Tricuspid Valve Repair in Holt-Oram Syndrome

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ABSTRACT

Holt-Oram syndrome is a disease with autosomal dominant inheritance characterized with cardiac defects and upper extremity skeletal anomalies. The prevalence of this syndrome is reported as 1 in 100.000 births. A 13-year-old female patient with Holt-Oram syndrome also called as Heart and Hand syndrome was presented. Tricuspid valve repairment was made in addition to the congenital cardiac defect repairment in the patient who also had tricuspid valve deficiency. The patient underwent a successful surgery.

Key Words: Holt-Oram syndrome; tricuspid valve; valve repair.

Holt-Oram Sendromunda Triküspid Kapak Tamiri

ÖZ

Holt-Oram sendromu kardiyak defektler ve üst ekstremite iskelet anormallikleri ile karakterize otozomal dominant geçişli kalıtsal bir hastalıktır. İlk olarak 1960 yılında tariflenen bu sendromun sıklığı 100.000 doğumda 1 olarak bildirilmektedir. Burada, kalp-el sendromu olarak da adlandırılan Holt-Oram sendromuna sahip 13 yaşındaki kız olgu sunulmuştur. Beraberinde triküspid kapak yetmezliği de mevcut olan hastaya konjenital kardiyak defekt tamiri yanında triküspid kapak tamiri de yapılmıştır. Hasta başarılı bir kardiyak cerrahi düzeltme geçirmiştir.

Anahtar Kelimeler: Holt-Oram sendromu; kapak tamiri; triküspid kapak.

INTRODUCTION

Holt-Oram syndrome is a disease with autosomal dominant inheritance. It includes congenital cardiac defects and upper extremity anomalies[1]. Cardiac transfer defects were also reported in this syndrome which was defined in 1960 for the first time.

In this syndrome which can be seen in a wide clinical spectrum, atrial septal defect or ventricular septal defect among congenital cardiac defects and right hand thumb deformities among upper extremity skeletal anomalies are generally observed. Concurrence of right branch block among transfer defect is frequent in this syndrome[2].

Tricuspid valve deficiency accompanied Holt-Oram syndrome in the 13-year-old patient in our case. Our aim was to present the successful cardiac surgical correction we made on the patient.

CASE REPORT

Thirteen-year-old female patient referred to our clinic with dyspnea complaint. The functional capacity was detected as NYHA (New York Heart Association) class 2. Arterial blood pressure was 96/59 mmHg and pulse rate was 81/min. Right branch block was detected in the ECG of the patient who had a respiratory rate of 14/min. Right upper extremity first finger skeletal deformity was present in the patient who had sinus rhythm (Figure 1). Bilateral pulmonary hilar shadow was evaluated as significant in chest X-ray. Transthoracic echocardiography revealed, 32 mm secundum type atrial septal defect with a left to the right

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shunt, medium degree of tricuspid valve deficiency, right atrial and right ventricular dilatation and low degree of pulmonary hypertension. Good contraction function was detected in both ventricles. Operation preparations were made.

Pericardium was opened with a median sternotomy and pericardial patch was prepared. Cardiopulmonary bypass was initiated with bilateral selective venous and arterial cannulation from the ascending aorta. Antegrade blood cardioplegia was administrated from the aortic root after placing the aortic cross clamp. The operation was made in normothermia. After performing the right atriotomy, a wide atrial septal defect and the tricuspid valve were seen. 34 x 21 mm wide atrial septal defect with an inadequate inferior rim was closed with autologous pericardial patch treated with glutaraldehyde using 4-0 propilenesuture (Figure 2). Tricuspid valve leaflets were normal but there was annular dilatation. Saline test revealed a central deficiency was observed in the tricuspid valve. Tricuspid valve annulus Z value was detected as + 4.7. Annulus was narrowed between posteroseptal and anteroseptal commissures by De vega annuloplasty. After the repairmen, leaflet coaptation was increased and Z value decreased to + 1.8. When it was controlled by giving serum physiological to the right ventricle, deficiency was not observed in tricuspid valve. Cardiopulmonary bypass terminated under spontaneous sinus rhythm. Total perfusion time was 34 minutes and aortic cross clamp time was 23 minutes. The patient was extubated on the fourth postoperative hour of the intensive care follow-up. In the control transthoracic echocardiography taken on post-operative first day, the patch located on atrial septal defect was seen and no leakage was detected. Moreover, tricuspid valve deficiency or narrowness was not detected and tricuspid valve coaptation was evaluated as good. The patient was discharged on postoperative fifth day. Postoperative functional capacity improved to NYHA class 1.

