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RARE COEXISTENCE OF MITRAL AND PULMONARY ATRESIA WITH DOUBLE OUTLET RIGHT VENTRICLE. CASE REPORT AND REVIEW OF LITERATURE

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ABSTRACT

Double-outlet right ventricle (DORV) represents a rare group of cardiac anomalies characterized by both great arteries arising primarily from the RV. This disease is a conotruncal anomaly as a complex heart disease and comprises less than 1% of all congenital heart anomalies. The first diagnostic criterion for DORV is the rise of both the aorta and the pulmonary artery from the morphologic RV. Other cardiac malformations are always present with the frequent coexistence of multiple abnormalities. These may include 1 or more ventricular septal defects, left ventricular outflow obstruction, and atrioventricular valve abnormalities. DORV may occur with any atrial situs and many other variations; nonetheless, situs solitus and atrioventricular concordance are the most common findings. We herein present an extremely rare case of 13 day old neonatal male suffering from DORV alongwith mitral and pulmonary atresia.

KEYWORDS: DORV, Atresia of MV, Pulmonary atresia, Hypoplastic LV, Malposition of Great Arteries.

INTRODUCTION

DORV is a rare congenital heart disease where both major arteries are primarily or entirely connected to the right ventricle, constituting about 1% of all congenital heart diseases.[1-3] (Figure 1, 2).

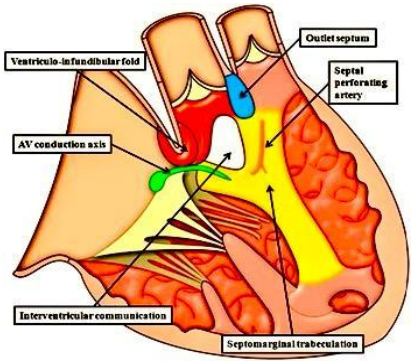


Figure 1: Diagrammatic portrayal of Double Outlet Right Ventricle.

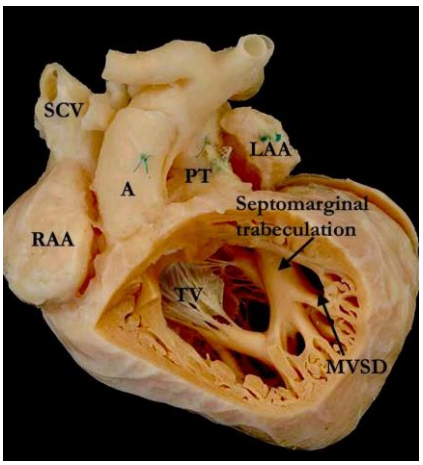


Figure 2: Pathological specimen of double outlet right ventricle with left ventricular hypoplasia. The aorta is anterior and to the right of the PT. The trabeculations within the right ventricle are prominent with a freestanding septomarginal

trabeculation. On the anterosuperior aspect of the ventricular septum and anterior to the septomarginal trabeculation, there is the opening of a MVSD. The ventricular septal defect has a posterior inferior muscular rim separating it from the TV. SCV, superior caval vein; RAA, right atrial appendage; LAA, left atrial appendage; A, aorta; PT, pulmonary trunk; MVSD, muscular ventricular septal defect; TV, tricuspid valve.

DORV was first described pathologically by Witham et al.[4] in 1957. Goor et al.[5] provided a definition of DORV in 1982, stating that it involves both great arteries arising from the morphologic right ventricle by 50% or more and the presence of an interventricular connection such as VSD or atrioventricular septal defect (AVSD).[6] The classification of DORV by Lev et al.[2], who classified DORV into four main groups as subaortic, subpulmonic, double committed and non-committed VSD types using the relationship of the VSD to the great arteries, remains the most widely used classification of DORV.

