



Prenatally Diagnosed Ventricular Inversion, Restrictive Ventricular Septal Defect, Pulmonary Stenosis, Hypertensive Left Ventricle and Double Outlet Right Ventricle: Case Report and Literature Review

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Abstract

We herein describe, for the first time, the fetal presentation of a case of ventricular inversion, restrictive ventricular septal defect, pulmonary stenosis, hypertensive left ventricle and double outlet right ventricle at 34 weeks of gestational age. Post-natal echocardiography confirmed the prenatal diagnosis. The patient was subsequently successfully palliated with a left ventricle to pulmonary artery conduit. This report illustrates the importance of detailed fetal echocardiography to ensure appropriate delivery and neonatal management, and to optimize outcome.

Keywords Fetal echocardiography · Double-outlet right ventricle · Ventricular inversion · Complex fetal heart disease

Introduction

Double outlet right ventricle (DORV) represents a wide spectrum of congenital heart defects, most of which include a nonrestrictive ventricular septal defect (VSD) that allows pulmonary venous return to exit the left ventricle and provide unobstructed systemic and pulmonary blood flow from the right ventricle (RV). This unusual fetal case of DORV, associated with ventricular inversion and a restrictive VSD, illustrates the prenatal presentation of an exceptional form of DORV that presents with a hypertensive right-sided morphological left ventricle. The unusual anatomy and pathophysiology of this case, both evident prenatally, led to an unusual but successful neonatal surgical palliation for this infant soon after delivery.

Case Report

A 29-year-old female, G2P0010 with unremarkable family history, was referred to the UCLA Pediatric Cardiology Department by her primary obstetrician at 33 weeks and 3 days gestation due to an abnormal appearance of the fetal heart.

Fetal echocardiography, at 34 weeks gestation, optimized image quality and angle of acquisition to perform a comprehensive, segmental evaluation of the fetal heart. Imaging confirmed the diagnosis of complex heart disease: {S,L,L} viscerotrial situs solitus with ventricular inversion, L-looped ventricles, double-outlet RV (Fig. 1a, b), pulmonary stenosis and a restrictive VSD (shunting from the right-sided, morphologic left ventricle to the left-sided, morphologic RV) (Fig. 2). There was significant subvalvar and valvar pulmonary stenosis and unobstructed aortic outflow, with both great arteries arising from the RV. The only egress from the right-sided, morphologic left ventricle was through the restrictive VSD. The hypertensive left ventricle generated mitral regurgitation (Fig. 2) with a peak velocity of 5 m/s, reflecting markedly elevated left ventricular systolic pressure over 100 mmHg. The hypertensive left ventricle had moderately reduced systolic function.

Ebstein's anomaly of the left-sided tricuspid valve was mild, with relatively well-formed leaflets and only mild regurgitation.

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Fig. 1 Prenatal echocardiography of a 34 weeks gestational age fetus showing double outlet right ventricle (DORV). **a** Pulmonary artery (PA); **b** Aorta (Ao) arising from the RV. Note the presence of atrioventricular discordance (arrows)

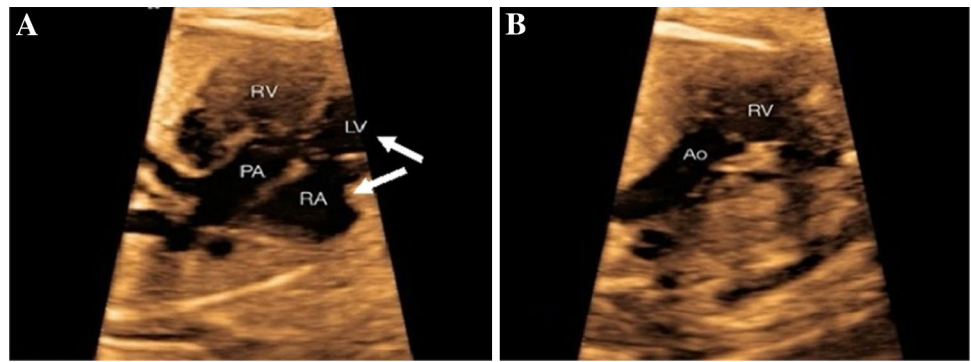
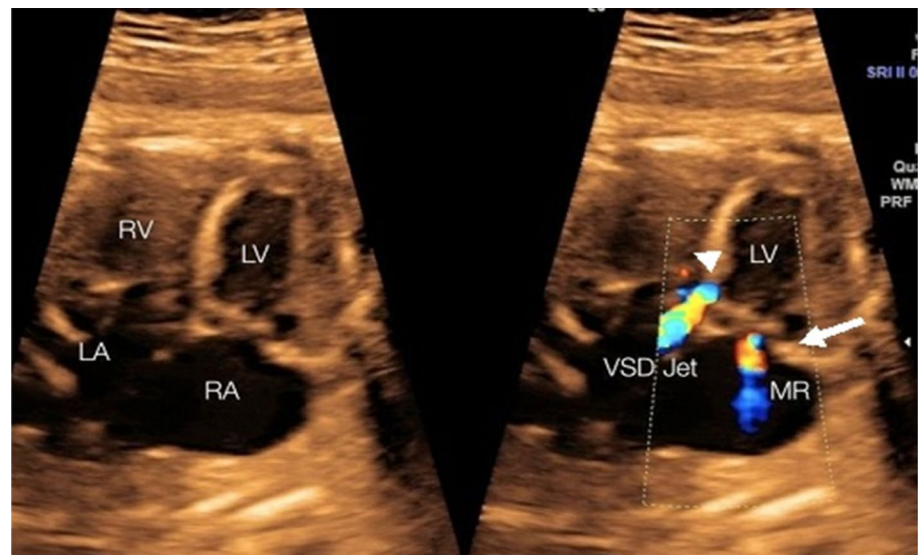


Fig. 2 Doppler ultrasonography of the fetal heart displaying mitral regurgitation (MR) (arrow) with a peak velocity of 5 m/s, secondary to a left ventricular hypertrophy instigating a ventricular systolic pressure exceeding 100 mmHg. Also shown, a restrictive ventricular septal defect (VSD) (arrowhead) with a peak velocity of 3.3 m/s



The mother had no chronic disease, no exposure to known teratogens, and no family history of congenital heart disease. Cell-free fetal DNA screen was low risk.

