Bilateral Primary Spontaneous Pneumothorax in a Young Male with Marfanoid Features: A Rare Case Report

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Abstract

Bilateral primary spontaneous pneumothorax (BPSP) is an uncommon clinical entity, accounting for only 1–2% of spontaneous pneumothorax cases. We report a rare case of a 19-year-old male with no known risk factors who presented with acute right-sided pleuritic chest pain and was found to have bilateral pneumothorax. He was managed with intercostal chest drainage and made a full recovery. Follow-up imaging confirmed pleural blebs, but genetic and systemic evaluations were unremarkable. At six months, he remained asymptomatic and had returned to normal activity. This case highlights the importance of clinical vigilance in identifying and managing BPSP, particularly in young patients without typical risk factors, and contributes to the limited regional data on this condition.

Keywords: Blebs Rupture, Pneumothorax, Spontaneous Pneumothorax, Bilateral Primary Spontaneous Pneumothorax, BPSP.

Introduction

Spontaneous pneumothorax is defined by the presence of air in the pleural cavity and occurs at an incidence of 1.2 to 28 per 100,000 individuals.[1,2] It is classified as either primary—seen in patients without underlying lung disease but often linked to risk factors such as male sex and smoking—or secondary, associated with conditions like COPD or malignancy.[3] Bilateral primary spontaneous pneumothorax (BPSP) is particularly rare, accounting for a small fraction of cases and posing risks of tension pneumothorax and respiratory failure.[1,2] Prompt diagnosis requires a high index of suspicion due to its subtle presentation.

We report the case of a 19-year-old male with no known risk factors who presented with acute right-sided pleuritic chest pain and was diagnosed with BPSP. Imaging confirmed bilateral pneumothorax, and whole-exome sequencing showed no evidence of connective tissue disease. The patient was successfully treated with an intercostal chest drain (ICD). This case contributes to the limited literature on BPSP in young adults, particularly from the Middle Eastern region, and underscores the need for early recognition of this rare condition. This report follows the 2020 guidelines for Surgical Case Reports (SCARE).

Case Report

A previously well 19-year-old male presented to the ED with acute, right-sided, stabbing pleuritic chest pain that was non-radiating, worsened with movement, and relieved at rest; notably, there was no

left-sided chest pain, chest heaviness, or radiating pain. He denied any associated palpitations, diaphoresis, dyspnea, pyrexia, cough, or recent trauma. The patient also reported no previous history of respiratory or cardiovascular disease, nor recent exposure to barometric pressure changes (e.g., air travel or diving). He was a non-smoker and had no history of substance misuse. The patient's growth and development were age-appropriate, with no reported delays or cognitive impairments. Family history revealed consanguineous parentage. His father passed away in his 40s from colon carcinoma, while his mother remains healthy. He has three younger siblings, all tall but otherwise healthy. No familial history of congenital disorders, cardiovascular disease, ocular pathology, malignancies or sudden death was reported, except for his maternal uncle, who is tall and thin. On clinical examination, the patient appeared comfortable, seated upright and in no apparent distress. Vital signs were within normal limits: blood pressure 118/70 mmHg, pulse rate 98/min, respiratory rate 18/min, temperature 36.7°C, and oxygen saturation 95% on room air. The patient reported a pain score of 7 out of 10. Auscultation revealed diminished air entry on the right side without adventitious sounds. Other general and physical examinations revealed no significant findings. His phenotypic features were notable for a Marfanoid habitus, characterized by tall stature, arachnodactyly (long, slender fingers), positive wrist and thumb signs, bilateral hallux valgus deformities, and pes planus (flat feet). There was no evidence of cutaneous hyper elasticity, joint hypermobility, or spinal deformities. His cardiac and ocular evaluations were normal, and therefore did not meet the Revised Ghent criteria for Marfan syndrome. His Beighton score was 4, which is below the threshold typically associated with systemic involvement. Genetic testing for FBN1 mutations was also negative. Taken together, these findings effectively rule out a diagnosis of Marfan syndrome. Chest radiography demonstrated bilateral pneumothorax, more pronounced on the right side 27.82*14.64 mm [Figure 1]. After consultation with the cardiothoracic surgery team, a right-sided intercostal drain (ICD) was inserted under sterile conditions with an underwater seal. Laboratory investigations, 12 leads Electrocardiogram and the genetic workup are outlined in Table 1.

