Rhabdomyoma Association With Atrioventricular Septal Defect in an Infant, A Rare Co-incidance

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Abstract

Cardiac rhabdomyoma is the most common primary heart tumor in childhood. This tumor, which is frequently associated with tuberous sclerosis complex, mostly disappears in childhood with spontaneous regression. Surgical resection is required in case of outflow obstruction, arrhythmia and protruding enough to disrupt the filling of the heart cavities. There are very few case series in the literature about rhabdomyom, whose relationship with other congenital heart defects has not been clearly demonstrated. In this case of tuberous sclerosis we presented to you, we reported our approach to the tumor during the corrective surgery of the infant, who was diagnosed with atrioventricular septal defect and patent ductus arteriosus, and rhabdomyoma accompanying these malformations.

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A Rare Co-incidance

Introduction

Cardiac rhabdomyoma is the most common primary heart tumor detected in infancy and childhood. The clinical course is generally benign. In studies conducted, spontaneous regression was observed in the majority of cases. The relationship of this tumor, which was first described in 1862, with tuberous sclerosis has been revealed as a result of researches[1]. Although it has a good clinical course, surgical resection is inevitable when it causes conditions such as ventricular outflow tract obstruction, myocardial dysfunction and malign arrhythmia.[2].

Despite many studies, very few congenital heart diseases have been reported in relation to cardiac rhabdomiomas. There are studies that argue that rhabdomyoma can cause any defect by causing a disruption in the embryological development steps of the heart. However, it is still controversial whether these two cardiac pathologies emerge as different pathologies [3] .

We report a rare case of tuberous sclerosis (TS) with cerebral and renal lesions, associated with cardiac rhabdomyoma and atrioventricular septal defect (AVSD).

Case Report

A girl, who referred to our clinic with symtoms of congestive heart failure, mild pulmonary hypertension (PAP : 30 mmHg) and resistant to medical treatment. The patient was 3 monts old and 3.800 grams weight. During the examination, S1 hard, S2 double, 3/6 pansystolic murmur was heard in the lower left sternal region. The electrocardiogram was a sinus rhythm at 150 beats /min and left axis deviation. Hemoglobin was 16.5 g/L and hematocrit was 52%. The patient with growt reterdation and tachipnea has oxygen saturation

95 % at room air. On chest x-ray, cardio-thoracic ratio was 0.6 and pulmonary plethora was seen. The patient has no signs of Down's syndrome.

In 2D transthoracic echocardiographic (TTE) examination, 7 mm primum ASD in the interatrial septum and 5 mm VSD in the interventricular septum were observed. It was observed that there was a second degree of insufficiency in the left atrioventricular (AV) valve. Intracardiac anomaly was named as AVSD transiotional type. At the base of the interventricular septum, close proximity to membraneus septum, a uniformly limited hyperechogenic mass of 1.5x1.0 cm was observed in the membranous septum. This mass minimally protruding into the cavitiy of right ventricle. **(Fig.1)** Doppler examination did not reveal any evidence of inflow or ventricular outflow tract obstruction. PDA was observed open and 6 mm in diameter. Cardiac mass seen by echocardiography was evaluated in more detail with multidetector computed tomography (MDCT). **(Fig.2)**

The patient who was considered rhabdomyom was examined for advanced tuberous sclerosis. With cerebral imaging, cortical tubers and subependimal nodules were observed. Multiple renal kists were detected by urinary ultrasonography. And after dermatological examination the diagnosis of tuberous sclerosis became definitive. The patients condition was discussed with pediatric cardiologists and surgical intervention is planned.

The patient was taken to the operation room. Following the median sternotomy, the thymus was excised subtotally and the pericardium was opened vertically. Arterial cannulation from ascending aorta and venous cannulations from both vena cava were performed. Double ligation and division of the ductus arteriosus was performed. The cardiopulmonary bypass (CPB) procedure was initiated. Cardioplegia was given from the aortic root and diastolic arrest was achieved. Right atiotomy was performed. Both primum ASD (7 mm) and secundum ASD (3mm) were observed in IAS. Left atrial vent was put through secundum ASD. Atrioventricular septal defect repair was done with using modified single patch tecnique. On the right ventricular side of the IVS, on the membranous septum, adjacent to the tricuspid valve anteroseptal comissures, well-circumscribed, shiny surface, primary mass was detected.**(Fig.3)** In addition, bright, scattered lesions of millimeter sizes were observed on the surface of the tricuspid valve. However, it was not excised because the primary lesion was rhabdomyoma, asymptomatic and did not cause functional and structural impairment. Additionally in the case of resection atrio-ventricular block risk was high. Subsequently, secundum ASD and right atriotomy was closed. Total CPB duration was 57 minutes and cross clamp time was 43 minutes. When CPB ended, the patient was in sinus rhythm. Dopamine supplementation at a dose of 5 mcg / kg / min was started for the patient who needed inotropic support in termination of CPB.