The diverse anatomical combinations of DORV can result in either decreased or increased pulmonary blood flow. Variations in pulmonary blood flow can lead to a wide range of clinical presentations, ranging from cyanosis to congestive heart failure and even pulmonary edema. In patients with subaortic or subpulmonary VSD, aggressive diuresis may be required due to pulmonary over-circulation, which can lead to the need for intubation. If pulmonary blood flow is very low, prostaglandin E1 infusion and/or atrial septostomy may be necessary. Some patients may have sufficient pulmonary blood flow and can be discharged for weight gain and growth until surgical intervention is required.[6]

Although differential diagnoses for a single cardiac outflow tract include truncus arteriosus, pulmonary atresia with VSD or intact ventricular septum, the association of DORV with pulmonary atresia is also seen rarely. When such rare anatomy is observed, obtaining information about which route supplies pulmonary blood flow, such as aortopulmonary collaterals or patent ductus arteriosus (PDA), is crucial for the management of the disease.[7]

DORV, a rare congenital heart disease with various anatomical features and consequently different treatment methods, has many subgroups. It should not be considered as a single disease, and morphology should be well defined. Detailed and unequivocal characterization of cardiac anatomy is crucial for appropriate diagnosis and surgical decision-making.

CASE REPORT

A 13 days old male neonate was referred to us for comprehensive clinical diagnosis of cyanotic congenital heart disease. The parents provided the history of cyanosis and breathlessness since birth.

The neonate was of average built with puffiness of the face and bluish discoloration of lips (Figure 3A). The characteristic intercostal retractions were present, accompanied by prominent precordial bulge (Figure 3B). All the fingers and toes demonstrated cyanosis. The neonate's weight was 3.3 kg, height was 25 cm, BP was 78/60 mmHg, HR was 134/min, respiratory rate was 84/min and SPO2 was 90% at room air. Cardiovascular examination revealed a grade 3/6 pansystolic murmur heard at the lower left sternal edge. IInd heart sound was single. Rest of the systemic examination was not significant.

(A) **(B)**

Figure 3: Photohraphs of our index patient. (A) Facies; (B) Intercostal retraction and prominent precordial bulge.

Xray chest (AP-view) revealed cardiomegaly with markedly decreased pulmonary blood flow (Figure 4).

Figure 4: Xray Chest (AP-view). Cardiomegaly with reduced pulmonary blood flow.

Resting ECG (Figure 5) displayed right ventricular hypertrophy, right axis deviation and sinus tachycardia with a ventricular rate of 150/min.

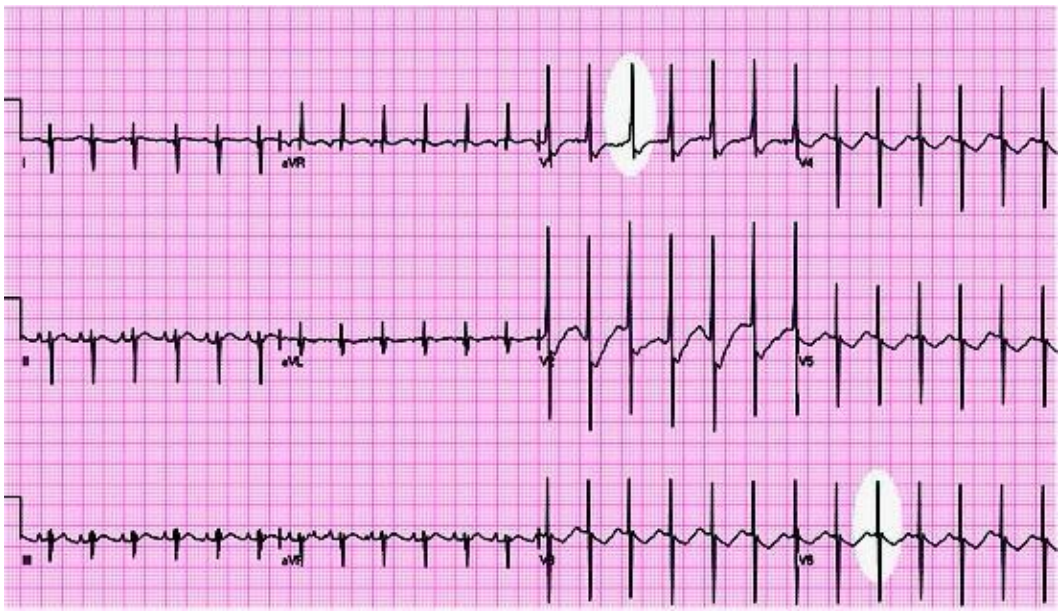


Figure 5: Resting ECG. Conspicuous right ventricular hypertrophy, right axis deviation and sinus tachycardia with a ventricular rate of 150/min was identified.

