An Unusual Association of Supra-annular Mitral Ring

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ABSTRACT

Supra-annular mitral ring is a very rare developmental anomaly of the supra-valvular area of the mitral valve with less than 100 cases reported in the literature [1,2]. Although, rarely it can be an isolated anomaly, in 90% of patients it is associated with other cardiac lesions [2,3,4]. Its coexistence is important to identify because it can alter the hemodynamics and surgical outcome significantly. We report for the first time an unusual association of supra-annular mitral ring and complete atrio-ventricular canal (endocardial cushion defect) in a 14-year-old boy with Marfan syndrome.

INTRODUCTION

Supra-annular mitral ring is a very rare developmental anomaly of the supra-valvular area of the mitral valve with less than 100 cases reported in the literature [1,2]. Although, rarely it can be an isolated anomaly, in 90% of patients it is associated with other cardiac lesions [2,3,4]. Its coexistence is important to identify because it can alter the hemodynamics and surgical outcome significantly. Marfan syndrome is a well recognised autosomal dominant disorder of connective tissue, commonly associated with cardiac lesions [5]. We report a 14-year-old boy with Marfan syndrome in whom the diagnosis of complete atrio-ventricular canal with supra-annular mitral ring was established by echocardiography. Such an association is not reported previously.

CASE REPORT

A 14-year-old boy, born of a nonconsanguineous marriage, presented to us with dyspnoea on exertion. His height was 174 cm (> 97th percentile), weight 48 Kg (50th percentile) and arm span of 180 cm. His arm span exceeded his height by 3%. He also had other characteristics of Marfan syndrome including decreased upper segment to lower segment ratio (0.93), the wrist (Walker-Murdoch) sign, thumb (Steinberg) sign, arachnodactyly, pectus excavatum, scoliosis, reduced extension of the elbows (approximately 150 degrees), high arched palate and stretch marks on the lower back. The eye examination was normal. The cardiovascular system examination revealed a diastolic gradient of 28 mm hg. There was severe atrio-ventricular valve regurgitation with a predicted right ventricular systolic pressure of 110 mm hg.

According to the Revised Ghent Nosology 2010, this case fulfilled the criteria for Marfan syndrome. His parents did not afford genetic study to demonstrate FBN1 mutation. The complete diagnosis was Marfan syndrome with complete atrio-ventricular canal with supra-annular mitral ring with pulmonary hypertension. We prescribed oral lasilactone, digoxin and enalapril. Clinical examination and echocardiography of parents and younger sibling did not reveal abnormality.

DISCUSSION

Supra-annular mitral ring was first described by Fisher in 1904 as a ridge of connective tissue above the mitral valve like a diaphragm of the microscope [6]. It is characterised by a crescentic shelf like structure, located in close proximity of mitral valve, often attached to the annulus. This shelf results in obstruction to the left ventricular inflow at that level, behaving like a stenosing ring. Consequently, this leads to elevated pulmonary venous pressure and pulmonary arterial hypertension. Hence, clinically children present with heart failure and failure to thrive.

Brown et al [3] reported associated cardiac defects in 92% of patients of supra-annular mitral ring, commonenest being ventricular septal defect (including tetralogy of Fallot), sub-aortic membrane, bicuspid aortic valve and coarctation of aorta (Shone’s anomaly). Atrio-ventricular canal defect accounts for about 5% of all congenital heart defects [7]. It is commonly seen in patients with chromosomal anomalies, notably trisomy 21, where concomitant congenital lesions, including Fallot’s tetrology, tricuspid atresia, coarctation of aorta and heterotaxia, are known to occur. The presence of supra-annular mitral ring with complete atrio-ventricular canal is most unusual and has not been reported previously.

The embryological origin of the supra-annular mitral ring is unclear. It has been postulated that, it could be secondary to
a developmental anomaly of the atrio-ventricular canal and that defective division of the endocardial cushion results in a fibrous band that forms the supra-annular ring [8]. Thus, embryologically it is possible that an endocardial cushion defect (such as complete atrio-ventricular canal) can result in formation of supra-annular mitral ring.

The incidence of Marfan syndrome is 2-3 / 10000 individuals [5]. It is an autosomal dominant disorder with nearly complete penetrance with variable expressivity. Cardiovascular abnormalities occur in 80-100% of these patients and commonly include aortic root dilatation, pulmonary artery dilatation, mitral valve prolapse, mitral regurgitation, descending aorta dilatation, left ventricular dysfunction, arrhythmia and atrial septal defect in decreasing frequency. Hence, while exact genetic mechanism is lacking presently, the combination of complete atrio-ventricular canal and supra-annular mitral ring in Marfan syndrome is more likely inherited disorder with a common genetic mechanism than a rare sporadic event. More reports of such an association in future would indicate a strong correlation.

Fig.1 (a) Apical four-chamber view still frame demonstrating complete atrio-ventricular canal and a discrete circumferential membrane (thick arrow). (b) Complete atrio-ventricular canal with bridging leaflet (thin arrow) and a discrete circumferential membrane (thick arrow) attached to the left atrial wall and the left lateral annulus. (c) Parasternal long-axis view still frame demonstrating a discrete circumferential membrane with a central orifice (thick arrow).

Fig.2 Color flow still frame demonstrating turbulent flow across the central orifice (thick arrow) of the membrane.

REFERENCES