Research Paper Medical Science

# An Unusual Association of Supra- annular Mitral Ring

**Mandar Bhausaheb Patil**

DNB (Ped) Asst. Professor, Division of Pediatric Cardiology, Department of Pediatrics, D.Y. Patil Medical College, Kolhapur, Maharashtra, India.

**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. Its coexistence with other cardiac lesions 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.

**KEYWORDS** supra- annular mitral ring; complete atrio- ventricular canal; Marfan syndrome; association

## 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 (> 97$^{th}$ percentile), weight 48 Kg (50$^{th}$ 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 excavitum, 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 pulse rate of 85/ min with normal peripheral pulses, central cyanosis, grade two clubbing, precordial bulge with a prominent right ventricular impulse, normally audible first heart sound and an accentuated pulmonary component of the second heart sound, a grade 3/6 harsh holo-systolic murmur at the apex and low pitched mid- diastolic rumbling murmur at the lower left sternal edge. The liver span was 6 cm. The lung fields were clear.

The chest X- ray showed cardiomegaly with a prominent right ventricle and right atrium, hilar pulmonary artery prominence, increased pulmonary vascularity and Kerley lines. ECG revealed bi- ventricular hypertrophy. Trans- thoracic echocardiography demonstrated situs solitus, levocardia and atrio- ventricular and ventriculo- arterial concordance. It revealed complete atrio- ventricular canal consisting of contiguous atrial and ventricular septal defects and a single atrio- ventricular valve common to both the ventricles with superior bridging leaflet (Fig. 1a & b). The color flow study demonstrated a right to left shunt across the defect and severe atrio- ventricular valve regurgitation. In addition, a discrete circumferential membrane with a single central orifice was visualised superior to the atrio- ventricular valve and attached to left atrial wall and the left lateral annulus of the atrio- ventricular valve (Fig. 1a,b,c). There was turbulence of flow across the membrane at the orifice indicating obstruction to the left ventricular inflow (Fig. 2). The left atrial appendage and the opening of the pulmonary veins was visualised above the level of the membrane. The ascending aorta was dilated with aortic root diameter at the sinus of valsalva= 3.3 cm (Z score of 2.95). There was no aortic regurgitation and the aortic arch and descending aorta were normal. The color Doppler examination confirmed obstructed flow across the membrane as evidenced by a continuous flow with 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 sib 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, commonest 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 tetrolgy, 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, arrthymia 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 an 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.

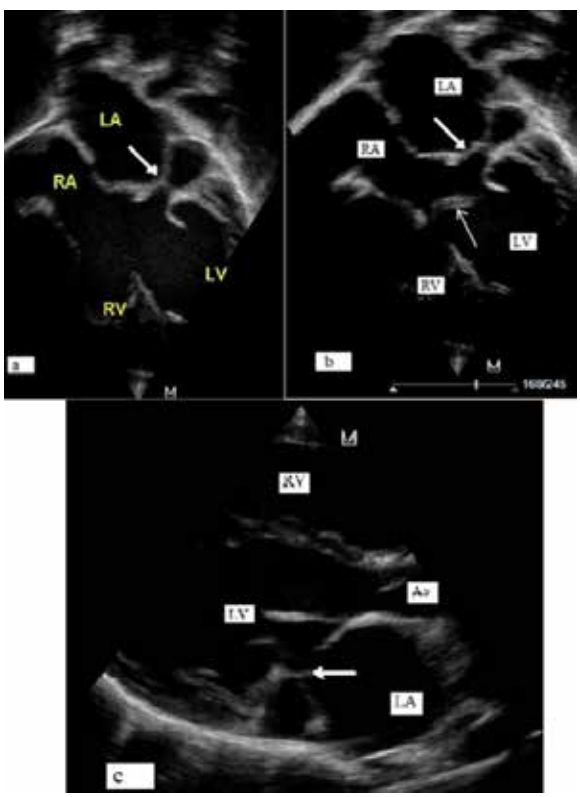


**Fig.1** (a) Apical four- chamber view still frame demonstrating complete atrio- ventricular canal and a discrete circumferential membrane (thick arrow). (b) Complete atrio- ventricular canal with bridging leaflet (thin arrow) and a discrete circumferential membrane (thick arrow) attached to the left 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).

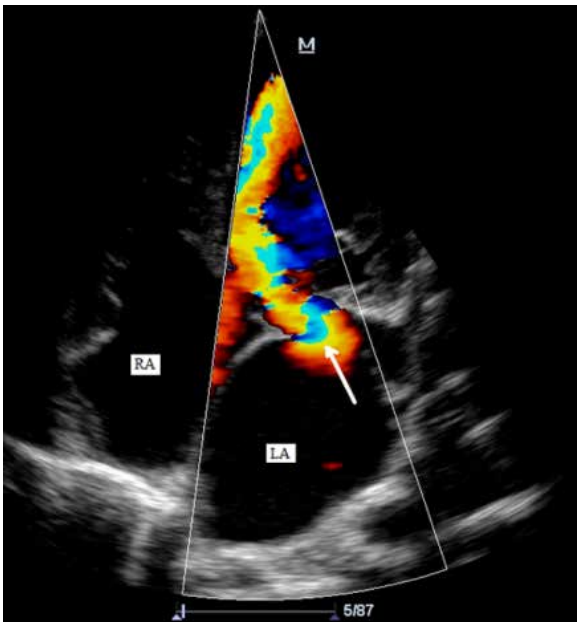


**Fig.2** Color flow still frame demonstrating turbulent flow across the central orifice (thick arrow) of the membrane.

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