VIA MEDICA

CASE REPORT

Cardiology Journal
2009, Vol. 16, No. 4, pp. 368–371

ISSN 1897–5593

Progressive discrete subaortic stenosis developed after successful primary repair of the supravalvular aortic stenosis in Williams syndrome

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Abstract

Discrete subaortic stenosis is a rare, late complication of the surgical repair of congenital heart defects. Secondary subaortic stenosis late after surgical repair of supravalvular aortic stenosis in Williams syndrome has not previously been described.

A 20 year-old female patient with Williams syndrome became symptomatic eight years after subaortic membrane resection. A discrete subaortic stenosis was identified by echocardiography. She was indicated for a Konno operation with a 19-mm Sorin SL valve inserted in the aortic position, along with ventricular septal defect closure with a pericardial patch. She produced a transient slight hypocalcemia after the operation. She was discharged home in a good condition on the fourth post-operative day.

Intracardiac morphological and hemodynamic factors have been suggested as responsible for such a late complication. Our case study suggests the need for regular follow-up in patients with Williams syndrome, even after primary surgical repair. (Cardiol J 2009; 16, 4: 368–371)

Key words: discrete subaortic stenosis, echocardiography, Konno operation, Williams syndrome

Introduction

Discrete subaortic stenosis is a rare, late complication of the surgical repair of congenital heart defects [1]. Subaortic stenosis can progress as children grow, and always necessitates a surgical intervention [2]. Secondary subaortic stenosis predominately develops secondary to the surgical repair of ventricular septal defect, patent ductus arteriosus, and coarctation of the aorta, etc. [1, 3]. Discrete subaortic stenosis late after surgical repair of supravalvular aortic stenosis in Williams syndrome has not previously been described.

Case report

A 20 year-old female had experienced shortness of breath on exertion for two years. She had been diagnosed with Williams syndrome and undergone a subaortic membrane resection eight years earlier. In this first operation, no significant subaortic stenosis was found. On current admission, her vital signs were normal except for a systolic murmur at the aortic valve region. Inspected by echocardiography, her left ventricular ejection fraction was 60%. The aortic annulus, root and ascending aorta extended 1.16 cm (Fig. 1), 2.5 cm and 2.1 cm respec-

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Received: 24.11.2008 Accepted: 1.10.2008

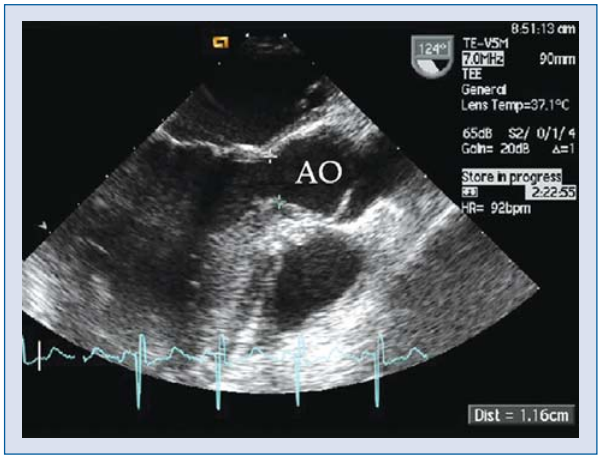

Figure 1. The aortic annulus extended 1.16 cm on long-axis view of transthoracic echocardiography; AO — aorta.

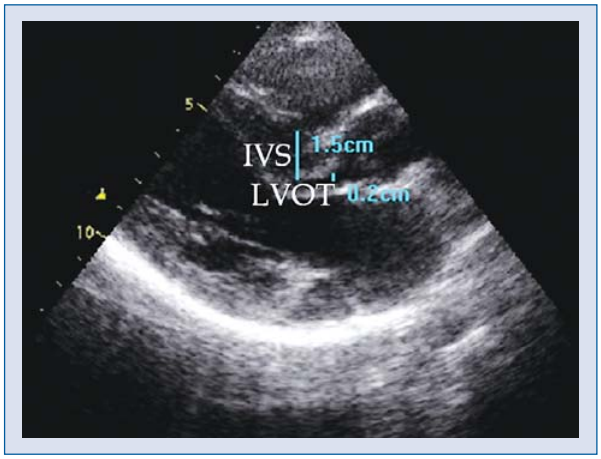

Figure 2. Left ventricular outflow tract (LVOT) became severely stenotic during systole caused by hypertrophic interventricular septum (IVS).

