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CLINICAL IMAGE

# Diffuse lung disease associated with neurofibromatosis type-1 can also affect children

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A 4-year-old child affected by neurofibromatosis type-1 (NF-1) was submitted to computed tomography (CT) of the spine for the assessment of severe scoliosis.

The CT scan confirmed scoliosis and revealed multiple rows of small sub-pleural air-filled lesions at the pulmonary upper lobes (Fig. 1). These pulmonary blebs present very thin walls (< 1 mm) and small diameters (2–7 mm). These findings suggested the diagnosis of diffuse lung disease associated with NF-1 (NF-DLD). Other lung regions were normal, no basilar fibrosis was detected.

NF-1 is an autosomal-dominant disease with a prevalence of one in 3000 [1]. Most common clinical manifestations include "cafè au lait" skin macules, cutaneous and central nervous system tumours. NF-DLD is described as possible manifestation of NF-1 in adult patients. The disease is generally characterized by basilar fibrosis and/or upper lobes air-filled lesions such as bullae, blebs (as in the case) or cysts [2]. Symptomatic patients usually present with dyspnoea on exertion; pulmonary function tests show either an obstructive or a restrictive defect [2]. Sub-pleural air-filled lesions represent a risk of pneumothorax which could be the first clinical manifestation of the disease [3].

This is, to our knowledge, the only pediatric patient with documented NF-DLD. This case together with a young-adult patient reported in the literature (16 years old) [3], both with upper lobes air-filled sub-pleural lesions and no basilar fibrosis, suggests that these lesions may arise before fibrotic basilar involvement, even in childhood/adolescence.

**Author contributions** PS wrote the paper, diagnosed the disease described. GF helped in writing paper, checked and studied literature about the disease. AB helped in image processing and writing paper. UA supervised and approved all the work.

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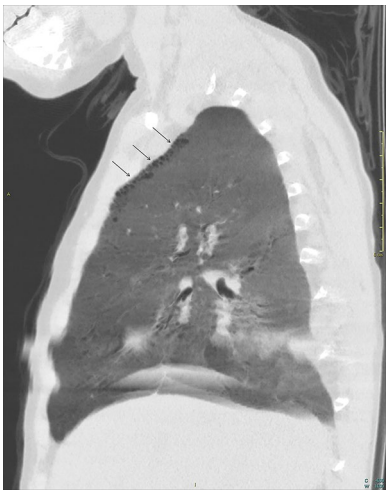

**Fig. 1** Computed tomography of the spine, visualized with "lung window" on minimum intensity projection (sagittal view), shows multiple sub-pleural blebs (arrows) at the upper lobes

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## Compliance with ethical standards

**Ethical approval** Parents of minor patient signed consent to use data for Research. The case report was conducted in line with ethical standard requested by our institution.

**Conflict of interest** The authors declared that they have no conflict of interest.

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