

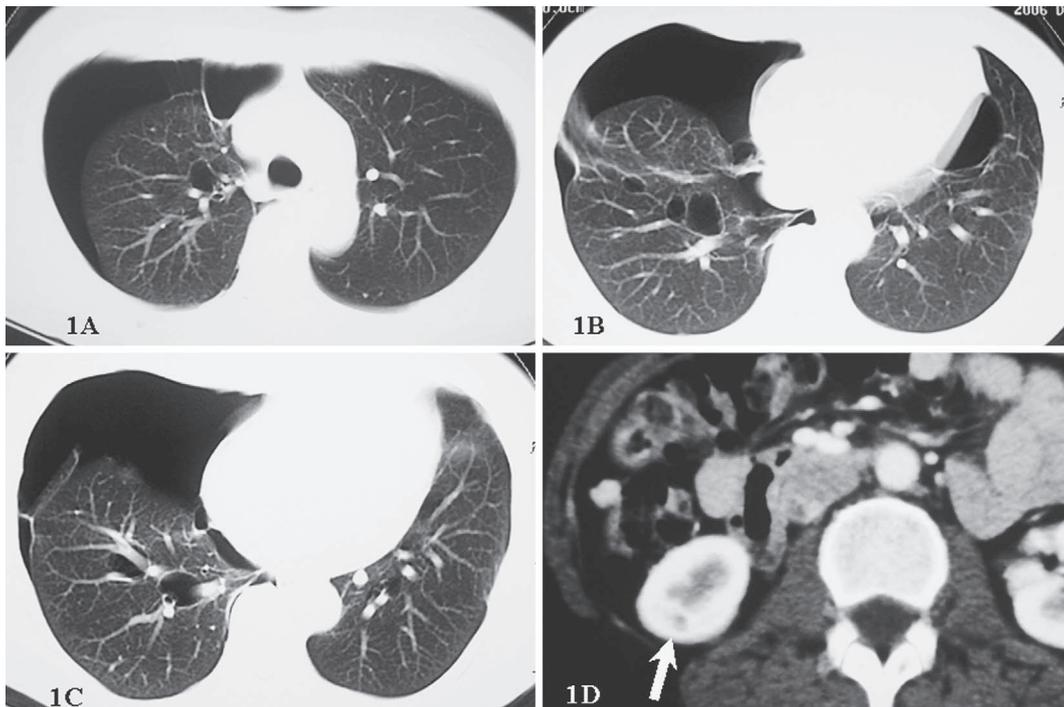
## Birt-Hogg-Dubé Syndrome with Renal Angiomyolipoma

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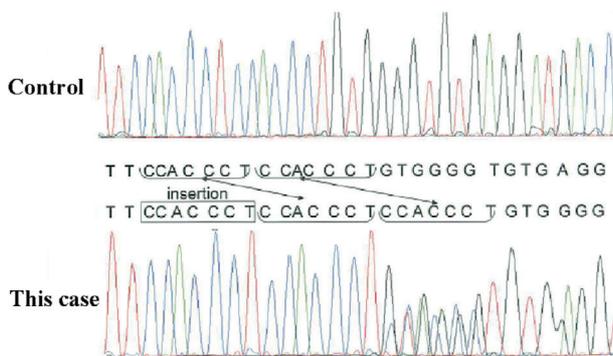
**Key words:** Birt-Hogg-Dubé syndrome, lymphangioleiomyomatosis, pneumothorax, cystic lung disease, angiomyolipoma

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Picture 1.



Picture 2.

A 43-year-old woman who was referred to our clinic had a right-sided pneumothorax, multiple pulmonary cysts and a right renal tumor. The preliminary diagnosis was possible lymphangioleiomyomatosis (LAM). Previously, she had a pneumothorax that recapitulated a similar family history in her son, grandfather and cousin. The patient's CT images revealed multiple pulmonary cysts with round or irregular shapes predominating in the lower-medial zone of both lungs (Picture 1A) and a right renal tumor containing fat density characteristic of angiomyolipoma (Picture 1D). A surgical lung biopsy contained cysts without LAM cells; subsequent genetic testing revealed a CCACCCT insertion

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in exon 12 of *FLCN* gene (chromosome 17p11.2) (Picture 2), confirming the diagnosis of Birt-Hogg-Dubé syndrome (BHDS). BHDS and LAM share some clinical features (pneumothorax with multiple pulmonary cysts, renal tumor and skin lesions), but a family history of pneumothorax and the characteristics of cysts in chest CT (multiple, irregular-shaped cysts of various sizes with lower medial lung zone predominance) facilitate their differentiation (1, 2).

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

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