

# Valvar Pulmonary Atresia in Afro-Caribbean Adolescents

## Echocardiographic Features

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### ABSTRACT

*Valvar pulmonary atresia is a rare congenital anomaly usually associated with ventricular septal defects or intact ventricular septum. The patent ductus arteriosus (56%) and systemic pulmonary collaterals maintain pulmonary blood flow. Presentation beyond childhood is unusual, as there is usually high morbidity in the first year of life. The two index cases highlight the rarity of adolescents with valvar pulmonary atresia and focus on the transthoracic echocardiographic features, whose diagnoses provide definitive and curative options with adequate size pulmonary dimensions. Options available are transcatheter interventional radiofrequency pulmonary valvotomy, which is both diagnostic and therapeutic, and intraoperative valve replacement, thus eliminating the need for right ventricle pulmonary artery external valved conduit during corrective surgery. They are the first cases of adolescents with isolated cardiac anomaly of valvar pulmonary atresia, documented in Afro-Caribbeans.*

**Keywords:** Patent ductus arteriosus, radiofrequency valvotomy, systemic pulmonary collaterals, valvar pulmonary atresia, ventricular septal defect

# Atresia Valvular Pulmonar en Adolescentes Afro-Caribeños: Características Ecocardiográficas

S Williams-Phillips

### RESUMEN

*La atresia pulmonar valvar es una anomalía congénita rara, generalmente asociada con defectos septales ventriculares o septo ventricular intacto. El conducto arterioso persistente (56%) y los colaterales pulmonares sistémicos mantienen el flujo sanguíneo pulmonar. Es inusual que se presente más allá de la infancia, ya que generalmente está asociada con una alta morbilidad en el primer año de vida. Los dos casos índice destacan la rareza de adolescentes con atresia pulmonar valvar, y ponen de relieve las características ecocardiográficas transtorácicas, cuyo diagnóstico proporciona opciones para una definición y cura, con dimensiones pulmonares de tamaño adecuado. Las opciones disponibles son la valvotomía pulmonar con radiofrecuencia intervencionista con catéter, que es a la vez diagnóstica y terapéutica; y el reemplazo de la válvula intraoperatoria, eliminando así la necesidad del conducto con válvula externa de la arteria pulmonar del ventrículo derecho durante la cirugía correctiva. Se trata de los primeros casos de adolescentes con anomalía cardíaca aislada de atresia pulmonar valvar, documentados entre los afrocaribeños.*

**Palabras claves:** Conducto arterioso persistente, valvotomía con radiofrecuencia, colaterales pulmonares sistémicos, atresia pulmonar valvar, defecto septal ventricular

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### INTRODUCTION

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Valvar pulmonary atresia is a rare structural cardiac defect exemplified by 25 cases with ventricular septal defect, noted in eight years at Guy's and St Thomas's Hospital (1–3). It has been diagnosed prenatally and has an incidence of 0.16% (1, 2). It was first mentioned and successfully repaired by Lille-

hei *et al* (1955), and is usually associated with ventricular septal defects or intact ventricular septum. It has been documented to also be associated with tetralogy of Fallot, transposition of the great arteries and double outlet right or double outlet left ventricle. Definitive investigation and treatment of these patients are dependent on the anatomy, morphology and differentiation of the right ventricle, right ventricle outflow tract and the pulmonary artery circulation. The site of atresia, from the subvalvar, valvar as in the index cases or supra-valvar defect, can be determined by two-dimensional echocardiograms. The anatomy of the main pulmonary artery, whether it is atretic, hypoplastic, confluent, or non-confluent, also guides the type of investigation and potential corrective treatment (1–4).

Gibbs *et al* and Lee *et al* have documented prerequisites for pulmonary valvotomy in isolated valvar pulmonary atresia with confluent pulmonary arteries, of tripartite right ventricle and normal tricuspid valve, facilitating biventricular repair, not universally accepted, but which the index cases have (5, 6).

