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CASE REPORT

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Ascending aorta dilatation for pulmonary atresia with ventricular septal defect: a report of three adult cases

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Abstract

Background Patients with pulmonary atresia and ventricular septal defect (PA/VSD) are prone to progressive aortic dilation. However, there are relatively few reports of progressive development of aortic aneurysm or aortic dissection in adult patients who missed early corrective surgery.

Presentation of cases Case 1: A 38-year-old man with PA/VSD and a bicuspid aortic valve (BAV), underwent VSD repair, aortic valve replacement, and PA correction at age 21. Seventeen years after surgery, an aortic dissection occurred in the ascending aorta, which subsequently underwent the Bentall procedure. Case 2: A 33-year-old male with PA/VSD, and a Nakata index of 31.24 mm^2/m^2, underwent a central shunt surgery at age 17. Sixteen years after surgery, an aortic root aneurysm and ascending aortic dilatation (AAD) developed. Case 3: A 42-year-old female underwent corrective surgery for PA/VSD repair at age 14. Twenty-eight years after surgery, an AAD developed.

Conclusions Adult patients with PA/VSD who miss the optimal age for surgery are more likely to develop dilatation of the ascending aorta and are at risk for aortic dissection. Therefore, long-term follow-up and monitoring is needed in this patient population.

Keywords Pulmonary atresia, Aortic dissection, Ascending aorta dilatation

Background

Pulmonary atresia with ventricular septal defect (PA/VSD) is a rare congenital heart disease (CHD), with approximately 7 cases per 100,000 live births, accounting for 1–2% of all children with CHD [1]. However, there are few reports on the long-term outcomes of adult PA/VSD patients who missed the age for corrective surgery. We report three cases of postoperative long-term progression to aortic root aneurysms in adult PA/VSD patients, one of which developed ascending aortic dissection.

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Case presentation

Case 1

A 38-year-old male with PA/VSD, BAV presented to our hospital at the age of 21 years. Preoperative physical examination showed cyanosis of the lips and skin, a grade III/6 systolic murmur in the third and fourth intercostal space at the left sternal margin, and a diastolic murmur in the aortic valve area. Echocardiography (Table 1, Fig. 1A) showed PA, VSD (33 mm in diameter), overriding of the aorta (more than 75%), and BAV. Cardiac catheterization (Table 1, Fig. 1B) showed major aorto-pulmonary collateral arteries (MAPCAs). The preoperative diagnoses were PA/VSD aortic regurgitation, BAV abnormality, MAPCs, tricuspid valve insufficiency, and patent foramen ovale (PFO). The Nakata index was 145.63 mm2/m2. The diseased aortic valve was resected and a 31# mechanical valve (St.

Table 1 Summary of clinical characteristics

Variables	Case 1	Case 2	Case 3
Age (years)	38	32	42
Gender(female/male)	m	m	f
Tchervenkov Classification [2]	B	B	
Age at the First Surgery (years)	21	17	14
Pre-operative ECHO(First Admission)			
LVEF(%)	41	60	
LVEDD(mm)	38	43	
Aortic Root Diameter (mm)	34	42	
Ascending Aorta Diameter (mm)	43	36	
Cardiac CATH(First Admission)			
MAPCAs Diameter (mm)	6	7	
LPA Diameter (mm)	12.3	6	
RPA Diameter (mm)	10.7	5	
CTA(Current Admission)			
Aortic Root Diameter (mm)	68	80	55
Ascending Aorta Diameter (mm)	102	63	59
MPA Diameter (mm)	32	10.7	44
LPA Diameter (mm)	13	13.2	14.9
RPA Diameter (mm)	12	10.8	14.6
ECHO (Current Admission)			
LVEF(%)	62	45	66
LVEDD(mm)	40	62	64
Interventions	Bentall procedure	Medication	Medication

ECHO: Echocardiography; LVEF: left ventricular ejection fractions; LVEDD: left ventricular end-diastolic dimension; MAPCAs: major aortopulmonary collateral arteries; LPA: Left pulmonary artery; RPA: Right pulmonary artery; CTA:Computed tomography angiography

