

## A Case of Marfan Syndrome Presenting as Tension Pneumothorax in A Tertiary Care Setup

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### ABSTRACT

Marfan Syndrome is a rare connective tissue condition that affects several systems, including musculoskeletal, cardiovascular and ocular systems. Although less frequent, pulmonary involvement can nevertheless lead to emphysema, bullae, apical blebs, and a higher risk of spontaneous pneumothorax. Another option is pectus excavatum, carinatum, or scoliosis-related restrictive lung disease. We will discuss a case of 18 year old girl, with marfanoid habitus, non-smoker with history of ATT taken on radiological grounds, with complains of shortness of breath on and off and presenting with sudden onset worsening of shortness of breath and dry cough for 3 weeks. Chest x-ray showed tension pneumothorax. After emergency management echo was done and it showed MVP with MR. Patient was diagnosed as a case of Marfan Syndrome following Ghent criteria. The case indicates that pneumothorax though rare can be first presentation of Marfan Syndrome.

**Keywords:** Marfan, pneumothorax, mitral valve prolapse, mitral regurgitation

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### INTRODUCTION:

Marfan syndrome is a rare but well-defined hereditary disorder of connective tissue system. Symptoms of Marfan syndrome (MFS) can be mild to severe in intensity and may vary because the condition can affect different parts of the human body. Prominent features of the syndrome include abnormalities of the skeletal, cardiovascular, and ocular systems that are manifested as either a congenital anomaly (microphakia, coarctation of the aorta) or a degenerative lesion of elastic tissue (aortic dissection, ectopic lens, hernia)<sup>1</sup> Lesser known areas of involvement are renal, dermatologic and pulmonary systems. This is one of the most prevalent hereditary connective tissue condition with autosomal dominant trait and no sex preference. Mutations occur in the FBN1 gene, which is found on chromosome 15q-21.1. The FBN1 gene codes for the matrix protein fibrillin-1, which is found in both elastic and non-elastic connective tissues.<sup>2</sup> Many individuals demonstrated distinct lung bullae changes that were most likely brought on by degenerating elastic fibres.<sup>3</sup> The diagnosis of MFS depends essentially

on the fulfilment of clinical diagnostic criteria as shown by the revised Ghent score. We report a case of MFS in which the patient's first presentation was an outcome of respiratory system.

### CASE

A 19 year old unmarried girl with thin built and above average height from Larkana presented to Ojha institute of Chest Disease, Dow University Health Sciences, Karachi in May 2021 with acute worsening of shortness of breath for 3 to 4 hours. She already had cough and shortness of breath for last 2 years and had been started on Anti-tuberculous therapy on radiological and clinical grounds. Despite treatment patient didn't improve. After 3 months of treatment patient stopped taking ATT herself and after a couple of weeks she developed worsening of her symptoms before presenting to OICD. On examination of the patient, she was in respiratory distress with a respiratory rate of 27/min and blood pressures of 90/60. Her saturations were 90% at room air. Examination of the chest revealed Pectus carinatum on inspection and hyper resonant percussion note on right side with diminished breath sounds on auscultation.

Urgent Chest radiograph was done and it revealed large right sided pneumothorax. Emergency chest intubation was performed and after stabilization of the patient detailed examination was done.

General physical examination revealed high arched palate, long arms with thin and long fingers and a hyper extendible thumb. Patient's chest CT scan with contrast showed bilateral apical blebs and right sided small pneumothorax as it was post intubation. Diagnostic bronchoscopy was done and all infective work-up including AFB smear, Xpert MTB and Fungal smear/culture all were negative. Eye examination

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was unremarkable. Her Echocardiography was ordered and it revealed mitral valve regurgitation (MR) along with mitral valve prolapse(MVP). Following Ghent Criteria Marfan Syndrome was diagnosed.

Thoracic Surgeon was taken on board to do Surgical Pleurodesis or if necessary surgical repair of the lung involvement. Patient was advised to follow Rheumatologist. Also she was referred to cardiologist for her regular follow up to look after her cardiac issues.

### DISCUSSION:

Marfan syndrome affects roughly 1 in 10,000 to 15,000 people, according to estimates<sup>4</sup> Marfan syndrome is a

systemic disease and may involve multiple systems simultaneously, although occasionally the diagnosis of Marfan's syndrome can be delayed due to a lack of identifiable symptoms.<sup>5</sup> A few studies previously demonstrated respiratory complications in Marfan syndrome<sup>6</sup>, and it wasn't until recently that Karpman et al. further studied the link between pneumothorax and Marfan syndrome. The prevalence of pneumothorax in Marfan syndrome ranged between 4.8% and 11%.<sup>7</sup> Although the frequency of apical blebs is relatively small in individuals with Marfan, the risk of pneumothorax is considerably increased in those with radiologically evident blebs or bullae.<sup>7</sup> Early recognition can help patient and the caregiver to provide the treatment accordingly. On account of first pneumothorax surgical treatment should be offered.<sup>8</sup> This patient presented with large pneumothorax which could be fatal and she was being treated as a case of Pulmonary Tuberculosis for the last 1 and a half year. Her CT scan revealed apical bleb which places her at higher risk of developing subsequent pneumothoraces. Our patient also had cardiac involvement and regular cardiac follow up is crucial as most of the times mortality is related to cardiac complications.

The revised Ghent criterion is used to make the diagnosis of Marfan Syndrome. The Ghent criteria, which encompass a set of major and minor findings across various body systems, have proven to function well because confirmation of the diagnosis can be obtained in over 95% of patients with advancing molecular techniques.<sup>9</sup>

Although effective therapy options, like routine cardiac monitoring and elective surgical surgery, have decreased the likelihood of life-threatening cardiovascular events, the vascular complications of MFS remain and they offer the biggest concern.<sup>10</sup> The quality of life can also be significantly impacted by the musculoskeletal manifestations of MFS, which have received less attention to date but are crucial for diagnosis.<sup>10</sup> Musculoskeletal system features include deformities in the limbs, craniofacial features, chest wall deformities and kyphoscoliosis. They can often cause significant changes in pulmonary physiology and mechanics of breathing thus affecting quality of life.

### CONCLUSION:

Marfan Syndrome affects persons in a variety of ways and the symptoms thus vary amongst individuals. Treatment varies depending upon the area of the body affected and may include medications, other treatments, and surgery to manage the condition and its complications. Advancement in treatments and surgeries allow people with Marfan syndrome to live long and productive lives. A multidisciplinary approach including a respiratory physician, thoracic surgeon, cardiologist and rheumatologist is required to manage a case of Marfan Syndrome. This might be helpful by providing patient the care they need by the expert team and then subsequently improving the quality of life of patient.



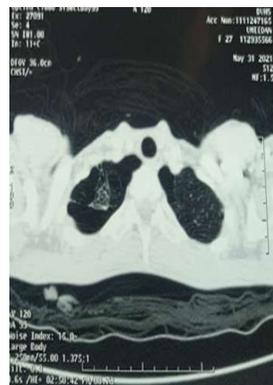
Right Sided Pneumothorax



Hyperextension of Joints



High Arched Palate



Elongated Limbs And Digits



HRCT Chest: Apical Blebs

In this regard, counseling of the patients and their caregivers about the disease and its expected outcomes is important so that the condition is managed properly and at right time to avoid any future mishaps.

**Authors Contribution:**

**Tehreem Ahmad:** Idea, data curation, writing of final manuscript

**Hina Asghar:** Data curation and visualization

**Areeba Hasan:** Writing of initial draft

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