Cystic lung disease in a young female patient

M S Abdool-Gaffar, MB ChB, FCP (SA), FCCP, CIME

Kingsway Hospital, Amanzimtoti, KwaZulu-Natal, South Africa

Corresponding author: *M S Abdool-Gaffar (sabs@docgaffar.co.za)*

A case report of a female patient with Birt-Hogg-Dubé syndrome is presented to illustrate the salient features of this condition. The 45-year-old woman was relatively asymptomatic when the chest X-ray was done. She was found to have bilateral pulmonary cysts without any other significant abnormalities. A differential diagnosis is presented with a final diagnosis of the folliculin gene-associated, Birt-Hogg-Dubé syndrome.

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Case report

A 45-year-old woman presented to her general practitioner (GP) with an upper respiratory tract infection, which had eventually settled on treatment, but her chest radiograph was abnormal. She had also had an abnormal chest radiograph 2 years prior to presentation at the GP, which had not been investigated, and she was subsequently referred to our unit. She was asymptomatic from a respiratory point of view, and denied any history of chest problems. As a food technologist, she travelled at least twice a month by air, both locally and internationally. Her history was negative for any medical problems and she was a lifetime non-smoker.

The patient's clinical examination was normal. Her chest radiograph (Figs 1A and B) demonstrated well-circumscribed, thin-walled cysts in both lower lobes, with a right-sided predominance.

A high-resolution computed tomography (HRCT) scan of the chest (Figs 2A - C) confirmed multiple thin-walled cysts involving both lower lobes, and the medial aspect of the right middle lobe. Localised left posterior basal atelectasis was evident, but there were no other parenchymal lesions. In particular, there were no features of ground-glass opacification, reticulation, or nodularity. At the time of publication of this manuscript, the patient was still being managed conservatively.

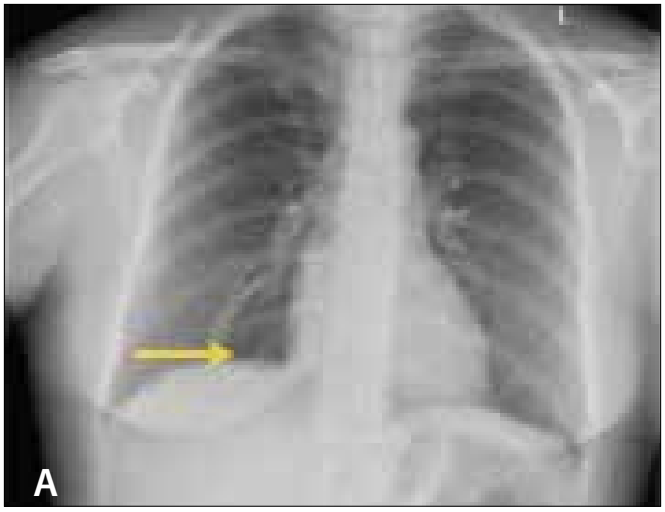

Fig. 1. (A) Posterior-anterior view of the chest radiograph showing cysts (yellow arrow) in the lower lobes.

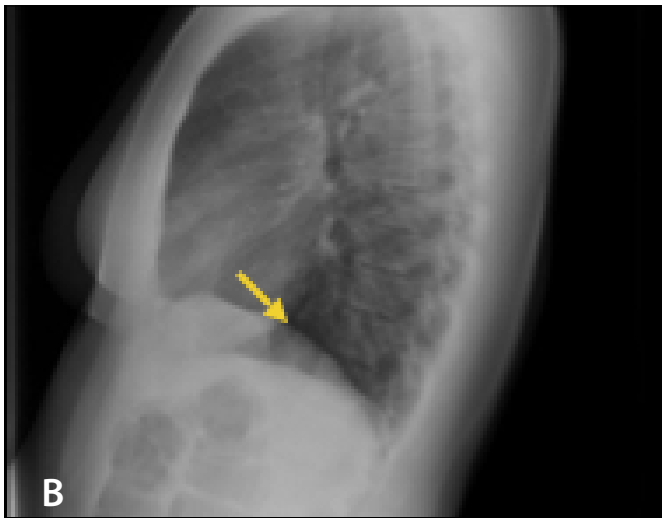

Fig. 1. (B) Lateral view chest X-ray showing cysts (yellow arrow) in the lower zones.

Discussion

The patient presented with multiple pulmonary cysts and the differential diagnosis was made based on the patient's history, clinical examination and radiological findings. The differential diagnosis in the index case included lymphangioleiomyomatosis (LAM), pulmonary Langerhans cell histiocytosis (PLCH), folliculin gene-associated syndrome (FLCN-S), which is also known as Birt-Hogg-Dubé syndrome, lymphocytic interstitial pneumonia (LIP), *Pneumocystis jiroveci* pneumonia (PJP), amyloidosis, light-chain deposition disease (LCDD), neoplastic disease, desquamative interstitial pneumonia, hypersensitivity pneumonitis, bronchiolitis, and a few other rare causes.[1] Bullae in patients with emphysema may mimic cysts, but this was not applicable to our patient. Other conditions that may produce cyst-like lesions include pneumatoceles secondary to an infective process, cystic bronchiectasis, and honeycombing, but these can be distinguished by associated primary aetiologies.