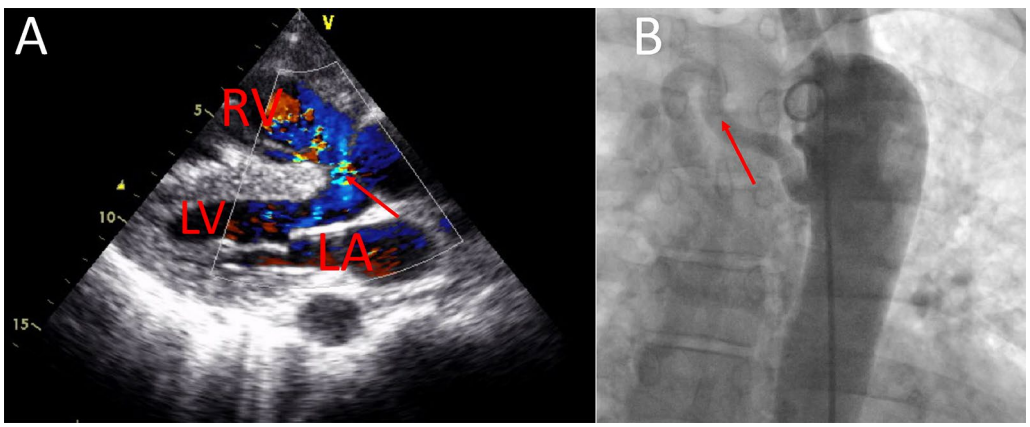

Fig. 1 Preoperative echocardiography and cardiac catheterization: **A** shows the VSD (red arrow), **B** illustrates the MAPCAs arising from the descending aorta (red arrow)

Jude Medical) was implanted. The VSD was repaired with a Dacron patch, and a 4/0 Prolene continuous suture established the connection between the left ventricle and the aorta. The right ventricular outflow tract was reconstructed with an autologous pericardial patch. The PFO was closed, and the tricuspid valve was reshaped using DeVega's method. Postoperative echocardiography showed no residual shunt in the VSD and no residual pressure gradient after right ventricular outflow tract reconstruction. The patient received

warfarin anticoagulation. After discharge, the patient had no regular follow-up.

Seventeen years post-surgery, the patient developed chest pain and dyspnea following physical labor. Echocardiography and computed tomography angiography (Table 1, Fig. 2 A and B) showed an aortic root aneurysm and dilatation of the ascending aorta with dissection. Surgery was performed via median sternotomy, with femoral artery, superior and inferior vena cava cannulation to establish extracorporeal circulation. The intraoperative exploration revealed severe dilation of the ascending aorta with a diameter of 15 cm (Fig. 3A) and dissection. A tear was found 10 cm above the valve ring on the right wall of the ascending aorta (Fig. 3B), and a 6 mm residual leak at the VSD. A 25 mm valved conduit replaced ascending aorta, and the coronary arteries were reimplanted (Bentall procedure). The VSD residual leak was sutured with 4–0 Prolene. Postoperatively, the patient was managed with warfarin anticoagulation and beta-blocker.

Case 2

A 33-year-old male with PA/VSD presented to our hospital at the age of 17. Physical examination revealed cyanosis of the skin and lips, A grade III/6 systolic murmur is present in the third and fourth intercostal space at the left sternal margin. Echocardiographic examination (Table 1) showed a tricuspid aortic valve, an aortic

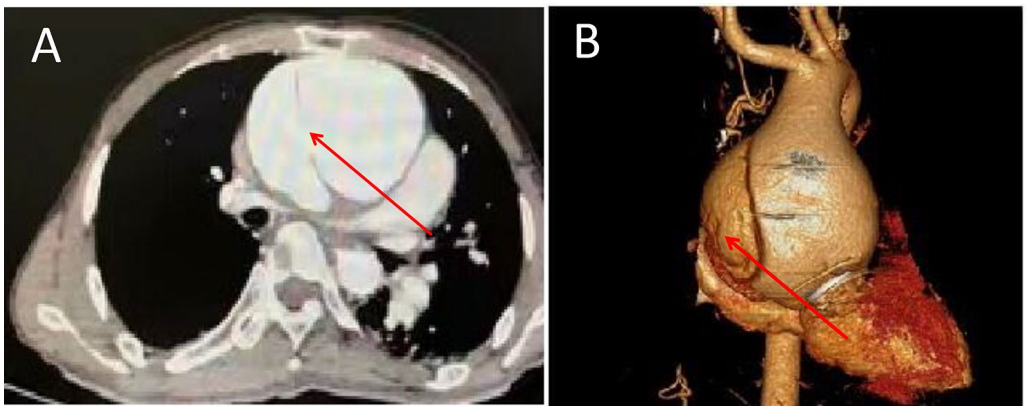

Fig. 2 Preoperative computed tomography angiography (CTA) of aortic dissection: **A** and **B** show the dilated ascending aorta and aortic dissection (red arrow)

