Double Outlet Right Ventricle

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Abstract

Double outlet right ventricle (DORV) is characterized by an abnormal connection between both great arteries and the right ventricle, leading to poor systemic oxygenation. The diagnostic criteria for this condition vary, as do the various morphologies that exist. Echocardiography is the primary imaging tool, requiring views from multiple planes to elucidate the specific morphology of DORV in each patient, along with any associated cardiac anomalies. Incorporating other imaging modalities such as CT to maximize diagnostic accuracy is essential in determining the proper surgical approach, as the survival rate for children who undergo surgical repair remains high.

Keywords: cardiothoracic, cardiac, congenital

Case Summary

A 6-hour-old female presented with cyanosis and tachypnea with oxygen saturations in the 70s despite supplemental oxygen. On physical exam, the neonate was tachycardic with a hyperdynamic precordium, a soft systolic murmur, mild hepatomegaly, and decreased femoral pulses with delayed capillary refill. The transthoracic echocardiogram confirmed the diagnosis, and a chest CTA was performed for surgical planning.

Imaging Findings

Chest CTA with contrast (Figures 1, 2) showed a double outlet right ventricle (DORV) with malposition of the great arteries, subpulmonic ventricular septal defect (VSD), and aortic arch hypoplasia. This constellation of findings is often referred to as Taussig-Bing anomaly. The patient had an arterial switch operation, aortic arch augmentation, and VSD closure at 1 week of life. A repeat chest CTA (Figures 3, 4) at 2 years of age showed repair of the aortic arch with left pulmonary artery stenosis.

Diagnosis

Double outlet right ventricle.

As DORV can present similarly to and occur in conjunction with other congenital heart anomalies, the differential diagnoses include Tetralogy of Fallot, transposition of the great arteries and VSD.

Discussion

DORV is a congenital heart defect in which both great arteries (the aorta and the pulmonary artery) arise predominantly or entirely from the right ventricle instead of one from each ventricle. This can lead to symptoms of poor feeding, fatigue, clubbing, and cyanosis.[1] During normal fetal development, the heart's outflow tract initially originates from the right ventricle. The common outflow tract later divides and spirals to form the aorta and pulmonary artery, aligning one with the left ventricle and the other with the right ventricle. Endocardial cushions facilitate the development of the semilunar valves and the conal septum that separates the ventricular outflow tracks—an abnormality in this process can lead to DORV. Epidemiologic studies indicate that DORV occurs in anywhere between 3 and 24/100,000 live births, accounting for approximately 1-3% of all congenital heart defects.[1] Studies have been conducted to determine any genetic basis for the disease, with common associations seen with Trisomy 18, Trisomy 13, chromosome 8 abnormalities, and less commonly, in patients with 22q11 deletions. Associations with mutations in the *CFC1* and *CRX* genes have also been found.[1-3]

The guidelines for what level of right ventricular outflow is considered DORV versus what constitutes another congenital heart anomaly are debated. The 50% rule (also commonly referred to as the 150% rule) is often used to delineate DORV. The 50% rule states that a great artery is considered to arise from the right ventricle when over 50% of its circumference is connected to it.[1] However, measuring the level of association between the aorta and the right ventricle is impractical due to the lack of a definitive boundary and

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Figure 1. (A) Chest CTA with contrast in an axial plane at the ventricular mass. There is a large VSD in a patient with double outlet right ventricle (DORV). DA, descending aorta; LA, left atrium; LV, left ventricle; MV, mitral valve; RA, right atrium; RV, right ventricle; TV, tricuspid valve; VSD, ventricular septal defect. (B) Chest CTA with contrast in an axial plane at the pulmonary valve. The VSD is subpulmonic. DA, descending aorta; LA, left atrium; LAA, left atrial appendage; LUPV, left upper pulmonary vein; PV, pulmonary valve; RA, right atrium; RV, right ventricle; RUPV, right upper pulmonary vein. (C) Chest CTA with contrast in an axial plane at the aortic valve. The aortic valve arises from the right ventricle consistent with DORV. The main pulmonary artery is posterior to the aortic valve. AV, aortic valve; DA, descending aorta; LPA, left pulmonary artery; MPA, main pulmonary artery; RPA, right pulmonary artery; RV, right ventricle; RUPV, right upper pulmonary vein.

