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Early Manifestation of Supravalvular Aortic and Pulmonary Artery Stenosis in a Patient with Williams Syndrome

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Williams syndrome (WS) is a developmental disorder characterized by vascular abnormalities such as thickening of the vascular media layer in medium- and large-sized arteries. Supravalvular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis (PPAS) are common vascular abnormalities in WS. The natural course of SVAS and PPAS is variable, and the timing of surgery or intervention is determined according to the progression of vascular stenosis. In our patient, SVAS and PPAS showed rapid concurrent progression within two weeks after birth. We report the early manifestation of SVAS and PPAS in the neonatal period and describe the surgical treatment for stenosis relief.

Key words: 1. Congenital heart disease
2. Stenosis
3. Aorta
4. Pulmonary artery
5. Williams syndrome

CASE REPORT

A male neonate born at 39 weeks gestational age and weighing 2,660 g was admitted for feeding difficulties along with abdominal distension and jaundice. Physical examination revealed a mid-systolic heart murmur at the left sternal border and a 20 mmHg lower blood pressure in the lower extremities than that observed in the upper extremities. Initial echocardiographic and computed tomographic (CT) findings showed a type A interrupted aortic arch arch (IAA) with a bicuspid aortic valve, a posterior malaligned ventricular septal defect (VSD), and an atrial septal defect (ASD). The annulus of the ascending aorta measured 4.5 mm (z-value=-3.7), and there was no stenosis of the pulmonary artery. The first surgery was performed nine days after birth when the patient weighed 2,700 g. IAA reconstruction by end-to-side anastomosis as well as VSD and ASD closure were performed. Unusual diffuse aortic wall thickening was observed during the surgery.

Systolic blood pressure was measured at 80–90 mmHg without difference between upper and lower limbs after first surgery. Extubation was performed on postoperative day 6 and respiration was stable without tachypnea. On postoperative echocardiography follow-up at one week, diffuse hypoplasia of the aorta with turbulent flow from the sinotubular junction to the IAA anastomosis site as well as peripheral pulmonary artery stenosis (PPAS) were observed. The diameter of the ascending aorta was 1.6 mm and that of the

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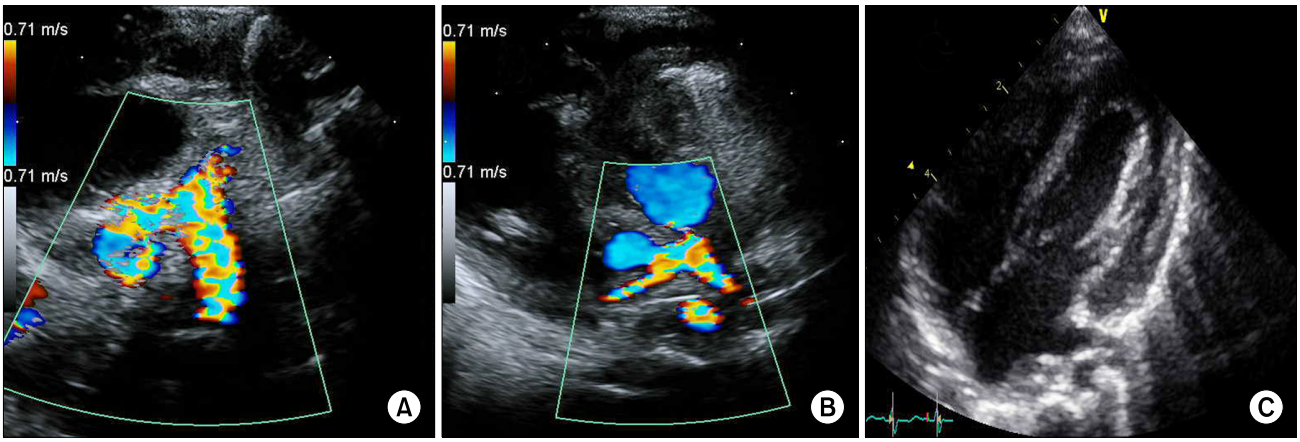

Fig. 1. Echocardiography images one week after the first surgery. (A) Turbulent flow from the ascending aorta to the descending thoracic aorta; the velocity of flow at the aorta ranged from 3.7 m/sec to 4.4 m/sec. (B) Stenosis of the MPA, and flow velocity of 3.5 m/sec at the MPA. (C) Left ventricular hypertrophy and hyperechogenic feature at the left ventricular endocardium. MPA, main pulmonary artery.