Transthoracic Echocardiography

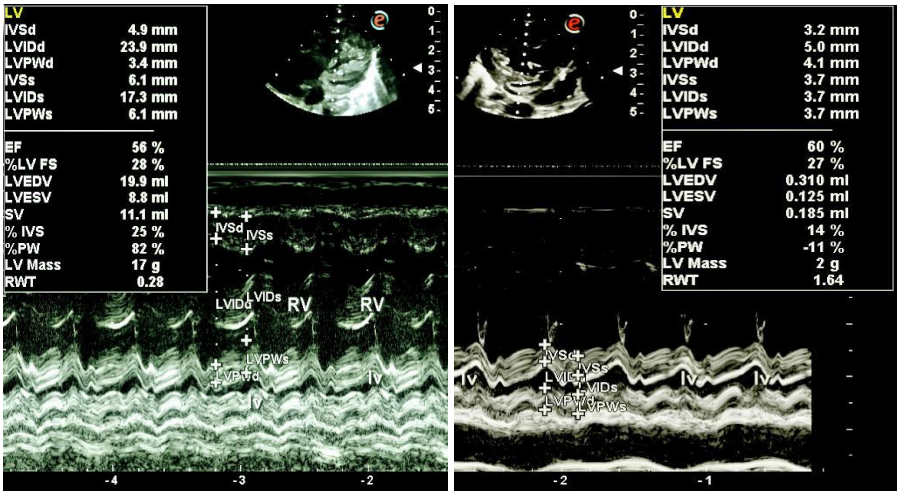
The echocardiography system - My Lab X7 4D XStrain, Esaote, Italy, was utilized for performing echocardiographic measurements and evaluations using a pediatric probe. Sequential segmental transthoracic echocardiography was performed in the classical subcostal, parasternal long axis (LX), parasternal short axis (SX), 4-Chamber (4CH), 5-Chamber (5CH) and suprasternal views.

M-mode Echocardiography

M-mode echocardiography of left and right ventricles was performed and the estimated measurements are outlined in Table 1, Figure 6.

Table 1: Calculations of M-mode echocardiography.

Measurements	RV	LV
IVS d	4.9 mm	3.2 mm
ID d	23.9 mm	5.0 mm
PW d	3.4 mm	4.1 mm
IVS s	6.1 mm	3.7 mm
ID s	17.3 mm	3.7 mm
PW s	6.1 mm	3.7 mm
EF	56 %	60 %
% FS	28 %	27 %
EDV	19.9 ml	0.310 ml
ESV	8.8 ml	0.125 ml
SV	11.1 ml	0.185 ml
Mass	17 g	2 g
IVS, interventricular septum, ID, internal dimension; PW, posterior wall, d, diastole; s, systole; FS, fractional shortening; EDV, end-diastolic volume; ESV, end systolic volume; SV, stroke volume; EF, ejection fraction.		



(A) **(B)**

Figure 6: M-mode Echocardiography. (A) RV volumetric estimation; (B) LV volumetric estimation.

Summary of M-mode echocardiography

M-mode echocardiography demonstrated massively dilated RV with rudimentary, hypoplastic LV. Biventricular ejection fraction was normal. RVEF %, LVEF %, RV mass and LV mass were 56 %, 60 %, 17 gm and 2 gm, respectively.

2-Dimensional-Transthoracic Echocardiography

Transthoracic echocardiography (TTE) was systemically performed by the sequential segmental approach (SSA) and the echocardiographic characteristics which were documented are enumerated below:

