A Window to Life: A Rare Association of a Small, Proximal Aortopulmonary Window With Pulmonary Atresia/Ventricular Septal Defect

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INTRODUCTION

Pulmonary atresia with ventricular septal defect (PA/VSD) is a complex congenital cyanotic heart disease that can lead to various clinical manifestations based on the central pulmonary artery anatomy and pulmonary distribution.[1,2] In this lesion, pulmonary blood flow is provided either from a patent ductus arteriosus (PDA) or major aortopulmonary collateral arteries (MAPCAS), which in turn contribute to the development (or lack thereof) of main and branch pulmonary arteries as well as the greater lung parenchyma.[3-5] However, to date, there has been no report of PA/VSD in the absence of a PDA or MAPCAS. Here, we present a case of PA/VSD with a small, proximal aortopulmonary window (APW) as the sole source of pulmonary blood flow and its respective management.

CASE PRESENTATION

A 3.0 kg, full-term male infant presented with respiratory distress and severe hypoxia refractory to 100% FiO_2 and nitric oxide shortly after birth. After prompt intubation, the patient was brought to the neonatal intensive care unit with an initial cord gas of 7.0/80.1/25.4/23.1/−9.1 and lactate 6.6 mmol/L. Initial transthoracic echocardiogram (TTE) on high-dose prostagladin E (0.1 mcg/kg/min) revealed a PA/VSD (Figure 1, Videos 1 and 2), with a small, proximal or type I APW (located just above the sinotubular junction) and right-to-left shunting across a secundum-type atrial septal defect (ASD; Video 3). There was a dysplastic aortic valve with severe aortic regurgitation (AR; Figure 2, Videos 4 and 5). The PA/VSD had minimal flow (Figure 3A, Video 6), and there was no evidence of a PDA or MAPCAS. Cardiac computed tomography (CCT) further delineated the presence of hypoplastic but also confluent main and branch pulmonary arteries. Genetic evaluation confirmed DiGeorge syndrome (22q11.2 deletion). Multiple surgical and catheter-based interventions were considered, including a shunt, a conduit, and radiofrequency perforation through the right ventricular outflow tract (RVOT), but due in part to the patient's severe AR and subsequent inadequacy for extracorporeal membrane oxygenation, the patient was felt to be too high risk of a candidate. After obtaining additional second opinions, which agreed with a comfort care approach, the patient was given a status of do not resuscitate.

After approximately 36 hours on 100% FiO_2 and 20 ppm nitric oxide, the patient's oxygen saturations rose to 55% to 60% from 30% to 40%. Repeat TTE demonstrated antegrade flow from the aorta to the pulmonary artery across the small, ~3 mm APW as well as antegrade flow through the main and branch pulmonary arteries (Figure 3B,

VIDEO HIGHLIGHTS

Video 1: Two-dimensional TTE, parasternal long-axis RVOT view with color-flow Doppler, demonstrates a membranous-type PA with no antegrade flow seen and a plate-like pulmonary valve and obligatory right-to-left shunting across the VSD into the left ventricle.

Video 2: Two-dimensional TTE, subcostal short-axis view, mid-to-basal sweep, demonstrates the narrow, atretic RVOT from the right ventricle.

Video 3: Two-dimensional TTE, subcostal long-axis view with color-flow Doppler, demonstrates a second outflow tract with antegrade flow from the right ventricle through a small, narrowed RVOT but no further flow past the atretic pulmonary valve into a main or branch pulmonary artery segment.

Video 4: Two-dimensional TTE, parasternal short-axis en face view of the dyplastic aortic valve, which appears thickened and bicuspid with fusion of right and left coronary cusps and to-and-fro billowing of the leaflets throughout the cardia cycle.

Video 5: Two-dimensional TTE, parasternal long-axis view with color-flow Doppler of the aortic valve, demonstrating the to-and-fro billowing of the leaflets with at least moderate to severe AR.

Video 6: Two-dimensional TTE, parasternal short-axis view, with color-flow Doppler of the APW with very limited, intermittent antegrade (*blue*) flow seen across the window into the hypoplastic main and branch pulmonary arteries.

