Cardiology in the Young

cambridge.org/cty

Tetralogy of Fallot with pulmonary atresia and aortopulmonary window may mimic common arterial trunk

Sudesh Prabhu[1], Manaswini Keshav[2], Prakash Ramachandra[1], Vimal Raj[1], Colin John[1] and Tom R Karl[3]

[1]Paediatric Cardiac Services, Narayana Institute of Cardiac Sciences, Bengaluru, India; [2]Medical Student, MS Ramaiah Medical College, Bengaluru, India and [3]Queensland Pediatric Cardiac Research, University of Queensland, Brisbane, Australia

Original Article

Cite this article: Prabhu S, Keshav M, Ramachandra P, Raj V, John C, and Karl TR (2022) Tetralogy of Fallot with pulmonary atresia and aortopulmonary window may mimic common arterial trunk. *Cardiology in the Young* **32**: 410–414. doi: 10.1017/S1047951121002298

Received: 31 January 2021
Revised: 18 May 2021
Accepted: 20 May 2021
First published online: 17 June 2021

Keywords:
Pulmonary atresia; ventricular septal defect; aortopulmonary window; common arterial trunk; sub-pulmonary infundibulum; pulmonary blood flow

Author for correspondence:
Sudesh Prabhu, Department of Pediatric Cardiac Surgery, Narayana Institute of Cardiac Sciences, 258/A Hosur Road, Bommasandra Industrial Area, Anekal Taluk, Bengaluru, Karnataka 560099, India. Tel: +919886899450. E-mail: sudesh006@gmail.com

Abstract

Tetralogy of Fallot with pulmonary atresia is a group of congenital cardiac malformations, which is defined by the absence of luminal continuity between both ventricles and the pulmonary artery, and an interventricular communication. Pulmonary arterial supply in patients with tetralogy of Fallot with pulmonary atresia can be via the arterial duct or from collateral arteries arising directly or indirectly from the aorta (systemic-to-pulmonary artery collaterals), or rarely both. The rarest sources of pulmonary blood flow are aortopulmonary window and fistulous communication with the coronary artery.

Herein, we describe an outflow tract malformation, tetralogy of Fallot with pulmonary atresia and aortopulmonary window, which was misdiagnosed as common arterial trunk. We emphasise the morphological differences.

The Congenital Heart Surgery Nomenclature and Database project defines pulmonary atresia with ventricular septal defect as a group of congenital cardiac malformations in which there is a lack of luminal continuity between both ventricles and the pulmonary arterial tree, with an interventricular communication.[1] In its severe form, there is an absence of either part or all of the intrapericardial pulmonary arteries. The pulmonary circulation in pulmonary atresia with a ventricular septal defect is heterogeneous and variable, and independent of the intracardiac anatomy. The authors of the paper proposed three subtypes of pulmonary atresia with ventricular septal defect, according to the anatomy of the pulmonary circulation and source of pulmonary blood flow. Type A has intrapericardial pulmonary arteries with pulmonary blood flow from an arterial duct. Type B has both intrapericardial pulmonary arteries and major aortopulmonary collaterals. Type C has no intrapericardial pulmonary arteries, and the pulmonary circulation is supplied by major aortopulmonary collaterals only.[1] Based on the site of origin and connection to the pulmonary circulation, systemic-to-pulmonary collateral arteries is categorised into three types, including direct aortopulmonary collaterals, indirect aortopulmonary collaterals, and true bronchial arteries. Only the first two are considered to be the major aortopulmonary collaterals.[2]

The above categorisations proposed by the Congenital Heart Surgery Nomenclature and Database project have been overtaken by those provided by the International Nomenclature Committee, which included most of those involved in preparing the documents that made up the supplement of 2000. The recommendations made by the International Nomenclature Committee, furthermore, have been adopted by the World Health Organization for the 11th iteration of the International Classification of Disease. This classification defines tetralogy of Fallot with pulmonary atresia (synonym: pulmonary atresia with ventricular septal defect, Fallot type) as a congenital cardiovascular malformation that is a variant of tetralogy of Fallot, in which there is no direct communication between the right ventricle and the pulmonary arterial tree (IPCCC code 01.01.26), and IPCCC code 01.01.57, when there are collateral blood vessels between the systemic and pulmonary arteries.[3]

