1. Introduction

Neurofibromatosis Type I (NF1), or Von Recklinghausen’s disease, is an autosomal dominant genetic disorder caused by a mutation in the NF1 gene. Its diagnosis relies on well-defined clinical criteria. Cutaneous involvement, including café-au-lait spots, axillary and inguinal freckling, and cutaneous neurofibromas, is prominent [1]. Pulmonary involvement, though less commonly reported, occurs in 10 to 20% of cases [2] and presents with apical air-filled bullae and/or cysts along with diffuse interstitial lung disease (DLD), typically bilateral with basal predominance. [3,4] Spontaneous pneumothorax, although rare in NF1 patients, can be a complication of underlying bullous lung disease. In our case, this complication serves as an indicator of pulmonary involvement.

2. Case Presentation

We present the case of a 33-year-old patient with a history of NF1 diagnosed in childhood, characterized by diffuse cutaneous neurofibromas, a right spontaneous pneumothorax in 2015 with isolated subpleural blebs noted on thoracic computed tomography (CT) at that time, and active smoking habit estimated at 11 pack-years. The patient presented with acute thoracic pain and dyspnea upon admission. He was clinically stable but chest X-ray revealed a complete compressive right pneumothorax, suggestive of a recurrent spontaneous secondary pneumothorax (SSP) in the context of NF1. Emergency thoracic drainage was performed, leading to favorable evolution with total lung re-expansion. Subsequent thoracic computed tomography scan (CT) showed numerous bilateral large air-filled bullae and cysts. Alpha-1-antitrypsin level was negative. Despite the theoretical indication for pleural symphysis, surgery was not considered beneficial in this case, due to technical challenges posed by severely pathological underlying lung and the risk of complications, making future lung transplantation difficult. Short-term evolution was favorable, with drain removal after 4 days, and follow-up will be ensured at a specialized NF center.

3. Discussion

Neurofibromatosis represents a heterogeneous disease affecting multiple systems such as the skin, eyes, lungs, bones, and nervous system. NF1, the most common form, is an autosomal dominant disorder, inherited in 50% of cases while the remaining 50% arise from de novo mutations. [3] Pulmonary involvement in NF1 typically manifests as apical bullous and cystic lesions and/or bilateral, symmetrical, predominantly basal interstitial parenchymal involvement. These lesions are associated with increased tissue fragility, raising the risk of air bleb rupture and subsequent pneumothorax development. [5] Spontaneous pneumothorax complication related to NF1 pulmonary involvement is reported in 5 to 10% of cases [4,6]. Histological studies also reveal neurofibromas affecting surrounding lung tissues, including bronchi and blood vessels, leading to structural alterations and increased susceptibility to respiratory complications, including spontaneous pneumothorax. The association between spontaneous pneumothorax and NF1 has been debated, largely due to smoking's impact on pulmonary parenchymal disease, particularly emphysema [3,4]. However, the incidence of SSP in NF1 appears to be independent of toxic habits. [3,7-10]. The diagnosis of pneumothorax should be suspected in a patient with NF1 presenting with acute respiratory signs such as dyspnea and thoracic pain. Therefore, a focused clinical and radiological evaluation is necessary given the diagnostic urgency and the potentially fatal risk of unfavorable progression. In our case, a recurrence of pneumothorax was necessary to establish the connection between this complication and the underlying
NF1 disease, in order to raise awareness regarding the imperative need for smoking cessation and to direct the patient towards specialized care. This observation underscores persistent gaps in the understanding of this association among healthcare professionals, thus highlighting the crucial role of publications such as this. Management of pneumothorax in NF1 patients often involves thoracic drainage to facilitate the evacuation of accumulated air and restore normal intrapleural pressure [11]. In cases akin to the one presented, the potential benefit of surgical intervention, sometimes required to repair underlying pulmonary lesions and prevent pneumothorax recurrences, should be should be deliberated in multidisciplinary discussions.

Figure

4. Conclusion
While spontaneous pneumothorax occurrences are uncommon among individuals with NF1, they may signal underlying pulmonary issues necessitating specialized care. A multidisciplinary approach involving genetics, pulmonology, and thoracic surgery specialists is crucial for early diagnosis and management of pulmonary complications in NF1 patients. Healthcare professional awareness of this clinical association is essential for early detection of pulmonary involvement and prevention of complications in NF1 patients.

References