Video 7: Two-dimensional TTE, parasternal short-axis view, with color-flow Doppler of the APW with improved, continuous antegrade (*blue*) flow seen across the window into the hypoplastic main and filling bilateral branch pulmonary arteries after the fall in PVR at 36 hours of life on 100% FiO_2 and 20 ppm iNO.

View the video content online at www.cvcasejournal.com.

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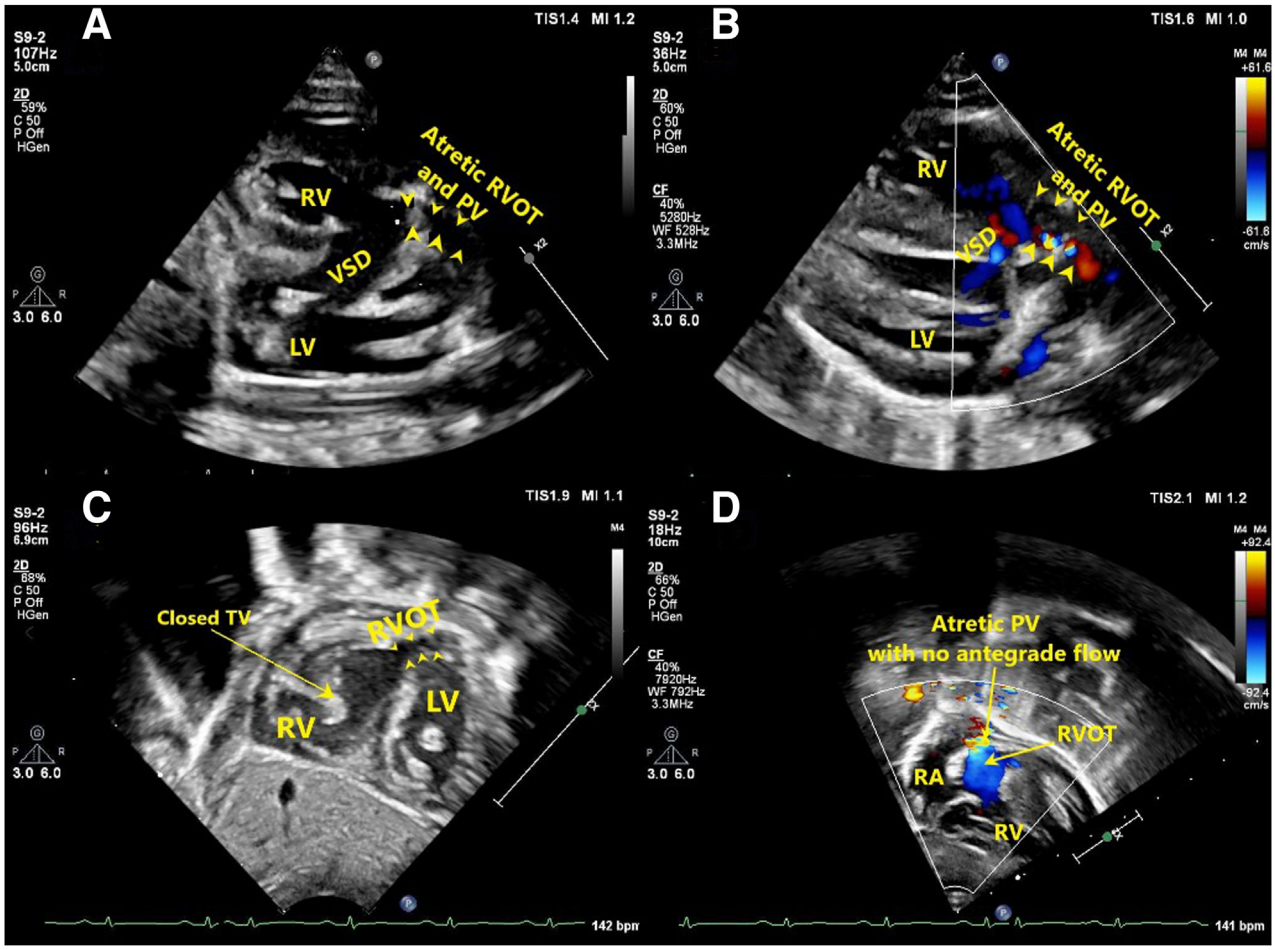