- Levocardia
- Situs solitus (Figure 7)
- Concordant d-bulboventricular loop
- D-Loop ventricles (Figure 8)
- Left aortic arch (Figure 9)
- Confluent pulmonary arteries
- Double outlet right ventricle (Figure 10)
  - Both the great arteries are arising from the anatomic RV
- Malposition of great arteries (Figure 11)
  - AO and PA are lying side by side
  - AO is on the right of PA
  - PA is in the left of AO
- Atresia of pulmonary valve and pulmonary valve annulus (Figure 12, 13)
  - PV annulus (D) 4.3mm, MPA (D) : 5.3 mm, LPA (D) : 2.7 mm, RPA (D) 4.4 mm.
- Patent ductus arteriosus (moderate) (Figure 14)
  - Size 3.1 mm.
  - Lt. to Rt. Shunt.
- Atresia of the mitral valve (Figure 15)
- Atrial septal defect (large) (Figure 16)
  - Size 7.7 mm
  - Ostium primum type
  - Lt. to Rt. Shunt.
  - Peak/mean gradient across ASD was 3.2/08 mmHg
- Tricuspid regurgitation (moderate) (Figure 17)
  - TV large & thickened
  - TR velocity = 4.11 m/sec (gradient 68 mmHg).
- Massively dilated RV, diminutive LV (Figure 18)
  - Normal biventricular systolic function.
  - Normal LVEF: 60 % & RVEF: 56 %
- No evidence of VSD, COA, AS.

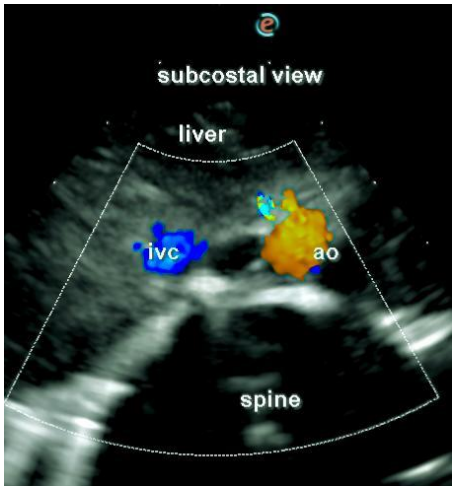


Figure 7: Situs solitus. In the subcostal view there is left sided aorta (ao) and right sided inferior vena cava (ivc).

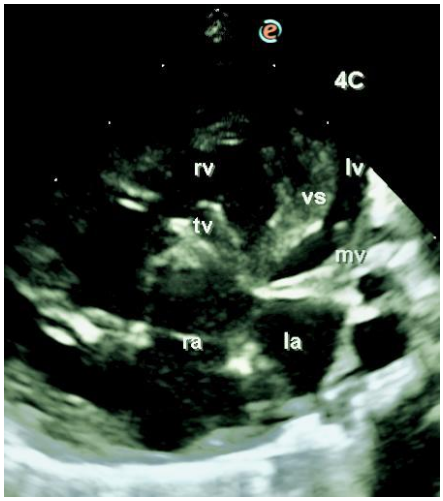


Figure 8: D-loop ventricles. In the apical four chamber view the dilated RV is to the right of diminutive LV, and LV is to the left of RV. la, left atrium; ra, right atrium; mv, mitral valve; tv, tricuspid valve; rv, right ventricle; lv, left ventricle; vs, ventricular septum.

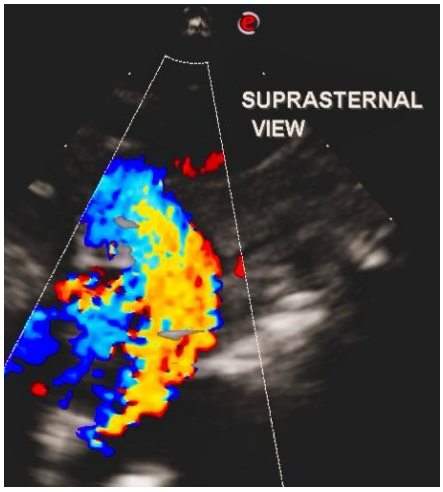


Figure 9: Left aortic arch. In the suprasternal view left aortic arch is delineated.

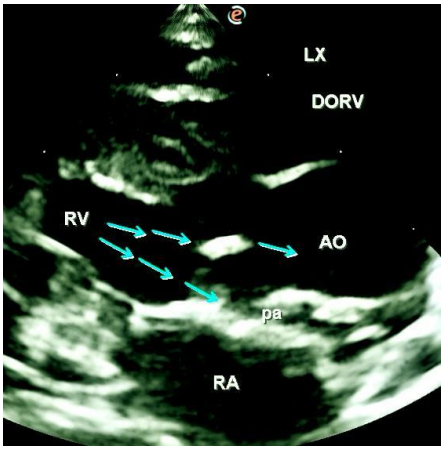


Figure 10: Double outlet right ventricle. In the parasternal long axis view, both the great arteries are arising from anatomical RV. RV, right ventricle; AO, aorta; pa, pulmonary artery; RA, right atrium.

