CLINICAL SNAPSHOT

Pulmonary Bullae as an Indicator of an Elevated Risk of Renal Carcinoma

This 46-year-old woman had sustained spontaneous pneumothoraces on four separate occasions since the age of 19 because of multiple pulmonary bullae. Conditions considered in the differential diagnosis had included lymphangioleiomyomatosis and histiocytosis X. Independently of this problem, she consulted a dermatologist because she had noted increasing numbers of whitish "millet seeds," 1-2 millimeters in diameter, on her face over the past 10 years. Histological examination revealed these to be fibrofolliculomata, which, in combination with the pulmonary bullae and spontaneous pneumothoraces, led to the suspected diagnosis of Birt-Hogg-Dubé syndrome (BHDS), a condition of autosomal dominant inheritance. The diagnosis was confirmed by the demonstration of a typical *FLCN* mutation (c.1285dupC). BHDS patients have a 15–20% lifetime risk of renal cancer; screening every six months is recommended, with MRI in alternation with ultrasonography.

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- a) Close-up of the face, with fibrofolliculomas.
- b) Coronal reformatted of a high-resolution CT (HRCT) of the lung reveals a caudal pneumothorax with septations. Multiple bullae can be seen, mainly located subpleurally.