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An isolated double-inlet right ventricular malformation: a case report and review of the literature

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Abstract

Background Double-inlet right ventricle is an extremely rare and complex cardiac malformation characterized by the opening of both atria into the right ventricle and is usually associated with other major cardiac malformations. In this case report, we have discussed an isolated case of double-inlet right ventricle without other malformations and conducted a literature review to compare our case, which is the third in the medical literature, with previously published papers on double-inlet right ventricle.

Case presentation A 2-month-old Syrian male infant was admitted to the cardiology department due to symptoms of central cyanosis and failure to thrive. Echocardiography revealed double-inlet right ventricle with a hypoplastic left ventricle from which the hypoplastic aorta emerged, while the pulmonary trunk emerged from the dilated right ventricle. The child was sent abroad for the Norwood procedure.

Conclusion Although double-inlet right ventricle is a very rare malformation, it can be encountered in clinical practice and should be taken into consideration by practitioners. Clinical researchers should document and publish cases of double-inlet right ventricle diagnosed to draw attention to this malformation and study it in greater depth.

Keywords Congenital heart defects (CHD), Double-inlet ventricle (DIV), Double-inlet right ventricle (DIRV), Norwood procedure, Case report

Background

Double-inlet ventricle (DIV) by definition is when both atria empty into a single ventricle [1]. The incidence of DIV is rare, affecting less than 1 per 10,000 live births, and makes up about 1% of all congenital heart defects (CHD) [1]. DIV classically presents with one large ventricle and one small ventricle, which usually communicate with each other via a ventricular septal defect (VSD) [1]. There are three morphological types of this malformation: double-inlet left ventricle (DILV; most common), double-inlet right ventricle (DIRV; sometimes), and solitary ventricle of indeterminate morphology (rare) [1]. DIRV is a rare cardiac malformation, and one study reported an incidence of DIRV in 1 (2%) of 50 patients with DIV [2]. Another study showed a higher prevalence of DIRV in Chinese populations than in Western populations [2]. It is noteworthy that DIRV is often related to double-outlet right ventricle (DORV) in literature, with one study finding that 71.4% of DIRV cases in the study were associated with DORV [1, 2]. This malformation can be diagnosed even before birth using fetal echocardiography [1]. DIRV can be treated surgically in three stages, with the first stage being the Norwood procedure, the second being the Glenn procedure, and the third and final stage being the Fontan procedure,

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with special attention given to this condition because the aorta arises from a hypoplastic left ventricle [1, 2]. In this case report, we have discussed an isolated case of DIRV without DORV in an infant.

Case presentation

A 2-month-old Syrian male infant was admitted to the cardiology department due to symptoms of cyanosis and failure to thrive. The patient's mother, who was 26 years old and mildly alcoholic, has a history of tension headaches that respond well to paracetamol, and she also has a healthy 2-year-old daughter and has no history of miscarriages. Her pregnancy with the patient was complete and uncomplicated, with a vaginal delivery at 39 weeks' gestation at a small health center in her remote area. On physical examination, the patient appeared to be in poor general condition, suffering from respiratory distress, central cyanosis that increased during crying, and a slightly swollen face. A 3/6 systolic murmur was heard in the aortic focus during cardiac auscultation. Most of his laboratory tests were normal. His pulse was 120 beats/minute, and his systolic blood pressure was 75 mmHg. Echocardiography showed both atrioventricular valves opening into the right ventricle (that is, DIRV), hypoplastic left ventricle (Fig. 1), inlet-type VSD (Fig. 2), moderate subaortic stenosis with a maximum velocity of 3.5 m/second, hypoplastic aortic arch (Fig. 3), and descending aorta, which is supplied via a patent ductus arteriosus (PDA) with reversed flow (right-to-left shunt) (Fig. 4); a well-functioning, dilated right ventricle; normal-sized right and left atria, with mild regurgitation of the mitral and tricuspid valves; and a small (4 mm) atrial septal defect (ASD) (left-to-right shunt). Normal pulmonary valve and dilated pulmonary artery showed no evidence of transposition of the great arteries (TGA) (that is, ventriculoarterial concordance). The infant's condition was stabilized, and he was initially treated with heart failure medications: metoprolol succinate 12.5 mg once daily, spironolactone 6.25 mg once daily, and furosemide at a loading dose of 1 mg/kg followed by a maintenance dose of 0.25 mg/kg once daily. The infant was then referred abroad for a Norwood procedure.