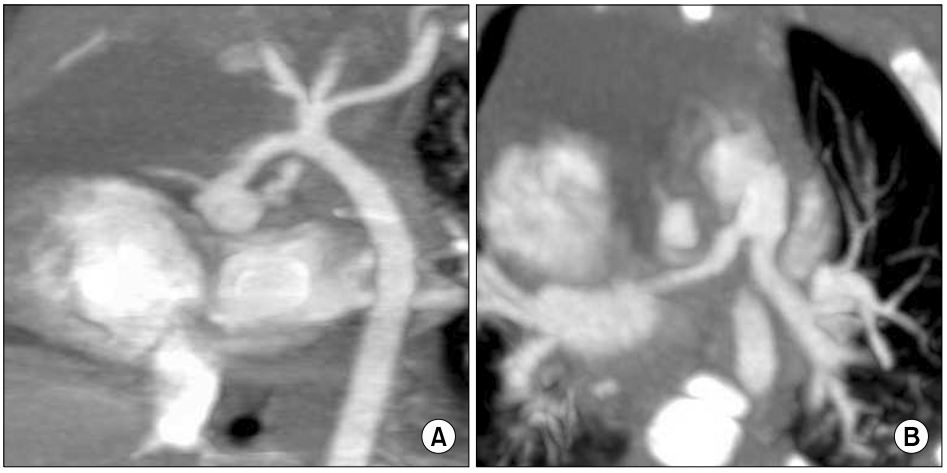

Fig. 2. Computed tomographic angiography images at two months after birth. (A) Hypoplasia of the aorta from the sinotubular junction to the descending thoracic aorta. (B) Peripheral pulmonary artery stenosis.

descending thoracic aorta was 2.8 mm at the anastomosis site. The ratio of the sinotubular junction diameter to aortic annulus diameter was 0.36. The flow velocity at the ascending aorta and IAA anastomosis site was 3.7 m/sec and 4.4 m/sec, respectively. The left and right peripheral pulmonary arterial diameters were 2.1 mm (z-value=-4.68) and 2.2 mm (z-value=-3.92), respectively. The flow velocity at the main pulmonary artery (MPA) was 3.5 m/sec. Left ventricular hypertrophy (LVH) with a hyperechogenic feature regarded as endocardial fibroelastosis was also observed (Fig. 1). The interventricular septal diameter of the left ventricle was 6.1 mm (z-value=2.5), and the left ventricular posterior wall diameter was 6.5 mm (z-value=4.4). On CT angiography before the second surgery, supravalvular aortic stenosis (SVAS) and PPAS were clearly observed (Fig. 2). Taking into account the patient's elfin face and rapid progression of aortic and pulmonary artery wall thickening, a chromosomal study was performed, and the finding of a micro-deletion on chromosome 7q11.23 led to a diagnosis of Williams syndrome (WS). On histopathologic examination, hyperplastic elastic lamina of media was observed at the stenotic aorta using elastic van Gieson's stain (Fig. 3). LVH, diffuse aortic hypoplasia and PPAS were aggravated on follow-up echocardiography.

The second surgery was performed at 77 days after birth when the patient weighed 4,370 g. The Brom's two-sinus patch enlargement technique using bovine pericardium was

performed for SVAS relief (Fig. 4). Two-thirds of the anterior ascending aortic wall was reconstructed using a bovine pericardial patch. The MPA and left peripheral pulmonary artery were widened with a 6-mm-long polytetrafluoroethylene vascular graft patch (Goretex; WL Gore & Associates Inc., Flagstaff, AZ, USA) angioplasty.

On postoperative echocardiography at two weeks after the second surgery, the flow velocity at the ascending and at the IAA anastomosis site was 2 m/sec and 3 m/sec, respectively. Furthermore, LVH deterioration was not observed. The flow velocity at the MPA was 1.1 m/sec, and the diameter of the left and right peripheral pulmonary artery was 3.2 mm and 3.2 mm, respectively. At the five-month follow-up after discharge, the patient was found to be free of symptoms. At follow-up echocardiography a further five months later, the flow velocity of the ascending aorta, IAA anastomosis site, and MPA was 1.3 m/sec, 2.1 m/sec, and 2.1 m/sec, respectively. The left ventricular posterior wall diameter was 7.5 mm (z-value=1.08), and the interventricular septal diameter of the left ventricle was 7.2 mm (z-value=1.44). However, a hyperechogenic feature on the left ventricular endocardium was still observed, although hypertrophy of the left ventricular posterior wall and the interventricular septum was improved.

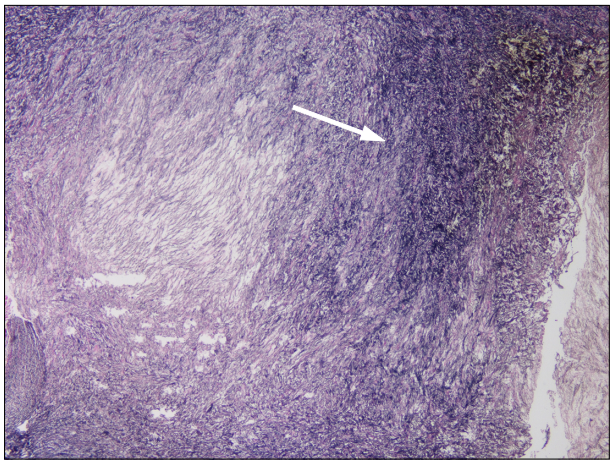

Fig. 3. Hyperplastic elastic lamina of media (arrow) was revealed in pathologic findings (elastic van Gieson's stain, ×40).