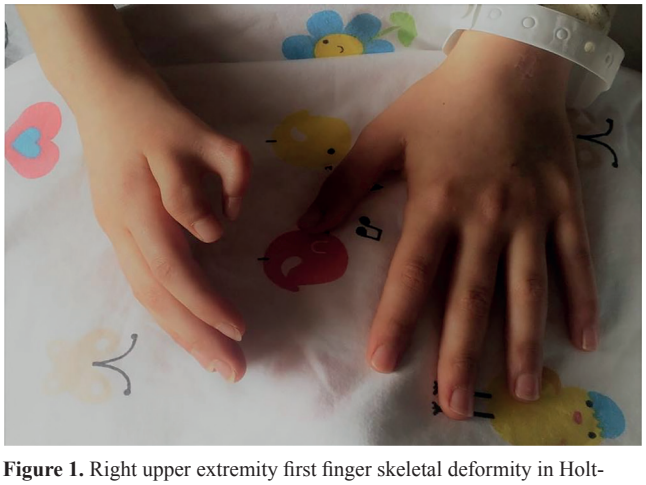

Figure 1. Right upper extremity first finger skeletal deformity in Holt-Oram syndrome.

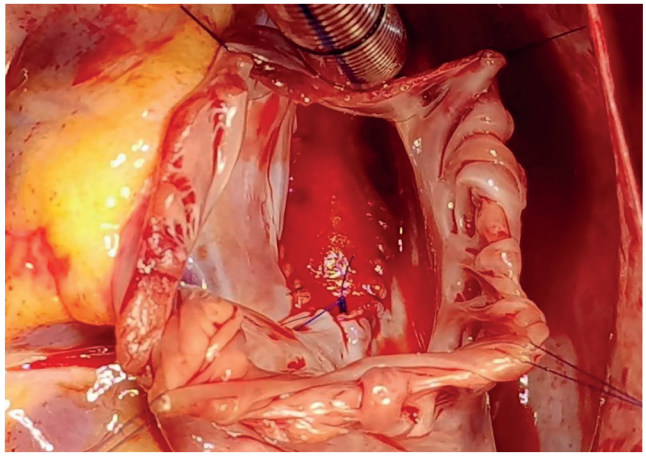

Figure 2. Operative image of wide atrial septal defect with an inadequate inferior rim covered with glutaraldehyde treated autologous pericardial patch.

DISCUSSION

Holt-Oram syndrome is a rare disease with autosomal dominant inheritance seen in 1 out of 100.000 births. Genetical defect was detected in the long arm of 12th chromosome of the patients. New mutations were also reported in addition to familial transition in literature[3]. We couldn't see the components of this syndrome when we searched the 1st degree relatives of our patient. We think that there may be a new mutation.

No relation was detected between the severities of cardiac defects and upper extremity skeletal anomalies in Holt-Oram syndrome[4]. In this syndrome, atrial septal defect among congenital cardiac defects and right hand first finger deformities among upper extremity skeletal anomalies are observed most commonly as in our case. But in addition to wide atrial septal defect, medium degree of tricuspid valve deficiency was observed in our case.

Tricuspid valve is the widest cardiac valve[5]. Dilatation can be observed in tricuspid annulus due to right ventricule widening and this condition may cause deficiency in tricuspid valve[6]. There was central deficiency in the tricuspid valve secondary to the tricuspid annulus dilatation in our case. De Vega defined a technique which can easily be applied for this kind of conditions and suggested annulus narrowing between anteroseptal commissure and tricuspid posteroseptal commissure where dilatation is most common[7]. In our case, we made a successful valve repairment by applying tricuspid valve annuloplasty defined by De Vega. We preferred 4-0 prolene stitch during this annuloplasty and used autologous pericardial pledget for durability in our first and last stitches. We also applied a controlled annuloplasty to prevent any narrowing in tricuspid valve.

In cases with Holt-Oram syndrome which is a rare hereditary disease, surgical cardiac repairment requirement changes according to the severity of cardiac defects. Successful cardiac valve repairment operations present great importance especially in congenital cardiac diseases.

In literature, we couldn't find concurrent tricuspid valve repairment during congenital cardiac defect closure in Holt-Oram syndrome which was presented in our case. We consider that multidisciplinary approaches are valuable in the diagnosis, follow-up and treatments of these rare syndromic diseases.

Informed Consent: Informed consent form was obtained from patient.

Peer-review: Externally peer-reviewed.

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Conflict of Interest: The authors have no conflicts of interest to declare

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