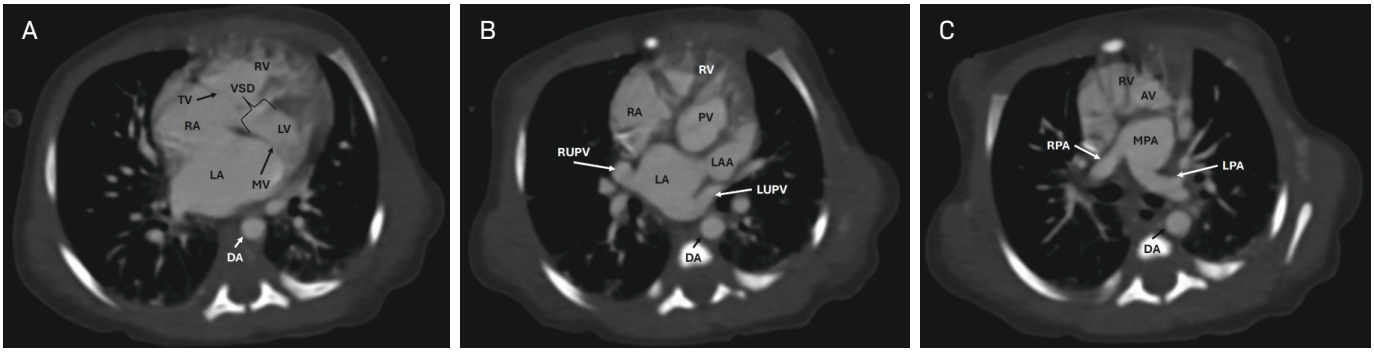


Figure 2. 3D chest CTA with contrast in a sagittal projection. The aorta is superior to the main pulmonary artery. The aortic arch is hypoplastic with coarctation. AA, ascending aorta; Coarct, coarctation; DA, descending aorta; Ductus, ductus arteriosus; IA, innominate artery; LCCA, left common carotid artery; LSA, left subclavian artery.

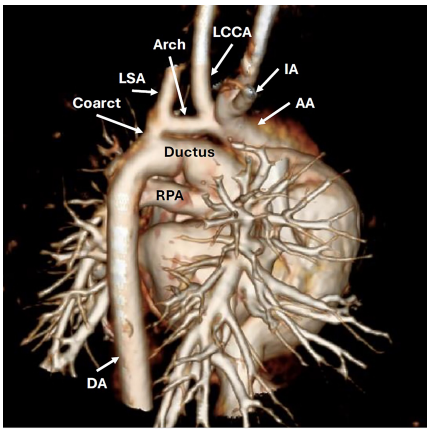


Figure 3. Chest CTA with contrast in an axial plane at the main pulmonary artery. The right pulmonary artery is anterior to the ascending aorta consistent with the arterial switch operation. AA, ascending aorta; DA, descending aorta; MPA, main pulmonary artery; RPA, right pulmonary artery; SVC, superior vena cava.

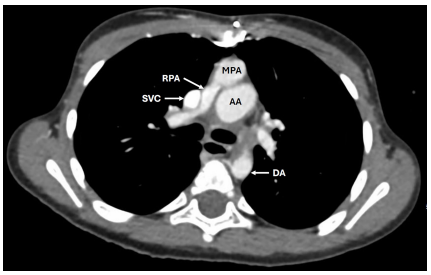
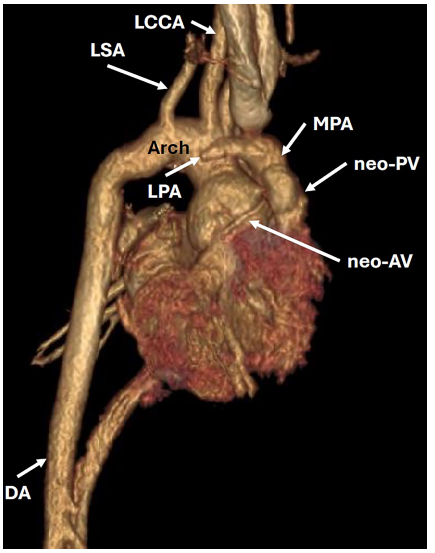


