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***Research paper***

# A Case of Spontaneous Pneumothorax: Don't Miss Birt-Hogg Dubé Syndrome!

**Faouzia Kambouche *, Ikhlas Belabbas**

*Department of Pulmonology, university hospital center of Mostaganem,*
*faculty of medicine of Mostaganem –Algeria*
**Corresponding author E-mail: ryadouzlifi@yahoo.fr*

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**Abstract**

Birt-Hogg-Dubé syndrome (BHDS) is a rare autosomal-dominant multiorgan systemic disorder manifesting as cutaneous fibrofolliculomas, lung cysts with or without spontaneous pneumothorax, and renal tumors. It results from mutation of the gene located on the short arm of chromosome 17 (17p11.2). The gene codes for the protein folliculin, which is believed to be an oncogene suppressor protein. we report a case of a 66-year-old male who was suspected to have the diagnosis of BHDS based on recurrent pneumothorax , family history of pneumothorax and characteristic features of lung cysts on the Chest CT, subsequently confirmed by genetic testing.

***Keywords***: *Birt-Hogg-Dubé Syndrome; Cystic Lung Disease; FLCN Gene.*

## 1. Introduction

First described in 1977 by three Canadian dermatologists, Birt-Hogg-Dube syndrome (BHD) is an autosomal dominant genetic disorder, often revealed by idiopathic pneumothorax. In its typical presentation, it combines cystic lung lesions, fibrofolliculoma-like skin manifestations, and a high risk of renal tumors. Although it is rare (probably around 1/200,000), its recognition is important in terms of monitoring and genetic counseling.

## 2. Observation

We report the case of a 66-year-old patient presenting with a spontaneous total right pneumothorax requiring chest drainage. His family history is notable for the occurrence of spontaneous pneumothorax in his two brothers. Clinical examination revealed the presence of wart-like skin lesions on the face and back. The chest CT scan identified multiple pulmonary cysts, predominantly in the apical and paramediastinal regions (Figure 1), and also led to the incidental discovery of a left renal tissue process (Figure 2). Fibrofollicular skin lesions on the face and neck had been present for many years.
The Genetic testing for the FLCN gene revealed heterozygous pathogenic mutations: c.1285del and p.His429Thrfs*39, confirming the diagnosis of Birt-Hogg-Dubé syndrome. The Management included intercostal drainage tube insertion for pneumothorax and symptomatic care. Post-discharge recommendations included pneumococcal and annual influenza vaccinations, pulmonary function monitoring, renal tumour screening, and genetic counselling for family members.

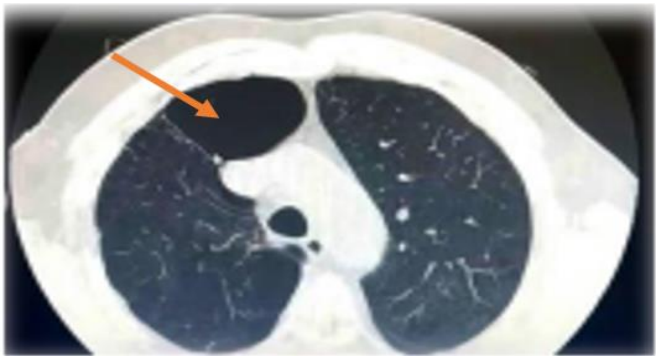

**Fig. 1:** The CT Chest without Contrast : Axial Image Showed A Multiple Pulmonary Cysts, Predominantly in the Apical and Paramediastinal Regions with Preserved Lung Volume and No Evidence of Interstitial Lung Disease.

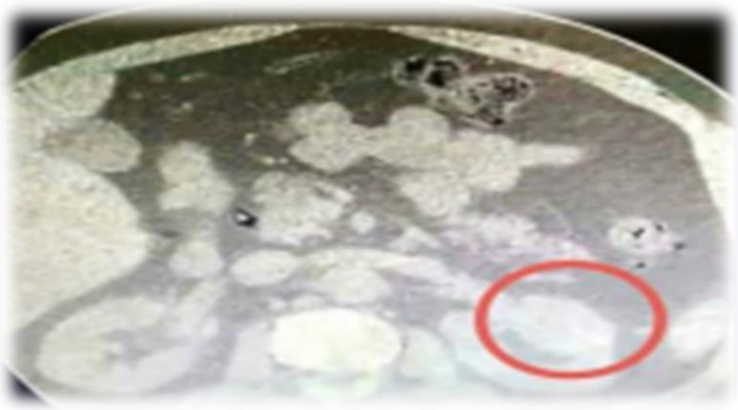

**Fig. 2:** The CT Abdominal without Contrast : Axial Image Showed A Left Renal Tissue Process

## 3. Discussion

Birt-Hogg-Dubé (BHD) syndrome, a rare genetic disorder with autosomal dominant inheritance, was initially described in 1977. This condition is characterized by the presence of lung cysts, spontaneous pneumothorax, renal cancer, and skin fibrofolliculomas [1]. The protein associated with BHD syndrome is extensively distributed across various tissues, including the kidneys, lungs, and skin [2 ]. The precise incidence of this syndrome remains unknown. This inherited condition results from mutations in the folliculin (FLCN) gene on chromosome 17p11.2, which codes for the folliculin protein. This protein plays a vital role in cellular signaling pathways, such as the mechanistic target of the rapamycin (mTOR) pathway, which is involved in cellular growth and multiplication [3]. Diagnosis of cystic lung disease is challenging. When coupled with dyspnea and a history of spontaneous pneumothorax, the differential can include pulmonary langerhans cell histiocytosis, lymphangioleiomyomatosis, lymphoid interstitial pneumonia, and BHD, among other etiologies. The European Reference Network for Genetic Tumour Risk Syndromes (ERN GENTURIS) and the European BHD Consortium have established diagnostic criteria for Birt-Hogg-Dubé syndrome (BHD) and consensus recommendations for its management. The diagnostic criteria involve either having a major criterion, such as one or more fibrofolliculomas, or meeting a combination of minor criteria. The consensus recommendations include offering genetic testing, performing lifelong surveillance for kidney cancer starting at age 20 using MRI, and not performing routine surveillance for other cancers unless there is a family history Recurrent pneumothoraces may be the prominent clinical manifestation of BHD, and consideration for early pleurodesis after the first spontaneous pneumothorax is indicated [4] .
When BHD has been identified in an index patient, a diagnostic work-up of family members is another important step in the management of this genetic disorder with dominant autosomal transmission.

## 4. Conclusion

When BHD is suspected, a multidisciplinary approach involving respiratory physicians, radiologists, geneticists, nephrologists, dermatologists, thoracic surgeons and pathologists is needed to confirm the diagnosis and to implement the appropriate follow-up, especially periodic kidney imaging for early detection of renal cancer.

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