

Supravalvar Aortic Stenosis in William Syndrome-Case Report and Review of Literature

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Abstract

Williams syndrome is a rare genetic disorder also called as Williams-Beuren syndrome. It causes many developmental problems. Parents may not have any family history of the condition. However, people with Williams syndrome have a 50% chance of passing the disorder on to each of their children. Williams syndrome occurs in about 1 in 8,000 births. Facial features frequently include a broad forehead, short nose, and full cheeks, an appearance that has been described as “elfin”. Mild to moderate intellectual disability with particular problems with visual spatial tasks such as drawing and fewer problems with language are typical. Problems with teeth, heart problems, especially supravalvular aortic stenosis, and periods of high blood calcium are common. SVAS may occur sporadically, as a manifestation of elastin arteriopathy, or as part of Williams syndrome, an autosomal dominant genetic disorder. The sporadic form of SVAS is the most common (> 50%) presentation. Here we report a case of 11 year old girl of William syndrome diagnosed with supravalvar aortic stenosis. Broms procedure was done and patient relieved of symptoms. Intraopt and postopt management dose not differ in William syndrome.

Keywords: Williams Syndrome; Supravalvular Aortic Stenosis; Sinotubular Junction

Abbreviations

WS: Williams Syndrome; SVAS: Supravalvular Aortic Stenosis; STJ: Sinotubular Junction

Introduction

Williams syndrome (WS), also referred to as Williams-Beuren syndrome (Online Mendelian Inheritance in Man 194050), is a congenital, multisystem disorder involving the cardiovascular, connective tissue, and central nervous systems [1]. WS occurs in ≈ 1 in 10,000 live births [2] as a result of the de novo deletion of ≈ 1.55 to 1.83 Mb on chromosome 7q11.23 [3]. Familial cases can occur but are far less common than de novo cases [4]. The deletion involves 26 to 28 genes, including the ELN gene, which codes for the protein elastin. Hemizygosity of the ELN gene coding for elastin has been demonstrated to be responsible for the vascular pathology in WS [5]. The remaining 25 to 27 deleted genes contribute to the phenotypic findings in patients with WS and have recently been reviewed in detail elsewhere. In 1961, Williams., *et al.* [6] reported their experience with 4 patients with supravalvar aortic stenosis (SVAS), mental retardation, and abnormal facial features. The following year, Beuren and colleagues reported similar findings in 5 patients, and they subsequently reported detailed cardiac and angiographic data from 10 such patients [7]. Their findings, combined with other characteristic features, led both groups to theorize that a previously unrecognized syndrome was the likely origin, a theory that led to the eponym Williams-Beuren syndrome.

Supravalvular aortic stenosis (SVAS) is a rare anomaly in which there is an exaggerated narrowing at the sinotubular junction. It can either be a localised narrowing of STJ or a diffuse variety affecting the ascending aorta, aortic arch and its branches. This anomaly is often

associated with Williams syndrome. There may be generalized hypoplasia of the ascending aorta and more distal arterial tree as well as stenoses in the pulmonary artery tree. Williams syndrome been associated with congenital cardiac malformations in approximately 10% of patients and symptomatic narrowing of arteries in up to 80%. Supravalvular aortic stenosis (SVAS) is reported to be the most common cardiovascular abnormality in Williams syndrome. There are few published reports of the results of supravalvular aortic stenosis correction with the use of Brom's three-patch technique. Nagre., *et al.* reported a case of four patch repair in his study [8]. Aortic repair involved the use of pericardium, Dacron, or both. Herein, we report our use of this procedure and the short-term followup results. We consider Brom's technique to be safe and effective in the repair of supravalvular aortic stenosis.

Case Report

A 11-year-old girl admitted with history of slowly increasing breathlessness since 3 years. On clinical examination ,She had a systolic heart murmur grade 3/6 in her right upper sternal border with radiation to right cervical region. The patient had a typical face with bulge forehead, broad nose, broad lips, increased interdentine distance (Figure 1A) and also she had a speech problem; although she had a good auditory rote memory and could follow the orders, she could not normally say the words and sentences. Echocardiography revealed a severe supravalvar aortic valve stenosis with gradient of 90 mm of HG ; then CECT done showing supravalvar aortic stenosis (Figure 1B) with small left SVC and normal size right SVC. After that, with suspicious to Williams' syndrome, the calcium level was done which was raised. Patient planned for Brom's three patch repair for the supravalvular aortic stenosis on CPB under GA.

