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Brief Report

An obstructive bicuspid aortic valve in the setting of tetralogy of Fallot with pulmonary atresia: a rare combination

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Abstract As far as we are aware, a bicuspid aortic valve has not previously been reported in the setting of tetralogy of Fallot and pulmonary atresia. We describe this association in a newborn who presented with a murmur and cyanosis. Echocardiography showed tetralogy of Fallot with pulmonary atresia, and also a moderately stenotic bicuspid aortic valve. The patient underwent open-heart surgery guided by transesophageal echocardiography. Postoperatively, there was only mild obstruction across both outflow tracts. We have also reviewed the pertinent data from our Jesse E. Edwards Registry of Cardiovascular Disease to establish the incidence of bicuspid aortic valve in the setting of obstruction of the right ventricular outflow tract, finding the association in no patients with pulmonary atresia and tetralogy of Fallot, in 0.7% of those with tetralogy of Fallot and pulmonary stenosis, but in 6.6% of those with pulmonary atresia and intact ventricular septum.

Keywords: Pulmonary atresia with ventricular septal defect; pulmonary atresia with intact ventricular septum; congenital cardiac malformations

THE BICUSPID ARRANGEMENT OF THE AORTIC valve is a congenital malformation that is seen relatively commonly, occurring in about 1% of the general population.[1] It occurs three to five times more frequently in males than in females.[2] In tetralogy of Fallot, however, a bicuspid aortic valve is a rare finding[3] and, to our knowledge, has never been described when tetralogy is associated with pulmonary atresia rather than pulmonary stenosis. We report here a child with atresia of the pulmonary valve and moderate stenosis of a bicuspid aortic valve in the setting of tetralogy of Fallot. Despite obstruction of both the outflow tracts, the patient was stable after birth, and the lesions were reparable surgically.

Since the finding was rare, we analyzed the data from our Jesse E. Edwards Registry of Cardiovascular Disease on the incidence of this phenomenon, and found no similar cases. In the Registry, nonetheless, we also found 428 cases of tetralogy of Fallot with pulmonary stenosis rather than atresia, and 122 cases of pulmonary atresia with an intact ventricular septum, so we established the incidence of bicuspid aortic valve in both of these settings.

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Case report

A 2-day-old female, born at 38 weeks of gestation to a healthy mother, presented with a loud systolic ejection murmur at the right upper sternal border with a precordial thrill and cyanosis. The patient showed no dysmorphism and was clinically stable in room air, with transcutaneous saturations via pulse oximetry in the 80s. There had been no significant perinatal complications, and there was no family history of congenital cardiac disease. The patient was commenced on prostaglandin E1, given intravenously.

An electrocardiogram showed sinus tachycardia with right atrial and right ventricular enlargement. The transthoracic echocardiogram demonstrated a large ventricular septal defect overridden by the aortic valve, with preservation of the fibrous continuity between the leaflets of the aortic and the mitral valves. The aortic arch was patent and left-sided. The aortic

valve, however, had two leaflets, with both leaflets being thickened and showing restricted motion in systole, producing moderate obstruction across the aortic outflow, with a mean gradient of 40 mmHg. There was no aortic insufficiency. The anatomy of the proximal right and left coronary arteries was normal. There was a moderate degree of subinfundibular hypertrophy, and the pulmonary valve was atretic, with no antegrade flow across it. The pulmonary trunk could not be identified using transthoracic echocardiography. A large aortopulmonary collateral artery was noted arising from the descending thoracic aorta. The ventricular systolic function was qualitatively normal. A small atrial septal defect was also found, but no patent arterial duct was demonstrated on the echocardiogram. The heart was imaged via a transesophageal echocardiogram in the perioperative period, which confirmed the bicuspid nature of the aortic valve (Fig. 1). An aortic angiogram prior to the surgery showed one major aorto-pulmonary collateral artery from the descending thoracic aorta that divided immediately into 2 branches (Fig. 2). The angiogram also revealed retrograde filling of diminutive confluent pulmonary arteries (Fig. 2).

