Recurrent pneumothoraces and facial papules: An insidious pulmonary cystic disease

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CASE PRESENTATION
A 30-year-old, Caucasian, nonsmoking woman presented with a six-month history of bilateral recurrent spontaneous pneumothoraces (PNXs). Previous treatments (bilateral chest drains and iterative chemical pleurodesis) had been attempted; however, only three months after the most recent surgical procedure, the patient presented once again with increasing dyspnea, chest pain, and radiological detection of a right PNX associated with bilateral multiple, thin-walled pulmonary cysts varying in size and shape and predominantly distributed in the lower pulmonary lobes (Figure 1). Physical examination revealed multiple, whitish, smooth, dome-shaped papules on her face (Figure 2) and upper trunk.

Surgery was, thus, indicated and a pulmonary bullectomy combined with a complete right pleural decortication was successfully performed via right thoracotomy.

Intraoperatively, multiple cysts were observed in the lung parenchyma surface (Figure 3A). Histopathological microscopic analysis of the pulmonary cysts (Figure 3B) revealed the presence of so-called ‘alveolar cysts’ or ‘few alveoli within a cyst’ (1), with a characteristic pattern marked by the inner surface lined by flattened respiratory epithelium/cuboidal cells (Figure 3C) with signs of chronic inflammation, interstitial bleeding and hyalinization. Ten months later, no radiological signs of recurrent PNX were observed.

Birt-Hogg-Dubé (BHD) syndrome was suspected and confirmed by genetic tests when the patient underwent (at a different centre) right partial nephrectomy for renal oncocytoma nine years earlier. The surgical management of recurrent PNX in such cases is an extremely challenging issue: first, because of the failure of standard surgical procedures (as in the present case) and, second, due to the fact that, because bilateral PNXs associated with pulmonary cysts are not extremely rare in young people, BHD syndrome may be confused with other more common entities causing PNX.
KEY LEARNING POINTS
- First described in 1977, BHD syndrome is a rare autosomal dominant multiorgan disorder (prevalence: 0.5 per 100,000) caused by mutations in the folliculin gene on chromosome 17p11.2.
- Patients affected by BHD syndrome present with the ‘diagnostic triad’: skin fibrofolliculomas/trichodiscomas, pulmonary cysts and renal tumours with early onset.
- Lung cysts have been described in 77% to 89% of BHD syndrome patients, while the occurrence of PNX could be predicted in approximately 33% to 38% of such patients. The right lung is more often affected, although both lungs are involved in approximately 23% of cases; pulmonary cysts, typically variable in size and shape, are generally located in lower lobes (2).
- The differential diagnosis of BHD syndrome should include other syndromes that may present with PNX and lung cysts such as alpha-1 antitrypsin deficiency, Marfan syndrome, Ehlers-Danlos syndrome, cystic fibrosis and lymphangio leiomyomatosis.
- In BHD syndrome patients, the treatment of PNX (primary occurrence) consists substantially of standard surgical procedures (chest drain placement); otherwise, in case of recurrent PNX, a pulmonary bullectomy combined with complete pleural decortication should be considered.

REFERENCES