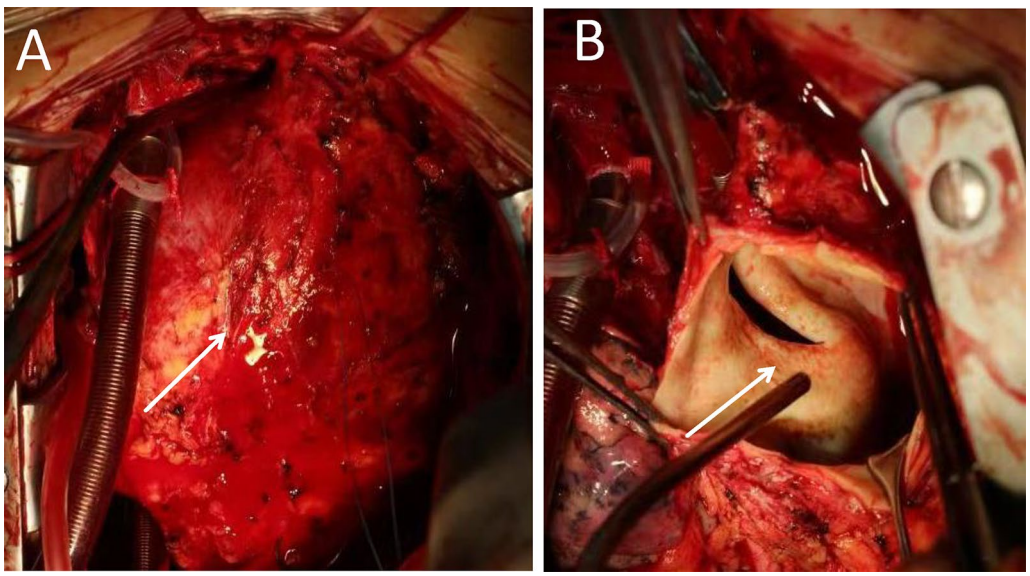

Fig. 3 Intraoperative findings: **A** shows the ascending aortic aneurysm (white arrow), and **B** illustrates the intimal tear of the aortic dissection (white arrow)

overriding rate of approximately 50%, and a subarterial VSD measuring 31mm. Cardiac catheterization revealed MAPCAs (Fig. 4A and B). The diagnosis included PA/VSD with MAPCAs. The Nakata index was 31.24 mm^2/m^2. Due to inadequate development of the pulmonary artery, the patient underwent a central shunt procedure with a median sternotomy, using a 6 mm Gore-Tex artificial graft to connect the aorta to the main pulmonary artery. Postoperatively, the patient was on long-term oral antiplatelet therapy. Sixteen years after surgery, the patient presented with chest tightness and dyspnea following activity. On evaluation in our outpatient clinic, CTA (Table 1, Fig. 5A and B) showed significant aneurysmal dilation of the aortic sinus and ascending aorta, with moderate to severe aortic valve regurgitation. The patient was treated with an antihypertensive agent and beta-blockers but declined further surgical intervention.

Case 3

A 42-year-old female with PA/VSD. At the age of 14, she underwent corrective surgery for PA/VSD in another hospital and did not have regular follow-ups. Twenty-eight years after surgery, the patient developed dyspnea, with progressively worsening symptoms. She visited our outpatient clinic for treatment. Physical examination revealed a grade III/6 systolic murmur in the third and fourth intercostal space at the left sternal margin and a diastolic murmur in the aortic valve area. Echocardiography and CTA (Table 1, Fig. 6A and B) revealed an AAD, a tricuspid aortic valve with insufficiency, and residual shunting at the level of the ventricular septum. Medication includes antihypertensive agents and beta-blockers, but the patient declined surgery.

