Pulmonary Arterial Hypertension and Aneurysm in a Patient with Marfan Syndrome

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Abstract

Cardiovascular involvement in Marfan syndrome is considered major criteria for its diagnosis and typically includes mitral valve prolapse, aortic root dilatation and aortic dissection. Pulmonary arterial trunk involvement in Marfan syndrome has rarely been reported. We report a case of a young Pakistani male presenting with dyspnea and fever who had morphological features of Marfan syndrome. On chest X-ray a left hilar mass was reported. Echocardiography of the patient revealed dilatation of pulmonary artery and moderate pulmonary hypertension. An aneurysm involving pulmonary artery and its left main trunk was confirmed on CT chest with IV contrast and on cardiac catheterization. Aneurysm of pulmonary trunk should be considered in the differential diagnosis of mediastinal masses in patients having Marfan syndrome.

Key Words: Marfan's Syndrome; Pulmonary Artery Aneurysm; Pulmonary hypertension.

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Introduction

Marfan syndrome is a hereditary disorder of connective tissue transmitted as an autosomal dominant trait. This ailment is characterized by molecular defects and mutation in FBN1[1] gene of fibrillin which is accountable for diminished structural integrity of ocular, skeletal and cardiovascular systems. Cardinal topographies of the condition consists of ectopia lentis, tall stature, aortic root dilation, mitral valve prolapse and aortic dissection[2]. Cardiovascular disease poses a main cause of mortality and morbidity in patients with Marfan syndrome[3]. It is classified as major criteria for diagnosis of Marfan syndrome[4]. However the involvement of pulmonary trunk is very rare. We report an unusual case of Marfan syndrome that had pulmonary artery aneurysm and primary pulmonary hypertension.

Case Report

A 17-year-old male patient was admitted from emergency department of Mayo Hospital, Lahore, Pakistan for complaints of low-grade fever for the last four years with recent aggravation of his fever for one month along with palpitations and NYHA class I dyspnea.

On physical examination he was a tall thin guy having an arm span to height ratio of more than one (figure 1). There was also a high arched palate, hyper extensible joints, positive thumb and wrist signs, pectus deformity of chest and scoliosis present. He had a pulse of 100/min, BP 100/60, temperature 98.6 F and respiratory rate of 16/min. His cardiovascular examination showed a well localized apex beat with visible pulsations in pulmonary area, a left parasternal heave with loud pulmonary component of the second heart sound and grade 3/6 systolic murmur localized to left sternal border. His eye examination was normal.

The electrocardiogram (figure 2) showed sinus tachycardia, with evidence of right ventricular hypertrophy and right axis deviation. The X-ray chest (figure 3) had a big left hilar mass presumed to be pulmonary artery shadow. Echocardiography (figure 4) of the patient revealed dilatation of pulmonary artery and moderate pulmonary hypertension. A CT chest with IV contrast (figure 5) was then performed that reported an aneurysm involving pulmonary artery and its left main trunk. Cardiac catheterization confirmed the findings, ruled out any shunt and reported a mean pulmonary artery pressure of about 80/50 mm Hg. CBC revealed hemoglobin of 8.7gm/dl, total leucocyte count was 9700/cm^3. Arterial blood gas analysis was normal at room air. Screening for rheumatic and autoimmune diseases as well as diagnostic evaluation for tuberculosis and other infections was negative.

No cause for his pulmonary hypertension was found and he was labeled as a case of primary pulmonary hypertension. The patient was then put on calcium channel blockers and sildenafil for his pulmonary hypertension and then referred to cardiac surgeon for further evaluation regarding the need for any surgical intervention.

Discussion

Marfan syndrome is a common Mendelian disorder having an estimated rate of incidence as 2-3/ 10000[5]. This syndrome is prevalent globally without any predilection for geography or ethnicity. In Marfan syndrome mitral valve prolapse and aortic dilatation are the main cardiovascular manifestations. The major reason of illness is presented by aneurysm or thoracic aortic dissection which accounts for more than 80% demises. However involvement of pulmonary trunk is very unusual and is considered under minor criteria for diagnosis of Marfan syndrome[6]. Similarly

incidence of Aortic dissection is common, but that of pulmonary artery aneurysm and dissection is a rarity[7].