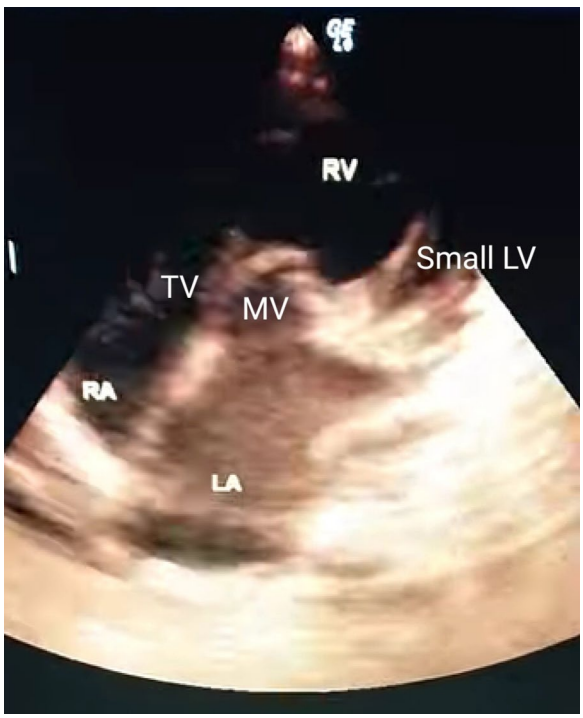

Fig. 1 Echocardiogram shows that both the right and left atria open into the right ventricle, while the hypoplastic left ventricle can be clearly seen. *MV* mitral valve, *TV* tricuspid valve, *LV* left ventricle, *RV* right ventricle, *RA* right atrium, *LA* left atrium

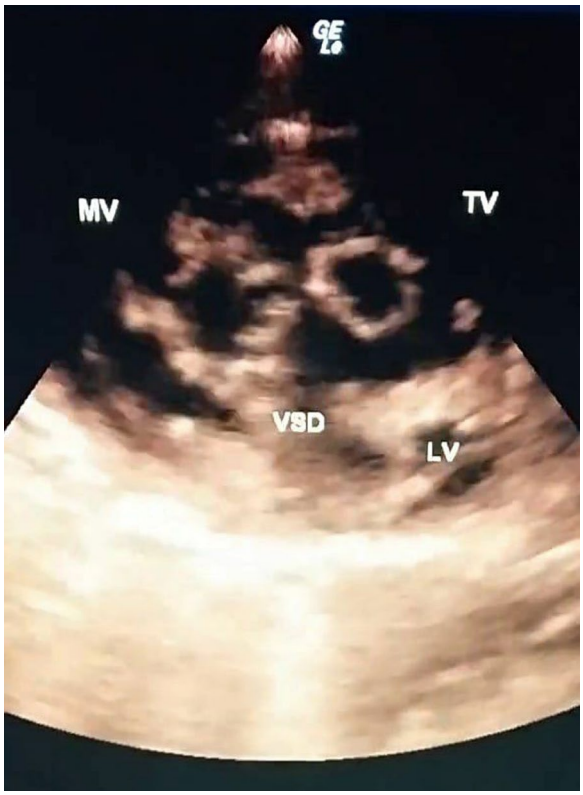

Fig. 2 Echocardiogram shows the ventricular septal defect in addition to the opening of the mitral and tricuspid valves on the right ventricle. *VSD* ventricular septal defect

Discussion

In normal cardiac anatomy, there are distinct left and right ventricles, each consisting of three segments: inlet, apex, and outlet [1]. The ventricular septum separates these chambers and houses the atrioventricular

