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## A Rare Cystic Lung Disease

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**Introduction:** Birt-Hogg Dube syndrome (BHDS) is a rare genetically inherited autosomal dominant disorder, characterized by pulmonary cysts, spontaneous pneumothorax, benign skin tumours (fibrofolliculoma, trichodiscoma) and renal tumors. It is caused by mutations in the folliculin coding gene (FLCN).

**Case Presentation:** A 77-years old female, was admitted with exertional dyspnea for the last two years. She was a never-smoker and operated for colon cancer 10 years ago. Physical examination revealed bilateral diminished breathing sounds and multiple papular lesions involving the nose and cheeks. Bilateral hyperinflation was evident on chest x-ray. Pulmonary function tests showed hyperinflation and airtrapping. Thoracic CT scan revealed bilateral multiple well-defined lung cysts various in shape and size. Histopathological examination of a dermal punch biopsy taken from multiple papular lesions on her face revealed trichodiscoma. Upon questioning, the patient described that her daughter had fibrofolliculoma-like tumors in the face and spontaneous pneumothorax history. Her son was diagnosed with clear cell renal cell carcinoma and had also ground glass appearances on chest CT-scan. Genetic screening revealed that the patient had a known heterozygote mutation in the folliculin gene, encoded by FLCN (c.1285dupC). BHDS is diagnosed on the clinical presentations and the identification of a FLCN germline mutation in the genetic study. The FLCN gene is expressed in skins, nephrons and pneumocytes. There is an increased risk of renal tumors in BHDS. For that reason, an abdominal ultrasound was performed, with no pathological findings. Patients with BHDS should be informed with the increased incidence of renal tumors.

**Conclusion:** BHDS is rare hereditary syndrome with an increased risk of benign tumours of hair follicles, multiple lung cysts and increased risk of renal cancer. It is important to evaluate BHDS in the presence of lung cysts with accompanying cutaneous lesions. Lung cysts are common, but they are usually asymptomatic unless a pneumothorax is present. The patients should be checked up to diagnose and treat renal malignancies before metastatic disease occurs. As a result when lung cysts are encountered on patients chest CT scan, BHDS should be also considered in differential diagnosis. In such patients, clinicians should keep in mind that accompanying kidney and other tumors should be kept in mind and such involvements be investigated. Moreover, family counselling and follow-up is required for the family.

**Keywords:** Birt-Hogg-Dubé-Syndrome, cystic lung disease, trichodiscoma, FLCN