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C A S E   R E P O R T

The double outlet right ventricle with additional cardiac malformations: an anatomic and echocardiographic study

M.T. Demir[1], Y. Amasyalı[2], C. Kopuz[1], M.E. Aydın[1], U. Çorumlu[1]

[1]19 Mayis University Medical Faculty, Anatomy Samsun, Turkey
[2]Anatolian Hospital, Cardiology Samsun, Turkey

Double outlet right ventricle (DORV) is a complex group of abnormal ventriculoarterial connections where both the aorta and pulmonary artery rise from the right ventricle morphologically. It is a rare condition affecting 1–1.5% of patients with congenital heart disease.

The patient's cardiac measurements were taken by two-dimensional echocardiography. The pathological findings of the defined case are illustrated in the figures. A 20-year-old male patient with palpitations, tachypnea, and tiredness with weak exercise capacity and back pain was diagnosed with heart murmur in different degrees; slight cyanosis, cachexia, and developmental retardation were found clinically. Echocardiography showed: DORV, perimembranous ventricular septal defect, pulmonary stenosis, dilatated right ventricle, and dilatated left atrium. However, the left ventricle was normal. In addition, the septum interatriale filled the left atrium like an aneurysm.

DORV is an important and rare congenital cardiac malformation. This complex condition may result in different clinical findings and require different therapeutic approaches. (Folia Morphol 2009; 68, 2: 104–108)

**Key words: heart, malformation, double outlet right ventricle, echocardiography**

INTRODUCTION

In normal cardiac anatomy, the aorta has its origin from the left ventricle and the pulmonary artery has its origin from the right ventricle. Double outlet right ventricle (DORV) is a complex group of abnormal ventriculoarterial connections in which both the aorta and pulmonary artery emerge from the right ventricle morphologically [1, 12]. It is a rare condition, affecting 1–1.5% of patients with congenital heart diseases with a frequency of 1 in each 10,000 live births [12]. A 20-year-old male patient with palpitations, tachypnea, and tiredness, with low exercise tolerance and back pain. Systolic heart murmur in different degrees (2°–3°/6°) in the mesocar-
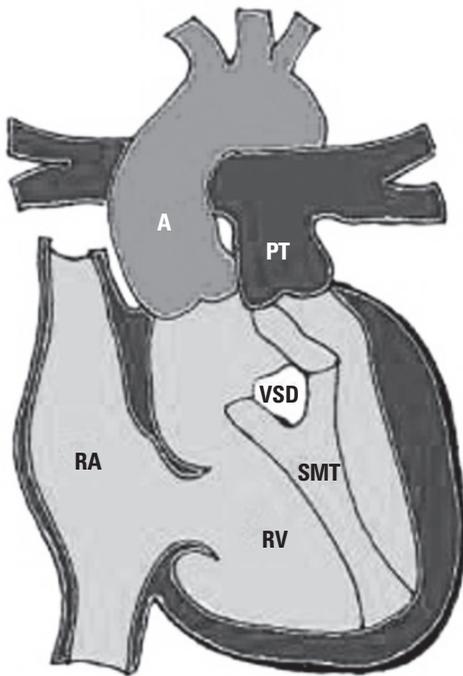
diac region, slight cyanosis, cachexia, and developmental retardation were found clinically.

