A family with a strong history of pneumothorax was referred to our genetic clinic for assessment. There were three siblings who had all developed spontaneous pneumothorax at the age of 30, 58, and 59 years. All were non-smokers with no pre-existing pulmonary disease. High-resolution computed tomography of the thorax for all showed multiple thin-walled pulmonary cysts of variable size on both sides, mainly located at the basal and peripheral lung regions (Fig). Lung biopsy was not informative. Physical examination revealed multiple smooth, dome-shaped papules over the face and ears in one of siblings (Fig b). There were no other features of tuberous sclerosis or history of renal

FIG. (a) Chest X-ray showing bilateral multiple pulmonary cysts over the basal region (arrows). (b) Clinical photo of fibrofolliculomas over pinna (arrows). (c and d) High-resolution computed tomography of the thorax showing lower-zone predominant pulmonary cysts of variable shapes and sizes (circles). The walls are thin, sharply demarcated and do not enhance with contrast (arrowhead)
Birt-Hogg-Dubé syndrome

416.e5Hong Kong Med J
Volume 23 Number 4
August 2017
www.hkmj.org

spontaneous pneumothorax and emphysema. Radiologically, the BHD-associated lung cysts are usually irregularly shaped, variable in size and number, and with sharply demarcated thin walls that do not enhance on computed tomographic imaging. Fibrofolliculomas are present in more than 80% of patients with BHD syndrome and typically appear after the age of 20 years. They are dome-shaped, white to flesh-coloured, non-painful and non-pruritic papules located on the facial, cervical, and upper truncal regions.

The most threatening complication of BHD syndrome is renal cell carcinoma. It occurs in approximately 15% of BHD patients by the age of 70 years. Therefore regular surveillance is mandatory. Physicians should be alert to the possibility of BHD syndrome in a patient who presents with diffuse cystic lung disease, particularly in the presence of a positive family history. Early referral to a clinical genetic service and multidisciplinary management is recommended. Early diagnosis and regular renal surveillance aim to greatly reduce renal cell carcinoma–associated morbidity and mortality.

HM Luk *, FHKAM (Paediatrics)
TMF Tong, MSc
IFM Lo, FHKAM (Paediatrics)
Clinical Genetic Service, Department of Health, 3/F Cheung Sha Wan Jockey Club Clinic, 2 Kwong Lee Road, Shamshuipo, Hong Kong

* Corresponding author: luksite@gmail.com

References