

Pneumothorax and Birt–Hogg–Dube syndrome: diagnostic and therapeutic aspects

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Date accepted for publication 21 January 2007

Abstract

We report a case of spontaneous recurrent pneumothorax in a 32-year-old male with a family history of Birt–Hogg–Dube syndrome. Specific aspects of the surgical treatment for a pneumothorax within this particular setting are discussed as well as the potential underdiagnosis of this complex genodermatosis. The literature linking the syndrome to spontaneous pneumothorax is reviewed.

Keywords

Pneumothorax; Birt–Hogg–Dube syndrome.

Case report

The 32-year-old patient, employed as a full time firefighter, was referred from a District Hospital for a left spontaneous pneumothorax. He had undergone an ipsilateral video-assisted thoracoscopy (VATS) and pleurodesis a year prior to his current admission in another hospital and a pneumothorax on the contralateral side had been treated with a thoracoscopy and pleurectomy 2 years before. His mother and aunt had previously been diagnosed with Birt–Hogg–Dube syndrome and both had undergone thoracotomies for recurrent pneumothorax due to failed VATS procedures. Two further relatives had also been treated for pneumothorax, but no information could be obtained about the specific type of treatment and its outcome.

In the light of a family history of previously failed VATS attempts and despite being offered a redo video-assisted operation the patient insisted this time on having his pneumothorax treated by thoracotomy. We performed a left axillary thoracotomy through the fourth intercostal space, a wedge excision of lung parenchyma including small and medium sized bullae of the upper and lower lung lobe respectively and an apical pleurectomy. No obvious adhesions indicating a previous pleurodesis were found. A thickened apical parietal pleura was noted.

Histology confirmed multiple bullae of different sizes within the resection specimens and unspecific pleural changes.

The patient made an uneventful postoperative recovery.

A chest CT arranged 7 weeks later for an assumed recurrence was unremarkable (Fig. 1). The patient remains well into his fourth year of follow up.

Discussion

The Burt-Hogg-Dube syndrome, an autosomal dominant genodermatosis characterized by benign tumours of the hair follicle (principally fibrofolliculomas and trichodiscomas, frequently associated with acrochordons) was reported for the first time in 1977^[1]. These benign dermal neoplasms derive from the mantle of the hair follicles found on the face, neck and upper trunk and usually develop in the third or fourth decade of life as multiple small raised lesions.

Clinical and genetic studies suggest that Birt-Hogg-Dube syndrome patients may have a predisposition to renal malignancies, lung cysts and pneumothorax^[2,3].

Spontaneous pneumothorax within the Birt-Hogg-Dube syndrome is caused by the rupture of pulmonary cysts (Fig. 1). Zbar *et al.*^[3] confirmed the presence of subpleural and intraparenchymal cysts by high resolution chest computed tomography (CT) scan in 83% out of 111 affected members of Birt-Hogg-Dube syndrome families. The author reported an age-adjusted odds ratio for pneumothorax in Birt-Hogg-Dube syndrome affected individuals of 50.3 (95% confidence interval, 6.4-392), and in agreement with earlier reports found that Birt-Hogg-Dube syndrome predisposed to renal tumours and therefore screening of affected patients should be considered^[3,4].