Harikrishman *et al* noted that the pulmonary blood flow for survival comes from the patent ductus arteriosus in over 56% of patients (1–4). Systemic-pulmonary collaterals are documented from all thoracic arteries, including the coronary artery as noted by Tfiza *et al* (7). Pulmonary blood flow usually requires augmentation and the six main methodologies are cardiothoracic surgery with B-T shunts, used by Huhta from 1987, right ventricle pulmonary artery extra-cardiac conduit, first completed by Rastelli in 1951, valved conduit by Ross from 1965, unifocalization initiated by Haworth and McCartney in 1981, transcatheter valve replacement used by Lock in 1983, or transcatheter therapy with radiofrequency pulmonary valvotomy of imperforate valve by Quershi *et al* from 1991 (1, 2, 4–6). Lee *et al* and Kawel *et al* documented lower perioperative morbidity and mortality in the latter two interventions (6, 8).

Valvar pulmonary atresia is the only physical abnormality of the two adolescents. This case series notes the rarity in adolescents and focusses on the transthoracic echocardiographic features of valvar pulmonary atresia, which would facilitate use of the transcatheter radiofrequency pulmonary valvotomy of the imperforate valve with transcatheter pulmonary valve replacement, or intraoperative valve insertion, which carries no need for potential thoracotomy extra-cardiac conduit revisions every 4–6 years. Kawel *et al* indicated that this group of valvar pulmonary atresia should be reserved for catheter interventions (8). They also excluded the need for computed tomography (CT) scan or magnetic resonance imaging (MRI), which are purely diagnostic and have no curative value in these specific index cases of valvar pulmonary atretic group, with main and confluent pulmonary arteries (4, 8).

## CASE REPORTS

### Case 1

The index case is a 13-year old Afro-Caribbean female with a height of 170 cm, weight of 47.5 kg and a body mass index of 16.4 kg/m<sup>2</sup>. She was diagnosed at four years of age with presentation of increasing cyanosis, with pulmonary atresia. She functioned at New York Heart Association (NYHA) II, on no medical therapy nor interventions until adolescence, when anti-failure treatment and phlebotomy for polycythaemia complicated by hyperviscosity syndrome commenced. She had plethoric mucous membranes, with signs of chronic hypoxia, cyanosis, clubbing of fingers and toes. Index case 1 is the only member of the family with congenital heart disease.

Cardiovascular examination revealed normal pulses, no pulse deficit, normal blood pressure, parasternal heave, displaced apex beat in the 6<sup>th</sup> left intercostal space in the anterior axillary line, tapping in character, single 2<sup>nd</sup> heart sound and a continuous murmur in the left mid-subclavicular area and both right and left lung fields. There were no diastolic murmurs, no additional heart sounds, nor signs of heart failure.

Haemoglobin was 19.0 g/dL. Packed cell volume was 62 L/L. Her steady state saturations were 78%. Electrocardiogram showed sinus rhythm and right axis deviation. Chest X-ray showed normal cardiothoracic ratio, pulmonary bay, with variable vascularity. There was plethora in the right mid and lower zones but the rest of the lung fields were normal, indicating multifocal pulmonary blood supply. There was a left-sided aortic arch.

Transthoracic echocardiography showed situs solitus, atrioventricular concordance, ventriculo-arterial concordance (Fig. 1A), confirmed tetralogy of Fallot with 50% override of the aorta (Fig. 1B) and valvar pulmonary atresia (Figs. 1C–F). There was a tricuspid aortic valve (Fig. 1C). Colour Doppler showed flow in the right ventricular infundibulum to the atretic pulmonary valve (Fig. 1D) and also showed flow in the main pulmonary artery, to the atretic valve (Fig. 1E). There was a patent ductus arteriosus with a left to right shunt, with retrograde flow into the main pulmonary artery confirmed on pulsed wave Doppler showing continuous flow (Fig. 1G), indicating low main pulmonary artery pressures. The pulmonary arteries were confluent with hypoplasia. There was a large ventricular septal defect extending from the trabecular area to the juxta-arterial outlet septum (Fig. 1C).

Systemic-pulmonary collaterals with left to right shunt were noted in the suprasternal view of the left-sided descending aorta. There were intact interatrial septum, normal tricuspid, mitral, aortic valves and normal ventricular function.

