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Case Report

Double Outlet Right Ventricle with Pulmonary Atresia and Antero-Posterior Malposition of Great Arteries: Transthoracic Echocardiographic Evaluation of a rare cyanotic congenital heart disorder

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Abstract

Double outlet right ventricle (DORV) is a heterogeneous group of abnormal ventriculoarterial connections where, by definition, both great arteries (pulmonary artery and aorta) arise primarily from the morphologically right ventricle, This condition affects 1-1.5% of the patients with congenital heart diseases, with a frequency of 1 in each 10,000 live births. We are reporting a case of seven year old deeply cyanotic child with DORV, subaortic VSD with rare anatomic features of antero-posterior malposition of great arteries (GA) and pulmonary vavular atresia, with conspicuous absence of patent ductusarteriosus (PDA) or major aorto-pulmonary collaterals (MAPCAs), despite the presence of atretic pulmonary valve.

Keywords: *DORV, Pulmonary atresia, TGA, VSD, subaortic VSD, overriding of Aorta, cyanotic congenital heart defects.*

Introduction

Double outlet right ventricle (DORV) accounts for about 2-3% of all congenital heart defects, with a birth prevalence rate of 1/10,000. In USA congenital heart disease (CHD) occurs in less than 1% of all newborns, and DORV is present in 0.5-1.5% of all patients with CHD[1].

Definition

DORV is defined as a form of abnormal ventriculo-arterial connection in which both GA arise completely or predominantly from morphologic right ventricle (Figure 1)[2].

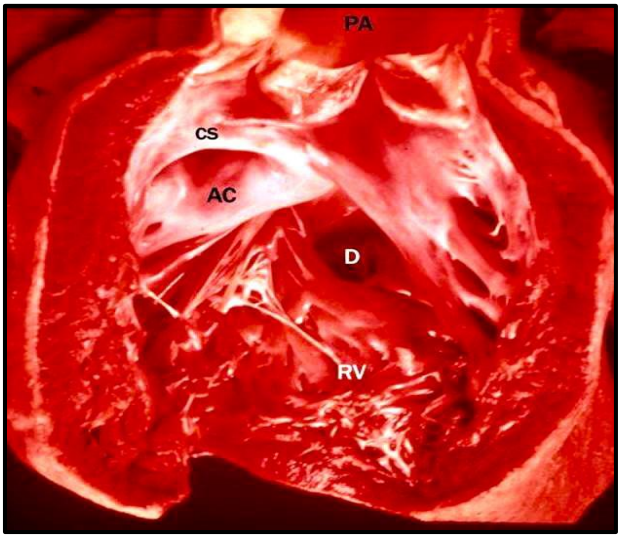


Figure 1: Pathologic specimen of double- outlet right ventricle. The right ventricle (RV) has been opened, demonstrating the origin of both great arteries from the RV. AC, aortic conus; CS, conus septum; D, defect (ventricular septal defect [VSD]); PA, pulmonary artery

Classifications

There are multiple classification systems describing DORV. The conventional classification groups the patients based on the location of the VSD and relationship of great arteries (Figure 2)[3]. Most VSD in DORV are conotruncal located below either the aortic valve or below the pulmonary valve or both. Rarely, they are remote from both great arteries.

Figure 2: Conventional classification of double outlet right ventricle [3].

Relation of great artery	Location of ventricular septal defect (%)			
	Subaortic VSD	Subpulmonic VSD	Doubly committed VSD	Remote VSD
Normal	3	0	0	0
Side by side	46	8	3	7
D-malposition	16	10	0	0
L -malposition	3	4	0	0

DORV: Double outlet right ventricle, VSD: Ventricular septal defect

DORV was classified by Lev et al[4] in terms of ventricular septal defect (VSD) positions (Figure 3):