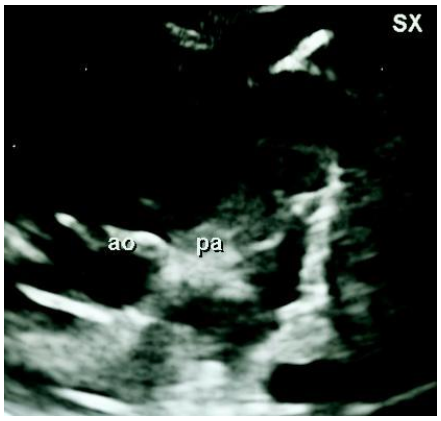


Figure 11: Malposition of great arteries, In the short axis view, both the great arteries are lying side by side. Aorta is to the right of pulmonary artery and pulmonary artery is to the left of aorta. ao, aorta; pa, pulmonary artery.

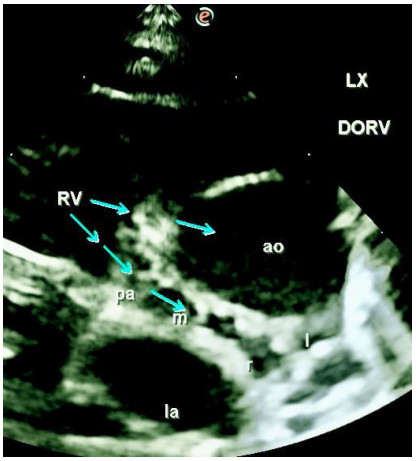


Figure 12: In the LX view, DORV is distinctly demonstrated accompanied with atresia of pulmonary valve and annulus alongwith hypoplasia of main, left and right pulmonary artery. RV, right ventricle; ao, aorta; pa, pulmonary artery; m, main pulmonary artery; L, left pulmonary artery; r, right pulmonary artery; la; left atrium.

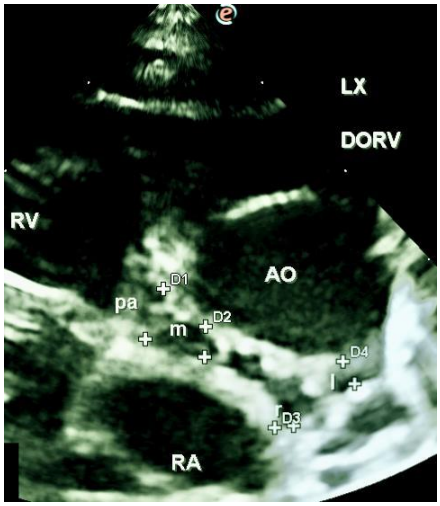
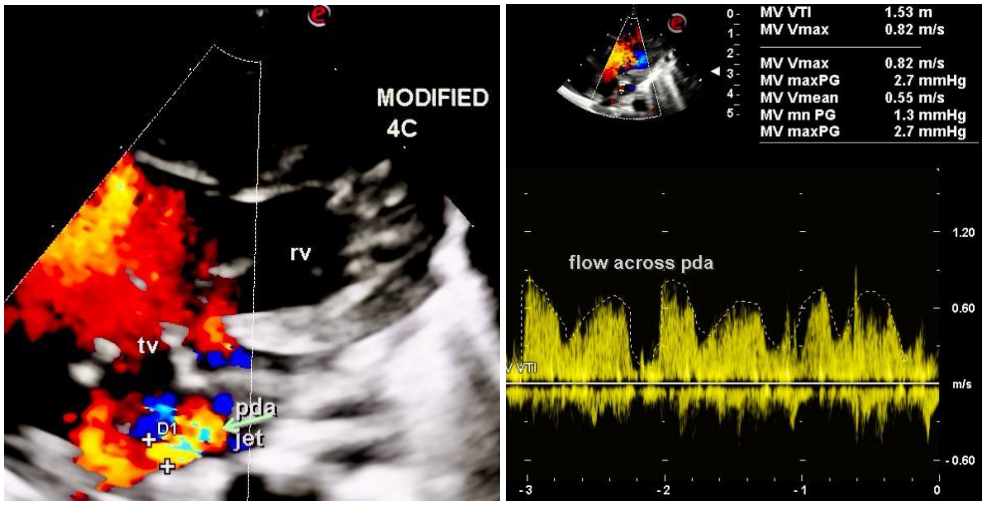