tively. The interventricular septum extended 1.5 cm in thickness. Left ventricular outflow tract became severely stenotic during systole (Fig. 2). Abnormal flow patterns were visible on color flow imaging (Fig. 3). The peak and mean pressure gradients across the aortic valve were 88 and 41 mm Hg, respectively. A ventricular septal defect and a persistent left superior vena cava were noted as well.

A Konno operation was performed with a 19-mm Sorin SL valve inserted in the aortic position, along with ventricular septal defect closure with a pericardial patch. During the first two post-operative days, her blood calcium appeared a transient slight decrease, with the total calcium of 8.0 mg/dL (normal 8.1–10.4 mg/dL), and the ionized calcium of

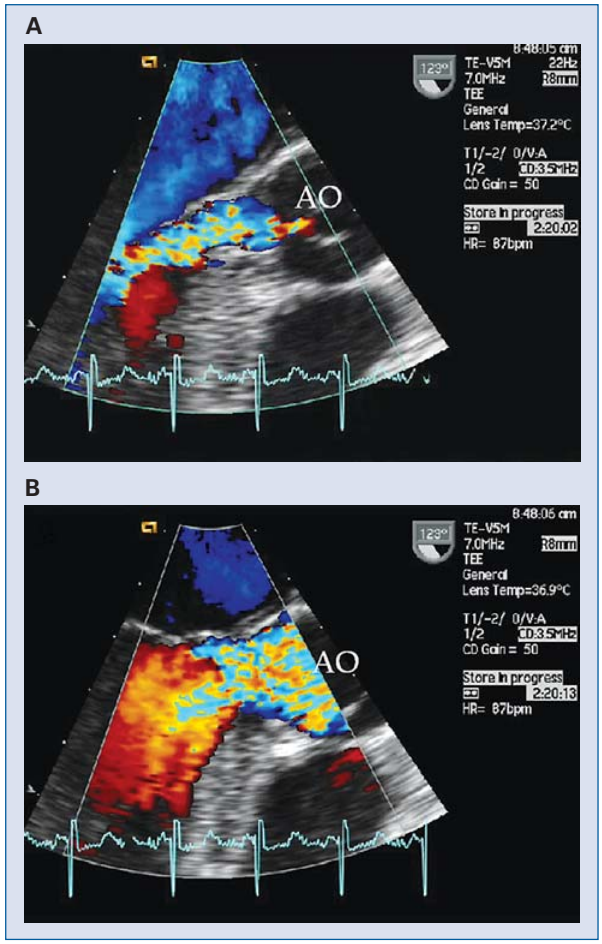

Figure 3. Abnormal flow patterns were visible on color flow imaging: **A.** Severe regurgitant flow across the stenotic region during diastole; **B.** Tunneling stenotic flow during systole; AO — aorta.

0.96 mmol/L (normal 1.00–1.20 mmol/L). Her post-operative course was uneventful and she was discharged home in a good condition on the fourth post-operative day.

Discussion

In Williams syndrome, the pressure gradient across the stenotic area is likely to increase over time [4]. Severe progression has been noted in patients with an initial pressure gradient > 20 mm Hg, but it remains stable in those with an initial pressure gradient < 20 mm Hg. Aortic arch hypoplasia could be associated with restenosis at the distal end of the patch in patients with patch aortoplasty [5]. In adult patients, the pressure gradient across the stenotic region tends to be higher, and the maximal and mean pressure gradients could be 104 and 73 mm Hg [6].