### Case 2

Index case 2 is a 13-year old Afro-Caribbean male with a height of 160 cm, weight of 35.7 kg and a body mass index of 13.9 kg/m<sup>2</sup>. He was diagnosed at three years of age with

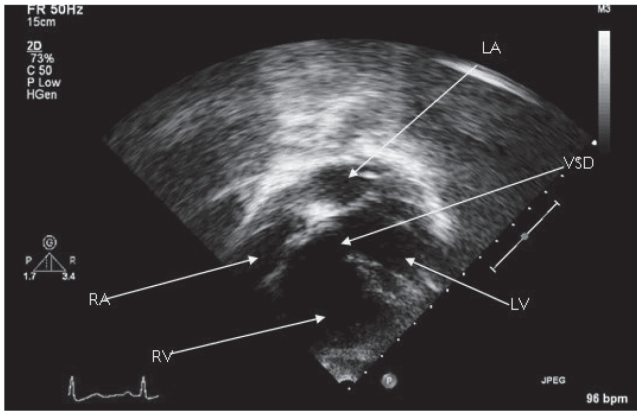


Fig. 1A: Four-chamber view showing right atrium (RA), right ventricle (RV), left atrium (LA), left ventricle (LV) and large ventricular septal defect (VSD).

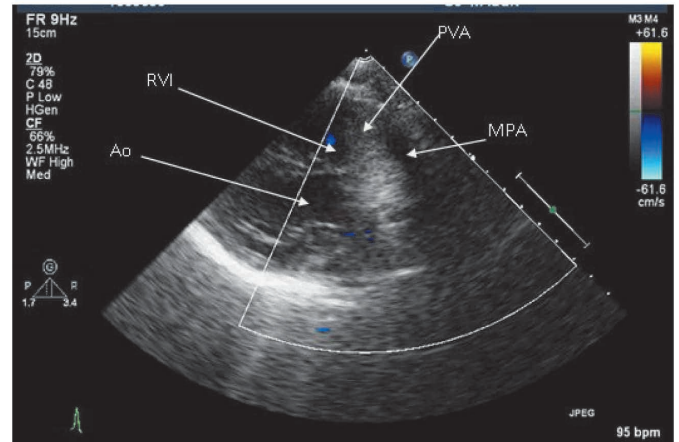


Fig. 1D: Parasternal short axis view showing right ventricle infundibulum (RVI) with colour Doppler showing flow up to pulmonary atresia, main pulmonary artery (MPA), pulmonary valve atresia (PVA) and aorta (Ao).

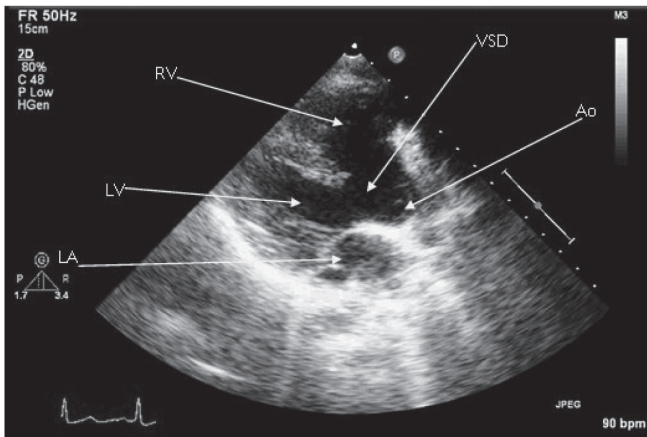


Fig. 1B: Parasternal long axis view showing right ventricle (RV), left ventricle (LV), aorta (Ao) override of RV and LV, large ventricular septal defect (VSD) and left atrium.

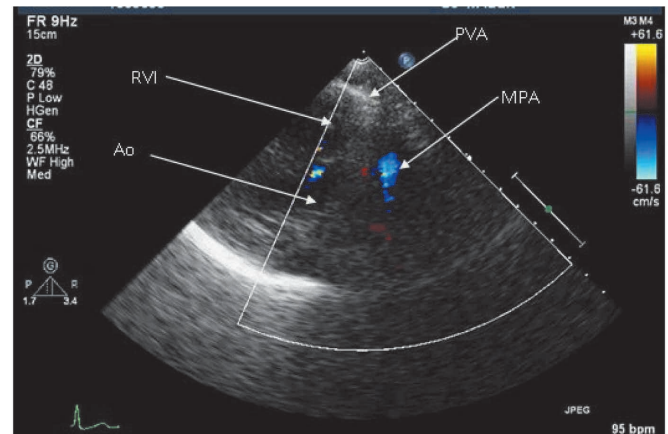


Fig. 1E: Parasternal short axis view showing right ventricle infundibulum (RVI), main pulmonary artery (MPA), with colour Doppler showing flow in MPA up to pulmonary atresia, pulmonary valve atresia (PVA) and aorta (Ao).

