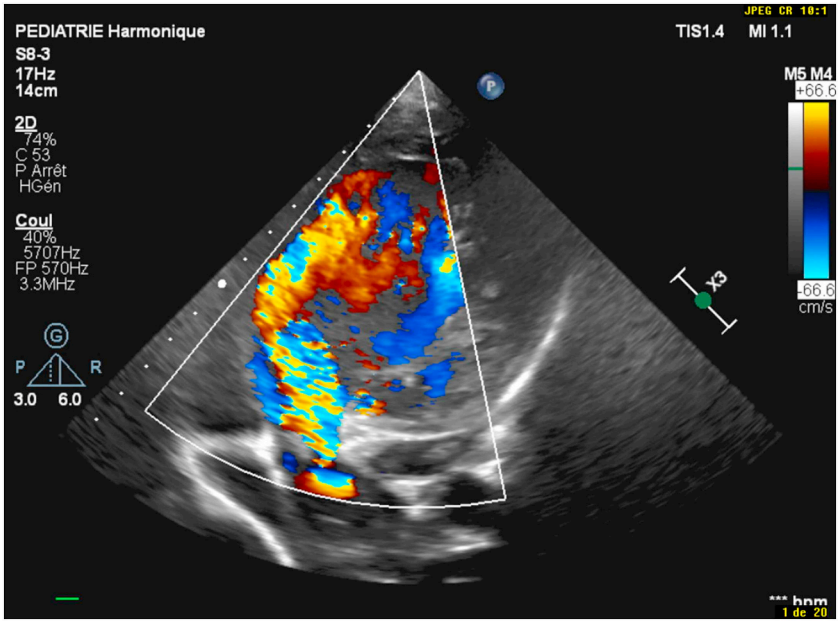

Fig. 1. Transthoracic echographic apical view of the aortic leaking jet with a sword shape.

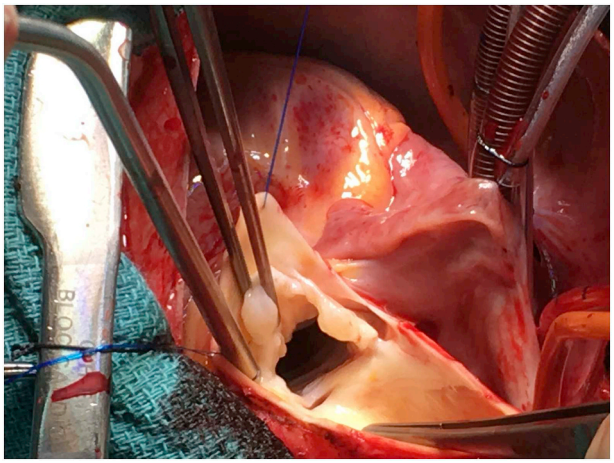

Fig. 2. Operative view: agenesia of the left coronary cusp with a low left ostium,

[Fig. 2].

Leaflets and aneurysmal aorta were excised and sent for anatomical pathology examination. Coronary buttons were delicately removed. A composite valved conduit 21 (Dacron composite graft with a mechanical valve; Medtronic Inc., Minnesota, MN, USA) was sutured to the aortic annulus using multiple interrupted pledgetted stitches (2-0 braided polyester). Left and right coronary buttons were reimplanted on the graft with a running suture of polypropylene 6-0 stitch. Finally, the distal portion of the graft was sutured end-to-end to the aorta with a polypropylene 5-0 stitch. Aorta was de-aired and cross-clamp was released after 98 min. Weaning from cardiopulmonary bypass was completed after 115 min. Two internal electrical shocks (15 then 20 joules) were required to halt ventricular fibrillation. Sinus rhythm and a mean blood pressure of 68 mmHg were achieved at the end of the procedure with inotropes: milrinone 0.5 μg/kg/min and adrenalin 0.1 μg/kg/min.

The patient was extubated 2 h after surgery. Inotropes were ceased 48 h after surgery and the patient was discharged from the ICU on postoperative Day 3. By postoperative Day 4, transthoracic echocardiography showed an ejection fraction of 40%. Ascending aortic diameter measured 22.1 mm (versus 34 mm before surgery). Postoperative Day 10, the patient was discharged on oral anticoagulation therapy.

Follow-up echocardiography at 20 days after surgery revealed a circumferential pericardial effusion of 23 mm with right ventricular compression requiring a surgical drainage to prevent tamponade. The boy recovered quickly in the weeks that followed.

