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Spontaneous pneumothorax as a manifestation of Marfan syndrome. A case report.

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Case Report

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ABSTRACT:
Marfan syndrome is a rare disease, it has a lot of manifestations, between them, pneumothorax, due to bullae. We present the case of a male patient, previously healthy, who was admitted into the thoracic surgery service, with spontaneous pneumothorax, with placement of a chest tube for 6 days, in a previous health service without improvement. In the physical exam he presented signs compatible with the Marfan Syndrome. Videothorascopy was performed with bullectomy and pleurodesis, no recurrence was observed. Additional studies were performed, it was found dilatation of the aorta and prolapse of mitral valve, compatible with Marfan.

KEYWORDS: Pneumothorax, Marfan syndrome, bullae, videothoracoscopy

Introduction

Marfan syndrome is an autosomal dominant disease of connective tissue, which affects two or three out of 10,000 people [1,2], both men and women, it is caused by a mutation in fibrinil alpha on chromosome 15, affecting the connective tissue, to the cardiovascular, skeletal and ophthalmic systems. In addition, it affects the pulmonary system, manifesting bronchial malformations, bronchiectasis, changes in the lower lobes of the lung, bullae, emphysema and spontaneous pneumothorax [3].

Two studies published in 1980 and the Karpman study in 2011 found that the prevalence of spontaneous pneumothorax in patients with Marfan syndrome is between 4-11% [4]. The risk of pneumothorax is attributed to the presence of bullae in the vertices, alterations of the connective tissue in the lung parenchyma or decreased mechanical stress in the vertices of tall and thin individuals; these changes may be due to alterations in the Alpha 1 fibrinil [5], which produces alterations in the pulmonary parenchyma, this produces deformities in the thorax, flaccid or small airway, as well as alterations at the level of the bronchi, which predisposes to premature closure of the airway, degenerative changes and emphysema which may be the mechanism of pneumothorax in these individuals [5,6].

There is a lack of consensus among different international guidelines regarding the management of pneumothorax in these patients, according to the British Thoracic Society, surgical treatment should be considered within the first 48 hours, the same as that performed in patients with air leak persistent and failure of pulmonary re-expansion [7,8].

Case report

A 16-year-old male from China, previously healthy, presented sudden pain in the right hemithorax 15 days ago, accompanied by dyspnea on moderate exertion that rapidly progressed to small exertions, he went to an external health home, a chest X-ray was performed (**Figure 1 A**) a grade III, right pneumothorax was found, so a chest tube was placed for 6 days without improvement (**Figure 1 B**)

In evaluation, it is evident: superposition of dental pieces, atrophic striae at the level of the lower limbs and dorso-lumbar region, in thorax: pectus carinatum, decreased vesicular murmur in the right lung field. He also presents aranodactyly, steinberg's sign, walker-murdoch's sign. An echocardiogram was performed with a report of an enlarged left ventricle, fev1: 53%, mitral valve prolapse, dilated ascending aorta. Chest tomography revealed persistence of right pneumothorax plus bullae at the level of the right pulmonary apex (**Figure 2**).

Patient is evaluated by cardiology, rheumatology, ophthalmology and internal medicine, confirming diagnosis of Marfan syndrome. Due to persistence of pneumothorax plus tomographic findings, a video-assisted thoracoscopy is performed, bullous complex is identified (**Figure 3A**), bullectomy is performed (**Figure 3B**) mechanical pleurodecis with propylene mesh (**Figure 4A-B**) and a right pleural drainage is place without complications.