Figure 1 Pulmonary atresia/VSD. **(A)** Two-dimensional TTE, parasternal long-axis (PLAX) view demonstrates a narrow, more anterior second outflow tract from the right ventricle with thickened, atretic pulmonary valve annulus. The large VSD is seen at the inferior margin. **(B)** Two-dimensional TTE, PLAX RVOT view with color-flow Doppler during systole, demonstrates a membranous-type PA with no antegrade flow seen and a plate-like pulmonary valve and obligatory right-to-left shunting across the VSD into the left ventricle. **(C)** Two-dimensional TTE, subcostal short-axis view at the level of the ventricular outflows during systole, demonstrates the narrow, atretic RVOT from the right ventricle. The VSD is not seen in this view, confirming that the outflow seen is not the aortic outflow from the LV that overrides the ventricular septum. **(D)** Two-dimensional TTE, subcostal long-axis view with color-flow Doppler during systole, demonstrates a second outflow tract with antegrade (*blue*) flow from the right ventricle through a small, narrowed RVOT but no further flow past the atretic pulmonary valve into a main or branch pulmonary artery segment. *PV*, Pulmonary valve; *RA*, right atrium; *RV*, right ventricle; *TV*, tricuspid valve.

Video 7), continued right-to-left shunting across the ASD (Figure 4A), and mild AR (Figure 4B). The patient was subsequently transferred to a higher level of care and underwent a left-sided Blalock-Taussig shunt off cardiopulmonary bypass on DOL4 with immediate improvement in oxygen saturations. A CCT was obtained postoperatively to evaluate the shunt, branch pulmonary arteries, and coronary artery anatomy, the latter of which was normal (Figure 5). Chest closure on DOL5 was well tolerated, and after undergoing a gastrotomy tube a few weeks later, the patient was discharged home with good clinical follow-up thereafter.

DISCUSSION

Pulmonary atresia/VSD has always been associated with a PDA or MAPCAS as obligatory sources of pulmonary blood flow.[5] As a result, a broad spectrum of pulmonary arterial anatomy and clinical manifestations have been described, ranging from well-developed, confluent main and branch pulmonary arteries with minimal cyanosis and congestive heart failure to complete absence of true pulmonary arterial structures with severe cyanosis.[6-8] Strategies for treating PA/VSD with a PDA or MAPCAS through surgical, interventional, and hybrid approaches have thus been the focus of research in this disease.[9-11]

Separately, an APW is an even rarer congenital malformation that constitutes 0.1% of all congenital heart diseases. It occurs as an isolated lesion as well as in association with other congenital heart defects including PDA, ASD, patent foramen ovale, tetralogy of Fallot, aortic arch interruption, aortic arch hypoplasia, and anomalous origin of coronary arteries.