Inotropic support was discontinued at the 4th postoperative hour and sedation support was discontinued at the 6th hour and extubated at the 8th hour. The patient was stayed in the intensive care unit for 22 hours after extubation was taken to the inpatient ward. The patient was followed up for 3 days in the ward and discharged on the 4th day with healing. Control TTE was performed before discharge. It was observed that IAS and IVS were intact and no color transition. It was confirmed that the PDA was completely closed and there was no regurgitation of AV valves.

Discussion

Primary cardiac tumors in childhood are extremely rare and detected tumors with a rate of 0.08% in autopsies, and are mostly benign. The most common histologically seen infantile cardiac tumors are rhabdomyoma (60% -85%) and according to their incidence, teratoma, fibroma, hemangioma and hamartomas, respectively. [4-6]. In the last two decades, an increase in the incidence of pediatric cardiac tumors has been reported due to the increase in noninvasive imaging methods, the technological development of TTE devices and the widespread use of them. Patients admission complaints depend on the size and localization of the mass, as they have an inlet or outlet obstruction and arrhythmogenic effect [7]. In general, the indications for resection are to eliminate hemodynamic and respiratory pathologies, if any, severe arrhythmia and a significant risk of embolization. These tumors are highly associated (60-80%) with TSC. It originates from the free wall of the ventricle and ventricular septum. The diagnosis of rhabdomyom is based on the characteristic TTE fea-

tures of the tumor, in addition to the cutaneous, central nervous system or renal manifestations of tuberous sclerosis. In TTE examination, they are seen as well-circumscribed, homogeneous and echo bright masses. Many studies have shown that rhabdomyomas that are medically followed and partially resected will regress spontaneously within 1 to 3 years.

The patient we presented had AVSD and PDA accompanying rhabdomyoma. In the literature, the patient with congenital heart defect associated with TS and rhabdomyom is very rare. In the study published by Jiang et al.[3], they mentioned that the heart may disrupt the growth pattern in the region where the mass appeared during the embryological development phase. In our case, since the intrauterine cardiac examination was not performed, it cannot be clearly revealed in which trimester the rhabdomyoma occurred. However, the atrioventricular canal defect and the area where the rhabdomyom is located are very close, suggesting that the tumor may have caused the supposed development defect.

In the literature, it has been observed that the rhabdomyoma associated with the TSC is largely multiple. However, we observed a solitary rhabdomyom in our case. In the study conducted by Bader et al. with 26 patients, the tumors in diagnosis were single in 5 patients (19%) including right or left ventricle and multiple in 21 patients (80.8%)[8]. In a study conducted by Sciacca et al. with 33 patients, multiple rhabdomyomas were detected in all tuberous sclerosis patients [9]. It is well known that AV channel defects often coexist with down syndrome. However, in our case, the absence of down syndrome is also interesting.

We did not excise rhabdomyoma in our patient, since it was asymptomatic, was largely intramyocardial, and would cause destruction in the surrounding structures during excision. Malign arrhythmias that may develop in the postoperative period after excision could be fatal. After birth, rhabdomioma cells lose their ability to divide, and tumor regression in infancy is an expected result regardless of tumor size. In more than 80% of tumors, full regression can occur in early childhood. In addition, it has been revealed by many studies everolimus, an mTOR inhibitor, have shown that this drug accelerates shrinkage in the size of rhabdomiomas by up to 12 times [10,11].

Conclusion

In rare cases with congenital heart defects accompanying rhabdomyoma, we think that resection should not be preferred to preserve structural integrity if the tumor is asymptomatic. We are of the opinion that excision of this cardiac tumor, which already tends to spontaneous regression, may negatively affect surgical success during the repair stages of other congenital defects.

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Figures

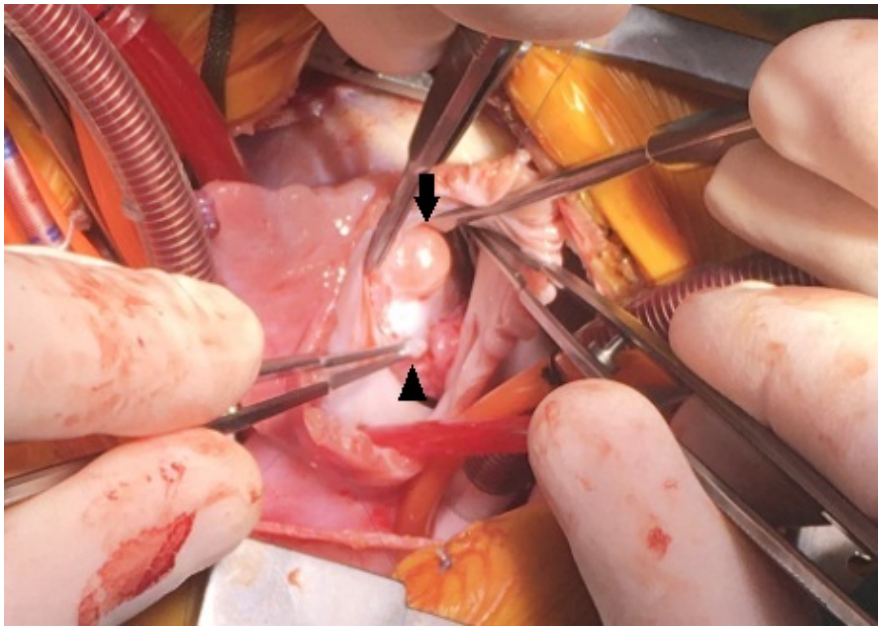

Figure.1 The appearance of rhabdomyoma through the right atriotomy (indicated by the blue arrow)