Main pulmonary artery aneurysms are themselves a rare entity with few available published data[8]. As reported in the literature, operative treatment is commonly recommended but the relation between the size of the aneurysm, its localization and risk of rupture is not as well defined as for aortic aneurysms[9]. Proximal lesions that involve the main branches of pulmonary trunk are usually apparent on chest radiographs and should be taken into consideration in differential diagnosis of mediastinal masses[10].

Usual causes are congenital heart diseases, pulmonary hypertension, infections (TB, syphilis), connective tissue disorders (Marfan syndrome, Ehler Danlos syndrome) and vasculitides such as Behcets syndrome. Intravascular or extra vascular trauma is a rare cause. Some cases are idiopathic and may be associated with Hughes Sovin syndrome, a rare syndrome with recurrent superficial and deep venous thrombosis, raised intracranial pressure and pulmonary artery aneurysm.

The main diagnostic modalities are chest radiographs, echocardiography, contrast enhanced computed tomography, magnetic resonance imaging and pulmonary angiography. Natural history is not known and possible complications include dissection, pulmonary embolism, intrapulmonary erosions, and compression of bronchi, trachea, superior vena cave and recurrence of laryngeal nerve

Pulmonary artery aneurysm is mainly managed by surgical intervention and described techniques include aneurysmectomy with Dacron graft placement or autologous pericardial replacement or arterial aneursymorraphy. Most peripheral lesions may be treated with aneurysmectomy, ligation, segmental resection, lobectomy, pneumonectomy or non-surgical embolectomy. However there are no clear guidelines about surgical indications[11].

The management of his pulmonary hypertension was started as we had sufficient guiding material regarding this. In general, high doses of CCBs are used in patients with primary pulmonary hypertension[12]. Other approved pulmonary vasodilators include Epoprostenol, Treprostinil, Bosentan, Iloprost and Sildenafil.

In conclusion our patient was not a typical case of Marfan syndrome with aortic involvement rather his disease showed a predilection for the pulmonary vasculature. Hence aneurysm of pulmonary trunk should be considered in the differential diagnosis of mediastinal masses. The other finding of pulmonary hypertension had no relation to his current ailment. Any other associations were not found as well so he was diagnosed a case of primary pulmonary hypertension and was managed accordingly.

