Pulmonary manifestations of Birt-Hogg-Dubé syndrome
(Gupta *et al*., 2013)

Lay summary

Who wrote this paper?

Three pulmonologists from the United States and Japan, who are world experts on the lung symptoms of BHD syndrome.

What is this paper about?

This paper describes the lung symptoms of BHD and recommends steps to diagnose and treat them.

What does the paper say?

9 in 10 BHD patients have lung cysts, which develop between the ages of 30 and 40, and are usually in the lower regions of the lungs. Cyst size is variable, but most (75.6%) are less than 1 cm in diameter and are irregularly shaped. 40% of cysts are located on the outside surface of the lung.

Cysts show no evidence of cancerous cell growth, inflammation, or fibrosis, meaning that they are histologically different from common blebs or bullae.

A quarter of BHD patients have at least one pneumothorax, and the average age for the first episode is 38. Of those BHD patients who have a pneumothorax, 75% will have multiple episodes. This means that roughly 1 in 5 BHD patients will have more than one pneumothorax. The presence of lung cysts is highly correlated with pneumothorax. However, gender, age, and smoking status do not seem to be linked to pneumothorax in BHD patients.

Lung function is not significantly affected by lung cysts, and BHD does not cause lung failure, like some other cystic lung diseases.

A diagnosis of BHD should be considered in a young patient with spontaneous pneumothorax, especially if they have a medical or family history of pneumothorax, skin lesions or kidney cancer. High resolution CT scans of the chest is the preferred imaging method for doctors to accurately differentiate the lung cysts from other cystic lung diseases. If BHD is suspected based on medical history, family history, and chest scans, patients should be referred for genetic testing.

Even if they have no lung symptoms, newly diagnosed BHD patients should have high resolution CT chest scans to determine how badly their lungs are affected.

There is no treatment for lung cysts. Video-assisted thoracoscopic surgery (VATS) and mechanical or chemical pleurodesis are most commonly used to treat recurrent cases of pneumothorax.

BHD patients with reduced lung function, particularly large numbers of cysts, or who have had previous episodes of pneumothorax should see a pulmonologist before travelling on a plane. Patients should not board the plane if they have any unexplained chest pain or shortness of breath.

Smoking is a known risk factor for pneumothorax, so should be strongly discouraged in BHD patients.

Pneumonia and flu vaccinations should be encouraged in BHD patients.