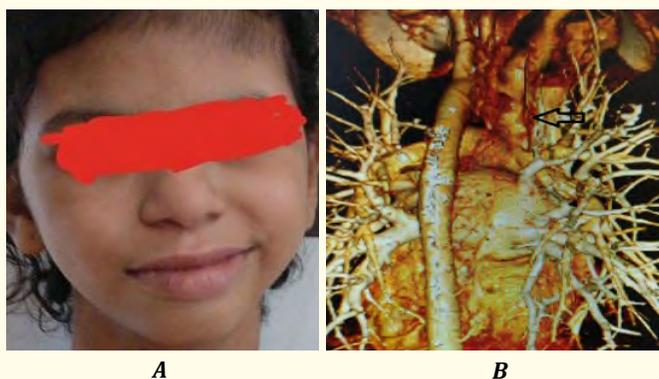


Figure 1: A. Facial features of William Syndrome like broad nose, broad lips and increased interdentine distance
B. CECT showing Supravalvar Aortic Stenosis.

The surgery was performed with cardiopulmonary bypass, cannulation of aorta and vena cava using antegrade cold blood cardioplegia (4°C) for myocardial protection, with the patient at 31°C. Pericardial patch harvested. Figure 2A shows the anatomical aspect of supravalvular aortic stenosis before surgical correction. Transverse incision was performed in the aorta just above the stenosis, which showed: significant stenosis of the sinotubular junction, extensive fibrosis of the aortic wall (Figure 2B). The aortic valve was normal with adequate coaptation.

In the case reported in this article, First we excised the excessive fibrotic tissue above the valve (Figure 3A) and then performed three sections: the first toward the non-coronary sinus and the second toward the bottom of the right coronary sinus, at left of the ostium of the right coronary artery and the third section toward the left coronary sinus, at right of the left coronary artery ostium (Figure 3B). Harvested pericardial patch was cut in three triangular pieces according to the size of supravalvar aorta we wanting (Figure 4A). In all the three sections of sinuses three triangular pericardial patch sutured with prolene 4-0 (Figure 4B, 5A and 5B). An end-to-end aortic anastomosis was made connecting the reconstructed aortic root to the ascending aorta (Figure 6A and 6B).

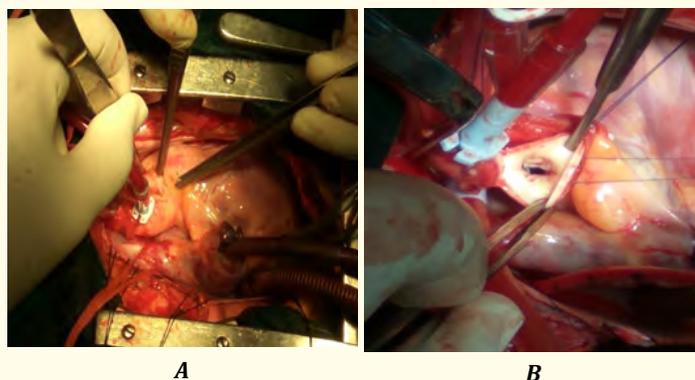


Figure 2: A. Supravalvar Aortic Stenosis. B. Transverse aortotomy showing supravalvar aortic stenosis with normal aortic valve.

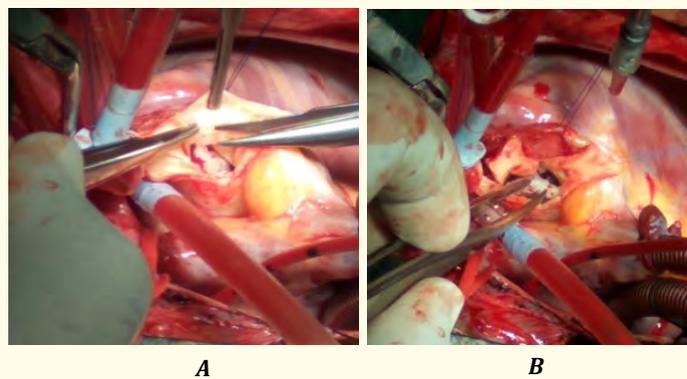


Figure 3: A. Excising excessive supravalvar tissue with scissor. B. Three longitudinal incisions extending in three sinuses without damaging coronary ostias.

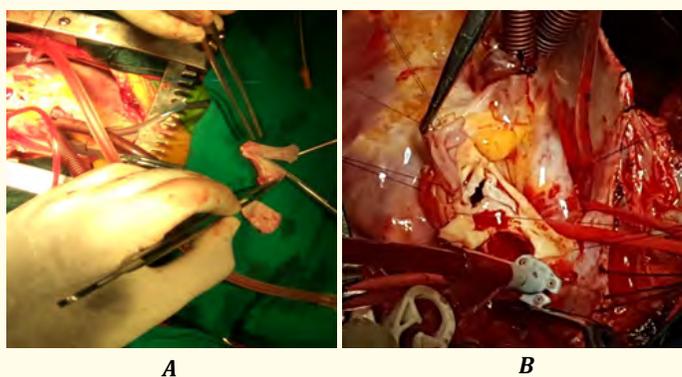


Figure 4: A. Pericardial patch cutting in three triangular pieces. B. Suturing first pericardial patch in left coronary sinus.

