

Double outlet right ventricle, atrioventricular canal defect with severe pulmonary artery stenosis: a rare cyanotic congenital heart disease

Shoukat Ali Baig,¹ Zara Saeed,² Arsala Fatima,³ Hasina Chagani,⁴ Sina Aziz⁵

Abstract

Double outlet right ventricle (DORV) is characterised by arousal of the aorta and the pulmonary vessel from the right ventricle, and is always accompanied by a ventricular septal defect (VSD). Our patient, a twenty days-old female child, presented to the Paediatrics Unit I of Civil Hospital, Karachi, with complaints of generalised cyanosis and reluctance to feed since birth. Apart from the fact that the neonate was cyanosed with increased respiration, the physical examination was unremarkable. Two-dimensional echocardiography revealed two atrial septal defects (ASD), a ventricular septal defect (VSD) in the membranous area, continuous with primum ASD, resulting in an atrioventricular canal defect. The aorta was dilated with the pulmonary artery stenosis. The left ventricle was rudimentary, and both of the great vessels were arising from the dominant right ventricle. Despite the rarity, DORV with complex anatomy should be considered among the probable differential diagnoses for infants presenting with generalised cyanosis since birth.

Keywords: Double outlet right ventricle, Cyanotic heart disease, Atrioventricular canal defect, Pulmonary stenosis.

Introduction

Double outlet right ventricle (DORV), being a rare entity, affects 1-3% of the infant population born with congenital heart disease.¹ Characterised by arousal of the aorta and the pulmonary artery from the right chamber of the heart, DORV is often associated with other septal or valvular defects, with ventricular septal defect (VSD) being the most common.² Congenital cardiac abnormality appears during the first eight weeks of gestation. A defective gene, or chromosomal abnormalities, are thought to be the proposed mechanism of pathogenesis, and are reported in up to 40% of the cases.¹ The rate of DORV was found to

be high among those infants whose mothers were placed on anti-depressants, like the selective serotonin reuptake inhibitors (SSRI), during their period of gestation.³ Classification of DORV is based on the position of VSD, with sub-aortic, sub-pulmonary, doubly-committed, and non-committed as the possible types. The severity of symptoms including tachycardia, tachypnoea, cyanosis, sweating and failure to gain weight⁴ are based on the type of DORV present, with any other accompanying defect. As the standard treatment is surgery at an early age, prompt recognition of the condition is essential for a favourable outcome.

Case Report

A twenty days-old, full-term, female infant, was admitted through the out-patient department (OPD), in the Paediatrics Unit I of Civil Hospital, Karachi, in the month of December 2012, with complaints of generalised cyanosis, reluctance to feed since birth, and continuous fever for the last three days, with temperature rising up to 102°F. Though the fever subsided for 6-8 hours with the help of an anti-pyretic, it recurred shortly after that. According to her mother, the baby was born in a cyanosed state, and was kept on a ventilator for one day.

An attempt to breast-feed the infant was accompanied with cyanotic spells and crying episodes, followed by reluctance to feed. The mother had a complete vaccinations' history, and no infections such as rubella or toxoplasmosis were reported during her pregnancy. There was no history of anti-depressants' use, nor was there any family history of any congenital cardiac anomaly.

On examination, the weight of the child was 2.8kg. She was very active, afebrile, moderately cyanosed, and well-hydrated, with no obvious abnormality. Her heart rate was 142 beats per minute, with a good volume pulse and no radio-radial or radio-femoral delay. The respiratory rate was 34 breaths per minute, and her blood pressure was 66/40mmHg.

Her cardiovascular system examination revealed the apex beat to be at the 4th left intercostal space, with tapping character. There was an ejection systolic murmur of grade

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¹⁻³Final Year Students, Dow Medical College, Dow University of Health Sciences,

⁴Department of Paediatric Cardiology, Dow University of Health Sciences, Civil Hospital Karachi, ⁵Department of Paediatrics, Karachi Medical and Dental College, Abbasi Shaheed Hospital, Karachi, Pakistan.