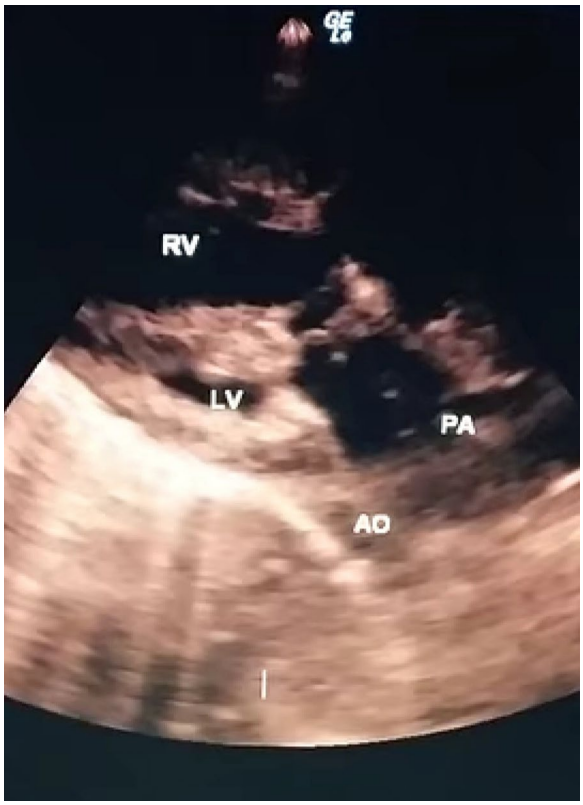

Fig. 3 Echocardiogram shows a hypoplastic aortic arch and dilated pulmonary artery. *AO* aorta, *PA* pulmonary artery

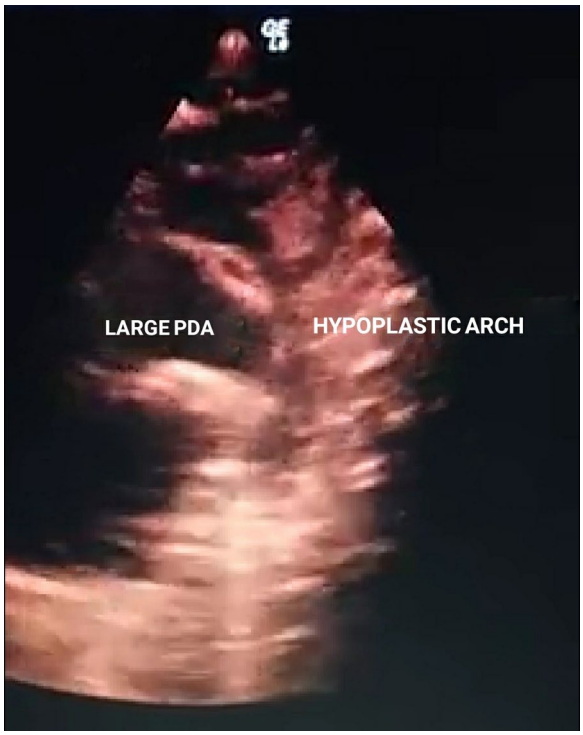

Fig. 4 Echocardiogram shows a large patent ductus arteriosus that supplies the hypoplastic descending aorta. *PDA* patent ductus arteriosus

conduction axis, but in DIRV, the left ventricle is incomplete and lacks all its typical components [1]. DIRV represents a less frequent morphological form among the complex congenital heart abnormalities characterized by univentricular atrioventricular junction, a group associated with significant mortality [1, 3]. This anomaly is defined as a rare congenital cardiac anomaly and characterized by both atria draining into a single morphological right ventricle through separate or common atrioventricular orifices, typically receiving the tricuspid orifice and, partially or completely, the mitral orifice [1, 4, 5]. This definition excludes conditions such as tricuspid or mitral atresia, atrioventricular canal defects, and other anomalies that do not constitute true atrioventricular orifices opening into the right ventricle [4]. The characteristic anatomical features of DIRV include the presence of an anterior papillary muscle, a grossly trabeculated surface of the interventricular septum, the crista supraventricularis formed by parietal and septal bands, and a muscular infundibulum [4]. In approximately two-thirds of cases with DIRV, there is a concurrent association with conditions such as DORV and malpositioning of the great arteries (MGA) [5]. Our isolated DIRV case is accompanied by a hypoplastic left ventricle, inlet-type VSD, small ASD, moderate subaortic stenosis, hypoplastic aortic arch, and descending aorta supplied via a PDA exhibiting reversed flow (right-to-left shunt). This anomaly is frequently associated with maternal pregestational diabetes, which increases the risk of its occurrence [1]. Epidemiologically, DIRV is more common in the Chinese population, accounting for approximately 2% of 50 patients reported to have DIV and 23 patients out of 1640 with congenital heart abnormalities [2, 5, 6]. Prenatal diagnosis of DIRV has significantly improved with advancements in imaging techniques, allowing for the detection of this anomaly through fetal echocardiography [1]. The four-chamber view on fetal echocardiography typically shows a single ventricle without a ventricular septum, necessitating the connection of both atria to this single ventricle for a definitive diagnosis [1]. Symptoms of DIRV include heart murmurs, cyanosis, and heart failure, which result from abnormal blood flow and pressure dynamics within the heart [1]. Our 2-month-old Syrian male infant presented with central cyanosis, respiratory distress, a 3/6 systolic murmur in the aortic focus, and failure to thrive. The echocardiography revealed partial or complete opening of both atrioventricular valves into the right ventricle, while the left ventricle was visible in all cases even though it was noticeably underdeveloped