Common arterial trunk is a congenital malformation in which a single great artery arises from the heart, overrides the interventricular septum, and supplies the systemic, pulmonary, and coronary circulations.[3–6] With common arterial trunk, there are no remnants of a separate pulmonary valve or ventricular-to-pulmonary artery continuity, distinguishing it from many forms of pulmonary atresia with ventricular septal defect.[7]

The pulmonary arterial supply in patients with tetralogy of Fallot with pulmonary atresia can be via the arterial duct or from collateral arteries arising directly or indirectly from the aorta (systemic-to-pulmonary artery collaterals) or rarely both.[8] The rarest sources of pulmonary blood flow are aortopulmonary window and fistulous communication with the coronary

artery.[8–12] The previously described fifth arch as a source of pulmonary blood flow has no scientific evidence, as the latest literature suggests that there is no embryonic fifth arch artery.[11,13]

Herein, we describe an outflow tract malformation, tetralogy of Fallot with pulmonary atresia and aortopulmonary window, which was misdiagnosed as common arterial trunk, despite definitive morphological differences. We do not suggest that this is a new morphological entity, and probably other teams have encountered such anatomy, diagnosed and managed as common arterial trunk.

Tetralogy of Fallot with pulmonary atresia is a subset of the overall group of patients who can be described as having pulmonary atresia with ventricular septal defect. This subset of tetralogy of Fallot with pulmonary atresia, however, is important because it is only this subset when associated with aortopulmonary window as the source of unifocal pulmonary arterial supply, produces the confusion with common arterial trunk. It would be unlikely that patients having congenitally corrected transposition, or regular transposition, or isomerism, in association with pulmonary atresia and a ventricular septal defect would be misdiagnosed as having common arterial trunk.

Case series

Over the past 3 years, three patients, with a referral diagnosis of common arterial trunk with aortic dominance (two with minimal and one with significant separation of intrapericardial pathways), were found to have tetralogy of Fallot with pulmonary atresia and aortopulmonary window. Echocardiography, cardiac CT scan, operative findings, and post-operative course details were reviewed. All three patients were presented with respiratory distress, feeding difficulty, and failure to thrive. All were tachypneic with chest retractions, systolic murmurs, cardiomegaly, and pulmonary plethora.

The diagnosis of tetralogy of Fallot with pulmonary atresia and aortopulmonary window was made echocardiographically (Fig 1), and confirmed with a CT scan in two patients (Figs 2 and 3). In one, the diagnosis was made intraoperatively.

One neonate underwent a patch closure of aortopulmonary window, and a central shunt with a polytetrafluoroethylene graft. This child underwent ventricular septal defect closure, transannular patch, and shunt takedown 2 years later. The remaining two infants underwent single-stage repair (separation of pulmonary trunk, closure of ventricular septal defect, and valved polytetrafluoroethylene right ventricle to pulmonary artery conduit).[14] All three patients survived surgery and are well currently.

Morphology

External anatomy

There was cardiomegaly with biventricular hypertrophy in all three patients. The pulmonary trunk was tense and dilated, arising from the right ventricle without luminal continuity (Fig 4). The source of pulmonary blood flow was an aortopulmonary window, which provided a unifocal pulmonary arterial supply (none of the three patients had a persistent arterial duct). The pulmonary trunk could be looped proximally and distally to the aortopulmonary communication. The coronary arteries were compared to those in a normal heart (right coronary artery and left main coronary artery dividing into left anterior descending artery and left circumflex artery) with origin having a counterclockwise rotation (Figs 2–4).

