



## Clinical case report based study

# A patient with Marfan's syndrome presented with severe rheumatic mitral stenosis and successfully treated with percutaneous transmitral balloon commissurotomy – Report of first case



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## ABSTRACT

Marfan's syndrome (MFS) is an inherited autosomal dominant disorder of connective tissue with mutation on the fibrillin-1 gene encoding for fibrillin. This frequently involves the cardiovascular system with prevalence is 1:5000–10,000. The clinical major criteria involve the skeletal and ocular apparatus and the cardiovascular and central nervous system. In Marfan's syndrome mitral valve prolapse and aortic dilation are the main cardiovascular manifestations. According to the literature database the prevalence of aortic dilatation is 76%, 62% for mitral valve prolapse, 29% for mitral valve regurgitation and 26% for aortic regurgitation, in adult patients with Marfan's syndrome. We are reporting a case who presented with severe rheumatic mitral stenosis & later on examination found to be a case of Marfan's syndrome. This patient was successfully treated with percutaneous transmitral balloon commissurotomy. In medline search, so far there is no other case of Marfan's syndrome reported to have rheumatic mitral stenosis.

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## 1. Introduction

Marfan's syndrome (MFS) is an autosomal dominant inherited disorder of connective tissue.<sup>1</sup> Its incidence is two to six per 10,000 individuals. Mutation on chromosome 15 (15q21) and other mutations related to the fibrillin-1 (FBN-1) gene have been reported.<sup>2</sup> Despite these defined mutations, diagnosis of MFS is still based on clinical evaluation and family history. In this disorder pleiotropic manifestations are exerted by *FBN-1* gene mutations in the classic triad of ocular, skeletal, and cardiovascular systems.<sup>1</sup> The Ghent criteria are used for the diagnosis.<sup>2</sup> Typical characteristics of the disease are aortic valve regurgitation (AR) with progressive dilatation of the aorta, which may cause dissection and rupture of the ascending aorta, mitral valve prolapse (MVP), mitral regurgitation (MR), lens dislocation, myopia, thin stature with long extremities, arachnodactyly, chest wall abnormalities and scoliosis.<sup>3</sup> Anomalies of the cardiovascular system account for a significant proportion of the shortened life span of patients with MFS.<sup>3</sup> We are reporting a case of MFS who presented with severe mitral stenosis (MS) due to rheumatic heart disease

(RHD) and he was treated successfully with percutaneous transmitral balloon commissurotomy (PTMC). There is no previous report of this association in the literature.

## 2. Case report

A 25-year-old male patient admitted in our hospital due to acute onset breathlessness for the last 7 days. He has fever & cough associated with breathlessness for this period. On past history, he was diagnosed of having RHD clinically 2 years back when he was admitted with left sided hemi paresis. Though he was put on penicillin prophylaxis but no echocardiography was done. He didn't give any history suggestive of acute rheumatic fever in the past. Family history of similar physical habitus was seen in father and younger brother. On general physical examination, he found to have marfanoid habitus with reduced upper and lower segment ratio, arm span to height ratio greater than 1.05, presence of wrist and thumb sign, arachnodactyly, clinodactyly, pectus carinatum, scoliosis and high arched palate, decreased skeletal muscle mass with thin built (Fig. 1). Ocular examination didn't reveal presence of ectopia lentis or other ocular disorder. On cardiovascular examination, he has engorged jugular vein, tachycardia (heart rate – 140/min), tapping apex, right ventricular heave, loud first & second

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Fig. 1. Demonstrated physical signs of MFS.

heart sound and mid diastolic rumbling murmur with pre systolic accentuation & opening snap suggestive of severe MS. On respiratory system examination, he had tachypnea (respiratory rate – 34/min), bilateral crackles and rhonchi suggestive of pulmonary edema. Central nervous system examination revealed presence of left sided hemi paresis. Per abdominal examination was normal. Routine blood investigations were within normal limit. Chest X-ray showed features suggestive of pulmonary edema. Electrocardiogram showed sinus tachycardia & left atrial enlargement. Echocardiographic examination showed features of severe rheumatic mitral valvular stenosis (mitral valvular orifice area (MVOA) – 0.9 cm<sup>2</sup>, mitral valvular gradient (MVG) – 34/22, Pulmonary artery systolic pressure (PASP) – 54 mm of Hg with fusion of both the commissure), no left atrial appendage clot & absence of aortic dilatation or regurgitation. As this patient had severe dyspnea, tachycardia and pulmonary edema & mitral valve were suitable for PTMC with Wilkins score was 8, we have planned for emergency PTMC with Wilkins score was 8, we have planned for emergency PTMC (Fig. 2). After emergency PTMC, he had improved markedly. His heart rate & tachypnea were also decreased. Post PTMC