1. Subaortic
2. Subpulmonary
3. Doubly committed
4. Uncommitted

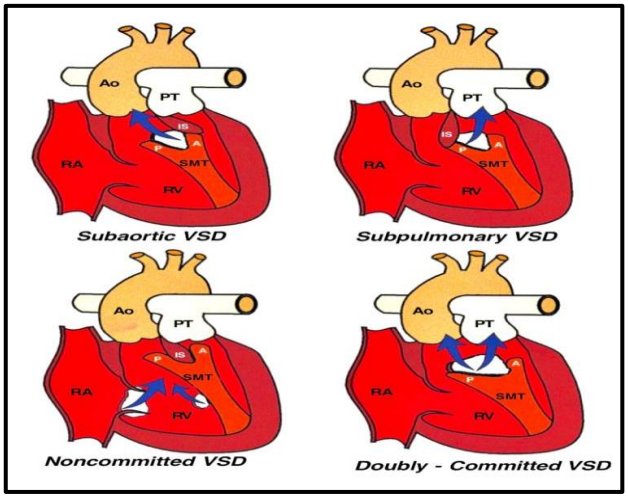
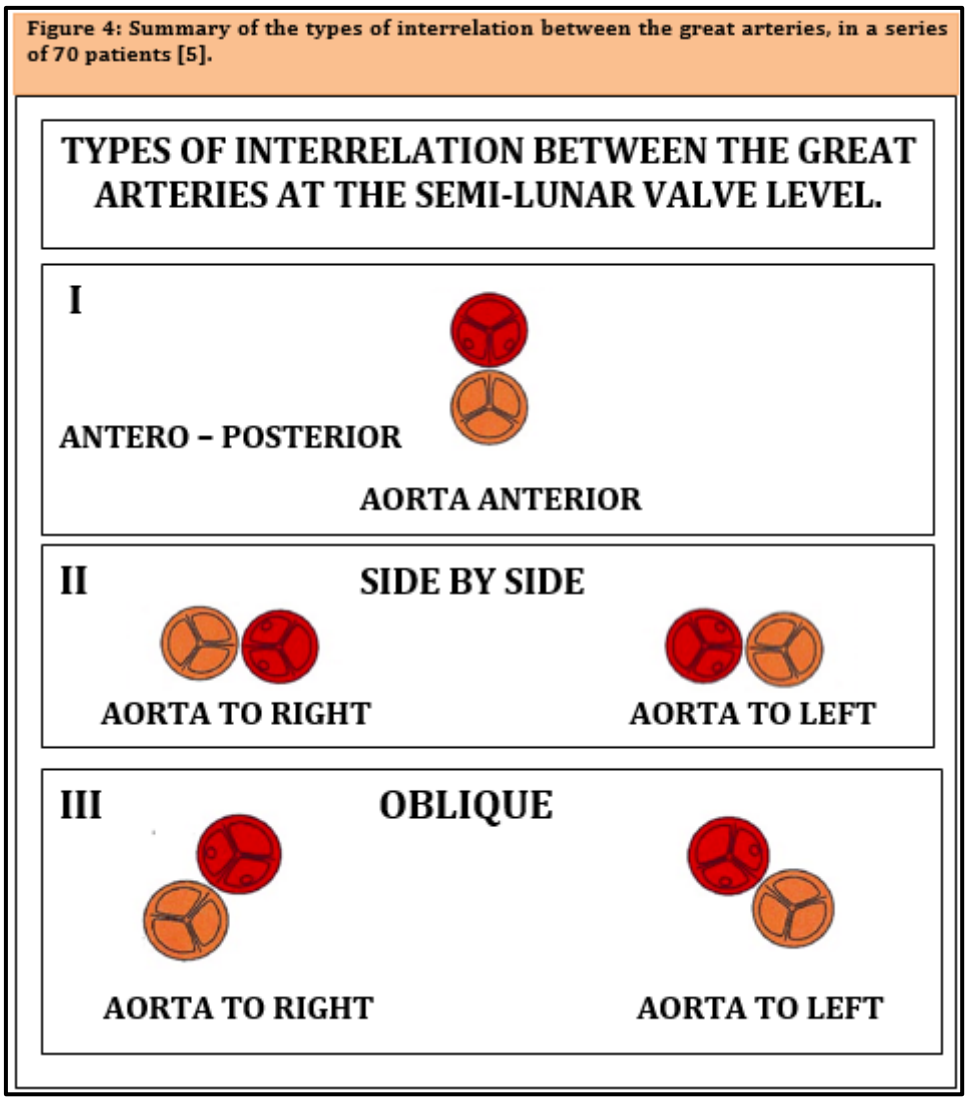


Figure 3: Illustration of the position of the ventricular septal defect (VSD) in hearts with double outlet right ventricle (DORV). A-anterior limbus; RA-right atrium; Ao-aorta; P- posteriorlimbus; IS- infundibular septum; PT-pul- monary trunk; SMT trabeculaseptomarginalis, RV-right ventricle.

However, Guerin et al[5] classified the DORV according to interrelation between the GA at the semi-lunar valve level in their series of 70 patients (Figure 4).



Modified Fuwai classification proposed by Pang et al.[6] after analysis of 500 patients with DORV is based on three parameters namely great artery relation, location of the VSD, and presence of outflow obstruction. These parameters determine the clinical features and provide guidance for the relevant surgery (Figure 5).

Figure 5: Modified Fuwai classification with double-outlet right ventricle adapted from Pang et al [6].