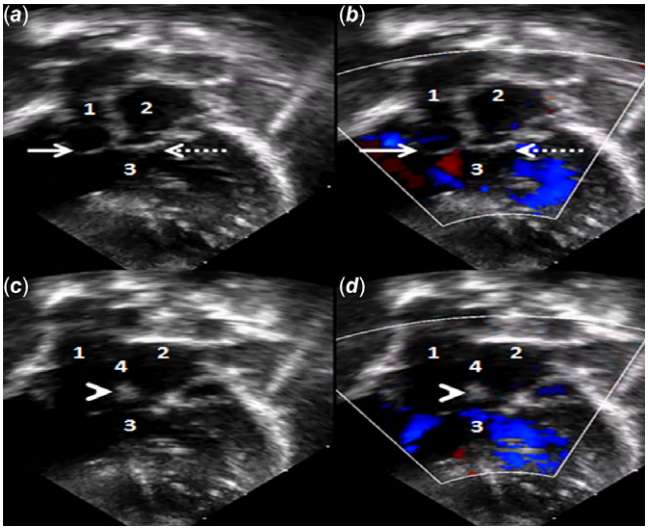

Figure 1. Transthoracic echocardiography of tetralogy of Fallot with pulmonary atresia and aortopulmonary window (1 – ascending aorta, 2 – pulmonary trunk, 3 – interventricular communication, 4 – aortopulmonary window, arrow – aortic valve, broken arrow – atretic pulmonary valve, arrowhead – inferior aortopulmonary septum).

Intracardiac anatomy

All three patients had a remnant of a separate pulmonary valve (Fig 4), but no communication with either ventricle. One patient had a bicuspid aortic valve (anterior and posterior aortic leaflets) (Fig 4). The remaining two patients had tricuspid aortic valves.

The aortopulmonary communication was proximal in two patients (Figs 2 and 4) and distal in one (extending into the right pulmonary artery) (Fig 3). The inferior aortopulmonary septum was present in all. During the embryological development of a normal human embryo, the aortopulmonary septum is a transient structure and does not persist in the postnatal heart.[15] This aortopulmonary septum gets transformed into the separate walls of the aorta and pulmonary trunk even in the setting of aortopulmonary window.

Interventricular communication was in the form of perimembranous ventricular septal defect in two patients with posteroinferior margin probably related to the conduction tissue. The third patient had a muscular border all around (the posteroinferior limb of the septomarginal trabeculation fused with the ventriculo-infundibular fold). A sub-pulmonary infundibulum (blindly ending sub-pulmonary outflow tract) was present in all. The ventricular septal defect was separated from the blindly ending infundibulum by a malaligned muscular outlet septum. Extensive malalignment of the outlet septum (fibrous or muscular) is the essence of Fallot-type pulmonary atresia with ventricular septal defect, and was present in all three of our cases. On the basis of this phenotypic anatomy, we have grouped our patients together.

Intrapulmonary anatomy

Rossi RN et al have mentioned that in patients with pulmonary atresia with ventricular septal defect, rare sources of pulmonary arterial supply normally do not usually bring problems since the segmental arterial supply to the lungs is normally distributed.[8] Our three patients had unifocal and symmetric pulmonary arterial supply with increased pulmonary blood flow status. CT scan in two patients demonstrated normal segmental distribution of pulmonary arteries without hypoplasia of intraparenchymal arteries.

https://doi.org/10.1017/S1047951121002298 Published online by Cambridge University Press