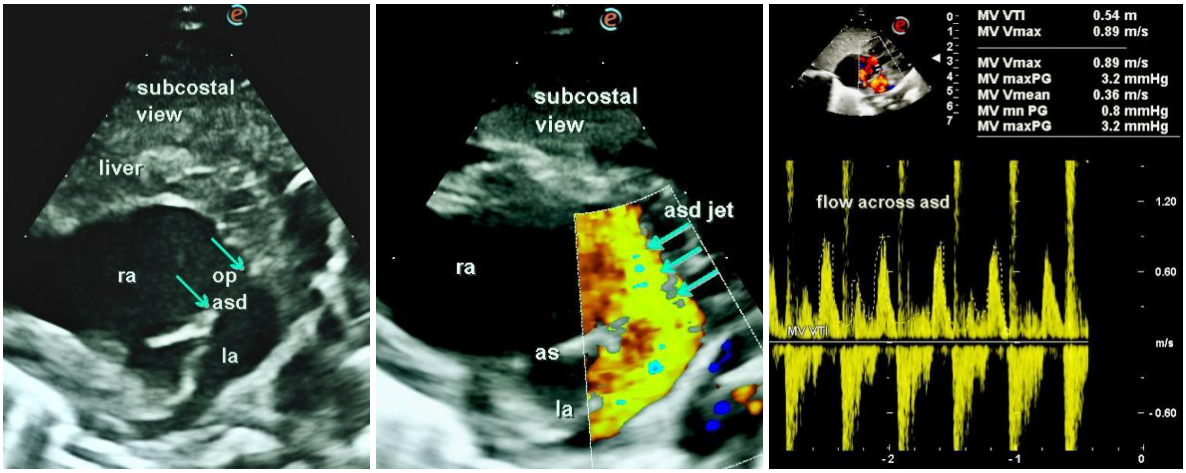
Figure 13: Dimensions of atretic pulmonary artery. pv annulus (D) 4.4 mm, mpa (D) 2.5 mm, lpa (D) 1.6 mm, rpa (D) 2.1 mm.



(A) **(B)**

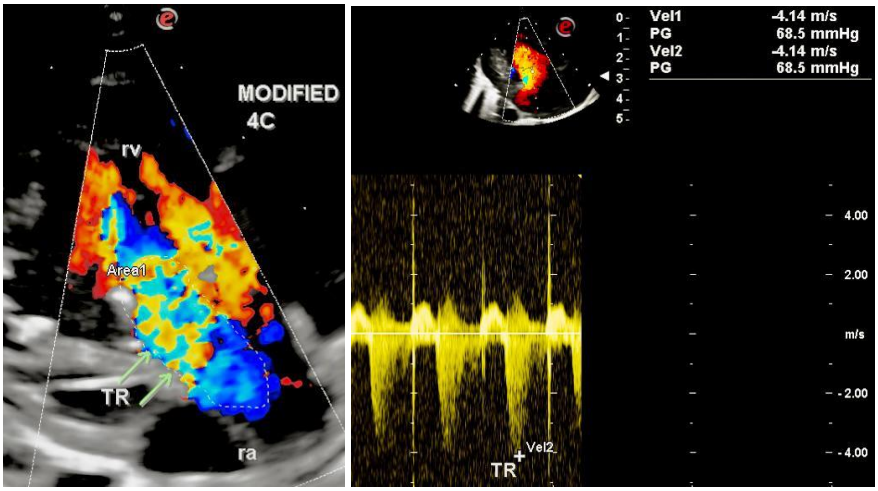
Figure 14: (A) Patent ductus arteriosus attaching to right pulmonary artery. pda, patent ductus arteriosus; TV, tricuspid valve; RV, right ventricle. (B) On continuous wave doppler analysis, peak and mean gradient across pda was 2.7/1.3 mmhg.