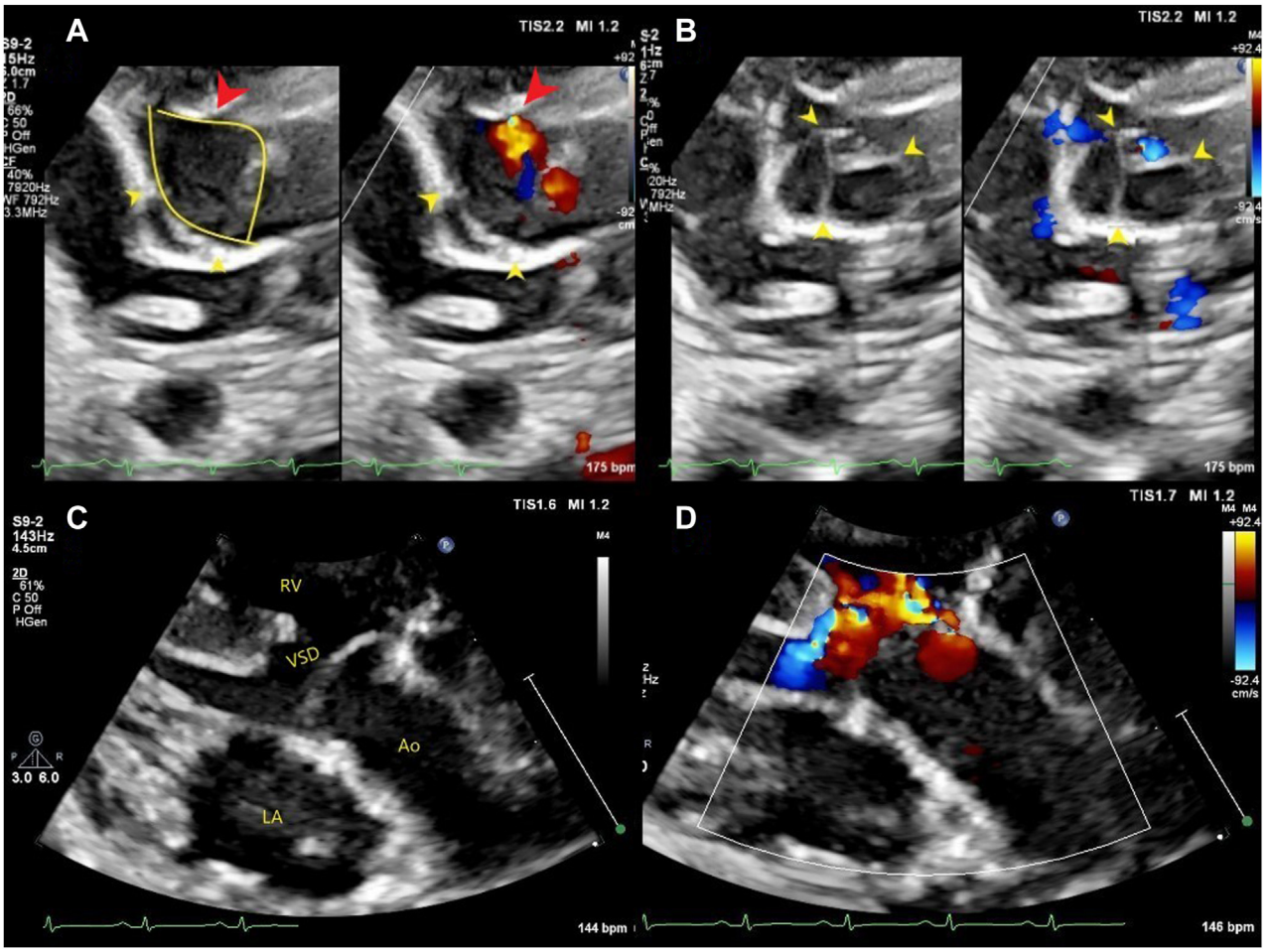

Figure 2 Dysplastic aortic valve. **(A)** Two-dimensional TTE, parasternal short-axis (PSAX) en face view of the aortic valve, without (*left*) and with (*right*) color-flow Doppler in systole, demonstrates a dysplastic, bicuspid valve with fusion of the right and left coronary cusps (*red arrows*). The two other *yellow arrows* demonstrate the right and noncoronary commissures. **(B)** Two-dimensional TTE, PSAX en face view of the aortic valve, without (*left*) and with (*right*) color-flow Doppler in diastole, demonstrates 3 separate commissures (*yellow arrows*). **(C)** Two-dimensional TTE, parasternal long-axis (PLAX) view of the aortic valve in diastole, demonstrates the billowing of the aortic valve leaflets as they prolapse backward into the VSD. **(D)** Two-dimensional TTE PLAX view with color-flow Doppler of the aortic valve in diastole demonstrates moderate to severe AR. *Ao*, Aorta; *LA*, left atrium.

This case demonstrates a case of PA/VSD associated with a proximal (type I) APW in the absence of a PDA or MAPCAS, which is very rare. While the presence of a severely dysplastic, aortic valve would prompt consideration of the alternative diagnosis of truncus arteriosus with a hypoplastic MPA segment, the presence of a second outflow tract (Figure 1, Videos 1-3) confirms PA/VSD. The pure right-to-left shunting across the ASD on initial serial TTEs was also a peculiar finding in the presence of normal pulmonary venous return, which we attributed to the relative difference in right and left atrial compliance secondary to poor pulmonary blood flow and subsequent poor pulmonary venous return. The initial coronary artery dilatation prompted the possibility of a coronary artery fistula, but this was resolved on CCT and ultimately felt to be related to the patient's initial hypoxia as repeat TTE postoperatively showed normal-caliber coronary arteries.