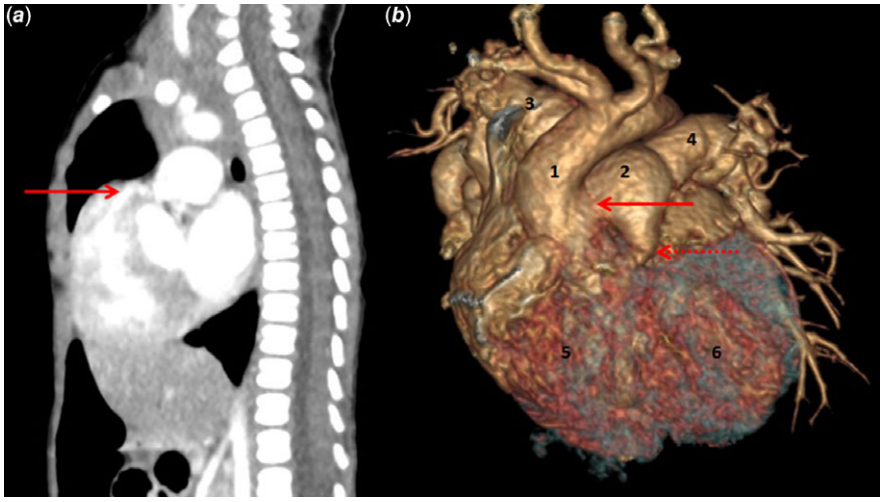

Figure 2. CT scan of tetralogy of Fallot with pulmonary atresia and aortopulmonary window. (***a***) Sagittal section demonstrating sub-pulmonary infundibulum with blindly ending sub-pulmonary outflow tract (arrow). This sub-pulmonary infundibulum is conspicuously absent in common arterial trunk. (***b***) Three-dimensional reconstruction of tetralogy of Fallot with pulmonary atresia and aortopulmonary window demonstrating separate aortic and pulmonary roots (with atretic pulmonary valve). 1 – ascending aorta, 2 – pulmonary trunk, 3 – right pulmonary artery, 4 – left pulmonary artery, 5 – right ventricle, 6 – left ventricle, arrow pointing aortopulmonary window, broken arrow pointing pulmonary root.

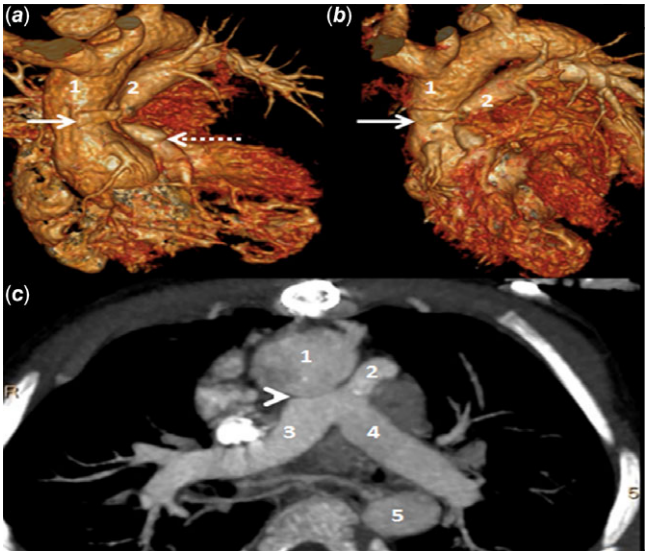

Figure 3. Post-operative (patch closure of aortopulmonary window and central shunt between ascending aorta and main pulmonary artery) CT scan of tetralogy of Fallot with pulmonary atresia and aortopulmonary window. (***a***) Three-dimensional reconstruction demonstrating separate aortic and pulmonary roots. 1 – ascending aorta, 2 – pulmonary trunk, arrow – central shunt, broken arrow – pulmonary trunk arising from the ventricle. The aortopulmonary communication is distal, extending into the right pulmonary artery and is not clear here (see below). The right coronary artery can be seen arising from the anterior sinus. (***b***) Same as Figure 3a. (***c***) Transverse section demonstrating the patch separating the distal main pulmonary artery and right pulmonary artery from aorta (arrowhead). Pulmonary trunk arising from the ventricle and right coronary artery can be seen clearly. 1 – ascending aorta, 2 – pulmonary trunk, 3 – right pulmonary artery, 4 – left pulmonary artery, 5 – descending thoracic aorta.

