JMSCR Vol||05||Issue||11||Page 30725-30727||November 2017

www.jmscr.igmpublication.org
Impact Factor 5.84
Index Copernicus Value: 71.58
ISSN (e)-2347-176x ISSN (p) 2455-0450
crossref DOI: https://dx.doi.org/10.18535/jmscr/v5i11.155

IGM Publication
Journal Of Medical Science And Clinical Research
An Official Publication Of IGM Publication

An Unusual Presentation of Marfan's Syndrome

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Introduction

Marfan's syndrome is a variable autosomal dominant hereditary connective tissue disorder caused by the mutation of the gene encoding the extracellular matrix protein fibrillin-1 (FBN 1) occasionally a mutation in TGFBR1 or 2.[1] The prevalence of Marfan syndrome is approximately 1 per 5000 2 population and 26% of the cases have no family history.[2] Its Characteristic clinical features include anterior chest deformities, long fingers, aortic root dilatation and dissection, lens dislocation and myopia.[3] It is characterized by skeletal, cardiovascular and ocular abnormalities. Pulmonary involvement is uncommon and occur in approximately 10%. The commonest pulmonary complication being spontaneous pneumothorax and emphysema. Marfan's syndrome associated with bronchiectasis is a rare presentation only few cases have been encountered so far.

Case Report

A 50-year-old farmer by occupation presented with a history of persistent cough associated with purulent sputum, breathlessness (grade 3), decreased appetite and generalised weakness for 1 month. No history of evening rise of temperature, hemoptysis, significant weight loss, chest pain, orthopnea and abdominal pain

He was a non-smoker and non- alcoholic with no relevant family history.

On admission He had marked clubbing of fingers and toes.

Examination of the chest revealed central trachea, normal percussion note and bilateral coarse crepitations all over lung fields.

Examination of the musculoskeletal system revealed Marfanoid features as follows: Arachnodactyly, positive wrist and Steinberg signs, dolichosternomelia, low ratio of upper segment to lower segment (height 169 cm, arm span 173 cm) and high arched palate.

Examination of the cardiovascular systems was normal whilst ECG showed right axis deviation.

Ophthalmoscopy showed no ectopia lentis.

Investigations

Patient's CBC showed haemoglobin 14 g/dl, total leucocyte count were 13300/cm. liver function test and renal function test were normal. Sputum acid fast bacilli reports revealed negative results. Sputum culture grew Haemophilus influenzae but no fungal or acid fast bacilli growth.

2D-Echo- showed evidence of mitral valve prolapse with dilated aotic valve root (55 mm).

Chest radiography demonstrated extensive cystic bronchiectasis of bilateral lower lobe.

In view of same CT thorax done which showed bilateral bronchiectatic changes in lower and middle zone.

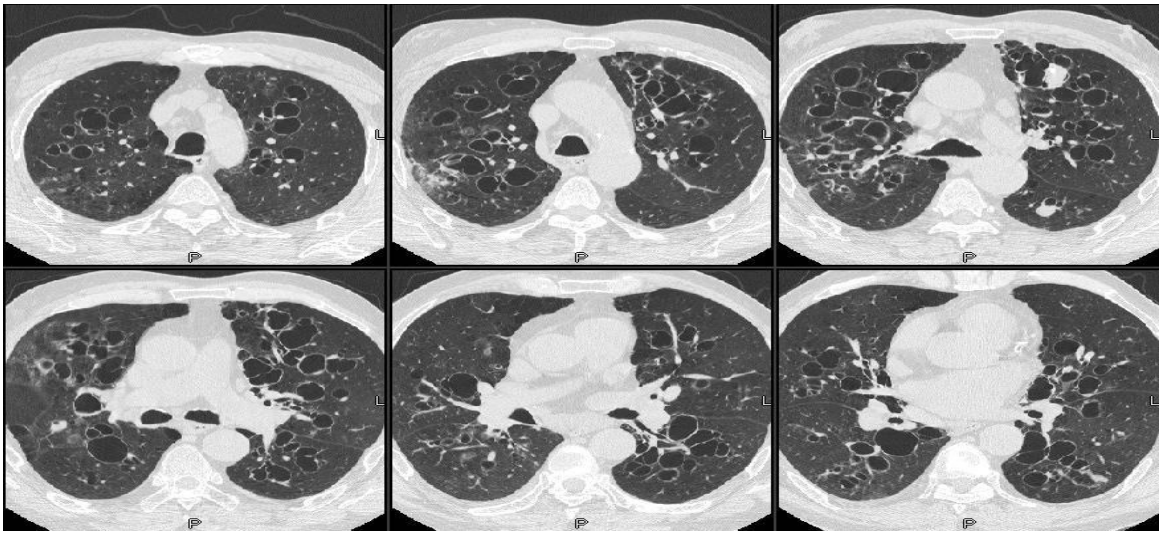
Slit lamp examination of the eyes was normal.

