CASE REPORT

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Successful Staged Surgical Repair of A Rare Association of Cardiac Rhabdomyoma, Tuberous Sclerosis and Tetralogy of Fallot

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ABSTRACT

We present a coexistence of cardiac rhabdomyoma, tuberous sclerosis and tetralogy of Fallot. A central shunt was emergently performed because of intractable cyanotic spells. After one-year of follow-up, total correction was performed as regression of the masses along with pulmonary artery growth was achieved. We suggest deferral of surgical total correction until regression of rhabdomyoma masses in these cases.

Keywords: Myocardial tumors; Rhabdomyoma; Tuberous sclerosis; Tetralogy of Fallot

Introduction

Although coexistence of cardiac rhabdomyoma and tuberous sclerosis has been well defined (1,2), the couple rarely accompanies with a congenital heart defect (CHD) (3,4). We aimed to present a very rare trilogy of a tetralogy of Fallot, tuberous sclerosis and cardiac rhabdomyoma in a young infant in whom an emergent surgical palliation was performed due to worsening cyanosis.

Case Report

An 85-day-old female baby weighing 5.5 kg was referred to our hospital due to cyanosis (SpO_2: 80%, on room air) and cardiac murmur. Along with 3/6 systolic ejection murmur on left upper sternal border, her physical examination revealed hypopigmented macular skin lesions (Figure 1A). Through transthoracic echocardiography, she was diagnosed with tetralogy of Fallot with a combined infundibular and valvar stenosis and a systolic gradient of 80 mmHg. There was pulmonary artery hypoplasia with a McGoon index of 1.4. She had multiple hyperechogenic cardiac masses which were not causing rhythm disturbance, ventricular dysfunction or any obstruction. They were mostly located at the right ventricular apical cavity and left ventricular outflow tract without causing obstruction (Figure 1B, C). The spot electrocardiogram and 24 hours Holter electrocardiogram were normal. With combined hypopigmented skin lesions and cardiac masses, the patient was suspected to have tuberous sclerosis. Further evaluation through brain magnetic resonance imaging revealed multiple tubers with subependymal nodules (Figure 1D) indicative of tuberous sclerosis, so the masses were concluded as cardiac rhabdomyoma.

As the patient had multiple episodes of hypoxic spell refractory to multi-drug medication (the lowest sO_2 being 30%) during intensive care unit follow-up, an emergent surgical palliation was decided. Through median sternotomy, cardiopulmonary bypass was initiated under normothermia, and a modified Waterston shunt between the ascending aorta and the right pulmonary artery was performed using a 3.5 mm

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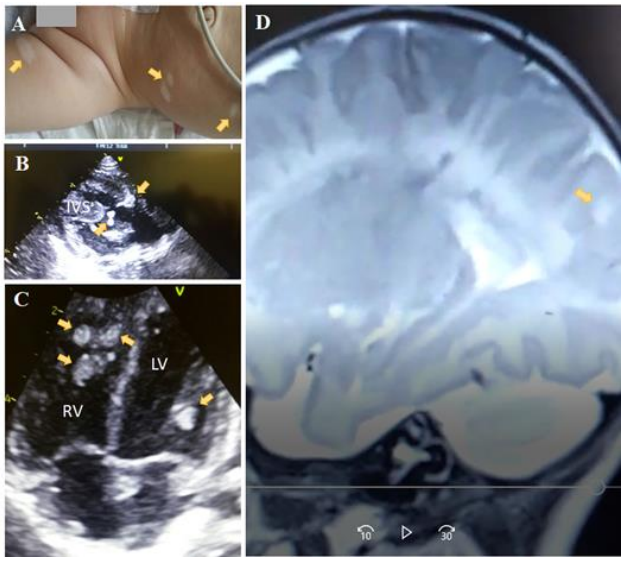

Fig. 1. A. Hypopigmented macular cutaneous lesions. 2D transthoracic echocardiographic evaluation through apical four-chamber (B) and parasternal long axis (C) views revealing multiple hyperechogenic masses indicative of rhabdomyoma. D. Magnetic resonance imaging of the brain (T2) revealing an occipital tuber (arrow) indicative of tuberous sclerosis.