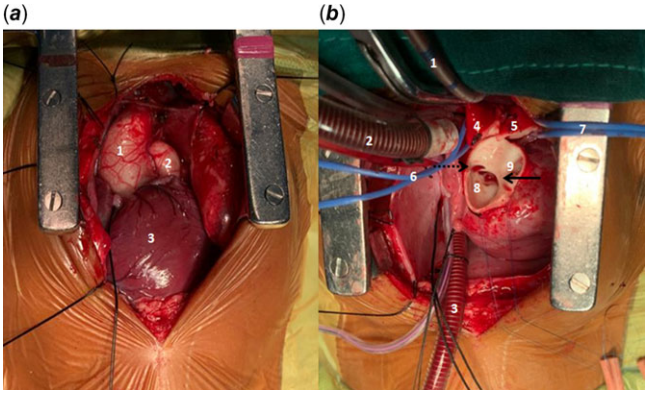

Figure 4. Intraoperative photographs of tetralogy of Fallot with pulmonary atresia and aortopulmonary window. (***a***) External appearance: 1 – aorta, 2 – pulmonary trunk, 3 – right ventricle. (***b***) After the division of great arteries: 1 – aortic cannula, 2 – superior caval vein cannula, 3 – inferior caval vein cannula, 4 – ascending aorta, 5 – pulmonary trunk, 6 – vessel loop around right pulmonary artery, 7 – vessel loop around left pulmonary artery, 8 – aortic root with bicuspid aortic valve, 9 – pulmonary root with atretic pulmonary valve, arrow indicates inferior aortopulmonary septum, broken arrow indicates ostium of left main coronary artery. Normal origin of right coronary artery can be seen.

Discussion

Patients with tetralogy of Fallot with pulmonary atresia and aortopulmonary window clinically and echocardiographically resemble those with common arterial trunk. Presentation can be late, as the patients are not overtly cyanotic. Surgical management is also similar to common arterial trunk and early surgical intervention (staged or total correction) is needed to avoid the establishment of irreversible pulmonary artery hypertension or Eisenmengerization.

In cases of tetralogy of Fallot with pulmonary atresia and aortopulmonary window, depending on the type of aortopulmonary window, there can be confusion with common arterial trunk with aortic dominance.[16–18] If the aortopulmonary communication is proximal or intermediate, it can be confused with common arterial trunk with aortic dominance and minimal separation of the intrapericardial pathways (two cases in our series). And if the aortopulmonary communication is distal or total, it can be confused with common arterial trunk with aortic dominance and significant separation of the intrapericardial pathways (one case in our series).

Patients with tetralogy of Fallot with pulmonary atresia and aortopulmonary window have a sub-pulmonary infundibulum, a

https://doi.org/10.1017/S1047951121002298 Published online by Cambridge University Press

Table 1. Morphological differences between common arterial trunk and tetralogy of Fallot with pulmonary atresia and aortopulmonary window.

