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Mid-term Results of Double Chamber Right Ventricle in Association with Genetic Syndromes

Genetik Sendromların Eşlik Ettiği Çift Odacıklı Sağ Ventrikülde Orta Dönem Sonuçlarımız

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Öz

Çift odacıklı sağ ventrikül (DCRV), hipertrofik kas bantlarının sağ ventrikülü ikiye böldüğü konjenital bir hastalıktır. Genetik sendromun da eşlik ettiği 6 hastamızın erken-orta dönem cerrahi sonuçlarını sunmaktayız. DCRV tanılı, genetik sendromun eşlik ettiği 6 hasta ile ilgili bulguları derlendi. Ortalama yaş 3.9±1.4 yıldı. Eşlik eden ek kalp anomalileri perimembranöz ventriküler septal defekt (n=3), atrial septal defekt (n=1), orta aort yetmezliği (n=1), diskret subaortik membran (n=1) idi. Eşlik eden genetik sendromlar Costello (n=1), Seckel (n=1), Down sendromuydu (n=4). Ortalama takip süresi 4.86±4.6 yıldı. Sağ ventriküldeki ortalama sistolik basınç gradienti 18.5 ± 11.5 mmHg idi. Takipte mortalite ya da tekrar operasyon gerekliliği olmadı. Çift odacıklı sağ ventrikül, Costello ve Seckel Sendrom birlikteliğinin literatürde ilk kez yayımlandığı düşüncesindeyiz.

Anahtar Kelimeler: Costello Sendromu, Double Chambered Right Ventricle, Down Sendromu, Seckel Sendromu

Abstract

Double chamber right ventricle (DCRV) is a congenital disease in which a hypertrophied muscle band divides the right ventricle chamber into two. The early-mid-term follow-up of 6 patients with DCRV and distant genetic syndromes is reported in this paper. A retrospective analysis was performed of 6 DRCV patients with a mean age of 3.9±1.4 years. Concomitant cardiac anomalies were perimembranous ventricular septal defect (n=3), atrial septal defect (n=1), mild aortic regurgitation (n=1), discrete subaortic membrane (n=1). Associated genetic syndromes were Costello (n=1), Seckel (n=1) and Down syndromes (n=4). The mean follow-up period was 4.86±4.6 years. Mean systolic pressure gradient in the right ventricle in the postoperative was 18.5±11.5 mmHg. No mortality occurred and there was no requirement for reintervention. To the best of our knowledge, this is the first report in literature of concomitant DCRV with Costello and Seckel syndromes.

Keywords: Costello Syndrome, Double Chambered Right Ventricle, Down Syndrome, Seckel Syndrome

Introduction

Double chamber right ventricle (DCRV) is a congenital disease in which a hypertrophied muscle band divides the right ventricle chamber into two, and constitutes 0.5-2% of all congenital heart diseases (1). An abnormal hypertrophic muscle band divides the right ventricle (RV) into two chambers (1). The defect between two chambers has a pressure gradient relative to the diameter (2). Although DCRV is usually asymptomatic, a progressive course (chest pain, heart failure symptoms) may occur. Ventricular septal defect (VSD) is the most associated congenital heart defect at the rate of 80-90% (2). In the absence of a co-existing defect, surgery is not indicated unless the intracavitary systolic pressure gradient is higher than 40 mmHg or the obstruction is progressive to maintain normal RV function (1). The early and mid-term follow-up is here presented of 6 cases with DCRV associated with different genetic syndromes, which were successfully treated with surgical correction.

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Material and Method

Patient Selection

A retrospective analysis was made of 6 patients diagnosed with DCRV who were operated on by the same surgeon in two separate centers, using data collected from the hospital database between 2016-2022. Demographic data of gender, age, weight, RV pressures, gradients measured with transthoracic echocardiography (Figure 1A), co-existing cardiac anomalies, and associated genetic syndromes were recorded. Transthoracic echocardiography was performed preoperatively, 10 days postoperatively, and during early and mid-term follow up. The diagnostic criteria for DCRV were: a. Echocardiographic or angiographic evidence of a mid-ventricular obstruction (a systolic pressure gradient between the RV proximal chamber (inflow) and distal chamber (outflow), and b. Absence of infundibular hypoplasia.