While diminutive main and branch pulmonary arteries were seen on follow-up TTEs, they were not well visualized on the initial study likely due to minimal flow across the APW. Hence, obtaining a CCT early on in this patient's course was critical in understanding not only the intracardiac anatomy but also the lung parenchyma and presence of absence of MAPCAS. That being said, true arborization of the pulmonary arterial bed would have been challenging to see in the first few hours even on CCT given the very limited antegrade flow.

Clinically, while this small APW, which functioned similarly to a Melbourne or "Mee-type" shunt, provided enough pulmonary blood flow for survival, it was certainly not sustainable. In each treatment strategy that was considered, the presence of moderate to severe AR remained a major barrier to any intervention. The degree of AR improved after the initial 36 hours, which we attributed to an

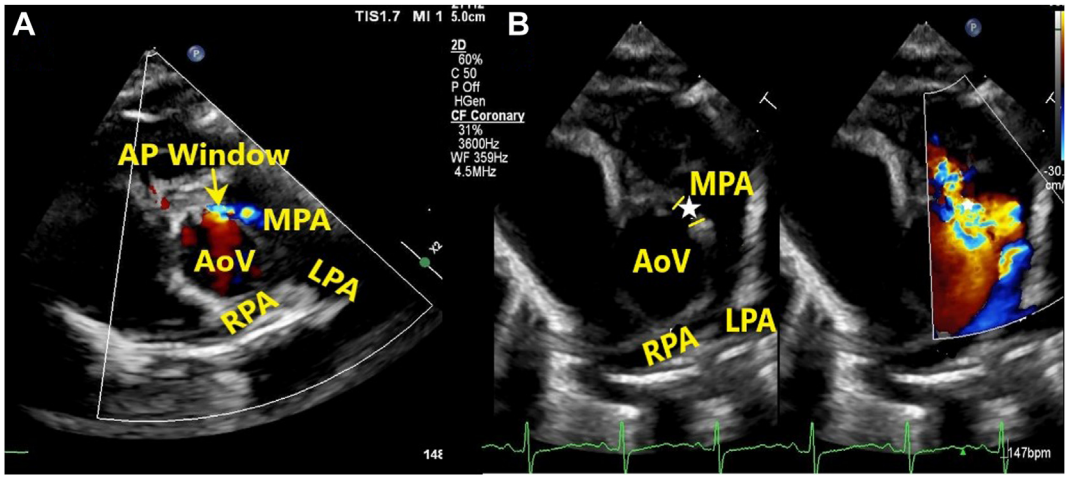

Figure 3 Small, proximal (type I) APW before and after drop in PVR. **(A)** Two-dimensional TTE, high parasternal short-axis (PSAX) view with color-flow Doppler of the aorta and main pulmonary artery at ~1 hour of life, demonstrates the limited aorta to hypoplastic pulmonary artery shunt (*blue*) across the small APW (*yellow arrow*). It is difficult to appreciate the true size of the APW at this time given the limited shunting in the setting of elevated PVR at birth. **(B)** Two-dimensional TTE, high PSAX view without (*left*) and with (*right*) color-flow Doppler of the aorta and main pulmonary artery at ~36 hours of life after the fall in PVR while receiving 100% FiO_2 and 20 ppm iNO, demonstrates antegrade flow across the APW into the hypoplastic main and branch pulmonary arteries. The size of the APW is now better appreciated as demonstrated by the *yellow lines* and *white star*. *AoV*, Aortic valve; *LPA*, left pulmonary artery; *MPA,* main pulmonary artery; *RPA*, right pulmonary artery.