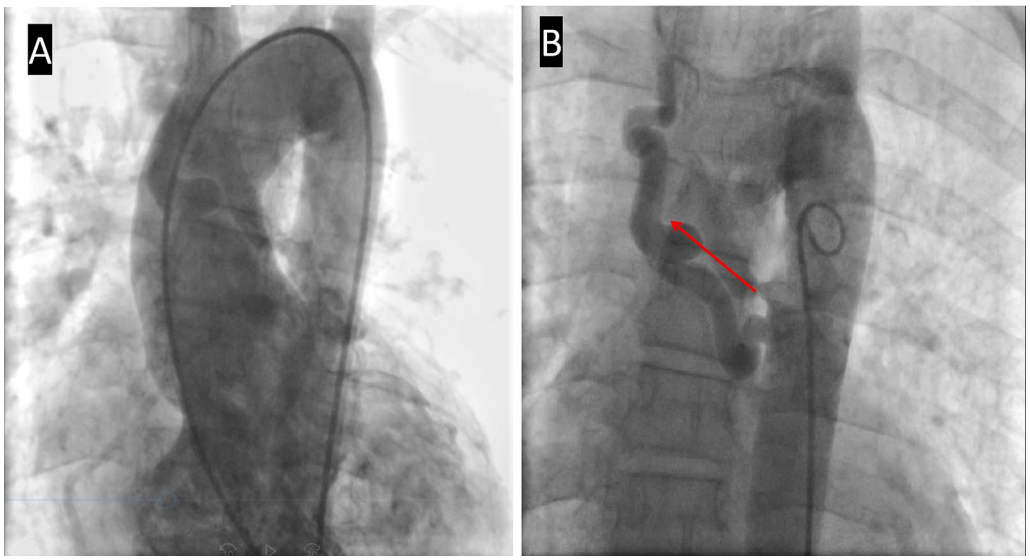

Fig. 4 Preoperative cardiac catheterization: **A** Right ventricular angiography showing no pulmonary artery visualization, **B** The MAPCAs (red arrow)

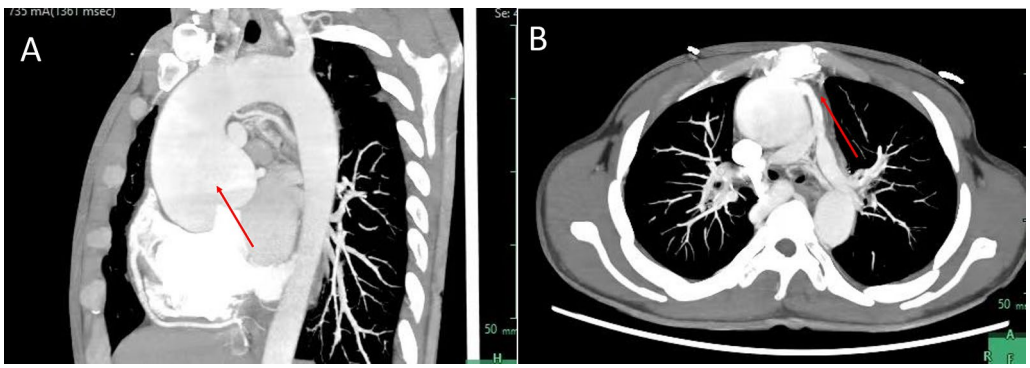

Fig. 5 Computed tomography angiography: **A** Aortic root aneurysm (red arrow) and AAD, **B** The artificial graft and the pulmonary artery (red arrow)

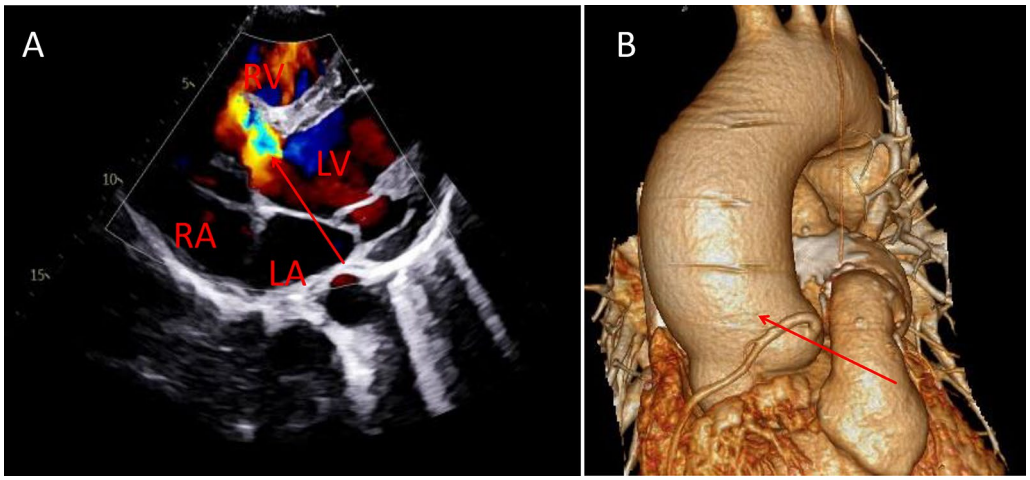

Fig. 6 Echocardiography and CTA: **A** shows VSD residual shunting (red arrow). **B** illustrates AAD (red arrow)

Discussion

We report three adult PA/VSD cases who missed the ideal age for corrective surgery. Of these three patients, two received corrective surgical intervention and one received palliative care. On long-term postoperative follow-up, aortic lesions included the aortic root aneurysms and AAD, which in one case developed into aortic dissection.

