In conclusion, open heart surgical operations are a feasible option for cardiac disease in patients with acceptable lung function after LTx. Continuation of adequate immunosuppression, appropriate antibiotic cover, and early extubation constitute the cornerstones for successful surgical management of these patients.

References

Radiologically Indeterminate Pulmonary Cysts in Birt-Hogg-Dubé Syndrome
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Birt-Hogg-Dubé (BHD) syndrome is an inherited disease characterized by recurrent pneumothorax. We report some unusual clinicopathologic features of the lung in a Japanese family with this syndrome presenting with recurrent pneumothorax. Radiologic imaging did not show...
detectable lesions; however, at video-assisted thoracic surgery (VATS), multiple diffusely distributed microcysts were visible on the pleura. This characteristic morphologic feature was common to all affected family members. The proband underwent genetic testing and BHD syndrome was diagnosed. Although many patients with BHD syndrome with pneumothorax show obvious pulmonary cysts, this case suggests that radiologically indeterminate cysts have the potential to cause pneumothorax.

Birt-Hogg-Dubé (BHD) syndrome is an autosomal dominant disease caused by mutation in the folliculin (FLCN) gene located on chromosome 17p11.2 [1]. The major features of this inherited disorder include recurrent pneumothorax, fibrofolliculomas, and renal cell cancer [2, 3]. All individuals with an episode of pneumothorax have multiple pulmonary cysts that can be identified by thoracic computed tomography (CT). Although previous studies have reported a possible association between the mutation patterns of FLCN and the frequency of pneumothorax [4, 5], the similarities in cyst morphologic features among the affected family members have not been well documented. In this report of familial recurrent pneumothorax, the family group consists of a female proband, who was diagnosed with BHD syndrome by genomic testing, and her 2 daughters. All 3 patients underwent video-assisted thoracic surgery (VATS). Thoracic CT did not demonstrate any cystic lesions; however, all 3 patients were found to have multiple microcysts on VATS, which was confirmed by pathologic examination.

A 52-year-old female nonsmoker presented to our institute with right-sided spontaneous pneumothorax. Five members of this family group had experienced spontaneous pneumothorax, including the patient’s mother, cousin, and 2 daughters (Fig 1). The only information in the patients’ medical histories that was of some significance was the repeated episodes of right-sided pneumothorax that generally resulted in mild symptoms. The patient consented to surgical intervention for diagnostic and therapeutic purposes. Preoperative thoracic CT showed no obvious cystic lesions (Fig 2A), which was considered unusual in the lung of a patient with spontaneous pneumothorax. At VATS, however, small thin-walled cystic structures were visible and were found to be diffusely distributed on the pleura of the right lung (Fig 2B). The patient underwent pulmonary wedge resection, cautery using an Endo-FB3.0 Floating Ball (Medtronic, Inc, Minneapolis, MN), pleural ablation, and chemical pleurodesis a few days after operation. These are our standard surgical procedures for common spontaneous pneumothorax. Histopathologic examination of the resected tissue showed microcysts 3 mm in diameter (Fig 2B).

Because of the characteristic familial history of pneumothorax, BHD syndrome was suspected. DNA was obtained from peripheral blood leukocytes of the patient, and exons of the FLCN gene were amplified by polymerase chain reaction: Direct sequences revealed a heterozygous mutation in exon 12. The patient was finally
diagnosed with BHD syndrome. Neither fibrofolliculomas nor solid renal tumors were detected.

Interestingly, 2 of the patient’s daughters had also undergone VATS for pneumothorax, and their clinical records are discussed here. Daughter 1 (D1) received VATS for right-sided spontaneous pneumothorax (Fig 3A) after 5 previous episodes of bilateral pneumothorax. She also presented postoperatively with recurrence of right-sided pneumothorax, which was treated with chemical pleurodesis. Daughter 2 (D2) received VATS for right-sided spontaneous pneumothorax (Fig 3B). Preoperative thoracic CT of D1 and D2 demonstrated no detectable pulmonary cysts, similar to their mother. In these cases, the surgical findings also resembled those seen in the mother, ie, small thin-walled cystic structures diffusely distributed over the pleura (Figs 3A, 3B). Histologic analysis of the resected pulmonary tissues from both daughters demonstrated microcysts. A catamenial cause of pneumothorax was ruled out, and although D1 and D2 did not consent to genetic testing for FLCN, the common clinicopathologic features in these cases strongly suggest that the FLCN mutation demonstrated in the proband was inherited by the daughters.