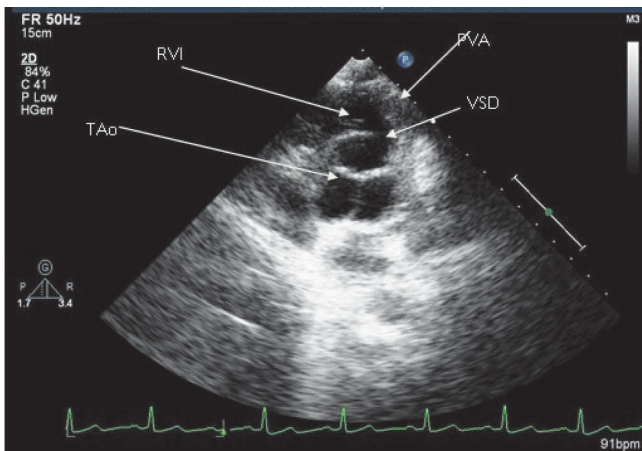


Fig. 1C: Parasternal short axis view showing trileaflet aortic valve (TAo) and juxta-arterial ventricular septal defect (VSD).

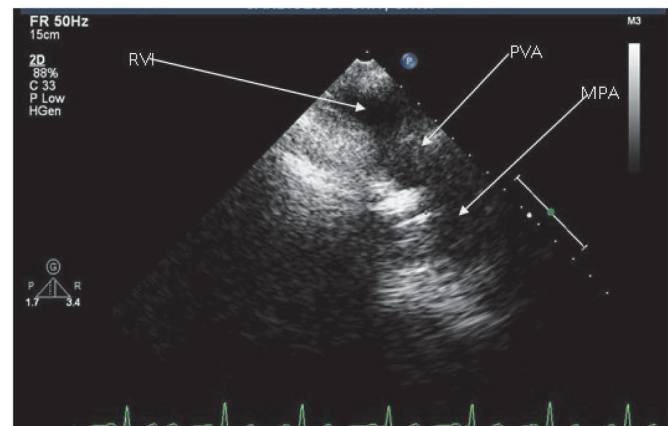


Fig. 1F: Parasternal short axis view showing right ventricle infundibulum (RVI), main pulmonary artery (MPA), up to pulmonary valve atresia (PVA).

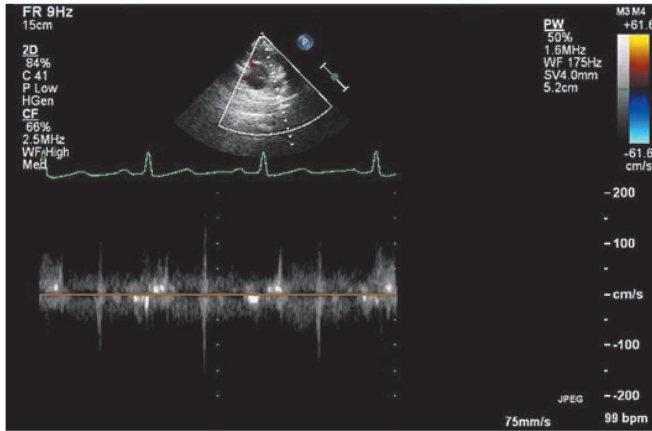


Fig. 1G: Parasternal short axis view of main pulmonary artery (MPA), with continuous pulsed Doppler flow in MPA.

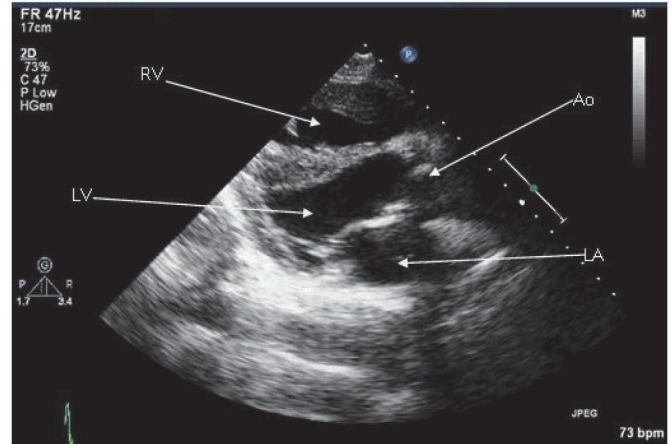


Fig. 2A: Parasternal short axis view showing right ventricle (RV), the aorta (Ao) arising from the left ventricle (LV) with no aortic override, and left atrium (LA).