DORV subtype	Relative position of great arteries	Relation between VSD and great artery	RVOTO
I-A	Normal	Committed	Absent
I-B	Normal	Committed	Present
II-A	Normal	Noncommitted	Absent
II-B	Normal	Noncommitted	Present
III-A	Abnormal	Committed	Absent
III -B	Abnormal	Committed	Present
IV-A	Abnormal	Noncomitted	Absent
IV-B	Abnormal	Noncomitted	Present

DORV: Double-outlet right ventricle, VSD: Ventricular septal defect.
RVOTO: Right ventricular outflow tract obstruction

The prefixes **a-, d- and l- transposition** describe the spatial relationship between the aorta and the pulmonary trunk, and should not be used to define this anomaly[7]. In d-transposition, the aortic valve lies to the right of the pulmonary valve. In congenitally corrected hearts, the aorta usually lies on the left (l-transposition). The a-transposition refers to the anterior position of the aortic valve in relation to the pulmonary trunk.

Associated lesions

The common associations of DORV include pulmonary stenosis in 50%, secundum atrial septal defect in 25% and atrioventricular septal defects in 8%. The other associations were patent ductusarteriosus, right aortic arch, persistent left superior vena cava, and mitral valve anomalies. Coronary anomalies seen in 10% include origin of the left anterior descending coronary artery from the right coronary artery. These carry significance during transannular patch repair or arterial switch surgeries for various forms of DORV[4,8].

Clinical Presentation

Patients with DORV have three different clinical presentations:

1. Commonest form is "like a Tetralogy of Fallot." This accounts for 40% of patients. There is a large unrestrictive subaortic VSD and the subpulmonaryconus is narrowed in its diameter.
2. Second form presents "like transposition of great arteries," seen in 20% of patients. The VSD is located in the subpulmonary region. The larger conal tissue below the aortic valve prevents left ventricular blood to stream into the aorta leading to lower aorticsaturation compared to pulmonary artery saturations. This is also named as Taussig Bing anomaly.
3. The third form is "like a large nonrestrictive VSD." They present with a large left to right shunt without any pulmonary outflow obstruction and are seen in 15% of patients.
4. Other less common variants are noncommitted VSD andassociations with atrioventricular septal defects with varying degrees of outflow obstructions[9,10].

Besides the invasive cardiac catheterisation and angiography technique there are other robust non-invasive imaging modalities currently available for a comprehensive anatomic diagnosis of DORV including Cardiovascular computed Tomography, Cardiovascular Magnetic Resonance and Echocardiography (Figures 6-8).

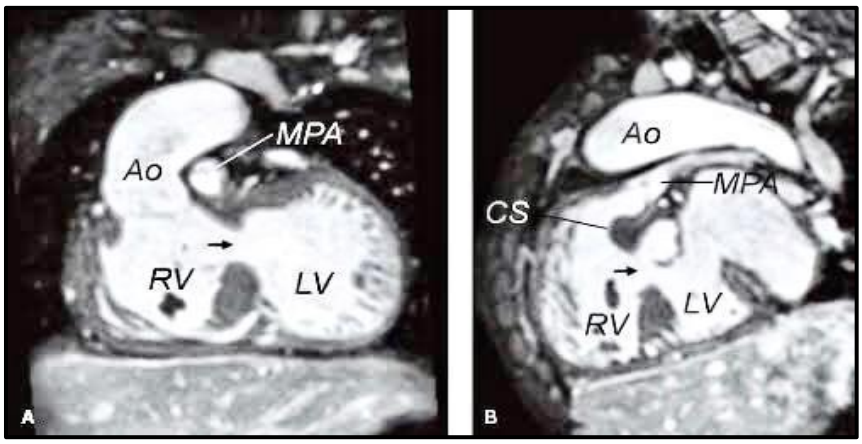


Figure 6: Cardiac MRI images of DORV- "tetralogy type". A: This image highlights the unobstructed pathway from the LV to the aorta, which will be surgically baffled for repair of this type of DORV. B: This sagittal image demonstrates prominent conal muscle beneath the pulmonary valve. The degree of obstruction at the subvalvar level determines the degree of cyanosis. Black arrow, Ventricular septal defect; Ao, aorta; CS, conal septum; MPA, main pulmonary artery; RV, right ventricle; LV, left ventricle.

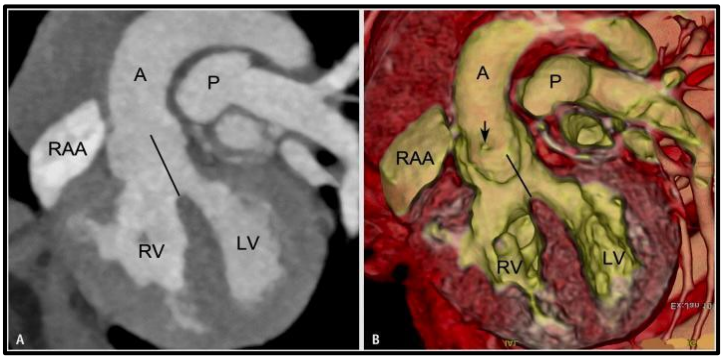


Figure 7: Cardiac CT images of DORV (A) Long axis CT image and (B) Transparent lumen volume-rendered CT image shows that a diameter > 50% of the annulus of the overriding aortic valve is connected to the morphologic RV based on an extension line of the ventricular septum. In clinical practice, measuring the diameter is more frequently used than the original method for measuring the circumference. The origin of the right coronary artery (arrow in B) was noted. A = aorta, LV= left ventricle, P = pulmonary artery, RAA = right atrial appendage, RV = right ventricle