Correspondence: Shoukat Ali Baig. Email: dr.shoukatali@hotmail.com

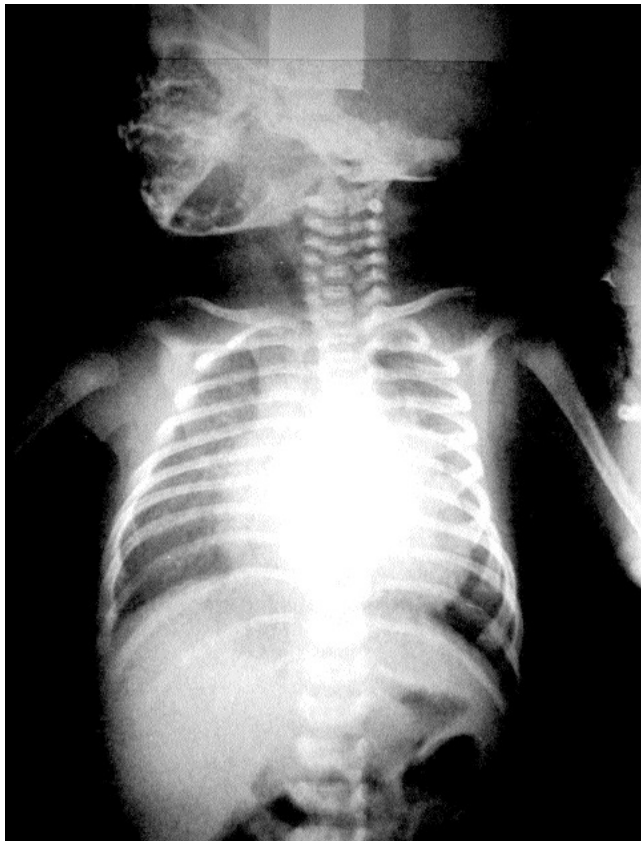


Figure-1: Chest radiograph of a 20-day old infant showing prominent cardiomegaly.

3/6 at upper left sternal border. Rest of the systemic examination was unremarkable.

The laboratory investigations revealed a haemoglobin level of 16mg/dL, while the other parameters were within normal limits. Chest x-ray revealed prominent cardiomegaly, as visible in Figure-1, with reduced vascular marking. Electrocardiography (ECG) was in favour of right axis deviation and right ventricular prominence. For further evaluation, a two-dimensional echocardiography was done, which revealed two atrial septal defects (ASDs) (9.4mm and 7mm in the secundum and primum region, respectively), and a VSD in the membranous area, continuous with primum ASD, resulting in an atrioventricular (AV) canal defect (Figure 2). The left ventricle was rudimentary, and both great vessels were arising from the dominant right ventricle. The aorta was dilated, and the pulmonary artery had stenosis, with a pressure gradient (PG) of 99mmHg. The tricuspid valve was regurgitant, with a PG of 54mmHg. There was no evidence of patent ductus arteriosus, coarctation, or endocarditis. The right ventricular systolic and diastolic function was excellent.

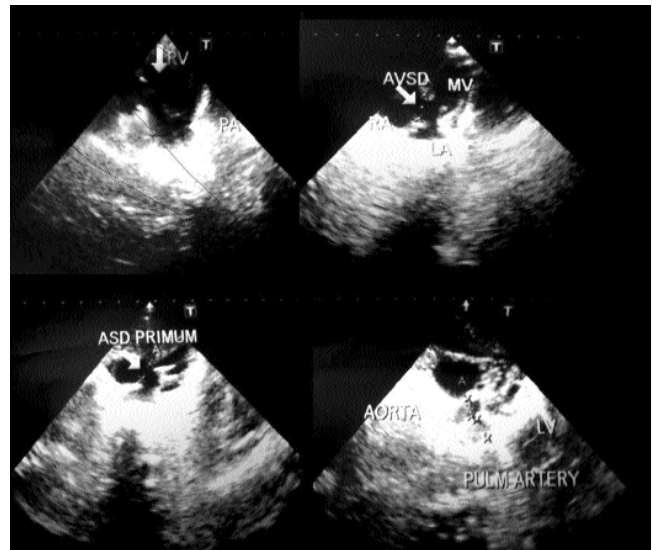


Figure-2: Echocardiographic findings of the infant.

In the medical setting of inadequate blood pulmonary blood flow, preserving ductal blood flow, that is maintaining patent ductus arteriosus, is vital. An infusion of prostaglandin E (that is, alprostadil) is the standard of care until repair can take place. The location of the VSD and its size are critical to the surgical repair. Repair of the double outlet right ventricle with pulmonary stenosis can be done by Blalock-Taussig anastomosis, Brock valvotomy, or Potts and a Blalock-Taussig shunt. Unfortunately, the baby had passed away before any surgical intervention could be carried out.

Discussion

Conotruncal abnormalities constitute the majority of defects in infants presenting with symptomatic cyanotic congenital heart disease during their first year of life.⁵ Being a complex conotruncal abnormality with substantial anatomic variations, DORV has been variably defined by different authors.⁶ DORV has been listed as a congenital cyanotic heart disease with an admixture of physiology (a cardiac defect which facilitates complete mixing of the deoxygenated systemic venous (SV) blood returning from the tissues, and the fully oxygenated pulmonary venous blood from the lungs, in a common receiving chamber⁷), and the degree of cyanosis depends upon the type of DORV present, being pronounced in the presence of pulmonary artery stenosis, as was seen in our case.