cyanosis and a cardiac murmur. He had infective endocarditis therapy at eight years of age with commencement of anti-failure therapy in childhood. His steady state saturation was 77%, and he had signs of chronic hypoxaemia as index case 1.

Cardiovascular examination revealed normal pulses, normal blood pressure, precordial bulge, parasternal heave, displaced apex beat in the 6<sup>th</sup> left intercostal space in the anterior axillary line, thrusting in character, single 2<sup>nd</sup> heart sound. There was a continuous murmur in the upper left sternal border and diastolic murmur in the aortic area. There were no additional heart sounds, nor signs of heart failure. Haemoglobin was 18.5 g/dL. Packed cell volume was 53.7 L/L. Electrocardiogram showed sinus rhythm with signs of biventricular hypertrophy. Chest X-ray showed increase in cardiothoracic ratio, normal lung fields and left-sided aortic arch.

Transthoracic echocardiogram showed situs solitus, atrioventricular concordance and ventriculo-arterial concordance with valvar pulmonary atresia. The aorta arose from the left ventricle with no aortic override (Fig. 2A). Valvar pulmonary atresia with confluent pulmonary arteries with continuous flow on pulsed Doppler was clearly seen between the right infundibular area and main pulmonary artery (Figs. 2B–D). There was a tricuspid aortic valve (Fig. 2B) with fusion of right and left commissures, functioning bicuspid valve with the classic fish mouth appearance when open (Fig. 2C). There was moderate regurgitation of the aortic valves, which was subsequently replaced. There was a large ventricular septal defect extending from the trabecular area to the juxta-arterial outlet septum (Fig. 2D).

There was a patent foramen ovale with left to right shunt. A patent ductus arteriosus (PDA) and systemic pulmonary collaterals were noted in suprasternal views of the descending aorta, with left to right shunt (Fig. 2E). Mitral and tricuspid valves were normal, with normal ventricular function.

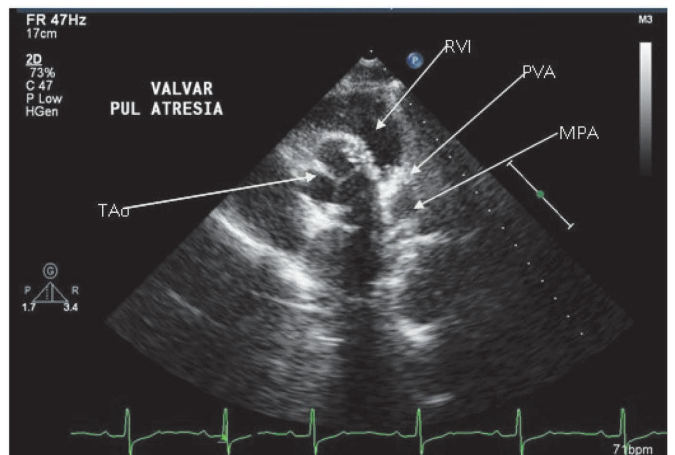


Fig. 2B: Parasternal short axis view showing right ventricle infundibulum (RVI), main pulmonary artery (MPA), pulmonary valve atresia (PVA) and trileaflet aorta (Ao).

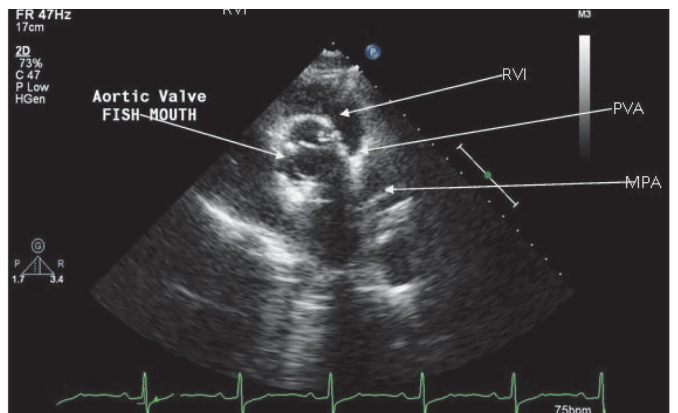


Fig. 2C: Parasternal short axis view showing right ventricle infundibulum (RVI), main pulmonary artery (MPA), pulmonary valve atresia (PVA) and aorta (Ao) open with fish mouth appearance.