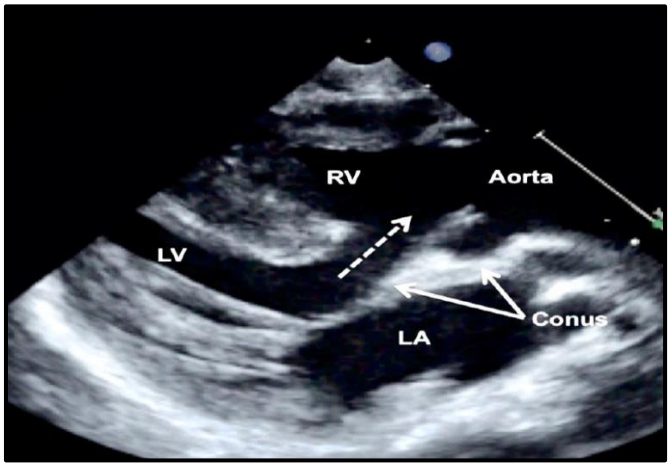


Figure 8: Parasternal LX view - Large subaortic VSD (shown by interrupted arrows) is discerned alongwith a large conal tissue (marked with 2 white arrows) causing aorto- mitral discontinuity.
LA; left atrium, LV; left ventricle, RV; right ventricle

Echocardiogram remains the mainstay in diagnosis of DORV for identifying the location of the VSD, its relation to the great arteries, relationship of great arteries, outflow obstructions, ventricular hypoplasia, atrioventricular valve anomalies, and other associated anomalies.

Case Report

A seven year old deeply cyanotic female child was referred to us for detailed cardiac evaluation. The clinical history of the patient was enumerated by the consenting parents. According to them the child was born at a private hospital as full term normal delivery and was found to be cyanotic at birth. The child was apparently sick looking, having deep cyanosis and clubbing of all the fingers and toes. A striking cyanosis was also obvious over lips and tongue (Figures 9 a, b, c, d).