[7]. The right ventricle was characterized by coarse apical trabeculations, while the left ventricle was in a posteroinferior position [5]. In our case, the echocardiography showed both atrioventricular valves opening into the right ventricle, moderate subaortic stenosis, and a hypoplastic aortic arch and descending aorta. DIRV must be differentiated from situations in which there is mitral or tricuspid stenosis, atrioventricular canal defects, or the presence of additional valves [7]. Surgical repair is required and consists of three stages. The first is the Norwood procedure, which is defined as a surgical procedure performed to establish sufficient outflow to the systemic circulation in individuals with systemic outflow obstruction [8]. Mortality risk is related to anomalous pulmonary venous drainage, moderate or severe atrioventricular regurgitation, lower weight, and prematurity. The increasing experience with the operation and prenatal diagnosis decreases the mortality [9]. The second stage is a bidirectional Glenn shunt, which is a connection of (the end of) the superior vena cava (SVC) to the superior aspect (side) of the ipsilateral pulmonary artery; the superior cavopulmonary shunt drains the single ventricle [10]. The third stage is the Fontan procedure, in which total right atrial or total caval blood flow is channeled directly into the pulmonary artery or into the small right ventricle that serves only as a conduit [11]. In our case, the patient was transferred abroad for a Norwood procedure. By reviewing the literature, we found that there are, to our knowledge, four case reports in the literature that record a case of DIRV, two of which differ from our case in the anatomical details of the malformation; the first reported TGA associated with DIRV [12], and the second reported a complete absence of the left ventricle, compared with our case, which was not associated with TGA and had a hypoplastic left ventricle from which the hypoplastic aorta arose [13]. The third and fourth cases reported cases of DIRV that are anatomically similar to our case [14, 15], making our paper the third paper in the literature that reports an isolated case of DIRV without TGA or DORV.

Conclusion

DIRV is a complex and extremely rare CHD that poses a major challenge in diagnosis and treatment. However, the rarity of this defect and the lack of research papers that record its diagnosis do not preclude the possibility of encountering it during the clinical approach and radiological diagnosis of an infant showing symptoms of severe heart defect, which highlights the importance of the correct clinical approach and the use of advanced cardiac imaging techniques by an experienced hand to develop an early surgical treatment plan. Continuous research and efforts to record DIRV cases and publish them in the medical literature are of utmost importance for a deeper understanding of this defect and to improve treatment methods and survival rates by enhancing cooperation between pediatric cardiologists, cardiac imaging specialists, and cardiac surgeons.

Abbreviations

DIV	Double-inlet ventricle
CHD	Congenital heart defects
VSD	Ventricular septal defect
DILV	Double-inlet left ventricle
DIRV	Double-inlet right ventricle
DORV	Double-outlet right ventricle
PDA	Patent ductus arteriosus
ASD	Atrial septal defect
TGA	Transposition of the great arteries
MGA	Malpositioning of the great arteries
SVC	Superior vena cava

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None.

Methods: This work has been reported in line with the case report (CARE) criteria [16].

Author contributions

William Borghol, Faisal Alfaksh, Saja Karaja, Lyne Barakat, and Saleh Takkem drafted the manuscript and revised it.

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Availability of data and materials

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Declarations

Ethics approval and consent to participate

Not applicable.

Consent for publication

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Competing interests

None.

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