Clinical picture

An inherited cause of pneumothorax—the Birt–Hogg–Dubé syndrome

A 38-year-old woman presented in 2004 a spontaneous pneumothorax (Figure 1a). Outcome was favorable with treatment with chest tube drainage. Eight years later, she complained of dyspnea. A computed tomography scan of the chest revealed lung cysts (Figure 1b). She presented 2–4 mm, white, dome-shaped papules involving the neck, which appeared during the last 2 years. Her brother also presented multiple skin lesions involving the head and neck. In both cases, biopsy of the dermatologic lesions showed fibrofolliculoma. A diagnosis of Birt–Hogg–Dubé syndrome was made, confirmed by identification of mutation in the folliculin gene FLCN.

Birt–Hogg–Dubé syndrome is a rare, autosomal dominantly inherited disease, characterized by multiple cutaneous hamartomas (mainly fibrofolliculomas and trichodiscomas) and increased susceptibility to renal neoplasms, pulmonary cysts and spontaneous pneumothoraces. It is caused by mutations in the FLCN gene, which encodes a tumor-suppressor protein, folliculin.

Lung cysts are mostly bilateral and multifocal. Most individuals with Birt–Hogg–Dubé syndrome and lung cysts are asymptomatic, but they have a high risk of developing spontaneous often recurrent pneumothorax. All individuals with a history of pneumothorax had multiple lung cysts identified by chest computed tomography imaging. Several inherited and non-inherited conditions can present with lung cysts and/or pneumothorax. A thorough history and physical examination help to differentiate these conditions from Birt–Hogg–Dubé syndrome. These conditions include Marfan syndrome, vascular Ehlers–Danlos syndrome, alpha 1-antitrypsin deficiency, Langerhans cell histiocytosis, lymphangioleiomyomatosis, which can occur as an isolated finding or as part of tuberous sclerosis complex.

In case of respiratory manifestations, it is important to consider the diagnosis of Birt–Hogg–Dubé syndrome, because individuals with Birt–Hogg–Dubé syndrome had an increased risk of renal tumors and lifelong renal surveillance is recommended.

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Conflict of interest: None declared.

Figure 1. (a) Chest radiography: right pneumothorax. (b) Chest computed tomography scan: lung cysts.
References