DISCUSSION

SVAS is a common feature of WS, found in approximately 70% of all patients with WS. The natural course of SVAS is variable. It may remain stable or progress during the first five years of life. Additionally, obstructions of the pulmonary vasculature have been described in up to 83% of patients with WS, familial elastin arteriopathy, or sporadic non-Williams SVAS. Patients usually show PPAS, and can have localized or diffuse stenosis in the central pulmonary arteries. However, PPAS often resolves spontaneously [1,2]. The mid-term outcome varies with regard to the relief of SVAS or PPAS. In studies on SVAS, the rate of freedom from reoperation or re-intervention ranges from 83% to 98% at five years and from 66% to 83% at 20 years. In studies on pulmonary arterial stenosis, the rate of freedom from reoperation or re-intervention ranges from 32% to 67% at five years [3-7].

In our patient, unusual aortic wall thickening was observed as one of the initial operative findings. Moreover, diffuse ascending aorta and pulmonary arterial stenosis with severe biventricular hypertrophy progressed one week after the initial surgery. In several reports, the mean age for vascular disease treatment in WS patients ranged from 3.3 years to 10.4 years and from 3.8 months to 14 months for SVAS and PPAS, respectively [3-7]. However, Albacker et al. [8] reported a case

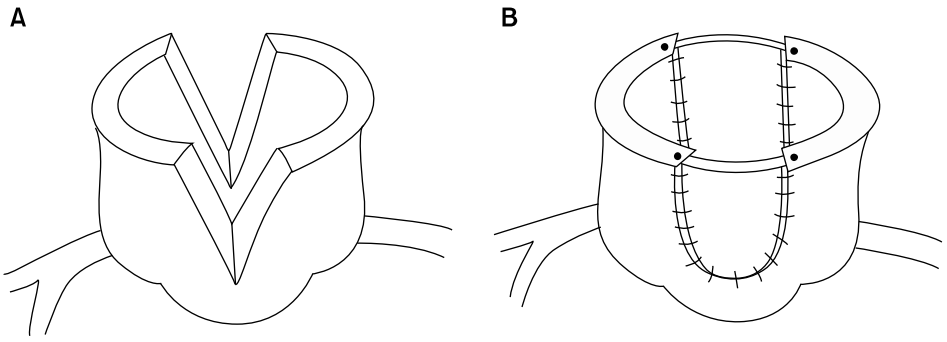

Fig. 4. (A) The two vertical incisions in the aortic sinuses. (B) Accommodation of the bovine pericardial patches in each sinus of Valsalva.

of neonatal presentation and repair of SVAS in a patient with WS. Thus, among WS patients, those with aortic hypoplasia, LVH, and PPAS should raise more concern and should undergo short-term follow-up with echocardiography, although rapid progression of concurrent supravalvular aortic and pulmonary arterial stenosis in the early period is rare.

Here, we report the early manifestation of supravalvular aorta and pulmonary artery stenosis in the neonatal period and appropriate surgical treatment for stenosis relief.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

REFERENCES

1. Stamm C, Friehs I, Ho SY, Moran AM, Jonas RA, del Nido PJ. *Congenital supravalvar aortic stenosis: a simple lesion?* Eur J Cardiothorac Surg 2001;19:195-202.
2. Kim YM, Yoo SJ, Choi JY, Kim SH, Bae EJ, Lee YT. *Natural course of supravalvar aortic stenosis and peripheral pulmonary arterial stenosis in Williams' syndrome.* Cardiol Young 1999;9:37-41.
3. Stamm C, Kreutzer C, Zurakowski D, Nollert G, Friehs I, Mayer JE, et al. *Forty-one years of surgical experience with congenital supravalvular aortic stenosis.* J Thorac Cardiovasc Surg 1999;118:874-85.
4. Kramer P, Absi D, Hetzer R, Photiadis J, Berger F, Alexi-Meskishvili V. *Outcome of surgical correction of congenital supravalvular aortic stenosis with two- and three-sinus reconstruction techniques.* Ann Thorac Surg 2014;97:634-40.
5. Geggel RL, Gauvreau K, Lock JE. *Balloon dilation angioplasty of peripheral pulmonary stenosis associated with Williams syndrome.* Circulation 2001;103:2165-70.
6. Monge MC, Mainwaring RD, Sheikh AY, Punn R, Reddy VM, Hanley FL. *Surgical reconstruction of peripheral pulmonary artery stenosis in Williams and Alagille syndromes.* J Thorac Cardiovasc Surg 2013;145:476-81.
7. Deo SV, Burkhart HM, Schaff HV, et al. *Late outcomes for surgical repair of supravalvar aortic stenosis.* Ann Thorac Surg 2012;94:854-9.
8. Albacker TB, Payne DM, Dancea A, Tchervenkov C. *Management of supravalvar aortic stenosis and severely depressed left ventricular function in a neonate with Williams syndrome.* Eur J Cardiothorac Surg 2009;35:915-6.