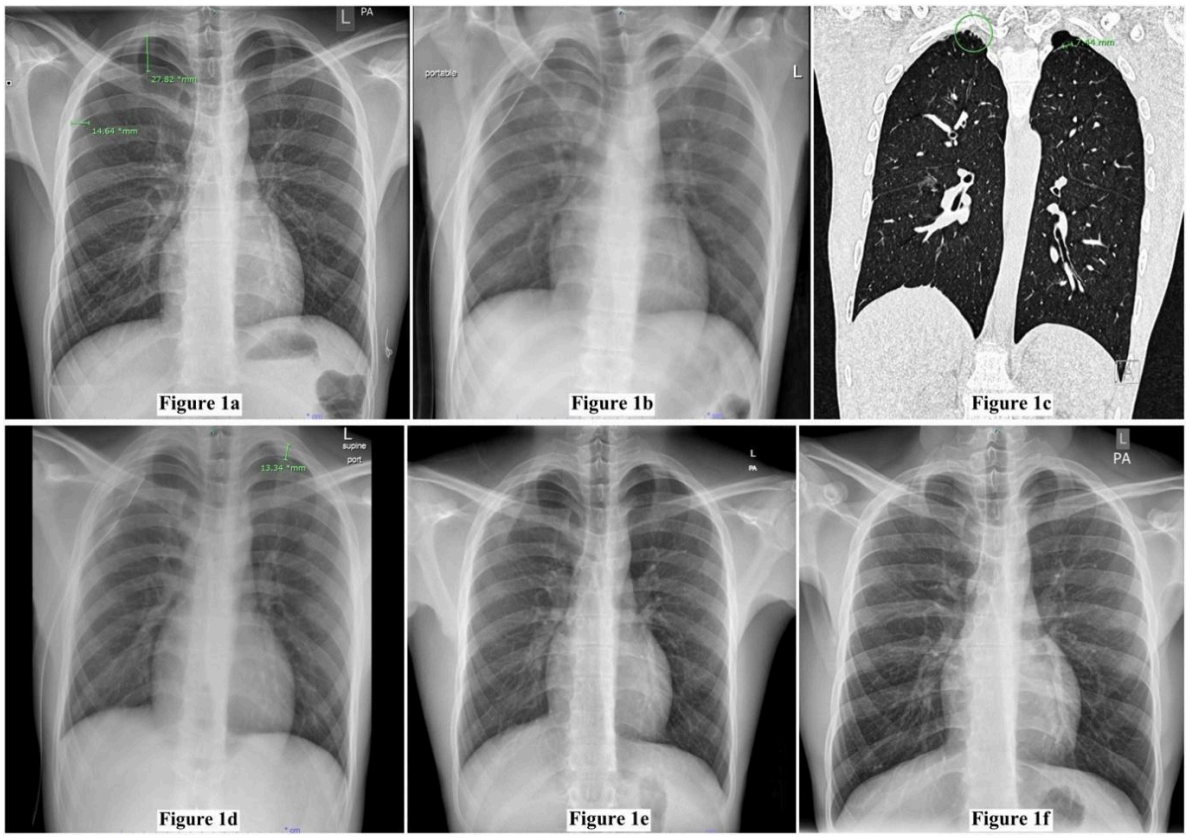

Figure 1: Serial Radiological Imaging Demonstrating the Resolution of Bilateral Pneumothorax with Interventional Management. ***Foot note:** Figure 1. Imaging progression: (a) Chest X-ray showing bilateral pneumothorax, larger on the right (27.82×14.64 mm; linear distance measurement), with normal cardiac silhouette, mediastinum, and bony structures; (b) post-insertion chest X-ray with right-sided chest tube showing reduced right pneumothorax and no significant change on the left; (c)*

CT thorax revealing right apical pleural thickening, a multiloculated bleb, minimal right pneumothorax, intercostal drain in place, and a small left apical pneumothorax, with normal lung parenchyma; (d) supine X-ray on day 3 showing stable right pneumothorax and decreased left pneumothorax (13.34 mm); (e) post-ICD removal X-ray showing marked improvement with expanded lung fields; and (f) PA X-ray at two months showing clear lungs with no residual pneumothorax and normal cardiac and skeletal structures.

