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Case Report Open Access

Surgery of mitral valve disease and coarctation of the aorta in Williams syndrome

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ABSTRACT

Although coarctation of the aorta is frequently diagnosed and treated in childhood, some cases are unable to be diagnosed until adulthood. Rarely, additional cardiovascular problems may accompany coarctation of the aorta in patients with genetic disturbances such as Williams syndrome. Herein, we report a case who presented to emergency service with symptoms of congestive heart failure and atrial fibrillation and underwent early, one-step surgery for severe mitral valve regurgitation and coarctation of the aorta after cardiac compensation. Genetic study confirmed the diagnosis of Williams syndrome.

Keywords: Coarctation of aorta; mitral valve regurgitation; one stage operation; Williams syndrome.

Williams syndrome is a hereditary, progressive, multi-system disease characterized by peripheral vascular disorders, mostly supravalvular aortic stenosis (SVAS) and peripheral pulmonary stenosis (PPS), dysmorphic "elfin facies", a characteristic cognitive profile, mild to moderate mental retardation and developmental disabilities.[1] Although supravalvular aortic stenosis (SVAS) tend to progress with age, PPS usually becomes milder.[1,2] We, herein report an interesting case demonstrating that cardiac abnormalities of Williams syndrome may not be solely confined to peripheral vascular stenosis and very rarely atypical presentations may also occur including tetralogy of Fallot, coarctation of the aorta (CoA), and severe mitral valve prolapse (MVP), as in our case.

Coarctation of the aorta constitutes 6 to 8% of congenital cardiac malformations.[1] Additional cardiac pathologies may accompany CoA, necessitating open cardiac surgery.[1] In patients with additional cardiac malformations and clinically unstable condition, two-step surgery may increase mortality and morbidity. Therefore, one-step procedure seems preferable and may decrease risk.

We, herein report a case who presented to emergency service with severe cardiac symptoms and was diagnosed with severe mitral valve regurgitation due to MVP. The patient underwent one-step successful surgery for accompanying CoA.

CASE REPORT

A 22-year-old male patient was admitted to the emergency service department with complaints of dyspnea, orthopnea, tachycardia, and syncope. The patient was hospitalized with the diagnosis of New York Heart Association (NYHA) Class III-IV acute decompensated congestive heart failure. His previous history included rheumatic heart disease, heart failure, and infective endocarditis. Blood pressure was 140/70 mmHg in both arms, the mean pulse rate was 160 bpm and in tachycardia. Physical examination revealed 4-5/6 systolic ejection murmur, bilateral rales up to the mid-lung fields and mild pretibial edema. Femoral pulses were not palpable and ankle-brachial index was 0.7. Electrocardiography was notable for atrial fibrillation with high ventricular response. Chest radiograph demonstrated pulmonary congestion and left atrial enlargement. Echocardiography showed severe mitral and tricuspid regurgitation, MVP, gigantic left atrium (10 cm), and a maximum systolic pulmonary artery pressure of 40 mmHg.

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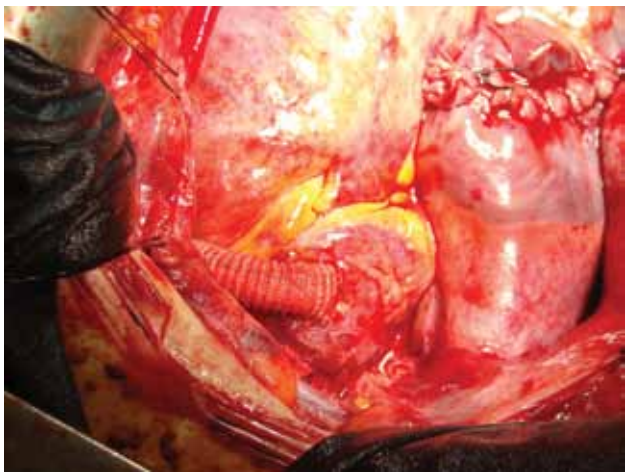

Figure 1. Graft placement during operation.

Suprajugular examination demonstrated systolic gradient in descending aorta with a peak value of 60-100 mmHg. Meanwhile, intravenous diuretics for congestion and digoxin and carvedilol for rate control of atrial fibrillation was initiated. The patient experienced polymorphic ventricular tachycardia and subsequent ventricular fibrillation. Defibrillation and brief cardiovascular resuscitation were performed. Amiodarone infusion converted rhythm to sinus and prevented ventricular tachycardia. Due to frequent ventricular premature beats and bradycardia, digoxin was withheld. Cardiac catheterization revealed normal coronary arteries, Grade 3 mitral regurgitation and coarctation of the aorta with a peak-to-peak systolic gradient of 60 mmHg, just distal of the left subclavian artery. A one-step surgery for correction of both pathologies was planned.