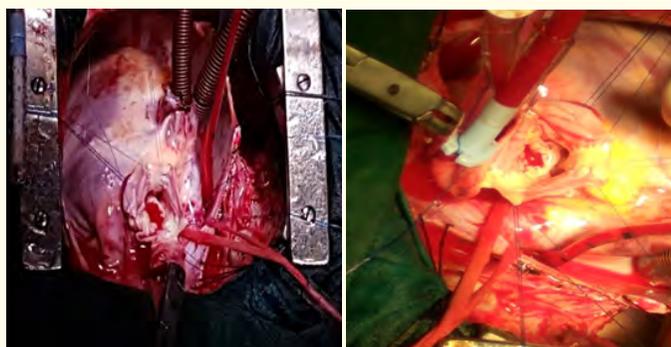
**A****B**

Figure 5: A. Second triangular pericardial patch sutured in right coronary sinus. B. After suturing third triangular pericardial patch in noncoronary sinus.

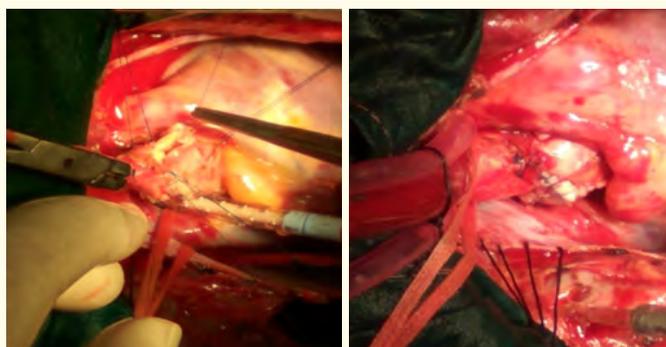
**A****B**

Figure 6: A. Closure of aortotomy. B. After complete closure of aortotomy.

We performed the rewarming of the patient at 37°C, removal of air from the left cavities and aorta, declamping of the aorta, heart rate return to normal, disconnection of cardiopulmonary bypass (CPB), decannulation, protamine administration and hemostasis. Postoperative transesophageal echocardiographic assessment was performed. The postoperative follow-up was uneventful, with a maximum systolic gradient of 15 mmHg and mean of 5 mmHg, good mobility of valves without reflux (transthoracic echocardiogram). The patient was discharged from hospital on the 9th day.

After nine month of follow-up, a patient was in functional class I, without cardiovascular symptoms. During this period the echocardiogram showed tricuspid aortic valve, slightly thickened with minimum reflux under color-Doppler, generating peak systolic valve gradient of 10 mmHg and supravalvular of 12 mmHg.

Discussion

Surgical techniques for repair of supravalvular aortic stenosis (SVAS) are numerous which include McGoon's one patch, Doty's two patch, and Brom's three-patch method [9]. Data definitively supporting one technique over another have been elusive. No technique is considered gold standard for SVAS repair. McGoon and colleagues described a single-sinus technique in which the noncoronary sinus and proximal ascending aorta were enlarged with a diamond-shaped patch. Doty and coworkers developed a two-sinus technique with inverted bifurcated patch aortoplasty of the right- and noncoronary sinuses. More recently a number of three-sinus repairs have been described including the Brom three-patch aortoplasty, the Myers all-autologous slide-aortoplasty, and a similar autologous method described by Chard [9].

Proponents of three-sinus repairs postulate that persistent geometric distortion and secondary effects on the leaflets may occur with more limited reconstructions and compromise long-term aortic valve function. Theoretically, three-sinus repairs restore more normal aortic root anatomy and will improve long-term outcomes. Stamm and colleagues demonstrated that the ratio between aortic leaflet free margin length and the sinotubular junction circumference is significantly higher in patients with SVAS than in control hearts. However, the ratio between the leaflet free margin and the circumference at the base of the leaflet hinge points was the same in patients with SVAS and control normals. This finding indicates that in most cases the leaflet dimensions are normal and that enlargement of all 3 sinuses can restore normal outflow tract anatomy. Most recent series favor the Brom repair for SVAS, and this is our preferred approach. In the presence of diffuse disease, the Brom technique is modified to include enlargement of the distal ascending aorta and aortic arch when necessary.

Conclusion

Brom's three patch repair produced good early postoperative results. Patient improved symptomatically with lower transaortic peak pressure gradients. Though the Brom's procedure technically looks challenging but if proper surgical steps are followed, it becomes easy with good results. William syndrome association with SVAS does not affect management plan and prognosis of patient.

Consent

Informed consent has been obtained.

Funding

No funding was required for this study.

Conflict of Interest

No potential conflict of interest exist.

Ethical Approval

All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

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