Comment

BHD syndrome was named after the study by Birt and colleagues [6] in 1977 describing a familial dermatologic disorder. The genetic test for this disorder was developed over the past decade [1]. The responsible gene has been identified as FLCN, which encodes the protein folliculin (FLCN). Repeated episodes of pneumothorax in adult patients aged 20 to 40 years is the key feature of BHD syndrome that warrants a thorough medical examination. In this unique study, 3 members of a family with BHD syndrome underwent surgical intervention and were followed at our institute, enabling comparison of detailed clinicopathologic findings. Notably, none of the individuals had pulmonary lesions determined by thoracic CT. Thoracoscopic and pathologic examinations identified microcystic lesions diffusely distributed over the pleura. To date, reports of BHD syndrome-associated pulmonary cysts and pneumothorax have not addressed whether the pulmonary lesions are similar in all affected family members. This study is the first to show that the pulmonary cysts in affected family members are pathologically similar. The pulmonary cysts of this family group were much smaller than those that have been reported previously [7]. This finding suggests that the size, morphologic features, and distribution patterns of the pulmonary cysts are inherited by the affected progeny. It has been reported that patients with BHD syndrome with a family history of spontaneous pneumothorax are significantly more likely to experience spontaneous pneumothorax than are those without a family history of spontaneous pneumothorax [4]. This indicates that the characteristic features of BHD syndrome differ between family groups. An important finding in this study is that microscopic cysts that are undetectable by thoracic CT have the potential to cause recurrent pneumothorax. In cases involving common spontaneous pneumothorax, we have often performed pleural ablation at VATS in combination with chemical pleurodesis a few days after operation, although these treatments would be not routinely carried out in other institutes. Chemical pleurodesis or pleural ablation in addition to standard surgical interventions are likely to be of even greater benefit in the treatment of pneumothorax caused by BHD syndrome, in which the cysts are diffusely distributed, than in common spontaneous pneumothorax.

In line with other recently published findings, these results help to further our knowledge about the pathologic characteristics and diagnosis of BHD syndrome–associated pulmonary cysts [7].

References

esophageal malformations symptoms may not present until adult life and is often intermittent. Congenital BEF in adult life is rare and most literature comprise of case reports [1, 2]. Most reported literature advocate fistulectomy and resection of the underlying parenchyma. We report the surgical treatment of 2 cases of adult onset BEF where we preserved the underlying parenchyma with both patients having a well expanded lung during follow-up.

The first patient was a 39-year-old male and the second, a 55-year-old female. Both patients had symptoms of bouts of cough when swallowing liquids since childhood. Barium swallow was diagnostic in both patients showing a fistula tract between the esophagus and the lower lobe bronchi (Fig 1). Chest computed tomography scan revealed consolidations and atelectasis in the right lower lobe superior segment, and infiltrations in the left lower lobe superior segment, respectively. Both patients were operated through a thoracotomy. In the first patient bronchoscopy and esophagoscopy confirmed the orifices of the fistula, whereas in the second patient only bronchoscopy revealed the fistula in the left lower lobe superior segmental bronchus. Both patients underwent thoracotomy and had significant adhesions in their lower lobes. Careful dissection of the posterior mediastinal pleura from the level of the carina to the inferior pulmonary vein was done. Large collateral vessels were noticed on the aorta in the female patient. The fistula tract was just above the inferior pulmonary vein between the esophagus and the lower lobe superior segmental bronchi (Fig 2).

**Two Unusual Cases of Adult Onset Congenital Bronchoesophageal Fistulas Treated With Fistula Division**

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Adult onset congenital bronchoesophageal fistula is a very rare entity. We report 2 cases of adult onset type II congenital bronchoesophageal fistula between the distal thoracic esophagus and the lower lobe superior segmental bronchi surgically treated through a right and left thoracotomy, respectively. In both cases the fistula was transected and sutured with no parenchyma resection. Both patients had an uneventful recovery. Resection of the underlying parenchyma during surgery for bronchoesophageal fistula is not always necessary as the lung can heal in time after performing just fistulectomy.


Bronchoesophageal fistula (BEF) is an abnormal communication between the esophagus and the bronchial tree. It can be acquired or congenital. Congenital BEF when associated with esophageal atresia presents early in life. In the absence of coexisting

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