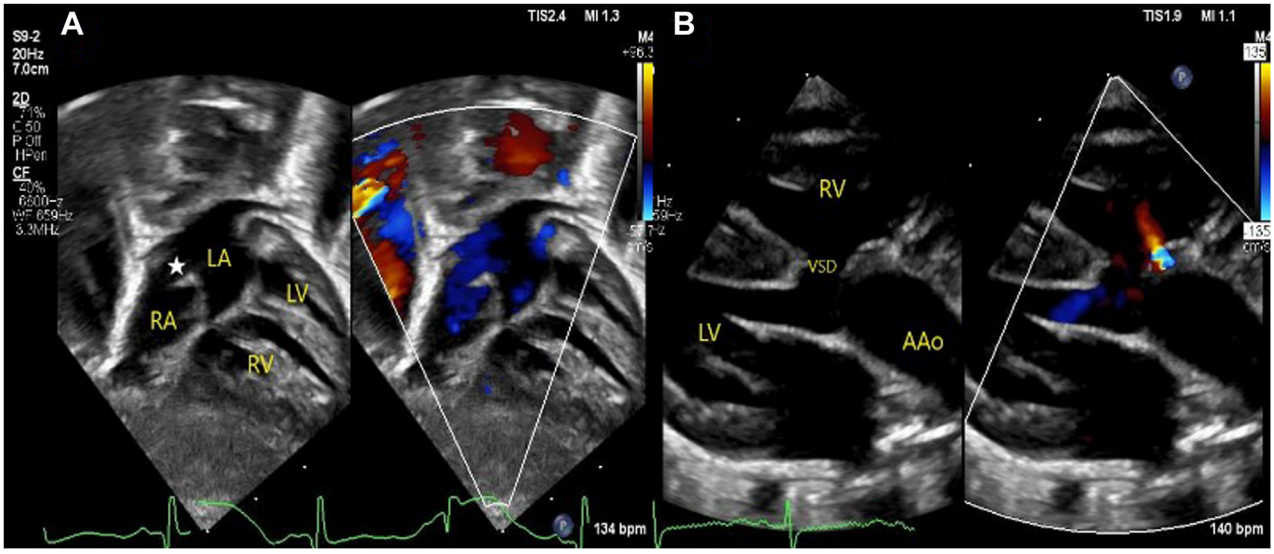

Figure 4 Atrial septal defect and AR after fall in PVR. **(A)** Two-dimensional TTE, subcostal long-axis view of the small ASD (*white star*) without (*left*) and with (*right*) color-flow Doppler, demonstrates a right-to-left shunt in the setting of normal pulmonary venous return related to the ongoing discrepancy between right and left atrial compliance from decreased pulmonary venous return. **(B)** Two-dimensional TTE, parasternal long-axis view of the aortic valve without (*left*) and with (*right*) color-flow Doppler, demonstrates mild AR in the setting of lower PVR. *AAo,* Ascending aorta*; LA*, left atrium; *RA*, right atrium; *RV*, right ventricle.

expediated fall in pulmonary vascular resistance (PVR) through 100% FiO_2 and 20 ppm iNO. This interdependence on PVR was further confirmed when the AR worsened after weaning off of iNO.

While the patient has clinically improved, the ultimate long-term outcome remains unknown but will surely be impacted by the initial prolonged period of hypoxia and his comorbid 22q11.2 deletion.

CONCLUSION

Pulmonary atresia/VSD in the absence of a PDA and MAPCAS with confluent main and branch pulmonary arteries is possible in the presence of a type I APW. When associated with a dysplastic aortic valve with severe AR, management strategies are limited and surgical shunt placement provides a prompt and successful restoration of pulmonary blood flow.