Lung function tests demonstrated non-reversible airway obstruction.

A diagnosis of Marfan Syndrome with bronchiectasis was made and patient was treated with Cefoperazone and tazobactam, deriphylline, nebulization with duolin and mucomix for 7 days and continuous non invasive oxygen therapy. He was discharged and advised for oxygen therapy at home and followed up for over 6 months with occasional infective episodes which did not require hospitalization.

Family Screening

Patient had two sibling and her sister has same respiratory problems since 5 years. He had been advised for screening of her sister.



Discussion

Approximately 10 % patients of marfan's syndrome are affected with pulmonary abnormalities, the most common being spontaneous pneumothorax, bullae, cystic lung disease, emphysema and recurrent infection of respiratory system.[4]

Bronchiectasis in marfan's syndrome is very rare association and so far one few cases have been encountered. About this abnormality few study has been done which suggested it is related to collagen disorder. The defect in collagen may be responsible for the reduced tensile strength of the connective tissue leading to both increased susceptibility to infection and to bronchiectasis.

Other congenital abnormalities such as Kartagener's Syndrome, congenital absence of bronchial cartilage and congenital kyphoscoliosis are associated with bronchiectasis.[5]

Unlike other pulmonary complications of this syndrome, the progress of bronchiectasis is amenable to modification with early institution of appropriate antibiotics and chest drainage manouvres.

Bronchiectasis in Marfan's syndrome was first described by Katz (1952). He recorded a 24-year-old male with the typical osseous and cardiac manifestations, including a depressed sternum and evidence of aortic incompetence. He also had extensive bilaternal cystic bronchiectasis, confirmed by bronchography. Katz postulated that the bronchiectasis in this case was either due to an intrinsic bronchial defect present in Marfan's syndrome or related to the high incidence of recurrent respiratory infection in patients with thoracic cage deformity.[6]

Teoh (1977) described a Chinese girl with Marfan's syndrome complicated by spontaneous

pneumothorax and bilateral cystic bronchiectasis. He suggested that these were separate pulmonary manifestations of the syndrome since pneumothorax is seldom reported as complicating bronchiectasis. In a review of pulmonary disease in Marfan's syndrome, Turner and Stanley (1976) postulated that the occurrence of pneumothorax and emphysema was related to weakness in the pulmonary connective tissue framework. Although it is difficult to exclude coincidental bronchiectasis in a patient with Marfan's syndrome, it appears likely that bronchiectasis represents an additional pulmonary complication of the syndrome. Bronchiectasis has been described in a number of other congenital abnormalities, the most well known being Kartagener's syndrome of bronchiectasis, dextrocardia and sinusitis or absent frontal sinuses. Other rare associations include congenital absence of bronchial cartilage, congenital heart disease and congenital kyphoscoliosis. The association of bronchiectasis with Marfan's syndrome can now be added to the literature.

In conclusion, bronchiectasis in Marfan Syndrome is as a result of weakness in the collagen structure and runs in the family. There is a need for screening family members with Marfan Syndrome and institution of early measures to avoid bronchial damage.

References

1. John C S Dean. Marfan syndrome: clinical diagnosis and management. Eur J Med Genet. 2007 May; 15:724–733
2. Lacro RV, Dietz HC, Wruck LM, Bradley TJ, et al. Rationale and Design of a Randomized Clinical Trial of Beta Blocker Therapy (Atenolol) vs. Angiotensin II Receptor Blocker Therapy (Losartan) in Individuals with Marfan Syndrome. Am Heart J. 2007 October; 154(4):624–631.
3. Dean JC. Management of Marfan syndrome. Heart 2002; 88(1):97–103
4. Wood JR Bellamy D, Child AH. Cilm KM. Wilmonary disease in patients with Martini's Syndrome. 'Iliorax 19X4: 39:7X0-4
5. M. E. FOSTER D. R. FOSTER. Bronchiectasis and Marfan's syndrome-case report. Postgraduate Medical Journal (October 1980) 56, 718-719
6. KATZ, H.L. (1952) Thoracic manifestation in Marfan's syndrome (arachnodactyly). Quarterly Bulletin of Sea View Hospital, 13, 95.