Figure 4. 3D chest CTA with contrast in a sagittal projection. The left pulmonary artery is anterior to the aorta and is stenotic. The aortic arch is normal in size. There is neo-aortic valve dilation. DA, descending aorta; LCCA, left common carotid artery; LPA, left pulmonary artery; LSA, left subclavian artery; MPA, main pulmonary artery; neo-AV, neo-aortic valve; neo-PV, neo-pulmonary valve.



the difficulty in quantifying the three-dimensional nature of this junction using standard imaging.[4] DORV can occur with varying physiologies as the great arteries can be in different positions in relationship to the VSD. VSD is found in almost 100% of cases of DORV, with atrial septal defects occurring in approximately 10-30% of cases.[4,5] When DORV has a subaortic VSD, the physiology is like Tetralogy of Fallot, and when there is a subpulmonic VSD, the physiology is like transposition of the great arteries. DORV with a subpulmonary VSD is often associated with aortic arch hypoplasia and called Taussig-Bing anomaly.

DORV can present as an isolated defect or in combination with other congenital anomalies; recognizing these variations is essential for guiding surgical management.[5] Commonly, the right ventricle will have an infundibulum that supports both the aortic and pulmonary valves. In some cases, both a subaortic and subpulmonary infundibulum are present, which may coexist with an overriding aorta or, less commonly, an overriding pulmonary artery. While VSDs are present in nearly all cases of DORV, bilateral infundibula occur in only about 10-15% of cases, making their value as defining diagnostic features controversial.[6,7]

Many different imaging techniques can be used to diagnose DORV by determining whether both the pulmonary artery and the aorta primarily connect to the right ventricle. Echocardiography is most frequently used as an initial screening tool to differentiate the ventriculo-infundibular fold from the conal septum using views from different planes.[8] Commonly, the parasternal long-axis imaging plane is used to identify a lack of communication between the mitral valve and the aorta.[8] Cardiac catheterization can also be

performed but has largely been phased out due to its invasiveness and associated risks. MRI has also been used, with CT emerging more recently with improved spatial resolution and reduced acquisition times.[4] These imaging modalities are used to guide surgical strategies by uncovering the orientation of the great arteries, the conus, the presence and location of a VSD, and other associated cardiac anomalies.[9]

Treatment for DORV depends largely on the specific morphology, as a specific surgical approach is indicated depending upon the subtype of DORV. Surgical approaches are almost always biventricular, but univentricular repairs are sometimes required.[5] The most common approach used is the intraventricular tunnel (baffle) repair, where a patch is placed within the right ventricle to direct left ventricular outflow through the VSD to the aorta, thereby establishing physiological separation of the systemic and pulmonary circulations.[6] If the communication between the ventricles is subpulmonary, intraventricular baffling to the pulmonary trunk is performed, and an arterial switch operation is performed (moving the pulmonary artery anterior to the native aortic valve and moving the aorta posterior to the native pulmonary valve). The presence of other cardiac anomalies, such as pulmonary stenosis or aortic arch hypoplasia, may also impact the surgical approach and warrant further intervention. In cases where the VSD is noncommitted to the left ventricle or is subpulmonary and exhibits pulmonary stenosis, a double-root translocation procedure involving the relocation of both the pulmonary and aortic roots to restore normal directional flow is pursued.[6,10]

The most common complication that can occur is outflow obstruction to either of the ventricles, occurring in 3-30% of cases. Other postoperative complications include residual or recurrent VSDs, arrhythmias, and right ventricular dysfunction.[1] The long-term prognosis after successful surgical repair is generally favorable, with survival rates estimated between 80% and 95%.[2] Mortality is most attributed to progressive ventricular failure, severe arrhythmias, or obstruction of reconstructed outflow tracts, necessitating lifelong cardiac surveillance in these patients.[10]

Conclusion

DORV is characterized by an abnormal connection between both great arteries and the right ventricle leading to poor systemic oxygenation. The diagnostic criteria for this condition vary, as do the various morphologies that exist. Echocardiography is the primary imaging tool, requiring views from multiple planes to elucidate the specific morphology of DORV in each patient, along with any associated cardiac anomalies. Incorporating other imaging modalities such as CT to maximize diagnostic accuracy is essential in determining the proper surgical approach, as the survival rate for children who undergo surgical repair remains high.

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