Supravalvular Aortic Stenosis and Peripheral Pulmonary Stenosis Coexisting With a Straight Thoracic Spine

Yoichi Uechi, MD; Kuniaki Kaneshiro, MD

Supravalvular aortic stenosis (SVAS) is recognized in cases of Williams syndrome and in sporadic cases not associated with other features of the syndrome. It is also well recognized as associated with peripheral pulmonary stenosis (PPS). A male patient was diagnosed as having PPS at the age of 1 year and 8 months, and was found at the age of 18 years to have SVAS. Cardiac catheterization showed that he had a localized type of SVAS and regression of the PPS. Chest X-ray showed that he did not have the normal thoracic curvature. His 19-year-old sister had also been diagnosed with PPS, and his 43-year-old mother was known to have a harsh systolic cardiac murmur of unknown etiology. Cardiac magnetic resonance imaging showed a localized type of SVAS in his mother also, though not in his sister, both of whom had a somewhat straight thoracic spine, most noticeably in the mother, though not to the degree observed in the patient. This case appears to be familial, though it is not clear whether this skeletal abnormality is an unknown phenotypic feature of this cardiovascular disease. (Circ J 2002; 66: 516–518)

Key Words: Peripheral pulmonary stenosis; Straight thoracic spine; Supravalvular aortic stenosis

Supravalvular aortic stenosis (SVAS) is recognized in sporadic cases, with a dominant inheritance pattern in family members, or in cases of Williams syndrome together with a peculiar face (elfin face), mental retardation, and other phenotypic features. Further, its association with peripheral pulmonary stenosis (PPS) is well known.

A straight thoracic spine, with compression of the heart and great vessels by the chest deformity, has been designated as 'straight back syndrome' and it simulates organic heart disease by causing systolic murmurs and alterations of heart sounds in patients with a normal heart. We present a rare case of SVAS and PPS with a straight thoracic spine.

Case Report

In April 1983, a 13-month old boy was referred to hospital because of a cardiac murmur. He was not cyanotic, and showed no signs of distress. On the second sternal border, a harsh systolic murmur that radiated to both lung fields was audible. Electrocardiography and echocardiography indicated right ventricular hypertrophy. Chest X-ray film showed no pulmonary congestion. A tentative diagnosis of pulmonary stenosis was made at that time. Cardiac catheterization was performed 7 months later. The right ventricular pressure was 57 mmHg in systole, but pulmonary artery pressure could not be measured because of difficulty with insertion of the catheter. A pulmonary angiogram revealed PPS (Fig 1). An oxymetric study indicated no intracardiac shunt. The risk of heart failure was assessed to be minimal, and he was followed up without medication. Until the age of 15, he remained asymptomatic and regular examinations showed no interval changes. In May 2000, at the age of 18 years, his cardiovascular disorder was re-evaluated. He was of normal intelligence and had no complaints of cardiac problems. A harsh systolic murmur that radiated to the bilateral neck vessels on the second sternal border was audible. We realized later that his facial appearance was somewhat peculiar, and partly resembled that of Williams syndrome, including periorbital fullness, long philtrum, and a thick lower lip. He also had sloping shoulders and a long trunk. The lateral view of his chest on X-ray showed that he did not have the normal thoracic curvature (Fig 2).

Electrocardiogram showed no apparent left ventricular hypertrophy or ischemic change, but echocardiography revealed a systolic turbulent flow above the aortic valves. Valvular or subvalvular stenosis was not detected. At this time, left ventricular hypertrophy was not apparent (posterior wall thickness, 8 mm). Trivial mitral regurgitation was diagnosed, although prolapse of the mitral valve was not apparent. We performed cardiac catheterization 3 months later, and found a localized type of SVAS with a pressure gradient of 90 mmHg (Fig 3). No obstruction or significant stenosis was seen in the coronary ostium. The thoracic aorta, the abdominal aorta and the carotid arteries were not stenosed. The right ventricular pressure was decreased at 39 mmHg, but the pulmonary artery trees were of normal size. We recommended surgical treatment and continued his follow-up. A commercially available genetic study using fluorescence in situ hybridization (FISH) was performed with a probe for Williams syndrome (ELN, LIMK1, and D7S613; in combination, not respectively), and revealed no lack of a signal on chromosome 7q11.23.

His 19-year-old sister had been diagnosed with PPS at the age of 2 years and 6 months, and his 43-year-old mother was known to have a harsh systolic cardiac murmur of unknown etiology. Both the sister and the mother were asymptomatic and SVAS was not clear on an echocardiographic study carried out in both of them. We recommended that the sister and mother undergo cardiac catheterization, but they did not agree, so cardiac magnetic resonance...
imaging was performed and showed a localized type of SVAS in the mother (Fig 4), but not in the sister. Both women, and especially the mother, had a somewhat straight appearance of the thoracic spine, though it was not as notable as that in the patient.

**Discussion**

Genetic studies suggest that when the SVAS is familial, it is transmitted as an autosomal dominant with variable expression. The vascular pathology of familial SVAS and Williams syndrome results from mutations involving the elastin gene on chromosome 7q11.23. However, in the present case, the genetic study of Williams syndrome did not reveal a deletion on this chromosome and it is known that the degree of gene deletion in familial SVAS is more subtle than in Williams syndrome. The pathogenic mechanism underlying SVAS is not yet fully understood. Reduced elastin content in the media of developing vessels may lead to recurrent injury and fibrosis, which then increases the vulnerability of the endothelium to hemodynamic stress, leading to intimal proliferation of smooth muscle and fibroblasts, fibrosis, and luminal narrowing.

The association between SVAS and PPS is well recognized and experience with children and adolescents has shown that in untreated patients, the degree of left ventricular outflow obstruction increases with age, whereas the associated PPS may become less severe because of an increase in the systolic distensibility of the pulmonary artery rather than to true luminal growth. The same natural course occurred in the case; that is, regression of the PPS with reduction of right ventricular pressure, together with apparent progression of SVAS. Because of the progressive nature of SVAS, surgical treatment was recommended, either a standard patch aortoplasty or extended aortoplasty. The standard repair is an asymmetric reconstruction of the aortic root and may not completely relieve left ventricular outflow obstruction.

A straight thoracic spine does not have the normal thoracic kyphosis and there is a narrow interval between the vertebral column and sternum on the lateral view of the chest on X-ray. In the present case, we measured the anteroposterior diameter from the level of the anterior border of the body of the 8th thoracic vertebra to the posterior border of the sternum and the transthoracic diameter at the level of the diaphragm. The ratio of anteroposterior to transthoracic diameter was 31%, which was compatible with the criteria of ‘straight back’. In patients with this syndrome, mitral valve prolapse is frequently caused by asynchronous papillary muscle motion related to the ellipsoidal left ventricle, which results from the short internal dimension of the thorax. In the present case, there was trivial mitral valve regurgitation without mitral prolapse. A previous report indicated that a loss of the normal physiological curvature of the dorsal spine can be seen in 9.2% of cases of congenital heart disease. At present, it is unclear whether this skeletal abnormality is an unknown phenotypic feature of SVAS. In patients with Williams syndrome,
some musculoskeletal problems have been recognized, including joint limitation, kyphosis, lordosis, scoliosis etc. However, the straight spine has not been previously reported in patients with this syndrome or with familial SVAS.

Based on a review of previously published reports, this appears to be the first case of a combination of SVAS and regression of PPS, coexisting with a straight thoracic spine. Our findings suggest that this skeletal abnormality should be further researched, and considered during the medical examination of patients with this cardiovascular disease.

References