The patient underwent cardiac surgery, with successful reconstruction of the pulmonary trunk, excision of obstructing right ventricular musculature, and placement of a transannular pericardial patch to establish continuity between the right ventricle and the pulmonary arteries. The confluent pulmonary arteries were unifocalized with the major aorto-pulmonary collateral arteries. The obstruction across the stenotic aortic valve was relieved surgically, the mean transaortic gradient decreasing to 24 mmHg with a qualitatively normal left ventricular systolic function and residual trivial aortic insufficiency. The ventricular septal defect was left to be repaired at a later stage.

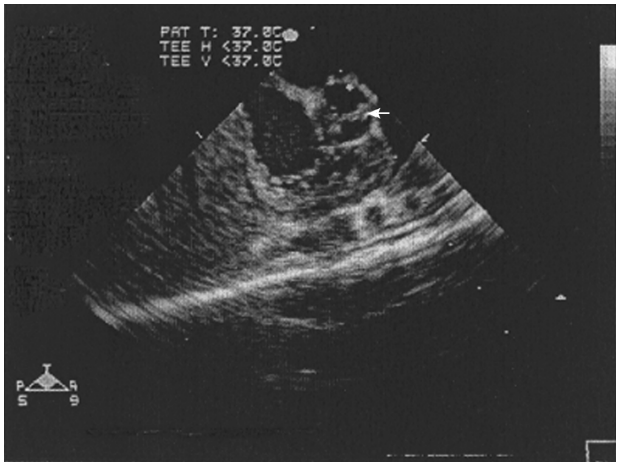

Figure 1.
Transesophageal echocardiogram showing the short axis view of the bicuspid aortic valve in diastole. Note the thickened leaflets (arrow). A raphe in the conjoined leaflets can also be seen.

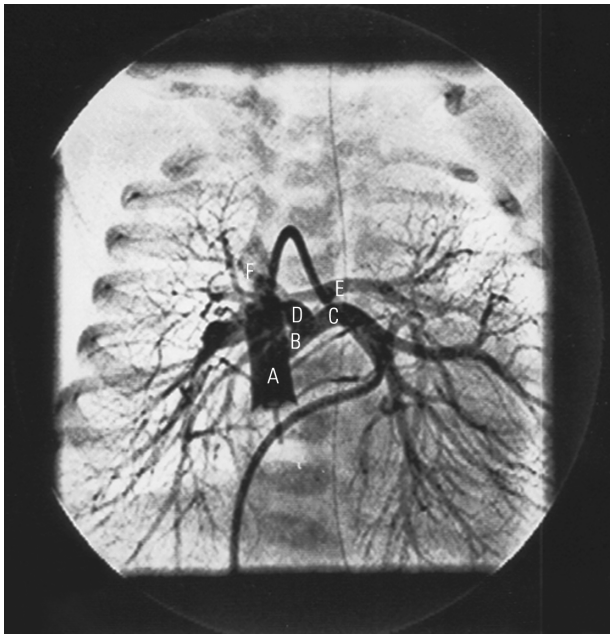

Figure 2.
An angiogram of the thoracic aorta (A) shows an aortopulmonary collateral artery (B) arising from the aorta, and its left (C) and right (D) branches. Note the retrograde filling of the confluent left (E) and right (F) pulmonary arteries.

Discussion

The association of a bicuspid aortic valve with tetralogy of Fallot in the setting of pulmonary stenosis is a rare finding.[3,4] Here, we report the presence of a bicuspid aortic valve in tetralogy of Fallot with pulmonary atresia. To our knowledge, this combination of anomalies has not previously been described.

In the Jesse E. Edwards Cardiovascular Registry of Cardiovascular Disease, of the 428 cases of tetralogy of Fallot without pulmonary atresia, three (0.7%) had a bicuspid aortic valve. In the Registry, we did not find any association of the bicuspid valve with tetralogy and pulmonary atresia. In contrast, the bicuspid aortic valve has been found more commonly in the setting of pulmonary atresia with an intact interventricular septum,[5] a finding confirmed by its presence in 8 of 122 cases (6.6%) with this malformation in our Registry. Thus, a bicuspid aortic valve can occur along with tetralogy of Fallot with pulmonary atresia, albeit exceedingly rarely, and can be treated successfully by surgery.

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