Classical Cardiovascular Manifestations of Marfan Syndrome

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Abstract
Presence of multiple cardiovascular manifestations of the Marfan syndrome in the same patient is not commonly encountered. We present a 49 year-old lady with this syndrome who presented with decompensated heart failure. Evaluation revealed presence of extensive Stanford type A aortic dissection alongwith severe aortic and mitral incompetence. However, the patient declined surgery and was discharged on medical management. At a year’s follow-up, she had dyspnea of NYHA class II with persistent cardiovascular findings.

Case Report
A 49 year-old lady presented with acute severe retrosternal pain and progressive dyspnea over three days. Morphologic features of the Marfan syndrome were observed: a height of 166 cm, arm span of 176 cm, thin built, scoliosis, arachnodactyly, high-arched palate and positive thumb and wrist signs (Figure 1). Left upper limb pulses were weaker than the right; all peripheral pulses were palpable with no bruit. Blood pressures were 100/45 mm Hg and 90/50 in the right and left upper limbs and 120/60 and 134/70 in the right and left lower limbs respectively. A short grade 3 early diastolic murmur in the 2nd right intercostal space and a grade 4 pansystolic murmur at the apex were auscultable. Routine hemogram, renal and liver function tests were within normal limits. Echocardiography demonstrated a dilated aortic root (54 mm), with a dissection flap extending from the non-coronary cusp to the visualized descending thoracic aorta with associated severe aortic incompetence. Presence of posterior mitral leaflet prolapse causing an anteriorly directed eccentric jet of severe mitral regurgitation was also noted (Figure 2). There was evidence of chamber dilatation with left atrial diameter of 43 mm and left ventricular (LV) systolic and diastolic dimensions of 44 mm and 60 mm, respectively. There was mild tricuspid regurgitation (TR) with severe pulmonary arterial (PA) hypertension (estimated PA systolic pressure by TR jet of 67 mm Hg). Biventricular function was normal with LV ejection fraction of 60%. A 64-slice multi-detector CT aortogram confirmed extensive dissection of the aorta extending from the aortic root to the abdominal aorta with involvement of the right brachiocephalic, bilateral common carotid and left renal arteries without frank luminal occlusion (Figures 3 and 4).

Urgent surgical aortic root replacement with aortic valve replacement (Bentall procedure) along with mitral valve repair was recommended. However, the patient declined surgery and was discharged on a conservative anti-failure regimen. At a year’s follow-up, the patient was doing well, with exertional dyspnoea of NYHA class II. Echocardiography showed similar findings with marginally worsened LV dilatation and function. She opted to continue
with medical management.

**Discussion**

The Marfan syndrome is a disorder of connective tissue caused by mutations in the fibrillin-1 gene, a major component of extracellular microfibrils, with multi-organ involvement. Our patient was diagnosed with the Marfan syndrome based on major involvement of skeletal and cardiovascular systems in accordance with the Ghent nosology. Cardiovascular manifestation of this syndrome include mitral valve prolapse, aortic root and pulmonary artery dilation, mitral and aortic incompetence, aortic aneurysm and aortic dissection. Anomalies of the cardiovascular system account for a significant proportion of the shortened life-span of patients with this disorder. The prevalence of aortic root dilation and mitral
valve prolapse is around 80% and that for aortic and mitral valvular incompetence is 24–40%. Our patient had involvement of the aortic root as well as the mitral valve. Acute aortic dissection can be life-threatening and hence should be the primary focus of evaluation of these patients. Marfan syndrome is one of the adverse predictors of mortality in patients with type A aortic dissection and accounts for up to 5% of all aortic dissections. The risk of dissection in Marfan syndrome is greatest in presence of aortic root dilation, reaching 10% in high-risk patients (aortic root diameter >40 mm or rapid dilation). Surgical correction is recommended for all patients with acute type A dissection. Root replacement with or without aortic valve replacement is the preferred choice. At present there is no role of endovascular therapy for type A dissection. The one year mortality in patients with type A aortic dissection can be as high as 58% in patients on medical therapy and up to 27% in the surgically treated group. The 3 year survival of patients with type A dissection who are managed medically and survive the initial hospital stay is 50-70%. In a recent series from the Johns Hopkins Medical Institutions on Bentall repair in patients with the Marfan syndrome, concomitant mitral valve repair/replacement was required in up to 20% of patients. This case was remarkable for the multiple, severe prototype cardiovascular manifestations of this syndrome and survival without surgical intervention.

References
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