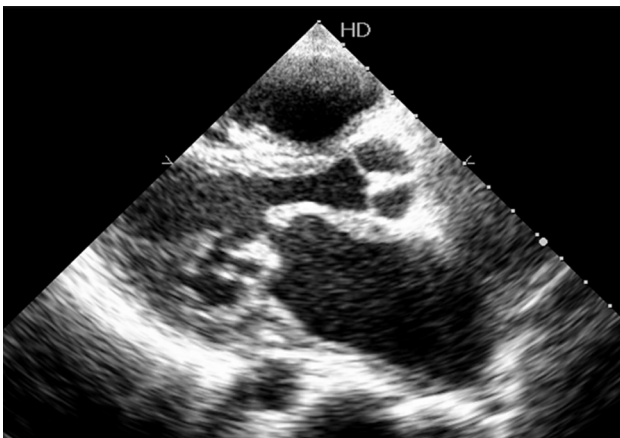


Fig. 2. PTMC done with Accura balloon.

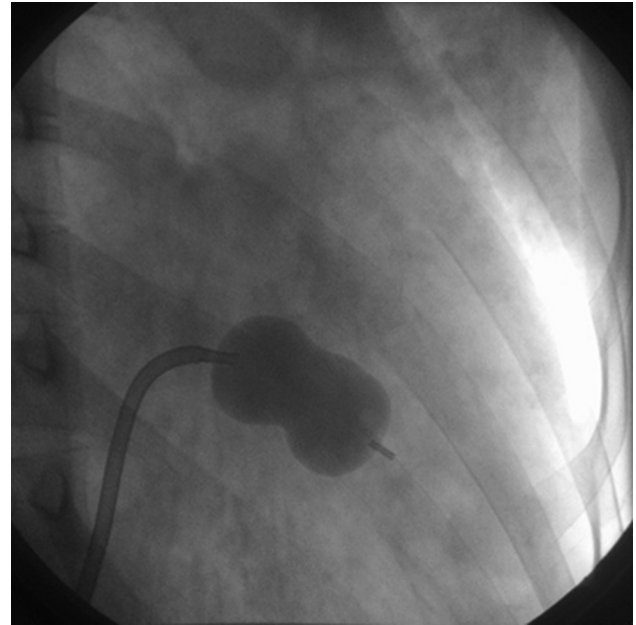


Fig. 3. Post PTMC echocardiography showed splitting of the commissure.

echocardiography revealed MVOA 1.7 cm<sup>2</sup>, MVG – 11/5 mm of Hg, PASP – 34 with splitting of both the commissure (Fig. 3). Patient was discharged third day after the PTMC.

### 3. Discussion

Marfan's syndrome was described for the first time by Antoine Marfan in 1896.<sup>4</sup> Family history may be negative like in our case, which is in agreement with the literature, which notes that sporadic cases may have a larger deletion or greater dysfunction of the gene, leading to a more severe phenotype.<sup>5</sup> In 1912, the German pediatrician Salle presented the first case with cardiac involvement.<sup>4</sup> Cardiovascular complications are the major cause of morbidity and mortality in MFS and the commonest cardiac lesions were MVP and aortic dilatation. MVP is present in around 75% of cases, with increased prevalence in adolescence.<sup>3</sup> The mean life expectancy for untreated patients with MFS is about 32 years with aortic dissection, aortic rupture or cardiac failure due to mitral and aortic valve regurgitation as the predominant cause of death in >90% of the cases.<sup>4</sup> There is a risk of sudden death in MFS. In most cases it results from aortic dissection or rupture, which may occur during exercise and be rapidly fatal. MR is generally a more benign pathology than AR, but rupture of the chordae tendineae can require immediate surgery. Some study revealed significant biventricular diastolic and biatrial systolic and diastolic dysfunction in a fairly large series of adult patients with MFS in the absence of valvular disease.<sup>6</sup> Congenital heart disease like atrial septal defect and aortic bicuspid valve had the same prevalence than in subjects without MFS.<sup>7</sup> Literature supports the existence of a primary cardiomyopathy in a subgroup of MFS.<sup>8</sup> But nowhere it was reported that MS was associated in patient with MFS. Our patient had RHD with severe MS along with the skeletal features of MFS along with family history of MFS. As in our country RHD is common acquired cardiac disorder so patient with MFS may also have RHD. There was no evidence of aortic root dilatation or MVP or MR. Though they are potential for development of MR due their genetic disorder. But in our case PTMC was successful and no significant MR developed after the procedure. So patient with MFS can have only MS as cardiac feature and they can be treated with PTMC as like patient without MFS with equal success.

### Contributors

Soumya Patra, Nagesh C.M, Ajitpal Singh, Srinivas B.C & Babu Reddy were involved in the management of this patient. Soumya Patra reviewed the literature & drafted the manuscript. Srinivas B.C & Manjunath C.N corrected the manuscript. All authors approved the final version of the manuscript.

### Conflicts of interest

All authors have none to declare.

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