The patient was monitored with arterial tracings in both radial and femoral arteries under general anesthesia. Simultaneous radial artery pressure was observed as 130/80 mmHg, when femoral artery tracings demonstrated 60/30 mmHg. After aortic and bicaval cannulation, cardiopulmonary bypass was started. The apex was lifted and pericardium was dissected to expose the coarctated segment of the descending aorta. Hypoplastic descending aorta (1 cm in diameter in a 10 cm segment) was seen. The distal end of the coarctation was anastomosed with an 8 mm Dacron graft and this segment was cannulated to perfuse the descending aorta after bleeding control. Following cross-clamping and cardioplegia, mitral valve was evaluated by the left atrial dissection. As mitral valve was degenerated and ineligible for repair, it was replaced with a 31 mm Carbomedics prosthetic valve (CarboMedics Inc., Austin, TX). Then, tricuspid De Vega annuloplasty was performed for functional tricuspid regurgitation and cross-clamp was removed. The proximal end of the graft was anastomosed to ascending aorta with a sided-clamp (Figure 1) and cardiopulmonary bypass was stopped. After the operation, simultaneous blood pressure readings were 110/70 mmHg in radial artery and 100/65 mmHg in femoral artery. Postoperative ankle/brachial index increased from 0.7 to 1. The patient remained asymptomatic and was discharged at the 10th postoperative day. Postoperative control

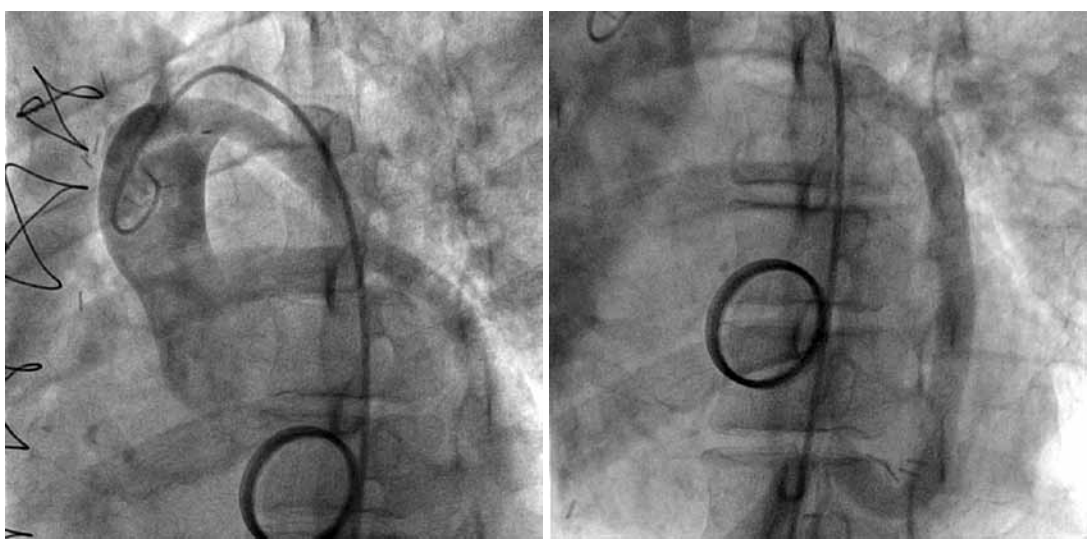

Figure 2. View of the graft in postoperative aortography.

aortography was normal (Figure 2a, b). His functional class was assessed as Class I in his first outpatient follow-up visit.

DISCUSSION

Williams syndrome is characterized with morphological facial features including curly hair, wide forehead, periorbital fullness, short nose with a bulbous tip, long philtrum, wide mouth, full cheeks and small, spaced teeth.[1] Our case also demonstrated same morphological characteristics signs since his toddler age.

Cardiovascular system is most frequently (80%) involved in patients with WS. Several published data reported the incidence of SVAS between 38-100%, and PPS between 16-100%. Moreover, MVP is also observed in patients with WS due to alterations in elastin gene.[2] Bruno et al.[3] reported the incidence of MVP as 27% in their series, the third most common anomaly following SVAS (71%) and PPS (38%), and these incidences were more frequent than previous reports. Our patient had severe mitral regurgitation due to MVP.

As another important pathology in WS, arteriopathy is a systemic disease caused by alterations of the elastin gene.[2] Coarctation of the aorta and renal artery stenosis apart from SVAS and PPS may also be seen. A thorough review reported stenosis of great arteries in 20% of patients without concomitant CoA.[2] On the contrary, Yau et al.[4] reported CoA in only 6% of cases and no arterial stenosis in other territories. Coarctation of the aorta in adulthood is usually an isolated condition. Surgical options include resection and end-to-end anastomosis, subclavian flap technique, reverse subclavian flap, patch-graft aortoplasty and graft interposition. Any of these techniques may be utilized in one-step or two-step surgeries in stabilized patients either with isolated CoA or co-existing anomalies. However, in patients with unstable cardiovascular condition, as in our patient, a one-step procedure correcting both anomalies may decrease associated morbidity and mortality.

Heinemann et al.[5] reported several surgical techniques to repair co-existing cardiac anomalies. Yilik et al.[6] and Bardakci et al.[7] used abdominal aorta as the distal site of anastomosis in a similar case with mitral valve disease and CoA. However, we did not prefer this method due to hypoplastic descending aorta and possible risk of intraperitoneal hemorrhage.

In conclusion, in the event of serious hemodynamic derangement in patients with coarctation of the aorta and coexisting cardiac anomalies, one-step surgery seems successful and feasible.

Declaration of conflicting interests

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