Figure 15: Atresia of mitral valve. In the 4C view Atresia of MV is delineated (**). Also visualized , are diminutive LV and small LA. RV, right ventricle; vs, ventricular septum; lv, left ventricle; TV, tricuspid valve ; ra, right atrium.



(A) (B) (C)

Figure 16: Ostium primum type, large atrial septal defect demonstrated. (A) Subcostal view shows ostium primum ASD of size 7.7 mm, with (B) left to right shunt, and (C) peak/mean gradient across ASD was 3.2/08 mmHg. op, ostium primum; ASD, atrial septal defect; as, atrial septum; la, left atrium; ra, right atrium.



(A) (B)

Figure 17: Tricuspid regurgitation. In the modified 4C view on color flow mapping a moderate tricuspid regurgitation jet was recognized. The jet area was 1.40 sqcm. On continuous flow doppler analysis tricuspid regurgitation jet velocity was 4.11 m/sec (peak gradient 68 mmHg). TR, tricuspid regurgitation; RV, right ventricle; ra, right atrium.

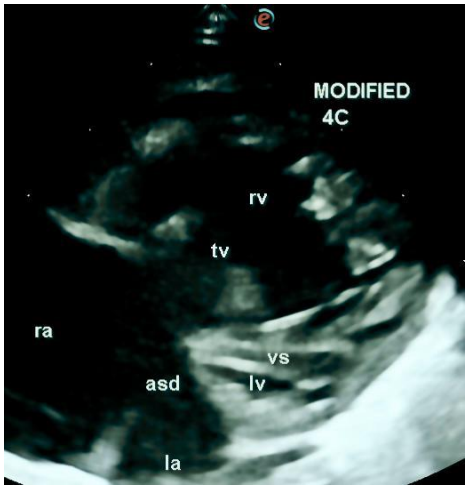


Figure 18: Massively dilated right ventricle with diminutive left ventricle is demonstrated in modified 4C view. There was normal biventricular systolic function. la, left atrium; lv, left ventricle; ra,right atrium; rv, right ventricle; tv, tricuspid valve; asd, atrial septal defect; vs, ventricle septum.

Summary of Transthoracic color Doppler Echocardiography

On Transthoracic echocardiography our index patient exhibited features of DORV, malposition of great arteries, pulmonary atresia, moderate sized PDA, atresia of the mitral valve, large ostium primum ASD with left to right shunt, massively dilated RV, rudimentary LV with normal biventricular systolic function.

DISCUSSION

Double outlet right ventricle (DORV) is extremely rare and occurs in approximately 3-9/1,00,000 live births. Conservative estimates project DORV accounting for about 1-3 % of all congenital heart defects.[8] DORV was first described pathologically in 1957.[4] This pathoanatomical constellation was initially referred to as partial transposition of great arteries (TGA) with only the aorta transposed. Goor et al. gave the definition of DORV as the cardiac condition wherein both great arteries arise from the morphologic right ventricle (RV). Each great artery should have 50% or greater of the valve diameter coming from the morphologic RV, and there is almost always an interventricular communication: ventricular septal defect (VSD) or atrioventricular septal defect (AVSD).[5] However, in rare cases, interventricular communication may be absent. In addition, DORV is frequently associated with an assortment of other cardiac malformations. The consensus definition by the International Society for Nomenclature of Pediatric and Congenital Heart Disease (ISNPCHD) ultimately settled on "the hearts with both arterial trunks supported predominantly by underlying morphologic RV" where both great arteries arise entirely or predominantly from the morphological RV.[9]

In 1949, Taussig and Bing described this entity as a "complete" transposition and left position of the aorta, with subpulmonic VSD[10] (i.e., Taussig-Bing anomaly). Later in 1957, Witham classified DORV into three groups: those with pulmonary stenosis (PS), those without PS, and those with an AVSD as well as other malformations.[4] However, only after 1972, Maurice Lev emphasized the importance of the position of the VSD, the great arteries, and the relationship between each other. This is the predominant classification that is most frequently used to classify this congenital anomaly in the modern era.[2]

Classification

The most frequently used classification of DORV is based on the location of the VSD and the position of great arteries[2], the relationship of great arteries to each other, and the presence or absence of outflow tract obstruction (Table 2).