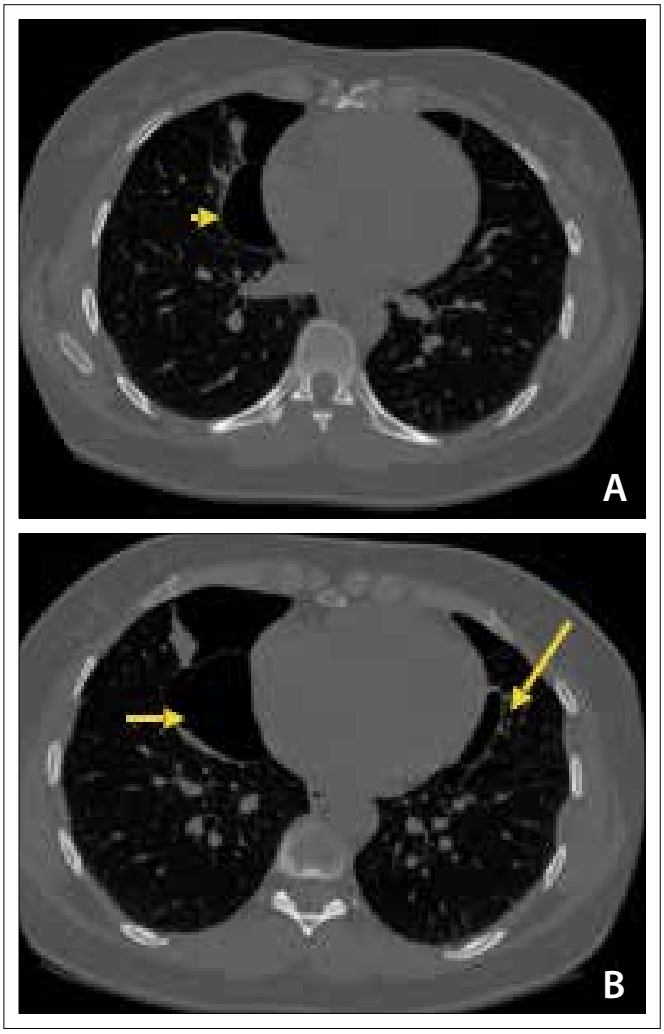

Fig. 2. (A) Computed tomography (CT) scan showing cysts in a paracardiac distribution (yellow arrowhead); (B) CT scan showing cystic changes (yellow arrows).

The patient was a lifetime non-smoker, was not immunocompromised and had no history of any pulmonary pathology. The chest X-ray and CT scan did not show any intervening pulmonary parenchymal changes and in the context of the clinical presentation, the differential diagnosis in this patient was narrowed down to LAM and FLCN-S. The CT scan in patients with LAM usually demonstrates symmetrical, bilateral, multiple thin-walled cysts throughout both lungs, which were not evident in this patient. Lymphatic abnormalities are identified radiologically and may also occur in patients with LAM. Furthermore, Raoof *et al.*[2] provide a very good diagnostic approach to the management of a patient with cystic lung disease.

FLCN-S is an autosomal dominant genodermatosis that manifests as a multisystem disorder. The clinical findings are often evident in the lungs, skin and kidneys;[1] however, patients may manifest with only pulmonary cysts. Affected patients may be asymptomatic, but the most common symptom is skin papules, especially on the face.[1] The most common pulmonary presentation is a spontaneous pneumothorax, which occurs in ~35% of cases.[3] The pulmonary cysts are usually bilateral, multiple, and vary in size. The intervening pulmonary parenchyma is normal. Cysts are further characterised by lower-zone predominant distribution; they may be lobulated and multi-septated.[1] Renal involvement includes cysts as well as benign and/or malignant tumours. The diagnosis is confirmed by genetic testing, which confirms a mutation in the *FLCN* gene. The genetic test may also be used to screen family members; however, no specific therapy for this condition is currently available. The most likely diagnosis in the index patient was FLCN-S (Birt-Hogg-Dubé syndrome). She had no extra-pulmonary manifestations of the condition; however, the patient remains at risk of a spontaneous pneumothorax that would be exacerbated by continued air travel. This may influence the patient's career path.[3] Genetic tests had not yet confirmed the diagnosis in this patient.

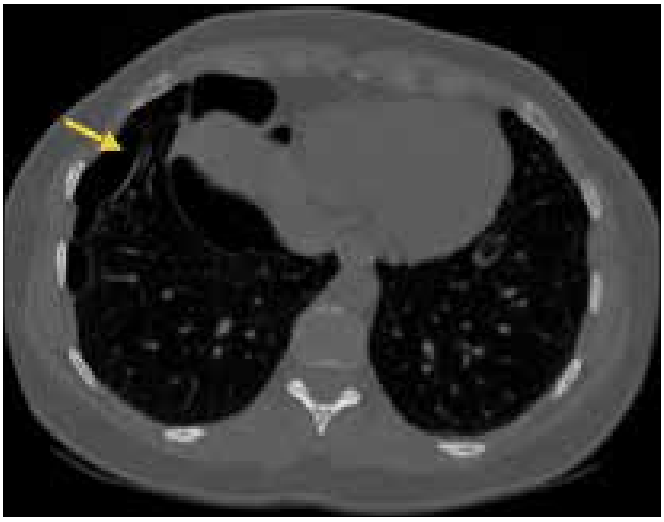

Fig. 2. (C) CT showing subpleural cysts (yellow arrow).

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