PA/VSD is a complex cyanotic congenital heart disease also known as tetralogy of Fallot (TOF) combined with PA [3–5]. Niwa et al. found that about 15% of adult cases develop significant aortic dilatation after undergoing corrective surgery for TOF [6]. A recent study involved 2261 patients with conotruncal malformations, 52% of whom presented with aortic aneurysms, but only 2.5% underwent surgical intervention. Notably, no cases of aortic dissection were observed over 7,984 patient-years of follow-up [7]. Additionally, literature highlights the importance of careful monitoring of aortic aneurysms in these patients, citing a case similar to our Case 1, where a patient underwent TOF repair at age 5, aortic valve replacement at age 38, and surgery for aortic dissection at age 61. This case emphasizes the need for close monitoring, and suggests that aortic replacement may be reasonable for TOF patients with an ascending aorta or aortic sinus diameter ≥ 55 mm [8].

Mechanisms of aortic root aneurysms, AAD, and aortic dissection in patients with PA/VSD, including histopathologic changes in the aortic wall, hemodynamic alterations, and genetic susceptibility. Chowdhury et al. analyzed the aortic wall tissue of 74 patients who underwent TOF repair and found that 78.4% of the aortic tissue samples exhibited significant medial layer defects, with 96% showing abnormal histopathological changes. Patients with histopathological alterations had an 8.83-fold increased risk of aortic dilation compared to those without such changes [9]. The hemodynamic changes in PA/VSD also play a significant role in developing of aortic pathology. PA combined with a large VSD increases aortic flow due to abnormal right-to-left shunting. Additionally, all three patients in this group had aortic override over both ventricles for a prolonged period, with aortic regurgitation causing volume overload. This volume overload generates maximal shear stress on the aortic root and ascending aorta [9]. A study has shown a significant correlation between the timing of complete correction and late aortic root dilation, with the age at complete repair being an important risk factor for late aortic root dilation [10].The three PA/VSD patients we reported missed the optimal age for corrective surgery, with follow-up times exceeding 15 years, and all developed aortic disease. PA/VSD has a notable familial tendency and is often associated with 22q11.2 chromosomal microdeletions [11, 12]. Although none of the three patients in our cases had a clear family history, genetic testing was not performed in any of them.

BAV malformation increases the risk of aortic dilation and aortic dissection [13]. Although the exact pathogenesis remains unclear. One patient in this group had PA/VSD combined with BAV and developed an aortic root aneurysm and aortic dissection 17 years after corrective surgery. Currently, there are no reported cases in the literature regarding long-term aortic dilation or dissection in PA/VSD patients with BAV. The development of aortic dilation and dissection in this case may be related to factors such as hemodynamics and histopathology.

For adult PA/VSD patients who miss the optimal time for corrective surgery, progressive aortic dilation is likely

to occur, with an increased risk of aortic dissection. Long-term follow-up and monitoring are essential for these patients to detect any potential complications.

Abbreviations

PA	Pulmonary atresia
VSD	Ventricular septal defect
AAD	Ascending aortic dilatation
CHD	Congenital heart disease
BAV	Bicuspid aortic valve
PFO	Patent foramen ovale
CTA	Computed tomography angiography
ECHO	Echocardiography
LVEF	Left ventricular ejection fractions
LVEDD	Left ventricular end-diastolic dimension
MAPCAs	Major aortopulmonary collateral arteries
LPA	Left pulmonary artery
RPA	Right pulmonary artery
TOF	Tetralogy of fallot

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Author contributions
ZJ and MK wrote the main manuscript text. ZJ collected and prepared the clinical data of the patients.H.X. and LX participated in patients care. MK revised the manuscript. All authors reviewed the manuscript. All authors read and approved the fnal manuscript.

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Data availability
No datasets were generated or analysed during the current study.

Declarations

Ethics approval and consent to participate
This work has received approval from the Ethics Committee of the First Hospital of Tsinghua University.

Competing interests
We declare that the research was conducted in the absence of any commercial or financial relationships that could be construed as a potential conflict of interest.

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