Table 1: Hematological and Biochemical Laboratory Investigations, Genetic workup, and Electrocardiogram Findings at Presentation, Interim, and Follow-Up.

Variables	At presentation	At Interim	At 6 follow up
Haemoglobin (11.0-14.5; 10^12/L)	13.0	12.8	14.1
Haematocrit (0.350-0.450 L/L)	0.438	0.418	0.476
Platelet count (150-450; 10 ^9/L)	257	236	289
White Cell Count (2.4-9.5; 10 ^9/L)	5.2	13.2	4.8
C- Reactive Protein (0-5 mg/L)	6	55	<1
Sodium (135-145 mmol/L)	138	141	138
Potassium (3.5-5.1 mmol/L)	4.3	5.1	4.3
Creatinine (45-84 µmol/L)	76	64	75
Estimated GFR-MDRD (ml/min/1.73 m^2)	>90	>90	>90
Troponin (< 14ng/L)	6	<3	-
Prothrombin Time (PT) (10.3 – 12.1 seconds)	11.3	-	-
INR (International Normalized Ratio) (0.90 - 1.10)	1.04	-	-
Activated Partial Thromboplastin Time (APTT) (25.8 - 36.2 seconds)	37.3	-	-
Fibrinogen – CLAUSS method (1.7 - 3.6 g/L)	2.2	-	-
Thrombin Time (12.8 - 17.6 seconds)	17.5	-	-
Whole Exome Sequencing - CentoXome® Solo* (*CENTOGENE GmbH. Am Strand 7. 18055 Rostock, Germany)*			
Folliculin (FLCN) gene: Disease-Causing Gene for Birt–Hogg–Dubé syndrome and familial spontaneous pneumothorax.	No clinically relevant variants related to the phenotype were seen.		
FBN1 (encodes the gene for fibrillin-1) gene: Disease-Causing Gene for Marfan Syndrome.	No clinically relevant variants related to the phenotype were seen.		
PRDM10 gene: involved in Neuronal development, Transcriptional control, Chromatin modification	No clinically relevant variants related to the phenotype were seen.		
12 leads Electrocardiogram	Normal sinus rhythm	Normal sinus rhythm	

**Footnote: Coverage of the coding regions ±10 base pairs at 20x nucleotide depth for the genes of interest (PRDM10, FLCN, FBN1) is 99.95%. PRDM10 is involved in neurodevelopmental disorders, FLCN is linked to Birt-Hogg-Dubé syndrome, and FBN1 is associated with Marfan syndrome. Sequencing is recommended annually or if there are any phenotypic changes*

Treatment: Following the insertion of an ICD tube, a computed tomography (CT) scan of the thorax was performed. The scan revealed bilateral multiloculated pleural blebs, with the right-sided ICD in situ, accompanied by a minimal bilateral pneumothoraces.

Outcome and follow-up: The patient tolerated the procedure without complications. Serial chest X-rays were performed (see Figure 1), showing no progression of the pneumothorax. The chest drain was removed on post-procedure day 3, and the patient was discharged on day 4 with oral antibiotics [Figure 1]. At the six-month follow-up, the patient remained clinically stable and resumed normal activities.