Double outlet right ventricle with complete AV canal is uncommon, and the VSD in this instance is usually subaortic.⁸ Our case shows a DORV with a complete AV canal defect, a non-committed type of VSD, accompanied

by pulmonary stenosis, and tricuspid regurgitation. The term non-committed VSD was introduced to define the location of the VSD in cases with DORV in which the VSD was distant from both the arterial valves. This subset includes DORV with an AV canal type, inlet (muscular), or trabecular VSD.⁹

The presentation of DORV with pulmonary artery stenosis seems to be similar to the clinical picture of the classical Tetralogy of Fallot (TOF), and is often a cause of misdiagnosis. Cyanosis is early in onset, progresses during the early months of life, and may be accompanied by hypoxic spells.¹⁰ Congenital cyanotic heart diseases considerably reduce the quality of life, as the babies get exhausted frequently on minimum efforts; this leads to poor feeding, poor weight gain, and eventually, failure to thrive. The lack of interest in feeding makes the baby lethargic, unresponsive, and further decreases the quality of life and development. The symptoms of tachypnoea and tachycardia worsen on slightest exertion.

For making the diagnosis in neonates and determining the treatment, studies reveal that it is significant to assess the following: the relationship of the great arteries (normal, side-by-side or transposed); the position of the VSD; the presence of pulmonary or sub-pulmonary valvular stenosis; and the absence of mitral-aortic or mitral-pulmonary valve continuity.¹¹ Findings vary greatly in the cases of DORV, and the management and treatment of different cases are tailored according to the specific requirements.

Echocardiography appears to be a promising tool to nail the diagnosis of this rare condition, which requires high quality 2D imaging, with high frequency transducer, for infants.¹⁰ Three-dimensional echocardiography has now emerged as a way to improve cardiac imaging, and to aid in the diagnosis of this complex cyanotic congenital heart disease.¹² The treatment often involves one or more surgical interventions conducted early in life.

Conclusion

Despite the rarity, DORV should be considered in the differential diagnoses of patients presenting with generalised cyanosis since birth. Early recognition and prompt management may improve outcomes in such patients.

References

1. Obler D, Juraszek AL, Smoot LB, Natowicz MR. Double outlet right ventricle: aetiologies and associations. *J Med Genet* 2008; 45: 481-97.
2. Double outlet right ventricle. (Online) (Cited 2013 March 10). Available from URL: <http://www.nlm.nih.gov/medlineplus/ency/article/007328.htm>.
3. Malm H, Artama M, Gissler M, Ritvanen A. Selective serotonin reuptake inhibitors and risk for major congenital anomalies. *Obstet Gynecol* 2011; 118: 111-20.
4. Double outlet right ventricle. (Online) (Cited 2013 March 12). Available from URL: <http://www.childrenshospital.org/az/Site1940/mainpageS1940P4.html>.
5. Graham TP, Gutgesell HP. Conotruncal abnormalities. In: Long WA, ed. *Fetal and Neonatal Cardiology*. Philadelphia: WB Saunders; 1990; 561-70.
6. Hagler DJ. Double-Outlet Right Ventricle and Double-Outlet Left Ventricle. In Moss and Adams' *Heart Disease in Infants, Children, and Adolescents: Including the Fetus and Young Adult*. 7th ed. Edited by Hugh D. Allen [et al]. Philadelphia: Lippincott Williams & Wilkins; 2008; pp 1100-20.
7. Tharakan JA. Admixture lesions in congenital heart disease. *Ann Pediatr Cardiol* 2011; 4: 53-9.
8. Bharati S, Kirklin JW, McAllister HA, Lev M. The surgical anatomy of common atrioventricular orifice associated with tetralogy of Fallot, double outlet right ventricle and complete regular transposition. *Circulation* 1980; 61: 1142-9.
9. Lev M, Bharati S, Meng CCL, Liberthson RR, Paul MH, Idriss F. A concept of double-outlet right ventricle. *J Thorac Cardiovasc Surg* 1972; 64: 271-80.
10. Double outlet right ventricle. (Online) (Cited 2013 March 12). Available from URL: http://en.wikipedia.org/wiki/Double_outlet_right_ventricle.
11. Smith RS, Comstock CH, Kirk JS, Lee W, Riggs T, Weinhouse E. Double-outlet right ventricle: An antenatal diagnostic dilemma. *Ultrasound Obstet Gynecol* 1999; 14: 315-9.
12. Chen GZ, Huang GY, Liang XC, Ma XJ, Chen WD, Tao ZY, et al. Methodological study on real-time three-dimensional echocardiography and its application in the diagnosis of complex congenital heart disease. *Chin Med J (Engl)* 2006; 119: 1190-4.
13. Gedikbasi A, Oztarhan K, Gul A, Sargin A, Ceylan Y. Diagnosis and prognosis in double-outlet right ventricle. *Am J Perinatol* 2008; 25:427-34.