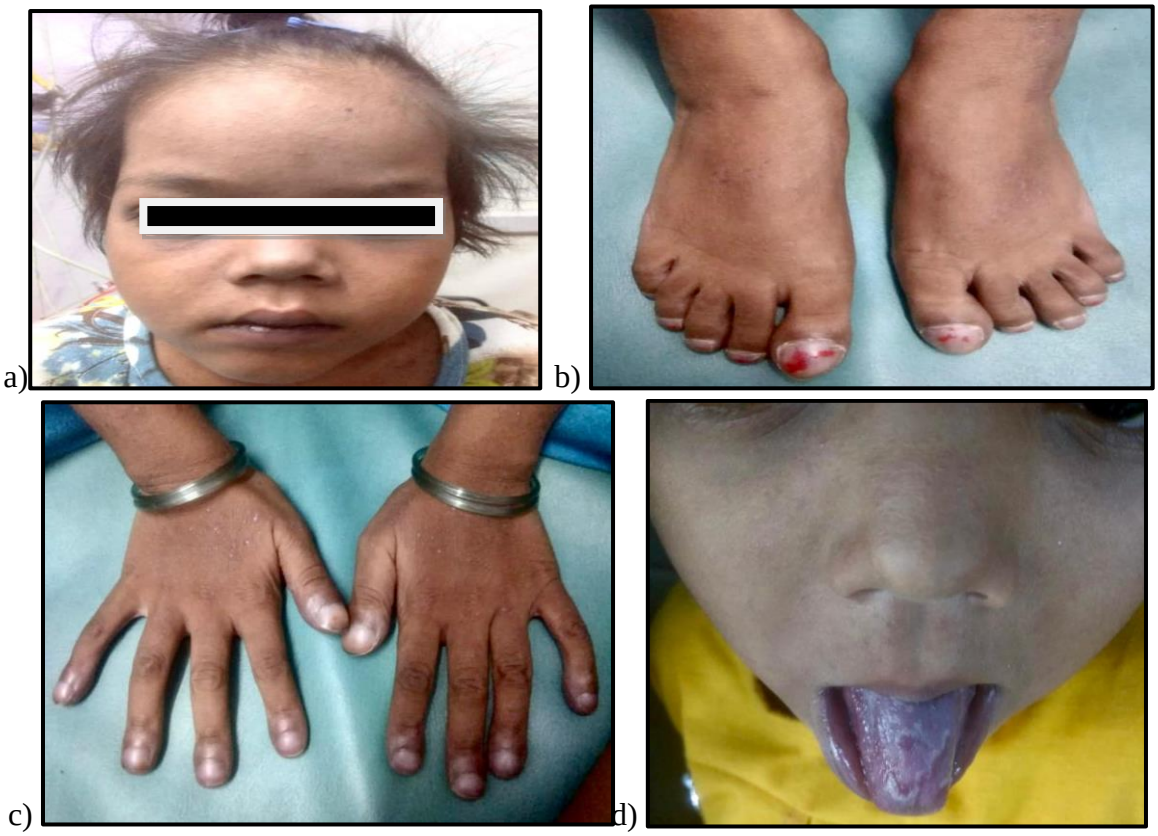


Figure 9: a) Sick looking deeply cyanotic child; b) striking presence of clubbing and cyanosis of bilateral toes of lower extremity; c) similar cyanosis and clubbing of bilateral fingertips of upper extremity; d) deep cyanosis of the tongue

On clinical examination weight was 14 kg, height was 100 cm, BP was 80/40 in the right upper extremity in the sitting position, pulse was 70/min SPO_2 was 58% at room air and all the peripheral pulses were normally palpable without any radio-femoral delay. On cardiac auscultation the second heart sound was single and loud. There was absence of any murmur clicks or gallop sounds.

Resting ECG (Figure 10) showed the presence of right ventricular hypertrophy (RVH), normal sinus rhythm and a ventricular rate of 82/mm. X ray chest (PA) exhibits (Figure 11) cardiomegaly with significantly decreased pulmonary blood flow.

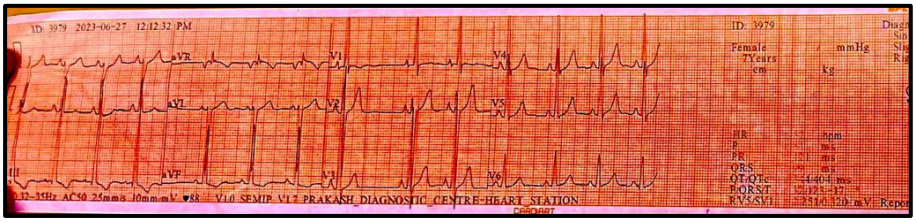


Figure 10: ECG was showing right ventricular hypertrophy with right axis deviation and a normal sinus rhythm.

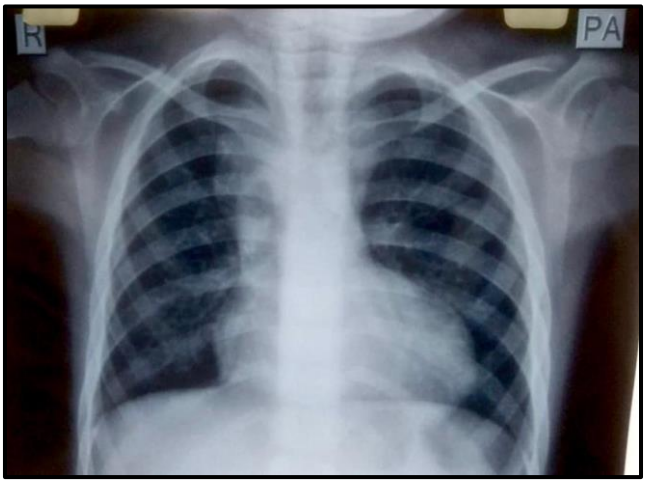


Figure 11: X-ray chest PA view was demonstrating cardiomegaly with significantly decreased pulmonary blood flow.

Transthoracic Echocardiography (TTE)

A detailed sequential chamber analysis by TTE was performed by the author in the supine and left lateral decubition position. 2Dimensional echocardiographic imaging was accomplished via parasternal long axis view (LX), short axis view (SX), 2 chamber view (2CH), 4 chamber view (4CH), 5 chamber view (5CH) and suprasternal views, to reach a definitive anatomic diagnosis. The summary of our TTE findings were:

There was situssolitus, levocardia, A-V concordance DORV, left aortic arch, normal systemic and pulmonary venous connections.

