Thoracic manifestations of segmental neurofibromatosis

Nader Chebib,1 Patrick Combemale,2 Denis Jullien,3 Vincent Cottin4

DESCRIPTION
A non-smoking 57-year-old female presented with long-standing mild dyspnea and dry cough. Her medical history was remarkable for a subcutaneous right cervical nodule which corresponded pathologically to a neurofibroma. There were no other signs for type 1 neurofibromatosis as she had no family history of neurofibromatosis, no «café au lait» macules, no axillary or groin freckling, no plexiform neurofibroma, no cutaneous/subcutaneous neurofibroma and no Lisch nodules. Genetic testing for the Neurofibromin 1 (NF1) gene mutations was negative. MRI of the brain, spine and pelvis was normal, except for the right cervical neurofibroma measuring 4.5 cm in greatest diameter. Pulmonary function tests showed no obstructive or restrictive pattern. The chest radiograph demonstrated enlargement of the upper mediastinum with bilateral non-cavitating lobulated opacities (figure 1). Thoracic CT scan showed multiple bilateral round and non-compressive nodules and masses in the superior, anterior and posterior mediastinum, along with mediastinal lymphadenopathies (figure 2). These lesions predominated in the upper zones. Because the patient had mild symptoms, an invasive procedure to have pathological proof of the thoracic lesions was initially delayed. Such procedure was not necessary during the 10-year follow-up because the lesions did not progress and the symptoms remained stable.

Neurofibromatosis is a group of genetic diseases characterised by the development of benign and malignant tumours of the central and peripheral nervous system, as well as characteristic cutaneous lesions.1 Mutations in the NF1 gene are found in 85%–95% of cases with neurofibromatosis type 1. Thoracic involvement is rare and includes thin-walled cysts, reticular lesions, ground glass opacities, pulmonary nodules and mediastinal masses, as well as subcutaneous nodules of the chest wall.2 3 The radiological thoracic lesions observed in our patient and their stability over several years are suggestive of non-NF1 segmental neurofibromatosis. A pathological confirmation by biopsy was not sought because symptoms were limited, with

Learning points
- Neurofibromatosis type 1 is a genetic disease caused mainly by mutations in the Neurofibromin 1 (NF1) gene.
- Characteristic clinical features include cutaneous lesions («café au lait» macules, axillary or groin freckling, cutaneous/subcutaneous neurofibromas), as well as benign or malignant tumours of the central and peripheral nervous system. However, limited or segmental forms of NF1 can also be found.
- Thoracic involvement is rare and includes thin-walled cysts, reticular lesions, ground glass opacities, pulmonary nodules, mediastinal masses and subcutaneous nodules of the chest wall.
cough considered to be related to masses in close contact to the trachea and to gastropharyngeal reflux.

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REFERENCE