A subpulmonary VSD in utero could lead to less flow along the LV and left ventricular outflow (LVOT) leading to subaortic/aortic stenosis, CoA and IAA. A subaortic VSD may mean that pulmonary stenosis and small branch pulmonary arteries are likely to develop. Not surprisingly, outflow tract obstruction can be seen, either left ventricular outflow tract obstruction (LVOTO) or right ventricular outflow tract obstruction (RVOTO). Some other common associations include AV valve anomaly: straddling vs. atresia, as well as common AV valves can be found with DORV. Coronary abnormalities may be similar to those seen in TOF and TGA, as well as the associated abnormalities with heterotaxy.

Table 2: DORV categories based on VSD location and the relationship with great arteries.[2]

Subaortic VSD	
Subpulmonary VSD	
Doubly committed VSD	
Non-committed VSD/remote VSD	
DORV categories based on great vessel relationship	
Right anterior aorta	Left anterior aorta
Right posterior aorta	Left posterior aorta
Right lateral/side-by-side aorta	
DORV categories by Van Praagh (9)	
Type I DORV as an isolated conotruncal anomaly	
Type II DORV with conotruncal anomalies and associated malformations of the atrioventricular valve(s) and ventricles	
Type III DORV associated with heterotaxy	
Functional categories by congenital heart surgery nomenclature and database project (3)	
DORV, VSD type (DORV-VSD)	
DORV with subaortic or doubly committed YSD and pulmonary stenosis, Fallot type (DORV-Fallot)	
DORV, with subpulmonary VSD, transposition type (DORV-TGA)	
DORV, non-committed VSD (nc-VSD)	

Cardiac morphology

In anatomically normal hearts, D-looping of ventricles brings the left ventricle (LV) to the left with the aorta moving right and posterior while the right ventricle (RV) moves to the right with the pulmonary artery (PA) moving anterior and leftward. A muscular tissue called subpulmonary conus follows beneath. Normally, the subaortic conus resorbs and brings the aorta in continuity

with the mitral valve (MV) or whichever atrioventricular (AV) valve is in that position. In DORV, the subaortic conus frequently persists. Furthermore, flaws in conotruncal development occur and there is a failure to achieve appropriate conotruncal maturation (inversion/rotation/looping).[2,4,5,9-13]

DORV - associations

A wide variety of cardiac malformations of the heart may accompany DORV including:

- Coarctation of the aorta (CoA).[14,15]
- Severe mitral valve malformations like mitral atresia.[14,15]
- Laterality defects such as the heterotaxy syndromes of isomerism of the atrial appendages.[14,15]
- Aortic and pulmonary stenosis.[16]
- Interrupted aortic arch (IAA) and aortic arch obstruction like hypoplasia or atresia of part of the aortic arch.[17,18]
- Bicuspid aortic valve.[19,20]
- Double inlet-double outlet right ventricle[21]
- Straddling tricuspid valve (TV).[22]

Survival of patients with DORV and mitral atresia (hypoplastic left heart syndrome) is rare.[22] In presence of mitral atresia, the only way DORV patients can survive long is by a univentricular pathophysiology and obligatory left-to-right shunting through an atrial septal defect as in the case reported here. Mitral valve anomalies may occur in DORV, including mitral stenosis and atresia with associated underdevelopment of the left ventricle.[23,24] Most cases of DORV with intact ventricular septum as in this case involve severe abnormalities of the mitral valve. In addition, cleft, double orifice, parachute, as well as straddling mitral valve can be associated.[23,25]

In the case reported, there was a physiologically univentricular atrioventricular connection to the dominant right ventricle via the tricuspid valve. Although pulmonary veins drained normally into the left atrium, there was an obligatory left-to-right shunting through the atrial septal defect. The mitral valve was atretic and there was a diminutive (hypoplastic) LV. Mitral atresia associated with an intact ventricular septum forms part of hypoplastic left heart syndrome and survival is possible only by a DORV connection and obligatory left-to-right shunt. Mitral atresia can be associated with chromosomal anomalies such as XO, trisomy 18 or 13.[23,24]

CONCLUSION

Double outlet right ventricle with its various variants is a rare congenital cardiac anomaly which could be fatal if not managed appropriately. Surgical correction offers good and long term benefits if the patient presents early and timely referral is warranted so as to avert death. The case report is important due to the rarity of this anatomical type of DORV.

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.

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