1. DORV (there is >50% overriding of Aorta).
2. Subaortic VSD (size 13.7mm, subaortic, perimembranous, non-restrictive defect)
3. Atresia of pulmonary valve (PV), Hypoplasia of PV annulus, main pulmonary artery (MPA), left pulmonary artery (LPA) and right pulmonary artery (RPA).
4. A-type malposition of great arteries;Aorta is anterior&Pulmonary artery is posterior.
5. Dilated right ventricle (RV) with concentric hypertrophy,
   Small left ventricle (LV),
   Normal biventricular systolic function,
   Normal LVEF 66%.
6. There was no evidence of PDA or MAPCAs feeding the hypoplastic branch pulmonary arteries. Moreover no additional anomalies were identified like coarctation of aorta (COA), aortic, mitral or tricuspid valvular abnormalities, atrial septal defect (ASD), remote muscular VSD or anomalous pulmonary venous drainage.

In the parasternal LX view (Figure 12), a distinctive DORV is visualized with > 50% overriding of Aorta.

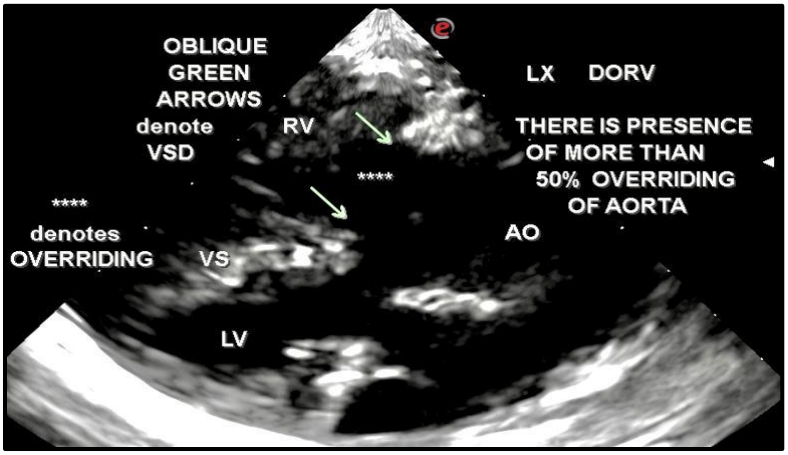


Figure 12: Parasternal long axis view revealed approximately 70% overriding of aorta and subaortic VSD. RV, right ventricle; VS, ventricular septum; LV, left ventricle; AO, aorta; **, denotes overriding.

A large subaortic, perimembranous, non-restrictive VSD of size 13.7mm was also envisaged with a left to right shunt. Additionally, a conspicuous sub-aortic conus was present (Figure 13), causing aorto-mitral discontinuity.

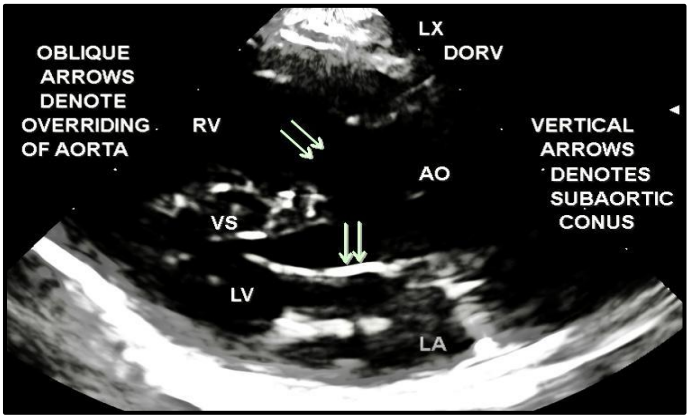


Figure 13: A subaorticconus was recognized in the parasternal LX view denoted by vertical green arrows.

In the subcostal view (Figure 14)the atretic pulmonary valve was clearly visible.

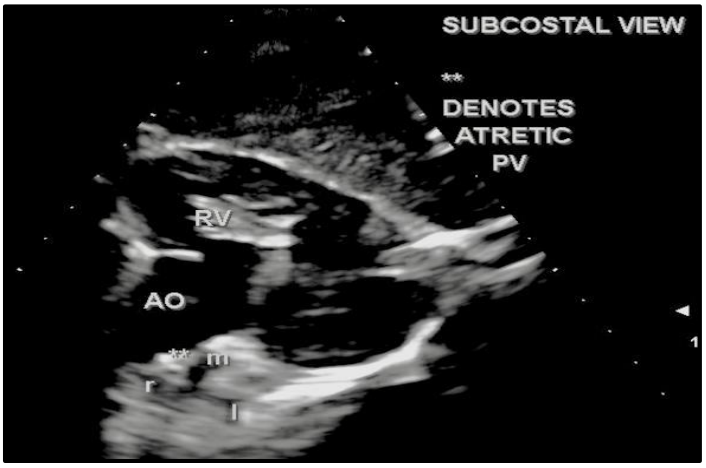


Figure 14: Subcostal view identified a typical atresia of the pulmonary valve with hypoplasia of pulmonary valve annulus, main, left and right pulmonary arteries.
RV, right ventricle; AO, aorta; m, main pulmonary artery; l, left pulmonary artery; r, right pulmonary artery; **, denotes atresia of the pulmonary valve.