MATERIAL AND METHODS

A 20-year-old male patient was admitted to the cardiology clinic of the Büyük Anadolu Hospital with palpitations, tachypnea, and tiredness, with low exercise tolerance and back pain. In the physical examination, slight cyanosis at the limbs and nails, developmental retardation despite his age, and cachexia were determined. At auscultation, 2°–3°/6° systolic murmur at the mesocardiac region and 1°–3°/6° systolic murmur at the pulmonary region were determined. Because of these findings, it was
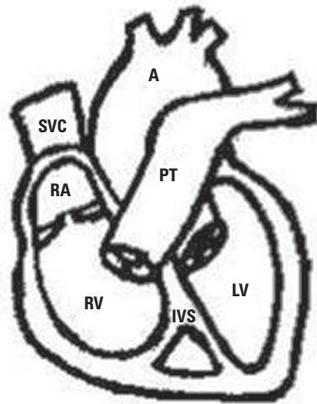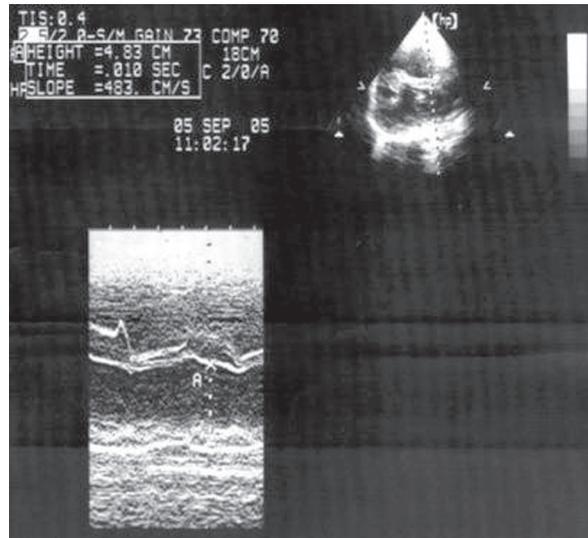
Figure 1. Schematic diagram of a normal cardiac structure; A — aorta, PT — pulmonary trunk, LV — left ventricle, RV — right ventricle, RA — right atrium, IVS — interventricular septum, SVC — superior vena cava.


Figure 3. Echocardiography showed dilatation of left atrium and double outlet right ventricle. The measurement was made in the position of parasternal long axis by M mode presentation.

Figure 2. Schematic diagram of our case; A — aorta, PT — pulmonary trunk, VSD — ventricular septal defect, RA — right atrium RV — right ventricle, SMT — septomarginal trabecula, IVS — interventricular septum, SVC — superior vena cava.

decided to evaluate the patient using echocardiography. The patient's cardiac measurements, taken by 2-dimensional echocardiography (HP Sonos 2000, 2-dimentional transthoracic echo-2.5 mHz cardiac prope) and the measured cardiac values, were compared with normal measurements. The causes of these conditions were discussed with knowledge of the patient's anatomy and embryology. The pathological findings of the defined case and the normal form of the cardiac values as our specimens are illustrated in Figures 1 and 2. The patient did not have a familial history of cardiologic disease.

RESULTS

In our case, the echocardiographic findings are explained as below:

— Double outlet right ventricle (Fig. 3).
— Dilatation of the left atrium (Fig. 3). The measurement was made on the position of the parasternal long axis by M mode presentation. The diameter of the left atrium was measured as 48 mm (normal value: 0–40 mm) and on the parasternal longitudinal axis it was seen that the aorta did not emerge from the left ventricle.
— Paradoxal motion of the interventricular septum (Fig. 4). The dimensions of the left ventricle were normal. In addition, the interventricular septum had a paradoxal condition at the ventricular systole. The ejection fraction (EF) of the left ventricle was normal at 67% (normal value of EF: 50–80%). The EF calculation was made according to Simpson's principle. Formula = end diastolic volume – – end systolic volume/end diastolic volume.
— Dilatation of the right ventricle. The diameter of the right ventricle was measured as 36 mm (normal value: 0–26 mm).
— Pulmonary stenosis (Fig. 5). The pressure gradient on the pulmonary valve was measured as
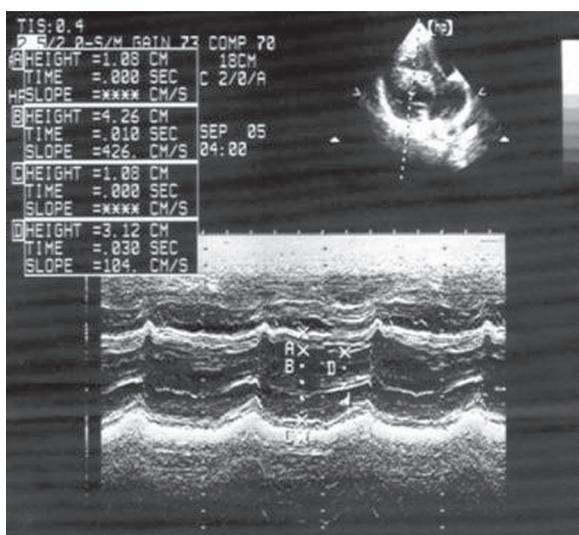
Figure 4. Echocardiographic image showed ejection fraction of left ventricle. The ejection fraction calculation was made according to Simpson's principle. Formula = end diastolic volume – end systolic volume/end diastolic volume.
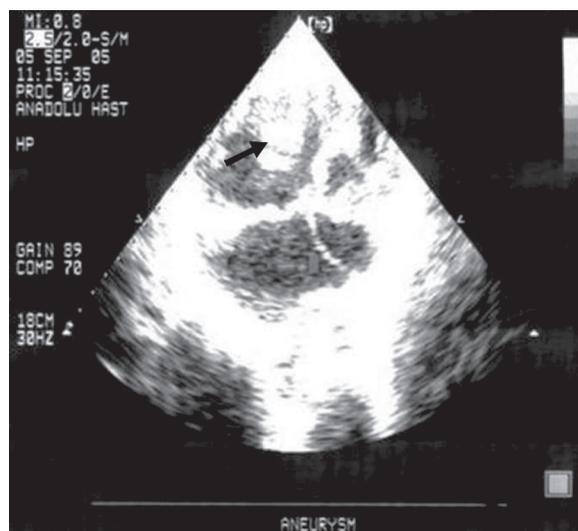
Figure 6. Echocardiographic image showing aneurysm on interatrial septum.
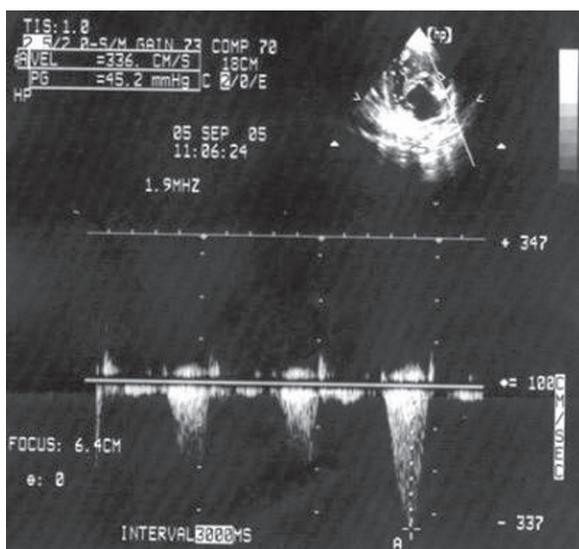
Figure 5. Echocardiographic view of pulmonary stenosis is in the presented case. Doppler (continuous wave) measurement elevated gradient in main pulmonary artery — 45.2 mm Hg. The visual images of diagnosis of pulmonary stenosis in transthoracic echocardiography on parasternal short axes were rendered. The gradient between right atrium and right ventricle was determined using a simplified version of Bernoulli's equation. Systolic pressure of the right ventricle, which is equal to the peak gradient of pulmonary stenosis by adding the pressure of right atrium to this gradient, was calculated.

45.2 mm Hg (slight stenosis: 0–20 mm Hg, medium stenosis: 20–50 mm Hg, severe stenosis: > 50). The visual images of diagnosis of pulmonary stenosis in transthoracic echocardiography on parasternal short axes were rendered. The

gradient between the right atrium and right ventricle was determined by using a simplified version of Bernoulli's equation. The systolic pressure of the right ventricle, which is equal to the peak gradient of pulmonary stenosis by adding the pressure of the right atrium to this gradient, was calculated.

— Perimembranous type ventricular septal defect (VSD). On echocardiography there was a shunt from left to right on the interventricular septum. The cause of this shunt was a defect in the interventricular septum. The type of VSD was perimembranous. In addition, the aorta emerged from the right ventricle.

— Aneurysm on the interatrial septum (Fig. 6). Aneurysm on the interatrial septum pouched to the left and filled up the left atrium.

The aortic arch had no obstruction; however, the pericardium had a normal appearance.

DISCUSSION

The origin of the aorta and pulmonary artery from the right ventricle is a complicated congenital cardiac malformation [4]. In normal cardiac anatomy, these great vessels have their origin of the aorta from the left ventricle as the pulmonary artery from the right ventricle [10, 14].