The indication for surgery was that the right ventricle intracavitary systolic pressure gradient did exceed 40 mmHg on the echocardiogram.

Approval for the study was obtained from the Institutional Ethics Review Board (16.03.2022 6/VII). All the study procedures were in compliance with the Helsinki Declaration. Informed consent was obtained from the parents or legal guardian of all the patients. All the patients had additional syndromes: Costello Syndrome (n=1) (Figure 1B), Seckel Syndrome (Figure 1C) (n=1), and Down Syndrome (n=4). The patients with Costello and Seckel

Syndromes did not have any other intracardiac defect.

Operation Technique

Corrective surgery was indicated according to the pressure values of the preoperative echocardiograms. All patients underwent cardiac surgery under CPB. Custodiol was used to establish diastolic cardiac arrest. Following right atriotomy, the membrane on the hypertrophic muscle bands dividing the RV, were resected (Figure 2A, 2B, 2C). Ventriculotomy of RV outflow was not needed for any further exploration or for resection of muscle bands. Three patients had perimembranous VSD, which was repaired via right atriotomy using a PTFE patch (Figure 2D). ASD repair was performed using an autologous pericardial patch in one patient. A subaortic ridge in one patient was considered to be non-significant, and did not require any ridge resection.

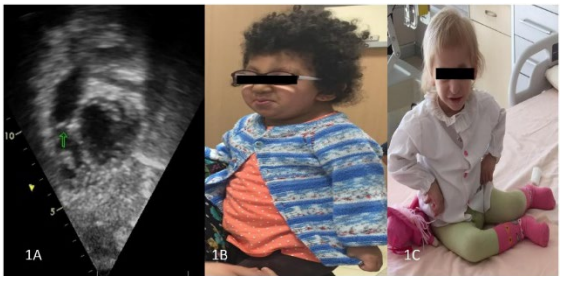

Figure 1A. Preoperative echocardiography of a patient with DCRV. B: Characteristic facial features of Costello Syndrome. C: Characteristics of 'bird head dwarfism' in Seckel Syndrome.

Results

Postoperative evaluation was made of 6 patients diagnosed with DCRV. The mean age at the time of diagnosis was 3.9±1.4 years, and mean weight was 12.1±3.4 kg at the time of the operation.

The mean value of the pressure gradient between the proximal and distal chambers in RV was 72±25 mmHg on the preoperative echocardiograms.

The mean postoperative length of stay in the intensive care unit was 2.1±1.4 days, and the mean length of hospital stay was 6.5±3.2 days.

The patient diagnosed with Costello Syndrome, had atrial fibrillation on the 5th postoperative day and was treated with amiodarone. No patient required a temporary or permanent pacemaker.

Echocardiography in the postoperative early follow-up revealed that the residual mean systolic pressure gradient in the RV was 18.5±11.5 mmHg. No residual VSD was detected.

Echocardiography showed residual discrete aortic membrane in one patient with a 10-mmHg pressure gradient in the early follow-up period.

The mean follow-up period was 4.86±0.6 years. According to the clinical and laboratory findings of the patients during the follow-up period, echocardiography and electrocardiography were performed in the first, third, and sixth months following the operation, then once a year thereafter. The mean systolic pressure gradient in the RV (on echocardiography) at mid-term after surgical intervention was 10.4±6.9 mmHg.

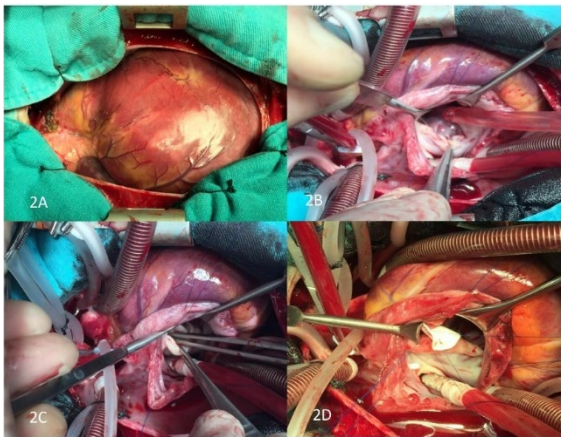

Figure 2A. Intraoperative view of RV in DCVR. B and C: Intraoperative view of membrane in the RV cavity. D: VSD closure through right atriotomy with PTFE patch.

