A rare association of tetralogy of Fallot and hypertrophic cardiomyopathy

Kang-Hong Hsu and Chung-I Chang*

Department of Cardiovascular Surgery, National Taiwan University Hospital, Taipei, Taiwan

* Corresponding author. Department of Cardiovascular Surgery, National Taiwan University Hospital, 7 Chung-Shan S. Rd., Taipei, Taiwan. Tel: +886-2-23123456; fax: +886-2-23934358; e-mail: joey@ntuh.gov.tw (C.-I. Chang).

Received 4 September 2011; received in revised form 6 November 2011; accepted 14 November 2011

Abstract

Tetralogy of Fallot (TOF) is the most common form of cyanotic congenital heart disease. It is often associated with other congenital cardiac or non-cardiac defects. However, its association with hypertrophic cardiomyopathy (HCM) is rarely reported. We reported two cases. The first case is a full-term girl receiving modified Blalock–Taussig shunt creation and the second case is an 8-month old boy receiving total correction for TOF. Although they tolerated the operation well, both of them developed congestive heart failure and died of malignant cardiac arrhythmia several months after the operation. We made a literature review and only 11 case reports were found. There is currently no treatment guideline for this group of patients. From our limited experience and case reports, the physiology of HCM and TOF should both be taken into consideration when managing these patients. Close echocardiography follow-up with early myectomy and preventive implantation of implantable cardioverter-defibrillator may be beneficial for them.

Keywords: Tetralogy of Fallot • Hypertrophic cardiomyopathy

INTRODUCTION

Tetralogy of Fallot (TOF) is the most common cyanotic congenital heart disease and is often associated with other congenital defects. On the other hand, hypertrophic cardiomyopathy (HCM) is a genetic disorder that is typically inherited in an autosomal dominant fashion. It is rarely associated with other congenital anomalies but carries a high incidence of sudden cardiac death. We reported two cases and briefly reviewed the literature.

CASE REPORT

Patient 1

A full-term girl weighing 3100 g was born after an uncomplicated pregnancy. There was no family history of cardiac disease from her parents. Cyanosis with poor activities developed soon after birth. Echocardiography revealed TOF with severe infundibular and valvular pulmonary stenosis (pressure gradient ≏ 40 mmHg). HCM was diagnosed with asymmetric left ventricular (LV) hypertrophy over the interventricular septum (IVS). The end-diastole septal thickness was 8 mm (Z-score = 14.44) without LV outflow tract obstruction (LVOTO). The pulmonary arteries were hypoplastic with a McGoon Index of 1.05. A modified Blalock–Taussig shunt was created when she was 5-days old. However, congestive heart failure (CHF) progressed after operation such as tachypnea, feeding intolerance and poor body weight gain. The follow-up echocardiography showed progressive hypertrophy of the IVS from 8 to 16 mm within 2 months. LVOTO also developed with pressure gradient raised from 15 to 43 mmHg. Oral propranolol 2 mg three times per day was prescribed with limited improvement. When she was 10-months old, she suffered from refractory pulseless ventricular tachycardia and was resuscitated with extracorporeal membrane oxygenation (ECMO). Unfortunately, she died of multiple organ failure 5 days later.

Patient 2

A full-term boy was born after an uncomplicated gestation. There was no family history of cardiac disease from his parents. TOF was diagnosed soon after birth without apparent cyanosis and propranolol was prescribed. When he was 5-months old, he was transferred to our hospital because of heart failure and frequent blue spells. DiGeorge syndrome was suspected but his parents refused further genetic study. Echocardiography showed TOF with severe infundibular pulmonary stenosis. The McGoon index was 2.6. HCM was diagnosed by echocardiography with concentric LV hypertrophy. IVS thickness was 14 mm (Z-score = 16.87) without LVOTO (Fig. 1a). The patient received total correction (TC) for TOF. We identified more severe right ventricle hypertrophy during operation (Fig. 1b). The pathology showed disarray of myocardial fibres with hypertrophy, which was compatible with HCM (Fig. 1c). During follow-up, CHF progressed with feeding intolerance, poor body weight gain and hepatomegaly. He was admitted 4 months later because of pneumonia. Refractory ventricular tachycardia and fibrillation developed and he died one day after admission because his family refused further intervention (operation or ECMO).
From 1978 to 2008, there were 11 cases reported [1–10] (Table 1). This rare association appears more often in males (nine boys and two girls). They were usually diagnosed in infancy with presentation of cyanosis and CHF. Seven patients (64%) were reported to have other congenital anomalies. Among the nine patients receiving operation, four patients received TC (two primary repair and two staged operation), four patients received palliative procedures and one operation unspecified by the authors. There was only one patient receiving LV myomectomy 8 years after TC due to progressive LVOTO [1].

Five of the nine patients (55%) receiving surgery died of either early or late cardiac failure.

Although several chromosomal aberrations had been observed in TOF, it is rarely a familial disorder. On the other hand, HCM is a genetic disease of the cardiac sacromere and rarely associated with other cardiac defects. More than 400 mutations of 11 different genes have been identified. Lewin et al. [2] first proposed the speculation about the possible genetic causes and reported two cases. In the animal study done by Kuribayashi et al. [3] with WKY/NCrim rats, they proposed the occurrence of TOF, pulmonary valve stenosis, VSD and HCM, isolated or in association, are genetically linked.

When planning surgical repair, there are several aspects that the surgeon should take into consideration. First, is the LV capable of pumping the increased pulmonary venous return after the repair? In HCM, the LV volume is decreased with diastolic dysfunction. After either palliative procedure or TC, it is questionable whether LV can adapt to the sudden increase of pulmonary venous return. Second, it is very rare to have LVOTO before TC because the aorta overrides the conoventricular VSD. But in patients with severe LV hypertrophy, the repair of VSD may create a new LVOTO. Therefore, intra-operative transesophageal echocardiography is very important to detect any evidence of new LVOTO. Third, with severe hypertrophy of both ventricles, myocardial protection must be carried out delicately to prevent any myocardial injury. When the above conditions occur, there may be a problem in weaning off cardiopulmonary bypass, or the patient could experience low cardiac output status post-operatively. Finally, the impaired ventricular diastolic function in HCM could result in increased left ventricle end diastolic pressure and subsequently elevated pulmonary artery pressure. This will exacerbate pulmonary regurgitation and cause right ventricular (RV) failure, especially after transannular patch repair. Therefore, surgeons should be more aware of the pulmonary valve function when performing TC. In both our patients, the operation was tolerated well. Neither post-operative LV dysfunction nor LVOTO was observed. However, LVOTO and ventricular failure gradually developed months later and may have caused the late death.

There is currently no guideline for managing this rare association. From the literature, we find that surgeons usually make their decisions according to the physiology of TOF. However, the mortality was high (55%) possibly due to the progression of HCM and heart failure. Because there was no long-term follow-up result available, it is inappropriate to make any premature conclusion on how to treat these patients or to improve the surgical outcome. We believe that we should take HCM into consideration when managing these patients. More competent pulmonary valve with less regurgitation may prevent RV failure and late cardiac arrhythmia. Close follow up with echocardiography and early myectomy may be indicated. Preventive use of implantable cardioverter-defibrillator may be considered to prevent sudden cardiac death.
Conflict of interest: none declared.

REFERENCES