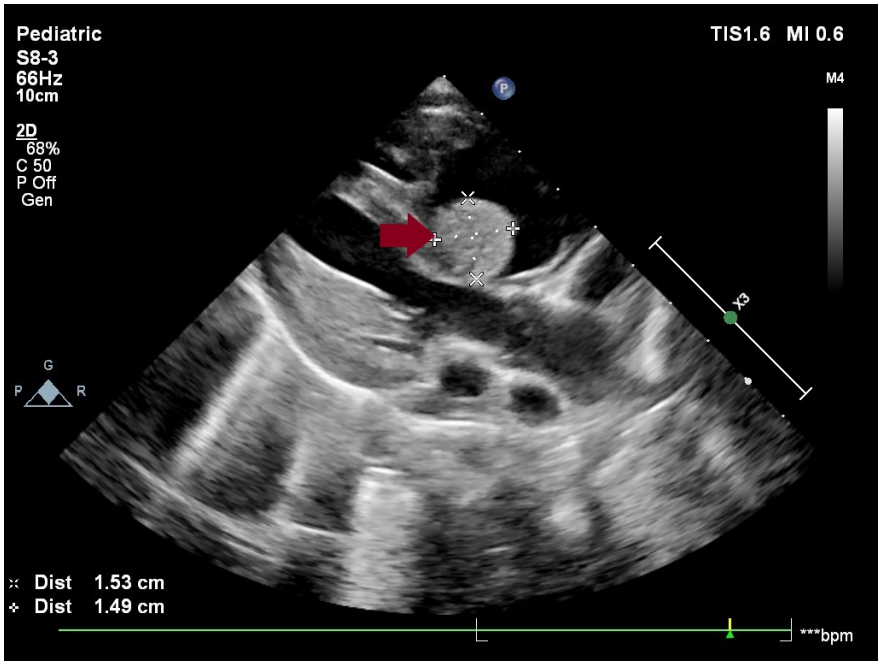

Figure.2 Modified parasternal long axis images of rhabdomyoma in TTE (indicated by the red arrow)

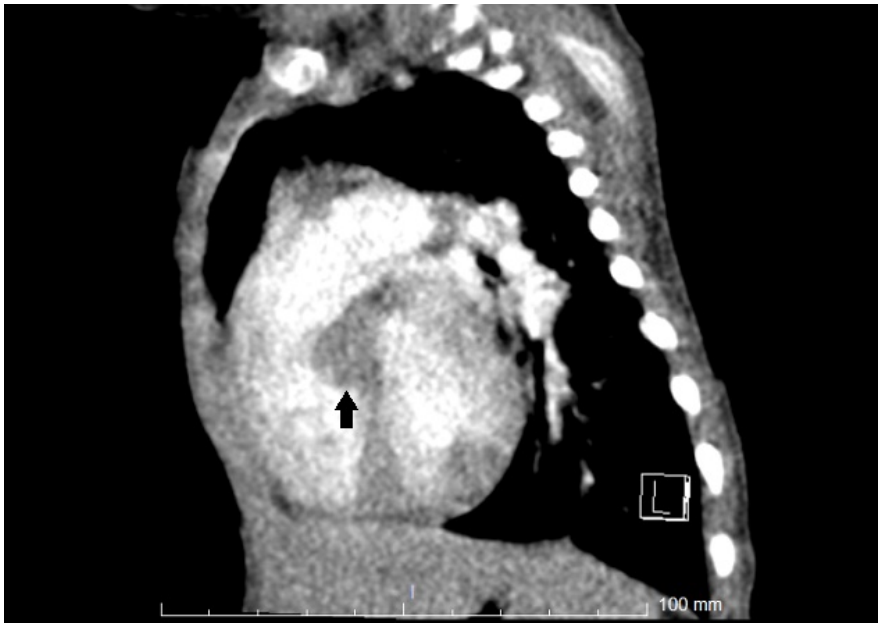

Figure.3 Vertical long axis reformatted multidetector computed tomography images of rhabdomyoma is well defined (indicated by the black arrow)

Abbreviations, acronyms & symbols

ASD : *Atrial septal defect*

AVSD : *Atrioventricular septal defect*

CPB : *Cardiopulmonary Bypass*

IVS : *Interventrcular septum*

MDCT : *Multidetector computed tomography*

PAP : *Pulmonary arterial pressure*

PDA : *Patent Ductus Arteriosus*

TS :*Tuberous sclerosis*

TSC : *Tuberous sclerosis Complex*

TTE : *Transthoracic Echocardiography*

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