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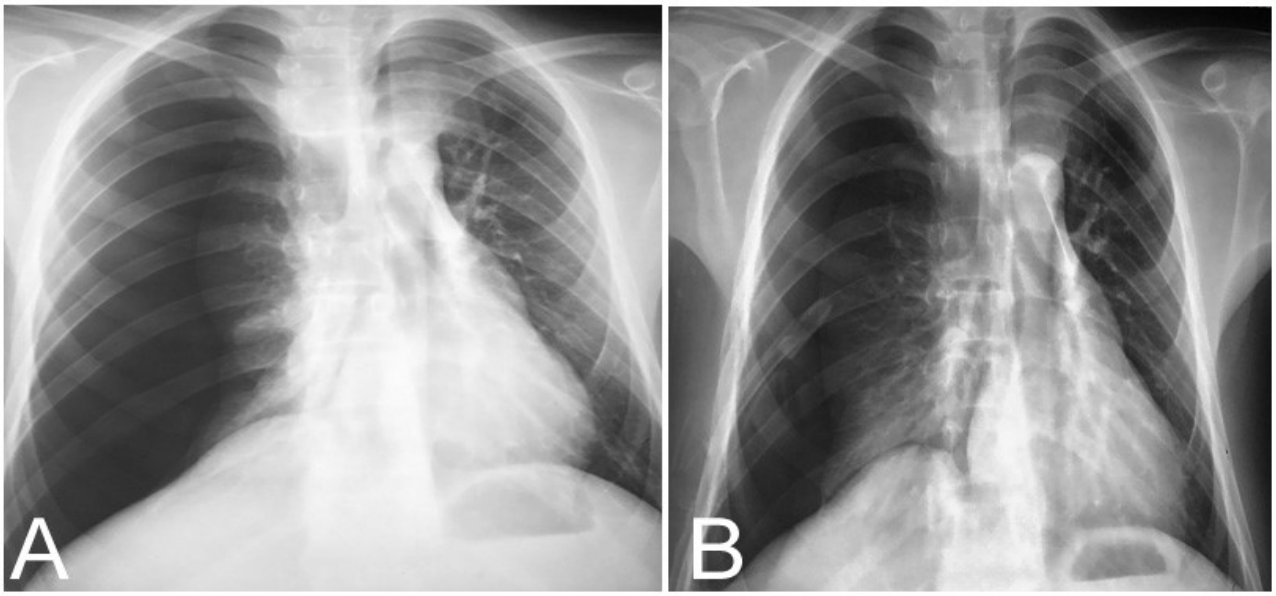

Figure 1. A Chest X rays showing pneumothorax grade III. **B** Chest X rays showing persistent Pneumothorax.

The patient remains hemodynamically stable, radiographic controls are performed twice (figure 5), observing lung expansion, after 6 days he is discharged without any complications and with controls by the outpatient clinic where there is no evidence of recurrence.

Discussion

Marfan syndrome involves an alteration in the synthesis of collagen fibers, which causes a reduction in elasticity and tensile strength at the level of the terminal bronchioles, which facilitates the appearance of bullae, blisters, and pulmonary cysts. These injuries predispose to a higher risk of spontaneous pneumothorax, bilateral pneumothorax, recurrence, and persistent air leak [8].

There is a lack of consensus between different international guidelines regarding the management of pneumothorax, in patients with Marfan syndrome with risk of recurrence, in general, conservative treatment is chosen, although there are authors who opt for surgical treatment or pleurodesis [9,10], according to the British Thoracic Society, surgical treatment should be considered within the first 48 hours, the same as that performed in patients with persistent air leak and lung re-expansion failure [10].

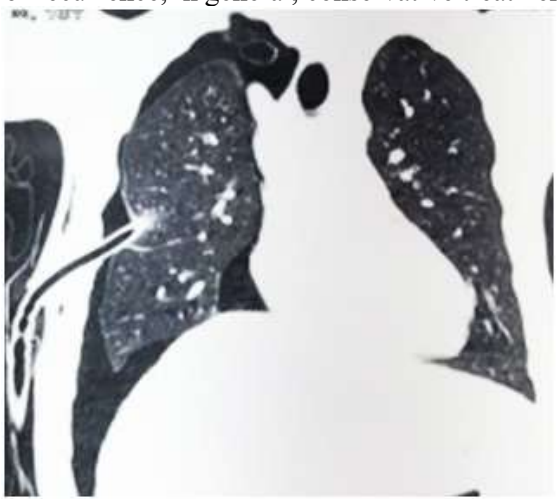

Figure 2. Chest tomography revealed persistence of right pneumothorax plus bullae at the level of the right pulmonary apex.