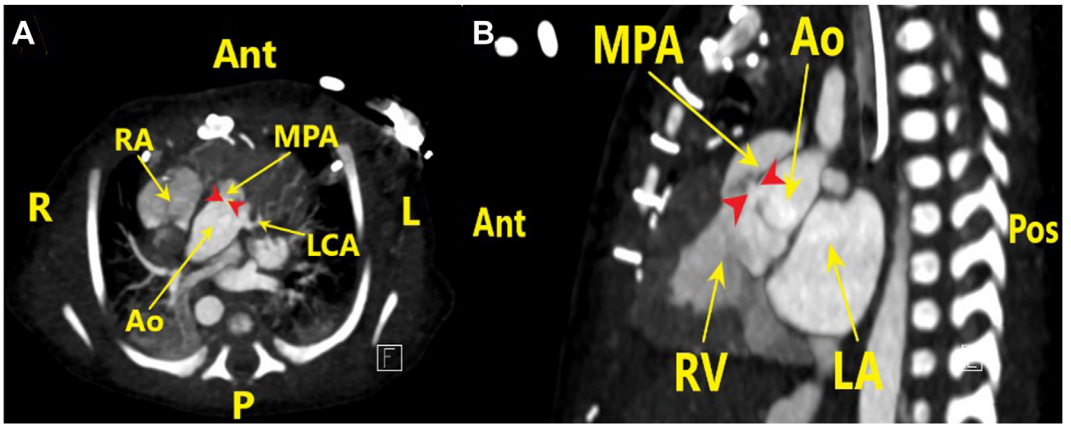

Figure 5 Cardiac computed tomography angiogram. Axial source image **(A)** and sagittal oblique multiplanar reformat **(B)** image showing the small APW (*red arrows*) between the aortic root posteriorly and the proximal, hypoplastic MPA anteriorly. *Ant*, Anterior; *Ao*, aorta; *L*, left; *LA*, left atrium; *LCA*, left coronary artery; *MPA*, main pulmonary artery; *P*, Pos; *RA*, right atrium; *R*, right; *RV*, right ventricle.

CONSENT STATEMENT

The authors declare that since this was a noninterventional, retrospective, observational study utilizing deidentified data, informed consent was not required from the patient under an IRB exemption status.

ETHICS STATEMENT

The authors declare that the work described has been carried out in accordance with The Code of Ethics of the World Medical Association (Declaration of Helsinki) for experiments involving humans.

FUNDING STATEMENT

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DISCLOSURE STATEMENT

The authors report no conflict of interest.

SUPPLEMENTARY DATA

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REFERENCES

1. Kumar SR. Collateral benefit in pulmonary atresia with ventricular septal defect? J Thorac Cardiovasc 2018;156:1205-6.
2. Van Puyvelde J, Meyns B, Rega F. Pulmonary atresia and a ventricular septal defect: about size and strategy. Eur J Cardiothorac Surg 2015;49:1419-20.
3. Bull K, Somerville J, Ty E, Spiegelhalter D. Presentation and attrition in complex pulmonary atresia. J Am Coll Cardiol 1995;25:491-9.
4. Carotti A, Albanese SB, Filippelli S, Ravà L, Guccione P, Pongiglione G, et al. Determinants of outcome after surgical treatment of pulmonary atresia with ventricular septal defect and major aortopulmonary collateral arteries. J Thorac Cardiovasc 2010;140:1092-103.
5. O'Byrne ML, Kanter JP, Berger JT, Jonas RA. Outcomes of patients with pulmonary atresia and major aortopulmonary collaterals without intervention in infancy. Pediatr Cardiol 2016;37:1380-91.
6. Lertsakulpiriya K, Vijarnsorn C, Chanthong P, Chungsomprasong P, Kanjanauthai S, Durongpisitkul K, et al. Current era outcomes of pulmonary atresia with ventricular septal defect: a single center cohort in Thailand. Sci Rep 2020;10:5165.
7. Kaskinen A, Happonen JM, Mattila I, Pitkänen O. Long-term outcome after treatment of pulmonary atresia with ventricular septal defect: nationwide study of 109 patients born in 1970–2007. Eur J Cardiothorac Surg 2015;49:1411-8.
8. Leonard H. Natural and unnatural history of pulmonary atresia. Heart 2000;84:499-503.
9. Amark KM, Karamlou T, O'Carroll A, MacDonald C, Freedom RM, Yoo SJ, et al. Independent factors associated with mortality, reintervention, and achievement of complete repair in children with pulmonary atresia with ventricular septal defect. J Am Coll Cardiol 2006;47:1448-56.
10. Mainwaring RD, Patrick WL, Roth SM, Kamra K, Wise-Faberowski L, Palmon M, et al. Surgical algorithm and results for repair of pulmonary atresia with ventricular septal defect and major aortopulmonary collaterals. J Thorac Cardiovasc 2018;156:1194-204.
11. Hribernik I, Jaber O, Bentham JR. Alternative hybrid approach to promote native pulmonary artery growth in pulmonary atresia VSD MAPCAs. J Am Coll Cardiol Case Rep 2022;4:1366-9.