Absence of an Aortic Valve Cusp, A Cause of Severe Aortic Regurgitation in Infancy

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SUMMARY. Congenital aortic regurgitation is an extremely rare cardiac lesion. This is a case report of an infant with a severe degree of valve regurgitation due to absence of the noncoronary aortic cusp. We show the echocardiographic, angiographic, and surgical aspects of the case, emphasizing the importance of a precise anatomical diagnosis and the problems of early valve replacement.

KEY WORDS: Congenital aortic regurgitation — Echocardiography — Angiography

Congenital aortic regurgitation in infancy is usually a consequence of primary malformation of the aortic root or aortic valve. Congenital malformation of the aortic root may involve either aneurysmal dilatation of the sinuses of Valsalva [2] or the presence of an aortic-left ventricular “tunnel” [4]. Primary valve lesions causing congenital regurgitation are extremely rare. Severe congenital aortic regurgitation in early infancy has been reported with absence of all three aortic cusps [1] and in older patients with absence of a single cusp [3]. This report describes the clinical, echocardiographic, angiographic, and surgical findings in an infant with severe congenital aortic regurgitation due to absence of one semilunar aortic cusp.

Case Report

The patient was an 18-month-old male child known to be in heart failure since the age of 6 months, referred with failure to thrive, dyspnea, diaphoresis, bounding pulses, a to and fro murmur, and refractory to digitalis, diuretics, and vasodilators. The electrocardiogram demonstrated a sinus tachycardia with left ventricular hypertrophy and diffuse ST-T wave changes. Cardiomegaly with dilatation of the ascending aorta was apparent on the chest X-ray. The two-dimensional echocardiogram revealed left ventricular dilation with hypertrophy. The aortic valve had two thickened cusps of unequal size with a large “fenestration” between two comissures. Pulsed Doppler interrogation of the left ventricle and aorta suggested severe aortic valve regurgitation (Fig. 1). Aortic angiography demonstrated severe regurgitation across a thickened aortic valve with marked dilatation of the ascending aorta. The innominate artery had its origin more leftward than usual. The left ventricle was hypertrophied with moderate dilatation and apical hypokinesis. A right ventricular angiogram revealed isolated stenosis of the left pulmonary artery (Fig. 2). The infant was referred for surgical management due to refractory heart failure. Surgical exploration revealed a rudimentary noncoronary cusp with a serrated margin with dysplasia of the remaining leaflets. Absence of the semilunar cusp prevented apposition of the left and right cusps, causing intense aortic regurgitation (Fig. 3). The aortic valve was replaced with an Omniscin 19 mechanical prosthesis following anterior augmentation of the annulus. The infant expired 18 h following surgery in low cardiac output. Autopsy was not available but histology of the valve revealed fibrosis and myxoid degeneration of the stroma in the native valve.

Discussion

Absence of one semilunar cusp should be considered in the differential diagnosis of isolated congenital aortic regurgitation. It is of special interest that in our case the symptoms and signs of severe aortic regurgitation occurred very early in infancy, contrary to what is mentioned in the few cases described in the literature, in which symptoms developed in childhood. This fact is of paramount importance because aortic valve replacement in infancy is a high risk situation, and often one needs to enlarge the aortic annulus so that it can accommodate a prosthesis. Also, severe left ventricular hypertrophy combined with aortic regurgitation compromises the effectiveness of the cardioplegic...
**Fig. 1.** Echocardiographic subxyphoid long axis projection displaying the left ventricular outflow tract in systole (upper left) and diastole (upper right). Parasternal short axis view of the aortic valve (lower left) demonstrating separation of the aortic cusps (arrow) with aortic regurgitation apparent on pulsed Doppler examination (lower right). Ao, aorta; AR, aortic regurgitation; LCA, left coronary artery; LV, left ventricle.

**Fig. 2.** Aortography in diastole showing massive regurgitation and thickened leaflets (upper left). Right ventriculography with moderate stenosis at origin of left pulmonary artery; the right pulmonary artery is normal (upper right) left ventricular angiogram in systole showing diffuse hypokinesis (lower left). High aortic root injection demonstrating a massively enlarged arch with the innominate artery arising more leftward than usual. Ao, aorta; AoV, aortic valve; IA, innominate artery; LV, left ventricle; MPA, main pulmonary artery; RV, right ventricle.

**Fig. 3.** Surgical view of the dysplastic aortic valve before its removal. There is a large posterior gap (asterisk) corresponding to the absence of the noncoronary cusp; the other two cusps show poor coaptation.