In 1984, a study was carried out in which the prevalence of spontaneous pneumothorax in patients with Marfan syndrome was 4.4%, male prevalent, but with less severity of signs and symptoms, compared to females. [More than 50% of patients with pneumothorax also had apical bullae visible on chest radiography. [11] Video-assisted thoracoscopy (VATS) is the gold standard for primary and secondary pneumothorax, allowing a faster recovery period, less pain, and significantly decreases hospital stay. This procedure can be three ports or less, which are manifest as a safe way to perform chemical or mechanical pleurodesis, bullectomies and pleurectomies. [11, 12]

Conclusion

Marfan Syndrome is a pathology that has cardiovascular, skeletal, and ophthalmological manifestations, and less frequently (4-11%) pulmonary manifestations, such as spontaneous pneumothorax secondary to bullae whose recurrence ranges between 50-60%, if opted for. for conservative treatment, so surgical treatment must be performed within the first 48 hours, such as video-assisted thoracoscopy plus bullectomy with pleurodecis, which thus prevents recurrences and reduces recovery time and hospital stay.

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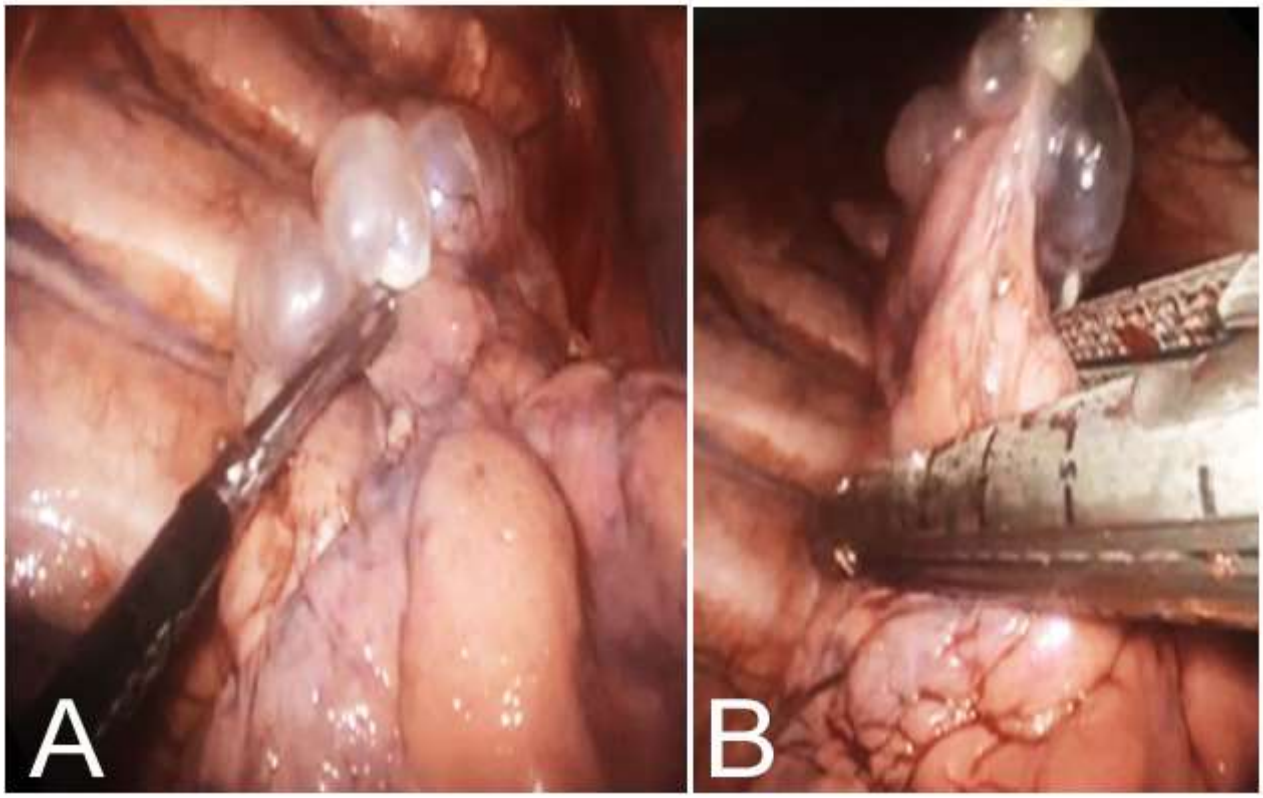