Trait	Common arterial trunk	Tetralogy of Fallot with pulmonary atresia and aortopulmonary window
External anatomy		
Pulmonary trunk	Present in CAT with aortic dominance with minimal separation of intrapericardial pathways. But pulmonary trunk originates from the truncal artery. Can be looped distal to the truncal artery communication.	Present, arises from the ventricle, without luminal continuity. Can be looped, proximally and distally to the aortopulmonary communication.
Coronary arteries	Origins and course of the coronary arteries are frequently bizarre.[6]	*Comparable to normal heart in our three cases.
Source of pulmonary blood flow	Truncal artery	Unifocal pulmonary blood flow from aortopulmonary window
Intracardiac anatomy		
Remnant of separate pulmonary valve	Absent	Present
Truncal valve	Two, three, four, or more leaflets.	*In our three cases, we had one bicuspid and two tricuspid valves.
Sub-pulmonary infundibulum (blindly ending sub-pulmonary outflow tract)	Absent	Present in all three patients.
Inferior (proximal) aortopulmonary septum (superior AP septum is present in both TOF-PA-APW and CAT with aortic dominance with minimal separation of intrapericardial pathways, but is absent in CAT with aortic dominance when there is significant separation of intrapericardial pathways)	Absent	Present. The embryonic aortopulmonary septum is transient and gets transformed into the separate walls of the aorta and pulmonary trunk even in the setting of aortopulmonary window.
Interventricular communication	Located in an anterosuperior position, lying between the two superior limbs of the trabecula septomarginalis remote from the conduction tissue. Occasionally extends to the membranous septum closer to the conduction tissue.[19] Upper border of the interventricular communication is truncal valve.	*Two patients had perimembranous VSD (postero-inferior margin related to the conduction tissue) and one patient had muscular border all around (the postero-inferior limb of the septomarginal trabeculation fused with the ventriculo-infundibular fold).
Intrapulmonary anatomy		
Normal segmental distribution of pulmonary arteries	Yes	Yes
Increased pulmonary blood flow status	Yes	Yes

*Data from more TOF-PA-APW patients is needed to categorise the ventricular septal defect type, morphology of ventricular outlet valve, anatomy of the arterial duct, and coronary artery pattern.

remnant of a separate pulmonary valve, and a pulmonary trunk arising from the ventricle (without luminal continuity). There is a true ventricular septal defect. These are four features that are conspicuous by their absence in common arterial trunk (in patients with common arterial trunk, the interventricular communication is bordered superiorly by the truncal valve, not ventricular septum).

Aortopulmonary window as a unifocal source of pulmonary arterial supply in tetralogy of Fallot with pulmonary atresia is considered to be a rare entity.[8,9,11,12] We suggest that this entity, rather than being rare, is misdiagnosed and managed as a common arterial trunk.

The morphological differences between the tetralogy of Fallot with pulmonary atresia and aortopulmonary window and common arterial trunk are shown in Table 1.

Conclusion

We have described an outflow tract malformation, tetralogy of Fallot with pulmonary atresia and aortopulmonary window, which can be misdiagnosed as common arterial trunk, but which has definitive morphological differences. Data from more such patients is needed to categorise the type of ventricular septal defect, morphology of ventricular outlet valve, anatomy of arterial duct, and coronary artery pattern.

Acknowledgements. None.

Financial support. None.

Conflicts of interest. None.

Ethical standards. The ethics committee of the Narayana Institute of Cardiac Sciences approved the study (NHH/AEC-CL-2021–636) and waived need for the individual consent.

References

1. Tchervenkov CI, Roy N. Congenital heart surgery nomenclature and database project: pulmonary atresia—ventricular septal defect. Ann Thorac Surg 2000; 69 (4 Suppl): S97–S105. doi: 10.1016/s0003-4975(99)01285-0.
2. Rabinovitch M, Herrera-deLeon V, Castaneda AR, Reid L. Growth and development of the pulmonary vascular bed in patients with tetralogy of

