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Clinical picture

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Renal tumor associated with pulmonary cysts: Birt–Hogg–Dubé syndrome

Case report

A previously healthy 66-year-old man presented with a 3-month history of prostatism symptoms. He had a familial history (in his daughter and sister) of renal neoplasia. Physical examination findings were normal, except for the presence of multiple white and skin-colored papules distributed over the face and neck, consistent with fibrofolliculomas. Laboratory test results were unremarkable. Ultrasound of the urinary tract showed a well-defined, heterogeneous exophytic mass measuring about 6.5 cm, related to the upper pole of the left kidney. Chest computed tomography for staging showed multiple elongated lung cysts, predominantly in the lung bases, and a rounded mass in the left kidney (Figure 1). The patient underwent complete surgical excision of the renal tumor. The histopathological and immunohistochemical diagnosis was renal cell carcinoma with oncocytic cells. The association of clinical and imaging findings with the patient's familial history allowed the diagnosis of Birt–Hogg–Dubé syndrome (BHDS). The patient recovered well, and he and his family members were referred for genetic counseling and oncological followup.

Discussion

BHDS is an autosomal dominant condition caused by germline mutations in the folliculin (FCLN) gene and characterized by skin fibrofolliculomas, multiple lung cysts, spontaneous pneumothorax and renal tumors.[1–3] BHDS is probably underdiagnosed because of the wide variability in its clinical

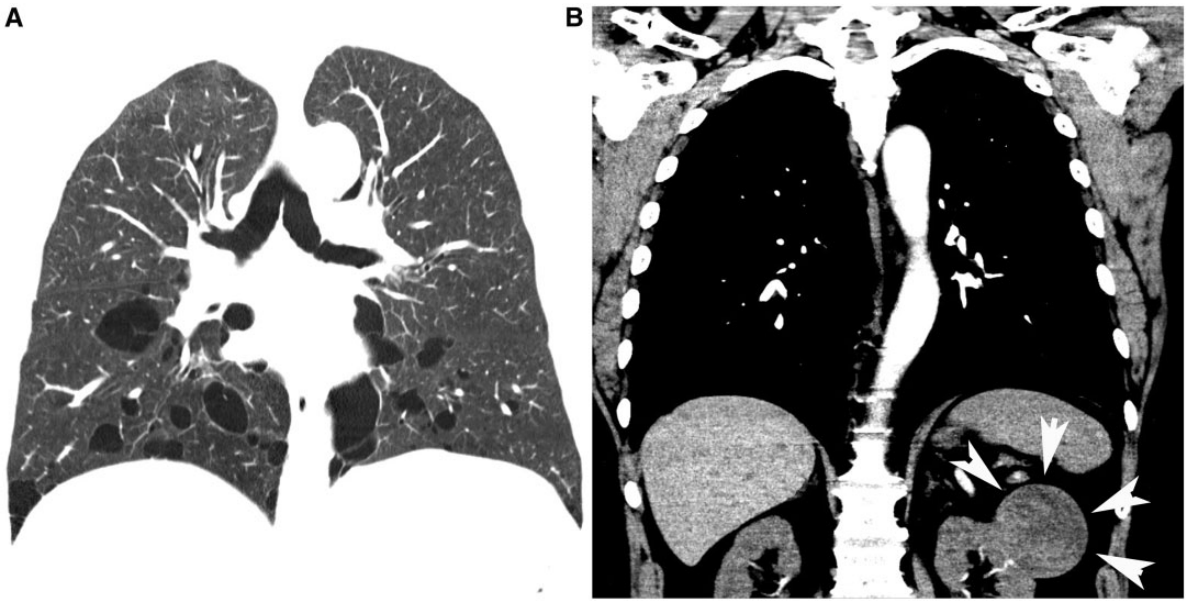

Figure 1. Reformatted coronal chest computed tomography image obtained in the lung window (**A**) demonstrates multiple elongated lung cysts, predominantly in the lung bases, and a reformatted coronal image obtained in the mediastinal window (**B**) shows a rounded mass in the left kidney (arrowheads).

expression. Patients may present with renal tumors or pneumothorax, which most often occur sporadically.[3] BHDS was formerly defined by the presence of at least 5–10 fibrofolliculomas and histological diagnosis based on at least one papule. In families with cystic lung disease, pneumothorax, or renal cancer alone, a definite diagnosis of BHDS can be made only by detection of a pathogenic FCLN germline mutation.[3] Detection of such a mutation not only confirms the diagnosis in the index patient but also allows pre-symptomatic testing of unaffected at-risk relatives.[3]

The most threatening complication of BHDS is renal cancer.[3] Renal tumors in patients with BHDS are often multiple and bilateral, and include hybrid chromophobe oncocytomas and chromophobe carcinomas. However, clear cell and papillary renal cell carcinoma have also been reported.[2,4] Fibrofolliculomas are the most characteristic skin finding of BHDS and typically appear as multiple dome-shaped papules on the head, neck, face and upper trunk.[3,4] Pulmonary findings in patients with BHDS are bullous emphysema, thin-walled cysts and pneumothorax. Pathologically, the cysts are often elongated and subpleural in distribution, most often in the basal lung regions.[1,4]

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Conflict of interest: None declared.

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