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【 PICTURES IN CLINICAL MEDICINE 】

Basal Lung Cysts in Birt-Hogg-Dubé Syndrome

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Key words: lung cysts, Birt-Hogg-Dubé syndrome, pneumothorax, folliculin gene

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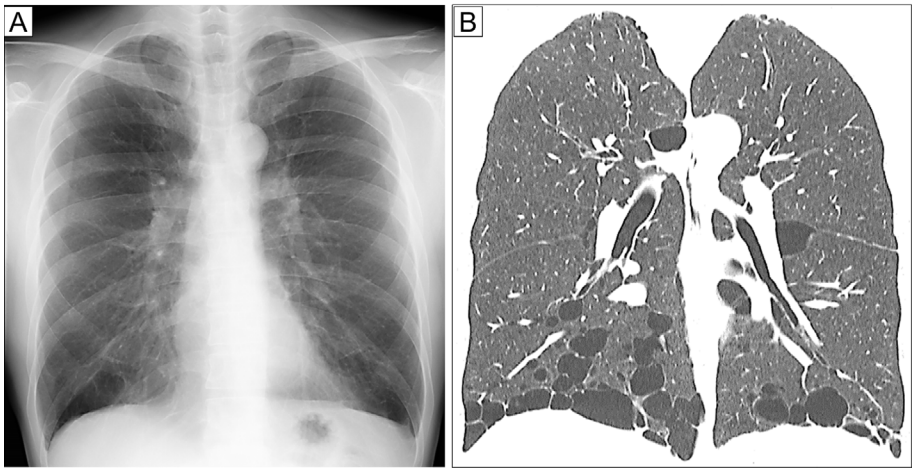

Picture.

A 45-year-old non-smoking man with a history of pneumothorax presented with repeated chest pain. Two of his family members also had a history of pneumothorax. Chest radiography showed decreased attenuation in the bilateral lower lobes (Picture A). Coronal computed tomography revealed multiple cysts in the bilateral lung bases (Picture B). He was diagnosed with Birt-Hogg-Dubé syndrome (BHDS) by an *FLCN* mutation analysis. BHDS is a rare autosomal dominant inherited disease associated with recurrent pneumothorax, cutaneous fibrofolliculomas, and renal tumors (1, 2). Familial pneumothorax is rare, and the differential diagnosis includes BHDS, Marfan syndrome, and Ehlers-Danlos syndrome (2). The key imaging finding for suspecting BHDS is the distribution of cysts. Cysts in patients with BHDS are predisposed to occur at the lung bases or around periacinar lesions, differing from the typical apical location of common causes of spontaneous pneumothorax, such as bullae/blebs or smoking-related emphysema (2).

The authors state that they have no Conflict of Interest (COI).

References

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