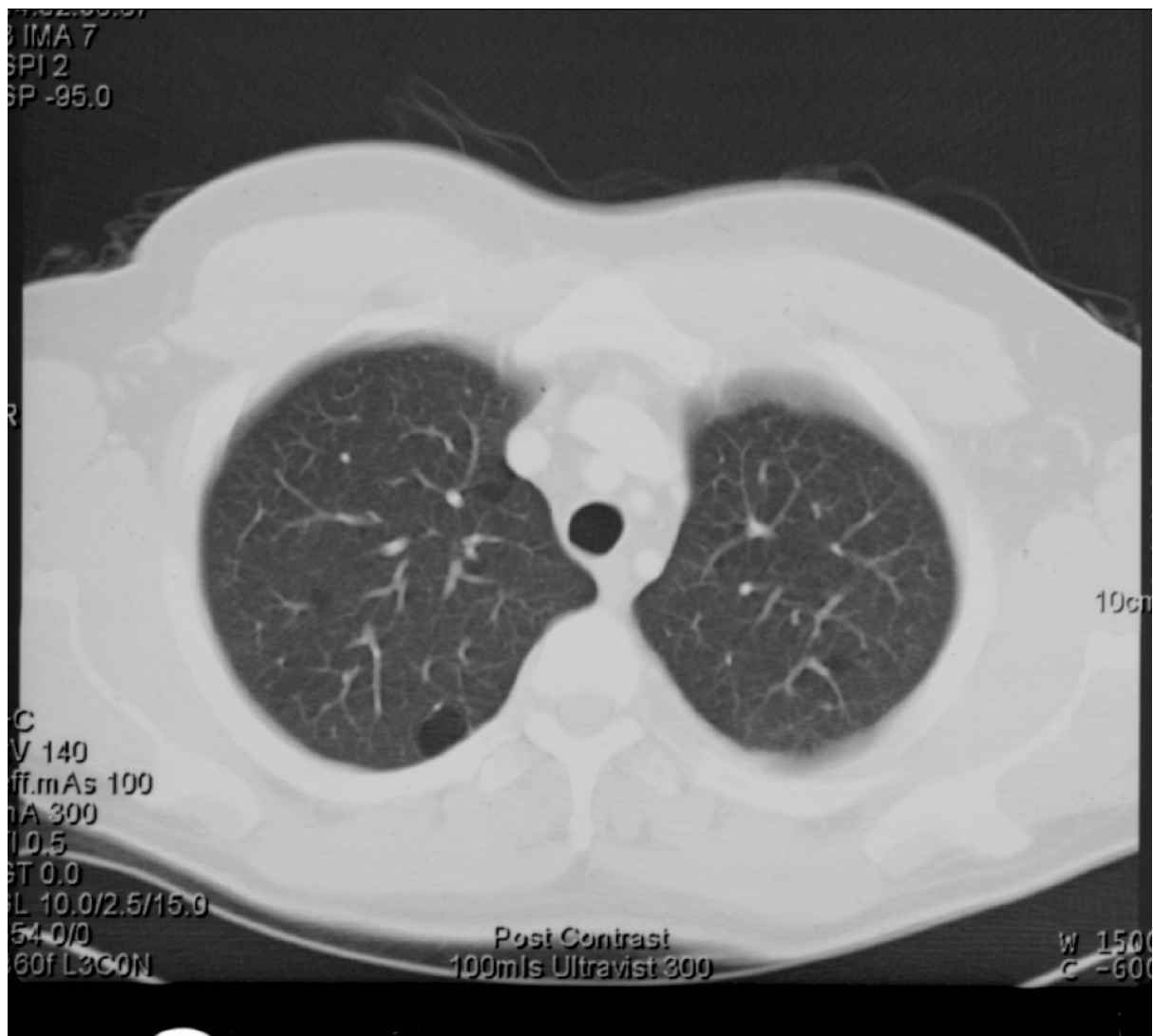


Fig. 1. Chest CT after left thoracotomy for recurrent pneumothorax in a patient with Birt-Hogg-Dube syndrome, showing right-sided subpleural cyst.

Does the high recurrence rate after VATS treatment of pneumothorax in our Birt-Hogg-Dube family (three out of three members) justify thoracotomy as the more appropriate approach for this particular patient group?

Mainly retrospective studies investigating both surgical techniques in the treatment of primary and secondary pneumothorax do not provide significant differences in outcome in terms of recurrence of disease. There is an obvious trend, however, favouring a thoracotomy in the case of secondary pneumothorax (7% recurrence rate after thoracotomy against 17% after VATS in a prospective study by Waller *et al.*^[5] including 60 patients) and in the event of pneumothorax as a second recurrence or more (recurrence rate 0% after thoracotomy against 32.2% after VATS in 56 patients in a retrospective study by Sawada *et al.*^[6]).

The reasons for the different outcomes are potentially due to more extensive adhesions caused by the open approach, and presumably a less thorough inspection of the lung during the VATS procedure with the consequence of overlooking bullae or blebs^[6].

Teaching points

Birt-Hogg-Dube syndrome is in the differential diagnosis of patients with pneumothorax, particularly in the context of a family history of pneumothorax amongst the other underlying lung conditions which need to be considered. In some patients with Birt-Hogg-Dube syndrome, pneumothorax occurs even in the absence of the typical skin lesions (due to the reduced penetrance or late age of onset of the disease)^[2].

For the group of Birt-Hogg-Dube syndrome patients with a pneumothorax, the low recurrence rate after thoracotomy should be critically balanced against the proven benefits of a VATS approach being less invasive and with a less protracted recovery time. Prevention of recurrence ought to be emphasised, because the current literature does not provide any information about potential progress of the cystic lung disease in these individuals.

References

1. Birt AR, Hogg GR, Dube WJ. Hereditary multiple fibrofolliculomas with trichodiscomas and acrochordons. *Arch Dermatol* 1977; 113: 1674-7.
2. Khoo SK, Giraud S, Kahnoski K, *et al* Clinical and genetic studies of Birt-Hogg-Dube syndrome. *J Med Genet* 2003; 39: 906-12.
3. Zbar B, Alvord G, Glenn G, *et al*. Risk of renal and colonic neoplasms and spontaneous pneumothorax in the Birt-Hogg-Dube syndrome. *Cancer Epidemiology Biomarkers Prev* 2002; 11: 393-400.
4. Schmidt LS, Warren MB, Nickerson ML, *et al* Birt-Hogg-Dube syndrome, a genodermatosis associated with spontaneous pneumothorax and kidney neoplasia. *Am J Hum Genet* 2001; 69: 876-82.
5. Waller DA, Forty J, Morrill GN. Video-assisted thoracoscopic surgery versus thoracotomy for spontaneous pneumothorax. *Ann Thorac Surg* 1994; 58: 372-6.
6. Sawada S, Watanabe Y, Moriyama S. Video-assisted thoracoscopic surgery for primary pneumothorax. *Chest* 2005; 127: 2226-30.