Fallot with or without pulmonary atresia. Circulation 1981; 64: 1234–1249. doi: 10.1161/01.cir.64.6.1234.
3. Franklin RCG, Béland MJ, Colan SD, et al. Nomenclature for congenital and paediatric cardiac disease: the International Paediatric and Congenital Cardiac Code (IPCCC) and the Eleventh Iteration of the International Classification of Diseases (ICD-11). Cardiol Young 2017; 27: 1872–1938. doi: 10.1017/S1047951117002244.
4. Jacobs ML. Congenital heart surgery nomenclature and database project: truncus arteriosus. Ann Thorac Surg 2000; 69 (4 Suppl): S50–S55. doi: 10.1016/s0003-4975(99)01320-x.
5. Jacobs ML, Anderson RH. Rationalising the nomenclature of common arterial trunk. Cardiol Young 2012; 22: 639–646. doi: 10.1017/S1047951112001606.
6. Anderson RH, Penny DJ. Common arterial trunk. In: Anderson RH, Baker EJ, Penny DJ, Redington AN, Rigby ML, Wernovsky G (eds). Paediatric Cardiology, 3rd edn. Churchill Livingstone Elsevier, Philadelphia, PA, 2010: 859–874.
7. Mavroudis C, Backer CL. Truncus arteriosus. In: Mavroudis C, Backer CL (eds). Pediatric Cardiac Surgery, 4th edn. Wiley-Blackwell, West Sussex, UK, 2013: 361–375.
8. Rossi RN, Hislop A, Anderson RH, Martins FM, Cook AC. Systemic-to-pulmonary blood supply in Tetralogy of Fallot with pulmonary atresia. Cardiol Young 2002; 12: 373–388. doi: 10.1017/s1047951100012981.
9. Liao PK, Edwards WD, Julsrud PR, Puga FJ, Danielson GK, Feldt RH. Pulmonary blood supply in patients with pulmonary atresia and ventricular septal defect. J Am Coll Cardiol 1985; 6: 1343–1350. doi: 10.1016/s0735-1097(85)80223-0.
10. Pahl E, Fong L, Anderson RH, Park SC, Zuberbuhler JR. Fistulous communications between a solitary coronary artery and the pulmonary arteries as the primary source of pulmonary blood supply in tetralogy of Fallot with pulmonary valve atresia. Am J Cardiol 1989; 63: 140–143. doi: 10.1016/0002-9149(89)91104-1.
11. Baker EJ, Anderson RH. Tetralogy of Fallot with pulmonary atresia. In: Anderson RH, Baker EJ, Penny DJ, Redington AN, Rigby ML, Wernovsky G (eds). Paediatric Cardiology, 3rd edn. Churchill Livingstone Elsevier, Philadelphia, PA, 2010: 774–793.
12. Anderson RH. Lesions with normal segmental connections. In: Anderson RH, Spicer DE, Hlavacek AM, Cook AC, Backer CL (eds). Wilcox's Surgical Anatomy of the Heart, 4th edn. Cambridge University Press, Cambridge, UK, 2013: 150–243.
13. Graham A, Poopalasundaram S, Shone V, Kiecker C. A reappraisal and revision of the numbering of the pharyngeal arches. J Anat 2019; 235: 1019–1023. doi: 10.1111/joa.13067.
14. Kasturi S, Prabhu S. Handmade trileaflet valved ePTFE right ventricle to pulmonary artery conduit – how do we do it? Multimed Man Cardiothorac Surg 2020. doi: 10.1510/mmcts.2020.023.
15. Anderson RH, Chaudhry B, Mohun TJ, et al. Normal and abnormal development of the intrapericardial arterial trunks in humans and mice. Cardiovasc Res 2012; 95: 108–115. doi: 10.1093/cvr/cvs147.
16. Mori K, Ando M, Takao A, Ishikawa S, Imai Y. Distal type of aortopulmonary window. Report of 4 cases. Br Heart J 1978; 40: 681–689. doi: 10.1136/hrt.40.6.681.
17. Ho S, Gerlis L, Anderson C, Devine W, Smith A. The morphology of aortopulmonary windows with regard to their classification and morphogenesis. Cardiol Young 1994; 4: 146–155. doi: 10.1017/S1047951100002092.
18. Jacobs JP, Quintessenza JA, Gaynor JW, Burke RP, Mavroudis C. Congenital heart surgery nomenclature and database project: aortopulmonary window. Ann Thorac Surg 2000; 69 (4 Suppl): S44–S49. doi: 10.1016/s0003-4975(99)01236-9.
19. Anderson RH, Wilcox BR. The surgical anatomy of ventricular septal defects associated with overriding valvar orifices. J Card Surg 1993; 8: 130–142. doi: 10.1111/j.1540-8191.1993.tb00363.x.