

Endotracheal neurofibroma in a patient with von Recklinghausen's disease

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ABSTRACT: Benign neurogenous tumours arising in the trachea are rare. We report on a patient with von Recklinghausen's disease who presented with shortness of breath caused by endotracheal neurofibroma.

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Von Recklinghausen's disease (neurofibromatosis) is a syndrome transmitted by an autosomal dominant gene. It is characterized by the presence of palpable neurofibromas of peripheral nerves, multiple soft cutaneous tumours and café au lait spots. Various manifestations of the disease are summarized elsewhere [1]. Respiratory complaints, such as dyspnoea and cough, are usually attributed to neurofibromatous tumours within the lung parenchyma or fibrosing alveolitis that accompanies this disease [2]. This case report describes a patient with a neurofibromatous tumour occurring endotracheally and reviews the other published case reports in the English literature.

Case report

A 45-old man with von Recklinghausen's disease presented to the Hadassah University Hospital in Jerusalem with an eighteen month history of shortness of breath on exertion, without chest pain or cough. The patient had a smoking history of twenty packs a year. Family history was positive for neurofibromatosis. Physical examination revealed multiple cutaneous neurofibromas, cutaneous tumours and café au lait spots, mainly on the face and the upper trunk. The remainder of the physical examination, including the chest, was unremarkable. Posteroanterior and lateral chest roentgenograms were normal. Spirometry showed vital capacity (VC) 3.49 l (112% predicted), forced expiratory volume in one second (FEV₁) 3.37 l (96% predicted) and FEV₁/VC 73%. His flow-volume loop was normal. Chest CT scan showed a small tumour protruding from the anterior wall of the trachea into the lumen and multiple extrathoracic cutaneous lesions. Indirect laryngoscopy was normal. Fibreoptic bronchoscopy revealed a small polypoid greyish tumour with a wide base located on the anterior wall of the trachea at a

distance of 22 cm from the teeth. It occupied approximately one third of the cross-sectional area of the trachea. No other endobronchial lesions were seen up to the subsegmental level in either right or left bronchial trees. Rigid bronchoscopy under general anaesthesia was performed and the tumour was completely removed. Pathologic examination of the tumour identified it as a neurofibroma. Following this bronchoscopic removal the patient has been well for 34 months without any respiratory complaints.

Discussion

Intratracheal tumours are relatively rare, and benign neurogenous tumours arising in the trachea are among the rarest [3, 4]. To our knowledge, only three other cases of endotracheal neurofibromas have been published in the English literature. The relevant clinical information from the three previously reported cases of endotracheal neurofibromas and our present case are summarized in table 1.

Three of the four endotracheal neurofibromas occurred in males. In two cases, the classic cutaneous von Recklinghausen's disease was present. The clinical presentation of these patients is similar to that caused by other tracheal tumours [3, 5-7]. In only two cases were endotracheal neurofibromas identified on chest roentgenograms. The other two cases had normal chest X-rays. This is understandable since on posteroanterior films overlying soft tissues obscure the trachea. Pulmonary function tests did not show an abnormal flow-volume loop in any of the four patients. Simple spirometry was completely normal in two cases and mildly abnormal in the other two. Pulmonary nodules, interstitial or parenchymal involvement were not noted in any of the cases with endotracheal neurofibromas. It therefore seems likely that the occurrence of endotracheal neurofibromas is

Table 1. - Reported intratracheal neurofibromas

Source	Yr	Age/sex	Clinical features	vRD*	X-ray findings	Spirometry findings	Treatment
Meredith [5]	1978	60/M	Wheeze on exertion. Exertional dyspnoea and cough attacks	No	Chest X-ray normal	Reduction in pulmonary function tests	Thoracotomy & excision of tumour
Thijs-Van Nies [6]	1978	36/M	Cough and blood-stained sputum, progressive dyspnoea, wheezing and stridor.	No	Tumour identified on tracheal radiograph; chest X-ray normal	FVC - normal FEV ₁ - normal DLCO - normal MEF ₂₅ - reduced MEF ₅₀ - reduced MEF ₇₅ - reduced	Endoscopic removal
J. Neal Rutledge [7]	1983	18/F	Cough, wheezing and shortness of breath.	Yes	Tumour shown on chest X-ray	FVC - reduced FEV ₁ - reduced FEV ₂₅ - 75% reduced	Thoracotomy & excision of tumour
Present case		45/M	Shortness of breath on exertion.	Yes	Chest X-ray normal	FVC - normal FEV ₁ - normal	Endoscopic removal

*Presence of von Recklinghausen's disease; FVC: forced vital capacity, FEV₁: forced expiratory volume in one second; DLCO: diffusing capacity of the lung; MEF₂₅, MEF₅₀, MEF₇₅: maximal expiratory flow at 25, 50 and 75% respectively; FEV₂₅: forced expiratory flow at 25%

independent of other pulmonary manifestations of von Recklinghausen's disease.

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RÉSUMÉ: Les tumeurs bénignes neurogènes de la trachée sont rares. Nous présentons l'observation d'une patient atteint de maladie de von Recklinghausen, qui a consulté pour une dyspnée provoquée par un neurofibrome endo-trachéal.