Parasternal SX view (Figure 15) unveils a notable presence of A-type spatial relationship of GA, whereby aorta is lying anterior to the pulmonary artery. Moreover the atretic pulmonary valve with hypoplastic PV annulus, MPA, LPA and RPA were also recognised (PV annulus (D) 5.1mm, MPA(D) 7.5mm, LPA(D) 5.6m RPA(D) 4.6mm). The RV was dilated with presence of severe concentric hypertrophy (Figure 16). The LV was small with a normal biventricular systolic function and LVEF was 66%.

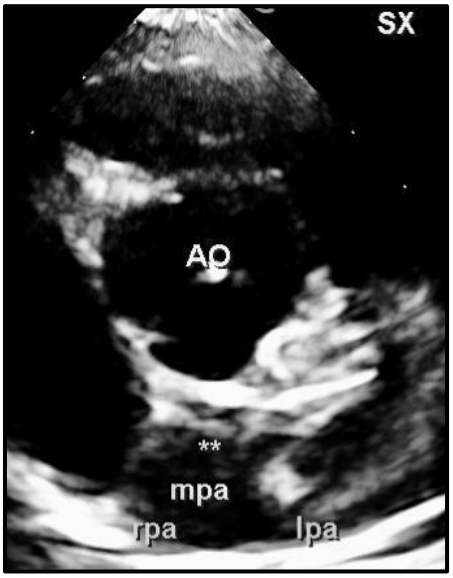


Figure 15: A-type malposition of great arteries was noted in the prasternal SX view with aorta being anterior to the pulmonary trunk. Atretic pulmonary valve along with hypoplasia of pulmonary valve annulus and branch pulmonary arteries were specially ascertained.

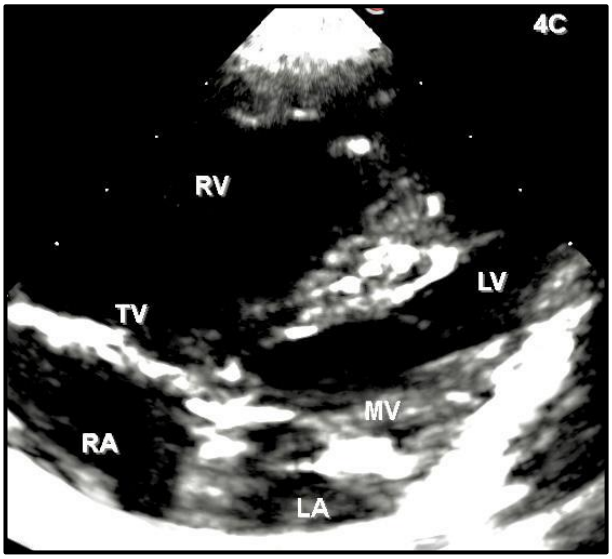


Figure 16: In the 4CH view right ventricle was dilated with severe concentric hypertrophy, conversely, the left ventricle was small. RV, right ventricle; TV, tricuspid valve; RA, right atrium; LV, left atrium; MV, mitral valve ; LA, left atrium.

In the suprasternal view a left aortic arch was present with unexpected absence of PDA or MAPCAs (Figure 17).

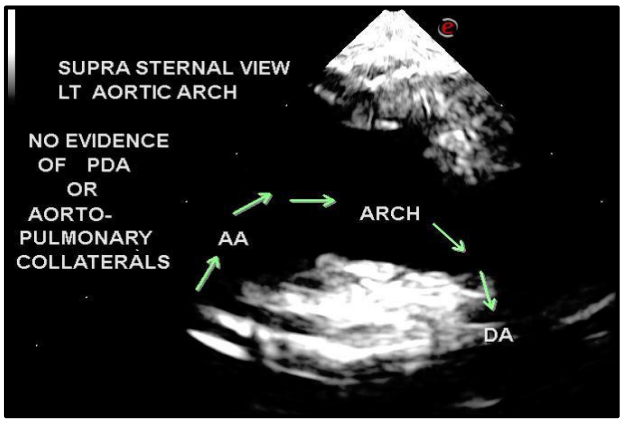


Figure 17: In the Suprasternal view there was a classical left sided unobstructed thoracic aorta with unexpected absence of PDA or MAPCAs.

Discussion

The wide spectrum of anatomic variations found in DORV may result in different clinical findings and require different therapeutic approaches.

The most frequent anatomic type is the subaortic VSD with posterior and right-sided aorta.

