Patch aortoplasty for supravalvular aortic stenosis with bicuspid aortic valve associated with Williams syndrome

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Citation

Abstract
We experienced a case in which a 7-year-old boy with a characteristic elfin face, heart murmur, and mental retardation, underwent extended patch aortoplasty using diamond shaped dacron patch for congenital supravalvular aortic stenosis. The aortography performed before operation demonstrated diffuse stenosis just above the aortic valve, which was a typical hour-glass type. The preoperative peak systolic pressure gradient between the left ventricle and ascending aorta was 86 mmHg, and was improved postoperatively. In this procedure no cusp was incised, resulting in no deformity of the aortic valve and no obstruction of coronary arteries. In conclusion this method was excellent for the diffuse type of supravalvular aortic stenosis.

INTRODUCTION
Williams syndrome is a rare neurodevelopmental disorder which occurs in 1/7500 births and is caused by a deletion of about 26 genes from region q11.23 of the long arm of chromosome 7. It is characterized by a distinctive, “elfin” facial appearance, along with a low nasal bridge; mental retardation coupled with unusual language skills and cardiovascular problems, such as supravalvular aortic stenosis and transient hypercalcaemia. The occurrence of bicuspid aortic valve associated with diffuse hourglass type of supravalvular aortic stenosis in Williams syndrome however is rare presentation.

CASE HISTORY
A 7-year-old boy was referred to our clinic because of a cardiac murmur. He had characteristic elfin face (Figure 1), mental retardation and grade 4 systolic murmur.

Figure 1
Figure 1: 17-year old male child showing characteristic “elfin” facial appearance and low nasal bridge.

Aortography, coronary angiography and echocardiography were performed before the operation, and all demonstrated a diffuse hour-glass type stenosis just above the aortic valve which was bicuspid (Figure 2).
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Figure 2
Figure 2: Aortography image showing supravalvular aortic stenosis.

The preoperative peak systolic pressure gradient between the left ventricle and ascending aorta was 86 mmHg. There was no coronary stenosis, other cardiac anomalies or renal artery stenosis. We performed successful surgical repair of patch aortoplasty using 30mm diamond shaped Dacron prosthetic patch. After standard cardiopulmonary bypass, vertical incision was made between the two cusps; and the ridge was removed with sharp dissection above the coronary sinus. Resulting defect on the aorta was then repaired with Dacron patch (Fig.3, Fig. 4).

Figure 3
Figure 3: Intraoperative view of completed repair with 30mm diamond shaped Dacron prosthetic patch.
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**Figure 4**

Figure 4: Schematic diagram showing completed repair with 30mm diamond shaped Dacron prosthetic patch.

The systolic pressure gradient between the left ventricle and ascending aorta improved to normal postoperatively. The patient was extubated 7 hours following surgery, and was discharged at the 5th postoperative day. Six months after the procedure, the patient was fine and had no complaints.

**DISCUSSION**

Supravalvular aortic stenosis is an unusual form of obstruction of the left ventricle outflow tract, which occurs in 3% to 6% of cases of various types of aortic obstruction. It may be due to the presence of a discrete fibrous membrane, an hourglass narrowing, or a diffuse narrowing. The anatomical variants of supravalvular aortic stenosis affect post surgery results and survival rates, e.g. the hourglass type of obstruction is the most common, whereas the diffuse type of stenosis mostly results in death [1,2]. The syndrome was first described in 1930 by Mencarelli, and its association with other facial defects and psychomotor retardation was established in 1961 [1]. In 1965, Logan described patients who had a familial form of supravalvular aortic stenosis with normal face and normal mental development, which may delay diagnosis [13]. Supravalvular aortic stenosis is a familial autosomal dominant disorder with variable expression and affects both sexes equally. The vascular pathology of familial supravalvular aortic stenosis and Williams syndrome results from mutations involving the elastin gene on chromosome 7q11.23 [14]. In 1961, McGoon et al. proposed widening the aorta diameter with a synthetic graft as a treatment option. In 1977, Doty used his well-known technique, which involves an inverted Y patch for the first time with success. When the stenosis is very close to the valves and the coronary ostia, the use of the Doty technique may be preferred [15]. There are several variations of the surgical technique for correcting this type of defect. In our case we successfully used the diamond shaped patch technique for diffuse type supravalvular aortic stenosis with bicuspid aortic valve.

In a study concerning the efficacy of several techniques, Hazekamp et al., did not find any significant differences in change in valve function, and found the efficacy of reducing the pressure gradient was similar and acceptable in all techniques [16]. In the cases of recurrent serious stenosis, an alternative is to use a valved conduit between the free wall of aortic root and the stenosis free aorta [17]. Similarly, autologous arterial graft from the pulmonary artery, as described by Al-Halees et al, can be used as another option not only for cases of recurring stenosis but also for complex cases with diffuse stenosis.

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**References**

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