Pathology results showed mucoid dysplasia of the aortic valve and discrete fibrosis of intima and media layers in the aortic wall. No specific features helped to define the underlying pathology. Genetic testing of the specimen was refused by parents.

3. Discussion

The first question is timing of surgical intervention, particularly relevant due to the lack of consensus and unclear paediatric recommendations.

Our patient was diagnosed with severe aortic regurgitation in combination with left ventricular dysfunction before birth. Antenatal diagnosis, now routine, allowed us to anticipate the need for intensive care. Four days of milrinone and diuretics were required to improve LV function after birth. Within the first postnatal month, medical management using amlodipine, then captopril, helped to postpone the need for surgical intervention and its high risk of morbidity and mortality in a 2.47 kg child (-2 Standard deviation; -3 Standard deviation).

European guidelines suggest that surgery should not be performed immediately in an asymptomatic patient [1]. Milk intake and growth were as expected for our patient, in accordance with WHO standards. Impairment of left ventricular function was reached between the ages of four and five years, with an ejection fraction of 32% in time motion or 15% by Simpson's method. Guidelines for asymptomatic severe aortic regurgitation indicate surgery when resting left ventricular ejection fraction $\leq$50% (Class I, Level B) [1]. In addition, between the ages of four and five years, left ventricle enlargement nearly reached the surgical cut-off ($>$ 70 mm) with a left ventricular end-diastolic diameter of 66 mm. All these arguments informed a decision by the cardiology team to schedule surgery at age five [1,2].

The second question is which surgical strategy should be adopted?

Several surgical options should be assessed in the case of a bicuspid valve with severe regurgitation. Despite the potential need for reoperation and dilatation of the autograft root, the Ross procedure remains the best choice for aortic valve replacement in infants with severe aortic insufficiency [3]. In this case, the Ross procedure would

have been ideal if it were not associated with pulmonary valve dysplasia and mild pulmonary stenosis (maximum peak gradient of 35 mmHg). Bioprostheses and homografts in children have been largely abandoned due to accelerated degeneration.

Mechanical valve replacement becomes inevitable despite small patient size and the risk of haemorrhage and thromboembolism. Aortic valve replacement in children is feasible with mechanical prostheses 2–4 sizes larger than the annulus measurement [3]. In our case, a 21 mm prosthesis sizer fit without any annulus enlargement. Shanmugam et al. [4] reported that no further replacement of prosthesis was required when the patient received a mean prosthesis size of 22.6 mm with freedom from reintervention at 20 years of 92% ± 4%.

Significant growth of the patient's ascending aorta was in favour of a growing aneurysm with +5 mm between the third and fourth years of age and +4 mm between the fourth and the fifth years of age. The main principle of surgery for ascending aortic aneurysms is to prevent the risk of dissection or rupture by restoring the normal dimensions of the ascending aorta [5]. Data on children are still unclear as they do not specify recommended thresholds for aortic aneurysms. According to ESC guidelines for adults, lower thresholds (below 50 mm) can be considered in patients with additional risk factors, such as increased aortic diameter > 3 mm/year or aortic regurgitation, and also according to age or body surface area (Class IIb, Level C) [5].

Performing a mechanical aortic valve replacement in addition to an ascending aorta replacement with a Dacron graft would have been ideal if the case were not associated with a particular anatomy of the left main coronary artery. The left coronary ostium was emerging from the left sinus in a lower position than usual, just above the annulus. A mechanical Bentall option was chosen to avoid obstruction of the left ostium [Fig. 2].

By postoperative Day 3, transthoracic echocardiography showed an improvement in left ventricular ejection fraction (40% by Simpson's method in comparison to preoperative 15%). Likewise, left ventricular end-diastolic diameter had decreased from 66 mm preoperatively to 44.6 mm postoperatively. In fact, Arnold et al. demonstrate that aortic valve replacement in children may lead to normalization of left ventricular function and structure in children and young adults [6].

4. Conclusion

Without prenatal diagnosis and appropriate medical management, severe aortic regurgitation in children induces progressive heart failure from birth to surgical correction. Timing of surgery and surgical options require multidisciplinary discussion, care, and follow-up. Here, we safely and successfully performed a rare procedure in a five-year-old boy with encouraging results. Nonetheless, paediatric valve replacement presents challenges for the patient into adulthood and further follow-up for this patient, in particular, will be necessary in terms of growth, thromboembolic/bleeding complications, and quality of life.

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Conflicts of Interest

There are no conflicts of interest.

Ethical Standards

The authors assert that this work does not involve human or animal experimentation.

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