Aoki et al[11] reported one of the largest case series of DORV.73 patients underwent heart surgery for correction of different types of DORV. A large majority of patients, the aorta and the pulmonary trunk were side by side, 18 (25%) showed a posterior and right-sided aorta, 12 (17%) showed an anterior and right-sided aorta, 5 (7%) showed an anterior and left-sided aorta, and only 2 (2.8%) showed an anterior aorta.

In the series of Aoki et al[11] only 2 (2.8%) out of the 5 patients with anterior left-sided aorta had subpulmonary VSD.

Surgical repair of the DORV depends basically on the anatomic type of the condition. Among the choices available for definite surgical repair, the intraventricular repair, connecting the aorta to the morphologically left ventricle through the VSD while maintaining the continuity between the right ventricle and the pulmonary artery, is the preferred operative approach, whenever possible. This technique yields the best results, with less in-hospital morbidity and a lower incidence of reoperation. It is the treatment of choice whenever the VSD is subaortic and may also be used in patients with other types of VSD[11].

Other forms of definite surgical repair include the anatomical repair (translocation of the great arteries or Jatene's arterial switch procedure), which directs the blood flow from the VSD to the pulmonary artery using a surgical flap; the Rastelli repair (which uses a conduit from the right ventricle to the pulmonary artery) and the insertion of a surgical flap directing the blood flow from the VSD to the aorta; the Damus-Kaye-Stansel operation, and the inversion at the atrial level, as performed by Senning or Mustard, which directs the flow from the VSD to the pulmonary artery[11].

When the VSD is subpulmonary, the preferred surgical approach is Jatene'soperation; however, this procedure is still a challenge for many surgeons, and the results obtained are less satisfactory than those achieved in patients who undergo the intraventricular repair.

Measurement of the distance between the tricuspid and the pulmonary valves and its comparison with the diameter of the aortic ring has been considered valuable for choosing the most appropriate surgical approach[12]. When the distance between the tricuspid and the pulmonary valves exceeds the diameter of the aortic ring, the intraventricular repair is indicated.

Our case of DORV with A-type malposition of great arteries with pulmonary atresia and absence of major aorto-pulmonary collaterals or patent ductusarteriosus is perhaps the first case reported

in the literature. We are reporting our case of DORV due to its rare anatomic anomalies. The presence of A-type malposition of great arteries in which aorta is anterior to the pulmonary trunk is extremely rare and the VSD is subpulmonary or non-committed[13]. Conversely in our patient there was presence of subaortic VSD. The existence of atresia of pulmonary valve along with absence of PDA or MAPCAs is rather unusual in our patient. Significantly, in pulmonary atresia with ventricular septal defect, the pulmonary blood supply arises from the ductusarteriosus or major systemic to pulmonary collateral arteries or from both[14-16]. The collateral vessels may be relatively large arteries (3 to 20 mm in diameter), varying from one to five in number and arising most commonly from the descending thoracic aorta and less commonly from the subclavian arteries, the abdominal aorta or its branches or the left coronary artery[15,17-19]. A third type of blood supply to the lungs, described by Jefferson et al. [18], comprises numerous, uncountable, small systemic collateral arteries that either follow the bronchi or spread over the pleurae; others [15,20] have considered these an enlarged bronchial arterial circulation.Earlier, we have mentioned that we failed to demonstrate the PDA or MAPCAs arising from the thoracic aorta in the suprasternal view and perhaps the reasons are that they maybe originating from other sites as mentioned above. These other locations can be adequately delineated by utilizing the invasive technique of aortic angiography.

Echocardiography is an effective and extremely useful method for diagnosing this complex malformation. This test accurately identifies the anatomical variables and guides the choice of the most appropriate surgical approach, either through the measurement of the distance between the tricuspid and pulmonary valves or through the detection of additional malformations. These malformations may include severe aortic and pulmonary stenosis, pulmonary atresia, obstruction of the aortic arch, coarctation of the aorta and coronary anomalies that hamper right ventriculotomy and contraindicate intra-ventricular repair.

In our index patient we were unable to demonstrate the presence of PDA or MAPCAs and moreover coronary arteries could not be appropriately visualized, hence we referred the patient to a tertiary care pediatriccardiovascular institute for cardiac catherization, angiography, coronary arteriography and the necessary surgical corrective procedure.

Conclusion

The presentation of double-outlet right ventricle, subaortic VSD, pulmonary valvular atresia with absence of PDA or MAPCAs is an extremely rare clinical scenario. The patients can be exceptionally ill and, with the interplay between many different defects, diagnosis and surgical treatment can be challenging. Familiarity with the anatomic appearance of each of the defects along with an in-depth understanding of the functional consequences of these deformities is critical for the clinical cardiologist who detects them in atypical combinations. Proper utilization of diagnostic imaging tools, such as echocardiography, cardiac CT and cardiac MRI, are important for establishing diagnosis and planning of surgical treatment.

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