During the third trimester, serial echocardiography was performed every 2–3 weeks. The fetal left ventricle became increasingly hypertensive, with worsening systolic function. Despite the significantly elevated left ventricular pressure, the fetus grew normally throughout gestation, and at no time developed heart failure or ectopy. Given the anticipated need for neonatal surgical intervention, and the mother's residence remote from a tertiary care center, the mother underwent elective induction of labor at 39 weeks and 3 days. Because of failure to progress, the baby was subsequently delivered via cesarean section with complication.

Following delivery, the diagnosis was confirmed by echocardiography. For decreasing saturation and pulmonary blood flow as the ductus arteriosus began closing, PGE was initiated. Cardiac catheterization demonstrated left ventricular pressure greater than twice that of the RV, and ruled out communications between the left ventricle and the coronary arteries. Genetic evaluation revealed absence

of any identifiable genotypic abnormality. The patient subsequently underwent placement of a Gore-Tex interposition graft (right-sided left ventricle to the pulmonary artery—PA) to decompress the left ventricle and to augment pulmonary blood flow. At 4 years of age, the patient has required upsizing of the interposition conduit, but continues to thrive clinically without the development of heart failure or arrhythmias.

Discussion

DORV has an incidence of 1 in 10,000 live births, or 1% of children with congenital heart disease [1]. DORV describes a range of congenital cardiac anomalies where the aorta and the PA both arise from the RV. This condition is usually associated with a nonrestrictive VSD [2]. The VSD ensures the flow of oxygenated blood from the left side of the heart into the right chamber to be pumped into the systemic circulation. Lev et al. [3] described the classification of DORV into different types depending on the location of the VSD

in relation to the great arteries. The VSD may be depicted as doubly committed, noncommitted, subpulmonary, or subaortic. Postnatal management is highly reliant on the type of DORV and the presence of associated cardiac abnormalities. In the vast majority of cases, the VSD is large and nonrestrictive.

In this case report, however, a highly unusual form of DORV with ventricular inversion, a restrictive VSD and a hypertensive left ventricle presented at 34 weeks gestation. DORV with a restrictive VSD, particularly in association with ventricular inversion, represents a highly morbid condition seldom seen postnatally, let alone before birth. Serratto et al. [4] and Cavalini et al. [5] described multiple such postnatally diagnosed cases of DORV. The postnatal presentation universally includes a hypertensive left ventricle, and typically some degree of pulmonary hypertension and atrial or ventricular arrhythmias. Surgical intracardiac intervention may introduce risks of atrioventricular node damage, particularly in newborns. The best intervention for DORV with a restrictive VSD is still highly controversial, and depends upon associated cardiac abnormalities. Nevertheless, in almost all cases, early relief of LV hypertension is essential. Previous surgical approaches have included atrial septectomy with mitral valve avulsion [6] or LV exclusion [7]. Apart from a surgical intervention, Lin et al. [8] reported the first trans-catheter approach that enabled a successful palliative relief of the VSD. Such an approach can be considered as a reasonable alternative in some cases with unacceptably high surgical risks.

In the current case, DORV and a restrictive VSD was complicated by ventricular inversion, in which the flow of blood drains from the morphologic right atrium into the morphologic left ventricle and from the morphological left atrium into the morphological RV. Ventricular inversion is most commonly associated with L-transposition of the great arteries (congenitally corrected transposition), but may also present with normal ventriculo-arterial alignments and connections (isolated ventricular inversion). Very rarely, as in the current case, ventricular inversion presents in association with L-malposition and DORV. This case was particularly unusual given the association of these abnormalities with a restrictive VSD.

In conclusion, this manuscript describes a rare fetal case of ventricular inversion, DORV, pulmonary stenosis, restrictive VSD and a hypertensive left ventricle. Postnatal evaluation included both echocardiography and cardiac catheterization, which confirmed the diagnosis and ruled out the presence of significant left ventricle to coronary artery sinusoids. The baby was successfully palliated by placement

of a Gore-Tex interposition graft (LV–PA) to decompress the left ventricle and to augment pulmonary blood flow. This graft was subsequently replaced with a larger conduit, and the patient continues to do well at 4 years of age.

DORV, usually accompanied by other heart defects [9], can rarely present with ventricular inversion, restrictive VSD and hypertensive left ventricle. Prenatal diagnosis in this case enabled planning for coordinated delivery at a tertiary care center, where prompt neonatal evaluation and management enabled successful surgical palliation and optimized the child's prognosis.

Compliance with Ethical Standards

Conflict of interest Drs. El-Asmar, Degheili, Al-Halabi, and Sklansky declare that they have no conflict of interest.

Ethical Approval This article does not contain any studies with human participants performed by any of the authors.

Informed Consent Informed consent was not performed on the single patient described in this case report; the patient's family was not available to obtain informed consent for this case report.

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