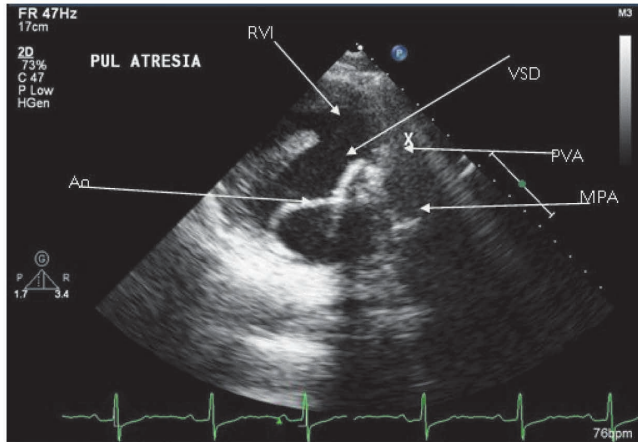


Fig. 2D: Parasternal short axis view showing right ventricle infundibulum (RVI), main pulmonary artery (MPA), pulmonary valve atresia (PVA), juxta-arterial ventricular septal defect (VSD) and aorta (Ao).

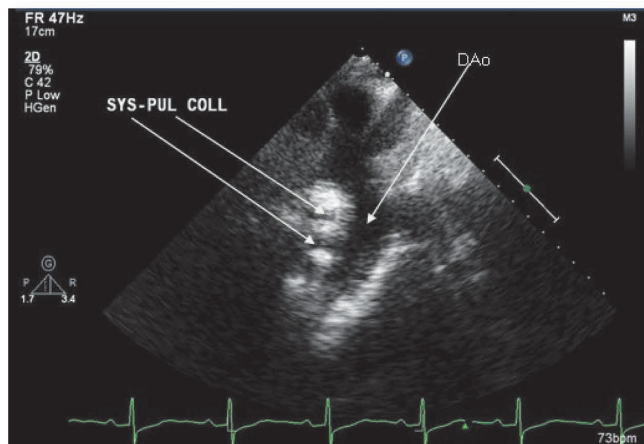


Fig. 2E: Suprasternal view showing systemic-pulmonary collaterals on anterior aspect of descending aorta (DAo).

## DISCUSSION

The two main non-echocardiographic, non-invasive cardiac structural anatomic diagnostic modalities are CT scan and cardiac MRI, which are effective in confirming the structural anatomic diagnosis; in the latter, it is 87%, as noted by Kawel *et al* (1, 2, 4, 8). They are not therapeutic and are costly investigations, not readily available in all cardiac centres, as is the case in the index country. Cardiac catheterization is invasive but can be both diagnostic and therapeutic, in cases with isolated pulmonary valve atresia, facilitating definitive treatment with transcatheter laser assisted pulmonary valvotomy and valve replacement. Pulmonary atresia with a concomitant ventricular septal defect may have pulmonary valve insertion

used during corrective cardiac surgery, thus avoiding the need for an extra-cardiac valved conduit, which may require open thoracotomy revisions, potentially every 4–6 years, with higher perioperative, interoperative morbidity and mortality. The replacement of defective, stenosed or incompetent intraoperative valves inserted can be done *via* cardiac catheterization in the future, if required (4, 6, 7, 9, 10).

Valvar pulmonary atresia is a rare congenital anomaly in adolescence, and its differentiation from other types of pulmonary atresia is important not only in determining the type of investigations, but ultimately the type of treatment that would be most suitable and successful (4–7). The rarity in adolescence and its transthoracic echocardiographic features are the focussed features of the index cases with valvar pulmonary atresia, which would, with good sized pulmonary dimensions, optimally lead to transcatheter interventional radiofrequency pulmonary valvotomy and valve replacement and intraoperative valve replacement (4–7, 9, 10).

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