A positive echo sign was defined as at least 2 m/s peak velocity or 16 mm Hg instant peak gradient [7]. Surgical indication for aortoplasty in relieving the supravalvular stenosis was mainly a high pressure gradient > 50 mm Hg, and focal supravalvular narrowing. Peak velocity in the ascending aorta could significantly be decreased by aortoplasty (3.8 ± 0.9 m/s *vs*. 2.4 ± 0.8 m/s, p = 0.02). Aortoplasty decreases the aortic gradient with no effect on resizing the ascending aorta, aortic arch and isthmus in short period [8]. Post-operative pressure gradient averaged 12–30 mm Hg, and persistent high gradient may indicate an incomplete surgical relief [9].

Acquired subaortic stenosis has been reported secondary to surgical repair of congenital heart defects including ventricular septal defect, patent ductus arteriosus, aorticopulmonary window, tetralogy of Fallot, supramitral membrane, ventricular septal defect and patent ductus arteriosus [1], coarctation of the aorta, pulmonary stenosis [3], ostium primum defect [3, 10–13], double-outlet right ventricle [12, 14, 15], and miscellaneous lesions [3]. The time of occurrence of late subaortic stenosis has been reported from two months to 22 years [10, 11]. Intracardiac morphological and hemodynamic factors have been suggested as responsible for such a late complication [10, 12]. The causes of the secondary subaortic obstruction were stenosis of the tunneling patch, subaortic fibrous ring, muscular septal hypertrophy, antero-lateral muscular hypertrophy, and relics of tricuspid tissue inserted on the infundibular septum [15]. Frequently associated initial anatomical conditions were coarctation of the aorta (40%), lesions of the mitral valve (32%), bicuspid aortic valve (21%) and left superior vena cava (14%) [11]. The mean pressure gradient across subaortic stenosis at the time of reoperation was 47 ± 29 mm Hg. Five patients developed a second subaortic stenosis after a mean period of 7.4 years. One patient developed a third subaortic stenosis [11]. Echocardiography remains a reliable tool for the diagnosis of secondary subaortic obstruction during follow-up [13, 15].

This condition secondary to surgical repair of supravalvular aortic stenosis in Williams syndrome has not previously been reported. Brown et al. [7] presented 101 cases of congenital supravalvular aortic stenosis over 38 years; 14 (14%) of them had Williams syndrome, and 14 (14%) had one or two reoperations. Left ventricular outflow tract obstruction was likely to be the only risk factor responsible for the reoperation by multivariate analysis. At long-term follow-up, left ventricle-aortic pressure gradient was lower in patients with localized supravalvular aortic stenosis than in those with diffuse supravalvular aortic stenosis (12 ± 10 mm Hg *vs*. 22 ± 17 mm Hg, p = 0.01). Residual stenosis after the surgical repair in Williams syndrome seemed to be related to the amount of the attachment of the aortic valve cusps to the fibrous intimal ridge and residual anterior supravalvular tissue [9].

Dimensions of the aorta are usually small in Williams syndrome. The mean diameter of the ascending aorta was 5.67 ± 1.97 mm in the local type, but the smallest (3 mm) in the diffuse [16]. In our patient, the aortic annulus, root and ascending aorta extended 1.16 cm, 2.5 cm and 2.1 cm respectively. Therefore, her clinical signs of obstruction prompted a Konno operation, and as a result the surgical outcome was promising. Severe infantile hypercalcemia and a defect in vitamin D metabolism are associated with Williams syndrome [17]. In contrast, the blood calcium of our patient was slightly lower during the first two days after the operation. This was probably a result of the cardiopulmonary bypass during cardiac surgery [18, 19].

A discrete subaortic stenosis eight years after surgical resection of supraaortic stenotic membrane in a patient with Williams syndrome is described. As indicated in the literature, the associated left superior vena cava and ventricular septal hypertrophy might be the risk factors predisposing to the late development of subaortic stenosis in the present patient. We suggest a regular follow-up in patients with Williams syndrome, even after primary surgical repair.

Acknowledgements

The authors do not report any conflict of interest regarding this work.

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