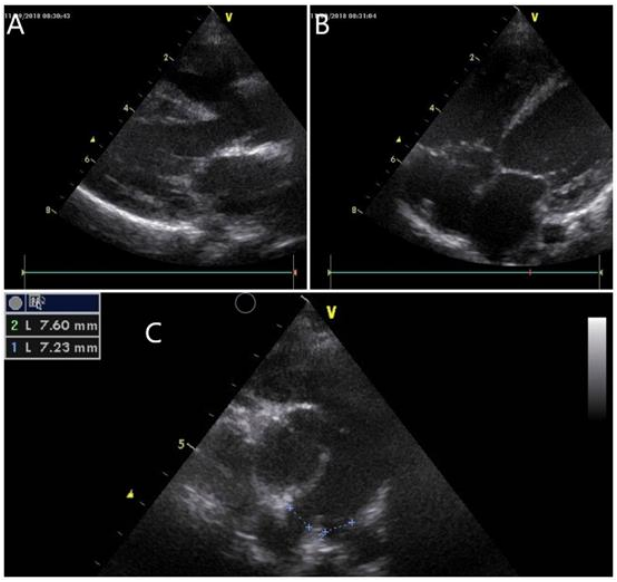

Fig. 2. Echocardiographic evaluation on 1-year age, revealing almost completely regressed cardiac rhabdomyoma masses (A,B) and increased pulmonary artery size (C).

polytetrafluoroethylene tube graft. The termination of the cardiopulmonary bypass was uneventful under mild inotropic support with the sO_2 of 80% under 40% of fraction of inspired oxygen. The postoperative course was uneventful. The patient was discharged on postoperative day 12 in good hemodynamic condition (sO_2 %85 in room air).

A heterozygous p.Arg611Trp change (c.1831 C>T) on exon 16 of TSC-2 gene was determined. The shunt was found to be effective and a fair amount of pulmonary artery growth was achieved (Figure 2A) with McGoon index of 1.8 on follow-up echocardiograms and no cyanotic spells occurred. The intra-cardiac rhabdomyomas were found to be almost completely regressed (Figure 2B, C) by the first year of age. Complete repair was performed when she was 13-month-old.

Discussion

Although the coexistence of cardiac rhabdomyomas with tuberous sclerosis has been well defined (1, 2), they rarely coexist with a CHD (3). In an asymptomatic neonate, conservative follow-up is recommended because many of the rhabdomyomas regress over time (2, 3). On the other hand, timing of complete repair in these patients is challenging as for surgery should inevitably be performed when there is inflow or outflow obstruction (2). The symptoms of rhabdomyoma mainly depends on the size and location of the tumor (2, 4). The rhabdomyoma masses in our patient were multiple but were not causing ventricular outflow obstruction. In fact, the hypoxic spell episodes and the pulmonary artery hypoplasia interfered with the performance of complete repair. For these reasons, a central shunt was performed to enable the growth of the pulmonary arteries and regression of the masses.

Being the most common form of intracardiac tumor in fetal life, the cardiac rhabdomyomas can be detected in-utero by ultrasonography. Cardiac rhabdomyomas completely regressed in our patient compatible with the literature (2, 3).

Coexistence of this trilogy is very rare. Apart from causing ventricular inflow or outflow obstruction, deferral of total surgical repair until sufficient growing of the pulmonary arteries and regression of the cardiac masses achieved, seems to be a good choice in these patients, which allows ease of surgical total correction with successful outcome.

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Conflict of Interest: None.

Ethical Standards: The authors assert that all procedures contributing to this work comply with the ethical standards of the relevant national guidelines on human experimentation and with the Helsinki Declaration of 1975, as revised in 2008.

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