Discussion

BPSP can be asymptomatic or present acutely with dyspnea, pleuritic chest pain, and anxiety, as seen in this case.[4,5] Larger pneumothoraces may produce more severe symptoms, such as tachycardia and hypotension. These indicate increased intrapleural pressure exceeding atmospheric levels, potentially causing mediastinal shift, decreased venous return, and cardiovascular collapse.[3] Notably, about 90% of PSP cases are linked to pulmonary blebs or bullae, often raising the need for bullectomy due to their risk of rupture and impaired lung expansion.[5,6] While unilateral PSP is more common, BPSP is rare, occurring in only 1-2% of cases, making this presentation clinically significant.[1,2] Pneumothorax occurring without trauma or iatrogenic cause, has a well-recognized genetic component. Familial spontaneous pneumothorax (FSP) accounts for 10–12% of cases, with FLCN mutations—associated with Birt–Hogg–Dubé syndrome (BHDS)—responsible for 17–50%.[7] Sporadic cases comprise the majority (88–90%), yet mutations in FLCN and FBN1 are still detected in up to 10% and 5% of cases, respectively.[7] Pneumothorax is also a feature of several genetic syndromes, broadly categorized into those involving tumour suppressor gene mutations (e.g., BHDS, tuberous sclerosis complex/lymphangioleiomyomatosis), connective tissue disorders (e.g., Marfan syndrome, vascular Ehlers-Danlos, Loeys-Dietz, homocystinuria, cutis laxa), and syndromes affecting lung architecture (e.g., alpha-1 antitrypsin deficiency, cystic fibrosis).[7] In Marfan syndrome, 5–11% develop pneumothorax due to apical bullae or chest wall deformities related to fibrillin defects. BPSP has also been linked to conditions such as pulmonary metastases, tuberculosis, Langerhans cell histiocytosis, and Chronic Obstructive Pulmonary Disease. Additional rare genetic associations include Sotos syndrome, spinocerebellar ataxia type 1, TERT mutations, and hereditary mucoepithelial dysplasia.[7] Additionally, BPSP may be iatrogenic or drug-induced.[1,3,7] In this case, these differentials were ruled out by history, WES and genetic testing. Imaging is essential for diagnosis. While upright posteroanterior and lateral chest radiographs offer around 70% sensitivity, CT provides 100%.[8,9] In this patient, CT revealed right apical pleural thickening, multiloculated right apical pleural blebs, a small left apical pneumothorax, and a 7-mm multiloculated left apical bleb. In patients without comorbidities, BPSP is usually due to ruptured subpleural blebs, as suspected here, though the exact pathophysiology remains unclear.[1,2,5] The absence of left-sided chest pain was likely due to the small size and apical location of the pathology. As the patient remained asymptomatic and serial chest X-rays showed a stable pneumothorax, conservative management was deemed appropriate, and an ICD was not required. ICD insertion remains the first-line treatment for symptomatic PSP, as was done here.[3,6]

Patients with BPSP have a higher risk of recurrence than those with unilateral cases—up to 50%, particularly when subpleural blebs are present. Despite this, prognosis is generally favorable in otherwise healthy individuals without underlying genetic or connective tissue disorders. Close clinical follow-up is essential. Surgical options such as video-assisted thoracoscopic surgery (VATS) with pleurodesis, bullectomy, or apical pleurodesis may be considered in recurrent or persistent cases, as they significantly reduce the risk of recurrence.[10] In patients with stable imaging and no progression, conservative management can yield good long-term outcomes. According to British Thoracic Society (BTS) guidelines, surgical intervention is recommended for recurrent ipsilateral or contralateral pneumothorax, synchronous bilateral pneumothorax, persistent air leak beyond 5–7 days, spontaneous hemothorax, and in high-risk individuals such as pilots, divers, or pregnant patients.[10] In this case, the absence of an air leak by day two permitted chest tube removal after an additional day of observation. Given the small size of the blebs and marked radiological improvement, surgery was not indicated. The patient has been thoroughly counselled regarding recurrence risk and the potential need for future intervention. He remains under close follow-up, and both he and his family are fully informed about the clinical course and available management options.

Key points:

- Bilateral primary spontaneous pneumothorax (BPSP) is rare, accounting for only 1–2% of PSP cases, and can present with unilateral symptoms, as in this patient.

- A Marfanoid habitus may suggest an underlying connective tissue disorder, warranting further genetic evaluation.

- Imaging, especially CT, is essential for identifying multiloculated blebs and guiding management decisions.

- Most BPSP cases without comorbidities are due to ruptured subpleural blebs; surgical intervention is reserved for persistent or recurrent cases.

- In this case, conservative management was successful following right-sided ICD insertion, with no recurrence at 6-month follow-up.

Conclusion

This case highlights the challenges of managing BPSP and stresses the importance of adhering to thoracic surgical guidelines for optimal outcomes.

Disclosure

No conflict of interest. We confirm that we obtained written consent from the patient to disclose clinical details in this report. The patient understands that their name and initials will remain confidential, and while we will protect their identity as much as possible, complete anonymity cannot be guaranteed.

Acknowledgement

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