Case report

Management of supravalvar aortic stenosis and severely depressed left ventricular function in a neonate with Williams syndrome

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Abstract

We report an interesting case of a patient with Williams syndrome who presented with moderate supravalvar aortic stenosis and bilateral pulmonary artery stenosis at one week of age. The supravalvar aortic stenosis became severe by the age of one month with severe depression of left ventricular function. The patient had a difficult postoperative course, developed an acquired aortic arch hypoplasia and required multiple interventions during the first two months of life with an excellent outcome. The management of this difficult patient is discussed with focus on the importance of close follow-up, early diagnosis and early surgical intervention in improving the outcome in this difficult group of patients.

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1. Introduction

Williams syndrome is a genetic disorder associated with a microdeletion in the chromosomal region 7q11.23 encompassing the elastin gene. Previous reports suggest that hemizygosity of the elastin gene is responsible for the typical vasculopathy of WS, namely supravalvar aortic stenosis and pulmonary arterial stenosis [1]. Early intervention and extensive aortic involvement at diagnosis is associated with poor outcome [2]. Progression of the aortic stenotic lesion can occur over a period of 3 months to 20 years and lower pressure gradients at diagnosis may predict stability of the stenosis during the follow-up [3]. We report a case of aggressive presentation and difficult management of supravalvar aortic stenosis in a patient with Williams syndrome during neonatal period with good outcome.

2. Case report

One-week-old neonate with Williams syndrome was found to have moderate supravalvar aortic stenosis (peak gradient = 56 mmHg) and bilateral pulmonary artery stenosis with normal aortic arch (Fig. 1) and intact left ventricular function (LVF). Follow-up echocardiogram at one month of age showed progression of the stenosis with a peak gradient of 64 mmHg and severely depressed LVF [ejection fraction (E.F.) = 20%]. The patient underwent urgent repair of the supravalvar aortic stenosis with a bifurcated pulmonary homograft patch using Doty's technique [4]. Postoperative echocardiogram showed that there was no gradient across the supravalvar area and peak gradient of 18 mmHg in the transition zone between the patch augmented ascending aorta and the aortic arch. The left ventricular function was followed with weekly echocardiograms, which showed rapid significant recovery over two weeks. During the third week, multiple echocardiograms showed rapid progression of the peak gradient at the proximal arch just at the origin of the innominate artery from 18 to 50 then 90 mmHg with mild to moderate depression of LVF again. The patient underwent angiography, which showed a long segment of stenosis at the aortic arch distal to the previous repair (Fig. 2) that was not amenable to balloon angioplasty. The decision was made to perform an urgent surgery next day to repair the arch hypoplasia and to prevent further deterioration of LVF. Unfortunately, the patient arrested the same night, was resuscitated and transferred to the intensive care unit where she stayed intubated and received inotropic support. After 24 h of support, the patient had good parameters of perfusion in both upper and lower limbs but with absent pulses. There was no improvement of LVF by echocardiogram and she was taken for urgent surgery. Intraoperatively the whole arch, which was normal in the initial surgery, appeared severely thickened and fibrosed. A pulmonary homograft patch was

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used to augment the whole arch and the proximal part of the descending aorta, just distal to the left subclavian artery, under deep hypothermic circulatory arrest. Postoperative echocardiogram showed no significant gradient across the ascending aorta, arch or descending aorta. The patient stayed on inotropic support for two weeks and achieved near complete recovery of LVF after one month. The patient was discharged home six weeks after the second operation in stable condition. The patient had a follow-up echocardiogram at 1, 4 and 12 months postoperatively with no evidence of recurrent stenosis in the ascending aorta or the arch and had normal LVF.

3. Comments

We describe a number of unique features in this case. First, the rapid progression of the disease with the need for multiple interventions within a short period of time which brings the attention to the importance of the close and frequent follow-up of those patients. Second, it has been described by Wessel et al. [3] that peak gradients less than 20 mmHg at presentation may predict stability or slow progression of the lesion during future follow-up. However, our patient developed rapidly progressing stenosis with arch hypoplasia despite an initial peak gradient of 18 mmHg in the first follow-up echocardiogram. This difference in behavior of the disease may be related to the age at presentation, which was during neonatal period in our patient compared to an average age of 4.4 years in the study published by Wessel. Third, patients with supravalvar aortic stenosis with aortic arch hypoplasia tend to develop restenosis distal to the patch after surgical repair. On the other hand, patients with normal aortic arch tend to be free from developing a restenosis at the distal end of the repair of supravalvar aortic stenosis [3]. On the contrary, our patients developed a new stenosis of the arch distal to the previous repair despite a normal aortic arch in the initial surgery. Reduced and abnormal elastin content in the media of the vessels in these patients may lead to recurrent injury and fibrosis. Vascular inelasticity may increase hemodynamic stress to the endothelium leading to intimal proliferation of smooth muscle and fibroblasts, fibrosis, and luminal narrowing of the vessels [1]. This suggests that those patients may react to any surgical procedure with exaggerated inflammatory and healing responses resulting in future restenosis regardless of the involvement of the aortic arch. This raises the question whether or not the aortic arch should be augmented with an extended patch during the repair of any supravalvar aortic stenosis in patients with Williams syndrome regardless of the gross involvement of the arch. Fourth, rapidly progressive arteriopathy in neonatal period in the setting of Williams syndrome has been reported only once before in the form of aortic coarctation [5]. Our patient represents the first reported case of neonatal presentation and repair of supravalvar aortic stenosis with severely impaired left ventricular function in the setting of Williams syndrome. The natural history of those patients is usually dismal with high risk of sudden death, the mechanism of which was described before [6]. Close follow-up and prompt intervention is warranted in those patients in order to improve their prognosis.

References