The mid-term follow-up examinations showed that the patient with preoperative mild aortic regurgitation had the same degree of regurgitation, and 2 patients who underwent pulmonary valvulotomy also had the same degree of regurgitation. The patient with discrete aortic membrane had no increase of pressure gradient in the mid-term follow up, and no additional surgery was required.

No mortality occurred in any patient, and there was no requirement for surgical re-intervention during the mid-term follow-up period due to residual VSD.

Discussion

Double chamber right ventricle is considered an acquired congenital heart defect caused by an abnormal membrane on the hypertrophic muscle band formation inside the RV (3). Various mechanisms of development of DCRV have been suggested. Superior displacement of the moderator band especially in association with a VSD and flow turbulence in the RVOT may lead to DCRV, or DCRV may occur due to anomalous muscular bands causing obstruction (3). The frequent associations are tetralogy of Fallot, perimembranous VSD or transposition of great arteries (3). Pulmonary valvular stenosis, ASD, aortic or tricuspid valve regurgitation, persistent left superior vena cava, ruptured sinus of valsalva aneurysm, and Ebstein anomaly are less common associations (4). Pulmonary valve agenesia, main pulmonary artery stenosis have also been reported (1,2).

In the absence of a co-existing defect, surgery is not indicated if the intracavitary systolic pressure gradient is not higher than 40 mmHg or the obstruction is not progressive to maintain normal RV function (1). Different surgical techniques have been described for removing the membrane on the hypertrophic muscle band in the RV, including transventricular, transatrial, and combined approaches (5). The transatrial approach is preferable to the transventricular approach depending on the RV dysfunction and risk of arrhythmia in long-term follow-up. However, in the presence of severe obstructing RV muscle bundles, right ventriculotomy permits adequate relief of the RV cavitary obstruction, allowing better exposure than the transatrial approach

The incidence of additional aortic valve insufficiency has been reported at the rate of 40% in adult DCRV patients and 5% to 20% in pediatric patients (6). In the current case series, aortic valve prolapse and/or mild aortic regurgitation were diagnosed in only 1 patient and no surgical intervention was required.

Williams Syndrome, VACTER-L Syndrome, and Noonan Syndrome may be seen in association with DCRV (7,8). Eltohami reported the high rate of 25% of associated Down's syndrome in a series of DCRV (9).

Costello Syndrome was first described in 1971, with findings of relative macrocephaly, curly-sparsely implanted hair, strabismus, downward slanted palpebral fissures, bulbous nose, large mouth, thick lips, low-set pinnae with large lobes with PS, VSD, ASD, bicuspid aortic valve, aortic stenosis, mitral stenosis, thickening of the intraventricular septum, and hypertrophic cardiomyopathy associations (10). Supraventricular tachycardia and atrial fibrillation have been reported. In the current series, DCRV was detected in routine echocardiographic examinations. Preoperative electrocardiogram showed sinus tachycardia, and the perioperative course was uneventful except for atrial fibrilation on the postoperative 5th day.

Seckel Syndrome was first determined in 1959 as a type of microcephalic primordial dwarfism-bird head dwarfism, neurodevelopmental defects and retinopathy (11). Concomitant congenital heart defects have been recorded as atrioventricular septal defect, ASD, VSD, PDA, tetralogy of Fallot, pulmonary atresia and overriding aorta, and tricuspid atresia (12-14).

In the current study, patient with Seckel Syndrome, the operation and postoperative follow up were uneventful.

Complete relief of the right ventricular obstruction demonstrated excellent functional and hemodynamic mid and long-term results. There was no death or surgical reintervention. In the current study, only 2 patients were followed up for 25 mmHg residual pressure gradient across the RV and for a discrete subaortic membrane with 10 mmHg pressure gradient postoperatively. No arrhythmia was detected.

This study had some limitations, primarily the retrospective design and that only 6 patients were evaluated. There is a need for further studies of larger populations to be able to obtain more information.

Conclusion

The surgical outcomes in this series were excellent due to the right atriotomy approach. Right atriotomy provides adequate exposure and less risk of postoperative arrhythmia. To the best of our knowledge, this is the first report in literature of concomitant DCRV with Costello and Seckel syndromes.

Ethics Committee Approval: Ethical approval for this study was obtained from the Mugla University Institutional Review Board (16.03.2022 6/VII).

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