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Figure 1 Height vs Arm Span

Figure 2 ECG

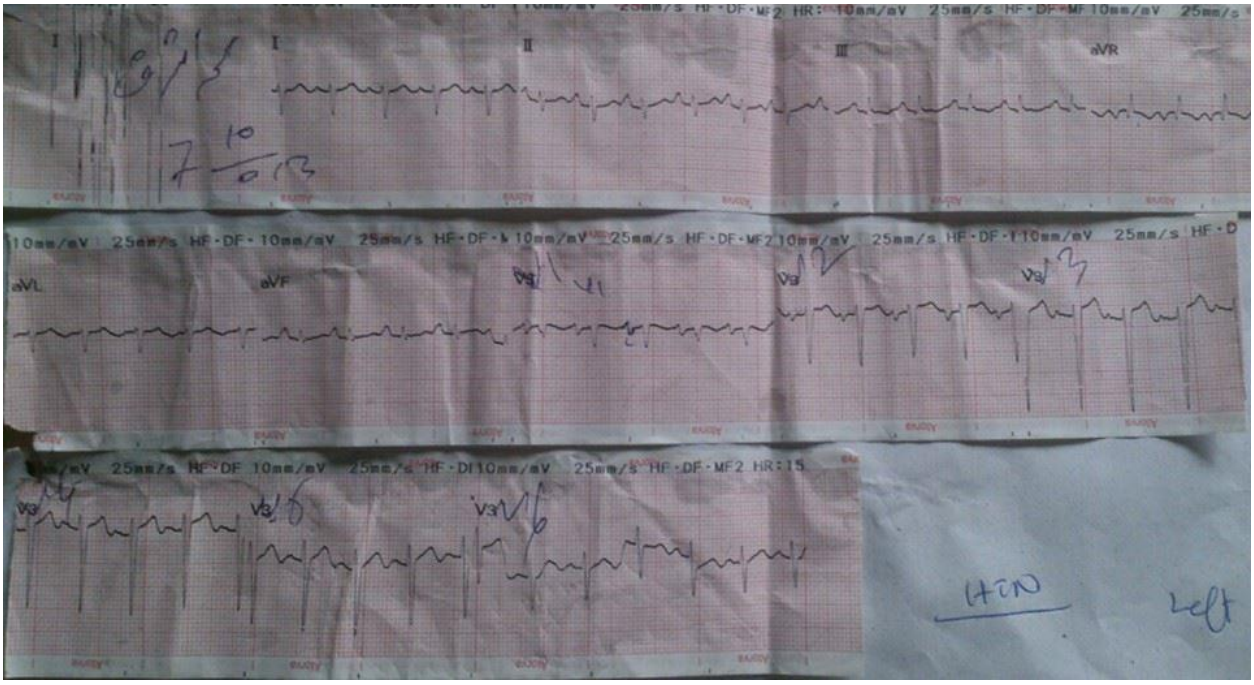

Figure 3 Chest X-Ray

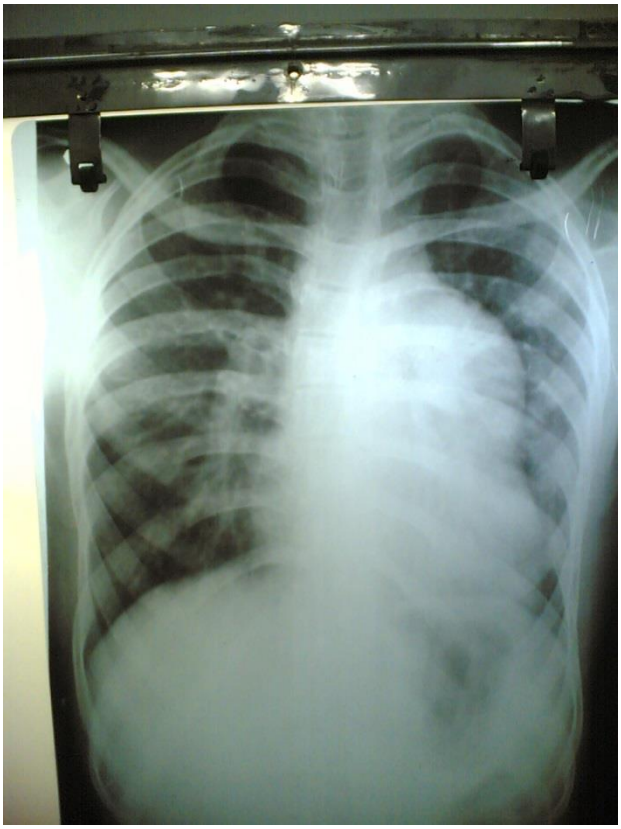

Figure 4 Echocardiography Parasternal Short Axis

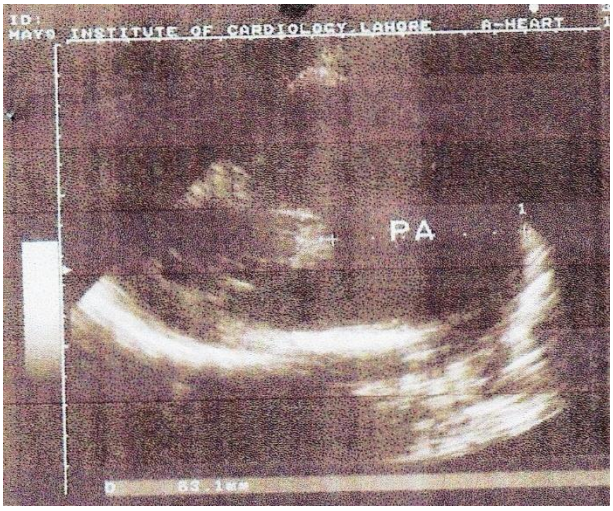

Figure 5 CT chest with IV Contrast