Figure 3.A. Identification of the bullous complex. **B.** Bullectomy with mechanical sutures

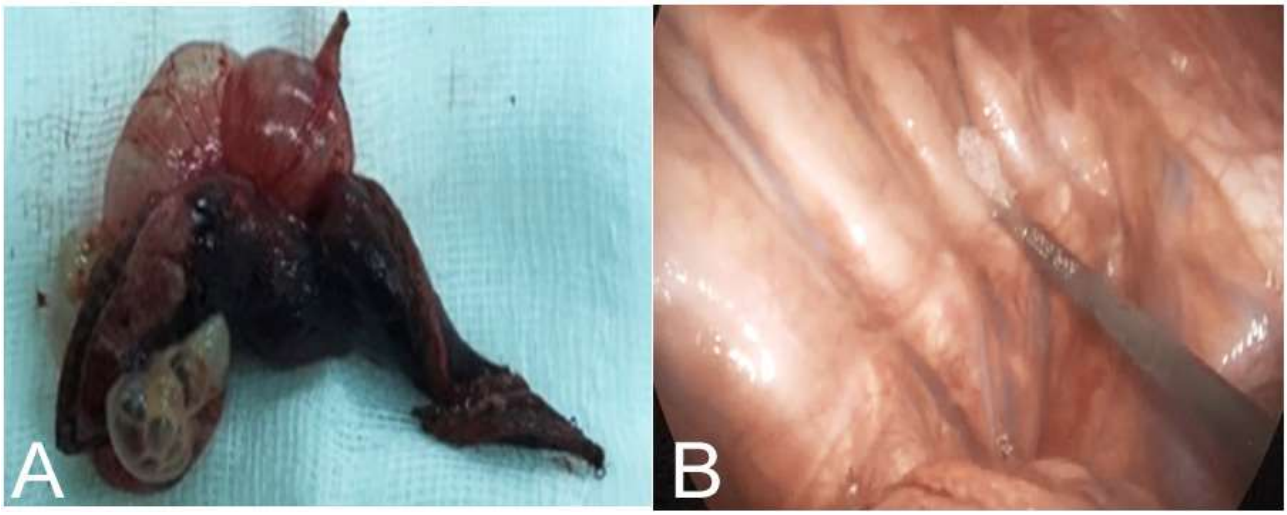

Figure 4. **A** Bullous complex extraction**. B**. Mechanical pleurodesis with propylene mesh

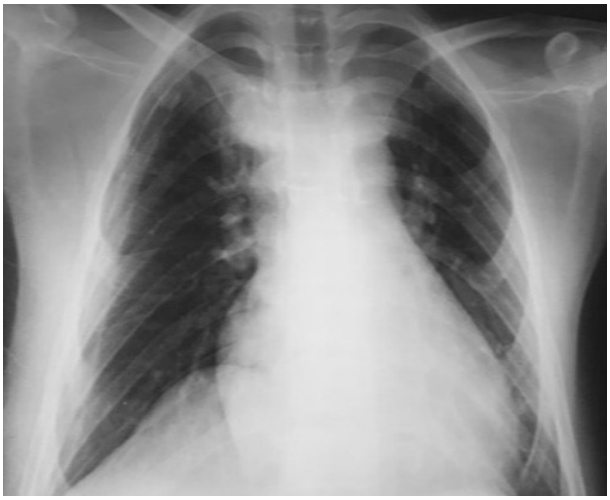

Figure 5. Chest x rays at discharge.

Conflicts of interests

The authors declare no conflict of interest.

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