DORV can be classified according to the relationship between the VSD and blood vessels as: DORV with subaortic VSD, DORV with subpulmonary VSD, DORV with doubly-committed VSD, and DORV with noncommitted VSD [12]. The most frequent anatomic

type is the subaortic VSD with posterior and right sided aorta [12]. The wide spectrum of anatomic variations found in DORV may result in different clinical findings and may require different therapeutic approaches [2, 3, 12]. DORV has traditionally been difficult to diagnose accurately [1]. In our case, DORV with perimembranous VSD has been described. Although many morphological variations of this condition have been described, DORV with normal left ventricle, perimembranous type VSD and aneurysm on the interatrial septum has not been reported

The additional anomalies were affected by the position of VSD, which was seen in DORV. When the VSD is subaortic; subpulmonary stenosis or subpulmonary-subaortic stenosis can be considered [12]. Appropriately to this knowledge, a perimembranous type VSD with pulmonary stenosis of middle degree was determined in our case

In any kind of VSD, atrial septal defect, or persistent ductus arteriosus can exist in this situation [12]. In support of this idea, a similar situation was considered in our case. Conversely, there was not a complete defect in the interatrial septum but the occurrence of an aneurysm which lay in the left atrium and filled the left atrium completely, which was determined by echocardiography.

The membranous part of the interventricular septum is formed by the extension of the tissue from the endocardial cushion. This tissue merges with the aorticopulmonary septum and the muscular part of the interventricular septum. After the formation of the membranous part of the interventricular septum, and at the same period after the closure of the interventricular foramen, as a result of these changes the pulmonary trunk communicates with the right ventricle and the aorta communicates with the left ventricle [11].

By the fifth week of development, mesenchymal cells proliferate in the walls of the bulbus cordis to form the bulbar ridges, and the truncal ridges are formed in the truncus arteriosus, which are the continuous structures of the bulbar ridges. Both truncal and bulbar ridges have a spiral orientation as a result of the blood flow from the ventricles, and after the ridges fusion it again causes the spiral form of the aorticopulmonary septum [11]. This septum takes the main role in the division of the bulbus cordis and truncus arteriosus into two arterial ways, as the aorta and pulmonary trunk. The spiral form of the aorticopulmonary septum is the cause of the placement of the pulmonary trunk around the ascending part of the aorta [11].

One single hypothesis cannot explain the origin of the defect during the embryological period, which may be the cause of this anomaly [1]. The late formation of the membranous part of the interventricular septum, and, as a result of this, the late communication of the great vessels with their original ventricles: aorta with the left ventricle and pulmonary artery with the right ventricle, may be the origin of the positional anomaly of these great vessels. And again, the late formation or deformation of the spiral form of the aorticopulmonary septum may be the cause of the inadequate division of the truncus arteriosus and bulbus cordis as the aorta and pulmonary trunk [11]. Additional anomalies may be the result of these changes of embryologic period, such as VSD and pulmonary stenosis in our case. The haemodynamic changes occurring as work against pressure may be the explanation of dilatation of the right ventricle, depending on these anatomic changes. In our case, the dilatation of the left atrium without dilatation of left ventricle may be the most interesting part of this variation.

The anatomic features of our case were clearly shown with two-dimensional echocardiography [8, 13, 15]. Two-dimensional echocardiographic findings have allowed the non invasive recognition of DORV, making haemodynamic studies unnecessary, and have the advantage of the demonstration of associated anomalies which were previously unrecognized preoperatively [4, 7, 9, 12]. The perimembranous VSD, the left ventricle with normal standard dimensions, and the interatrial septal aneurysm were the most interesting findings in our case. Surgical repair of this rare condition may be performed successfully, depending on the anatomic type of the condition, and the surgery may be planned by maintaining the normal anatomic origins of these great vessels, which may be obtained by echocardiography or by angiocardiography [5, 6, 12].

There it is only possibility that some abnormalities exist or not as a morphological changes. Shunts are necessary to maintain communication between the right and left sides of the heart. Various total neoplastic approaches are necessary for the treatment of this kind of patient with DORV with other cardiac problems. For this reason, this patient was sent to the cardiovascular surgeon for evaluation after the initial symptomatic treatment. We think that the evaluation, and the anatomical and embryological discussion of the cause of this kind of complicated pathological situation, can be useful for clinicians in their future experience.

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