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Challenges in the classification of fibrotic ILD: Patient case 1

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Patient presentation and clinical history

The patient is a 43-year-old male non-smoker who works as a farmer and cheese-maker. He complained of a dry cough for 6–12 months without fever or other clinical signs. His medical history was unremarkable with no use of drugs or alcohol and no signs of Raynaud's syndrome. In his family history he had two first-degree relatives with pulmonary fibrosis, one of whom also had rheumatoid arthritis.

Diagnostic work-up

The patient underwent chest x-ray as part of a routine health assessment, which revealed extensive cystic lesions in the upper lobes, infiltrates, widening of the mediastinum, loss of lung volume and a reticular pattern.

On clinical examination he had "velcro" sounds bilaterally and finger clubbing. He was hypoxaemic at rest and reached 79% desaturation on minimal exertion. Pulmonary function test results were compatible with severe restrictive disease with a DL_{CO} of 28% predicted. Cardiac ultrasound showed dilatation of the right ventricle and pulmonary arterial pressure of 40 mmHg. Serology for autoimmune disease, calcium levels and urinary calcium levels were all normal.

Chest CT revealed extensive cystic lesions, ground glass opacities, thickening of the pleura, and traction bronchiectasis (Figure 1). The patient was referred promptly to a thoracic surgeon by his family doctor; therefore bronchoscopy and BAL were not included in the initial evaluation. Instead, open-lung biopsy was performed from the right upper and lower lobe and the pleura. Histopathological findings included evidence of marked fibrosis and architectural distortion; honeycombing; patchy involvement of lung parenchyma by fibrosis; presence of multiple fibroblast foci; absence of granulomas, organising pneumonia or hyaline membranes; lymphoid aggregates with germinal centres; areas with cysts, fibrosis and non-specific inflammation in the upper lobes and congestive vessels and dense chronic inflamma-

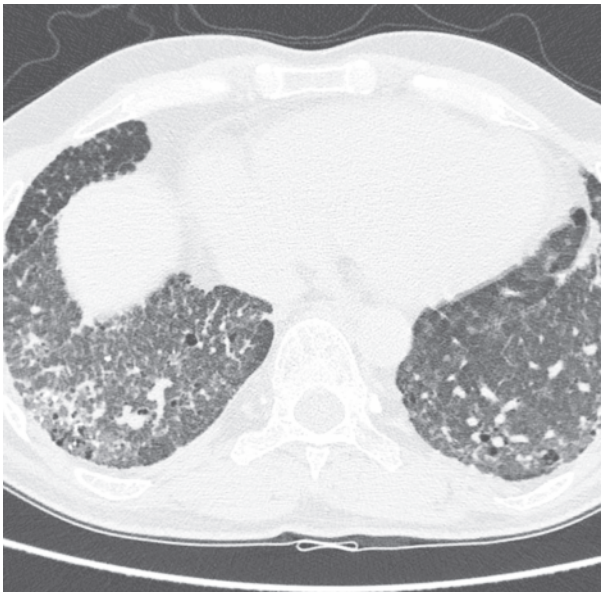

Fig. 1. Presenting HRCT undertaken in 2013

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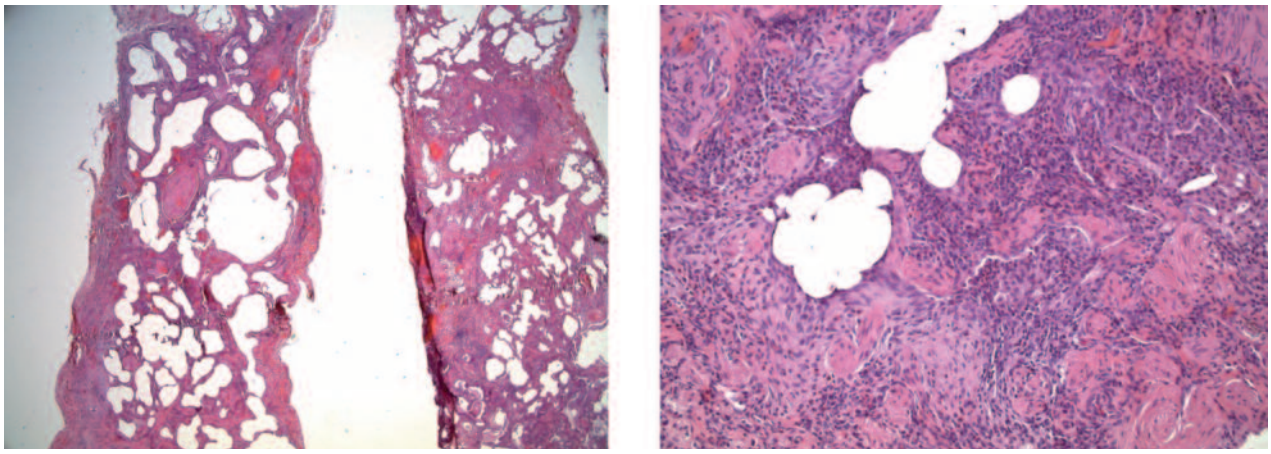

Fig. 2. Histology

tory infiltration in the pleura. There was no vasculitis or features of malignancy. Overall the features were compatible with a UIP pattern (Figure 2).

Diagnosis and classification

On the basis of the clinical, pathological and radiological findings, six possible diagnoses were considered: 1) chronic hypersensitivity pneumonitis mostly due to his environmental and professional exposures, 2) lung-dominant collagen tissue disease mostly due to his young age and the family history that included members with rheumatoid arthritis; 3) familial ILD due to the fact that two first-degree relatives had already deceased diagnosed with ILD; 4) idiopathic pleuroparenchymal fibroelastosis due to the excessive thickening of the pleura in the upper lobes; 5) sarcoidosis due to his young age and predominance of the lesions in the upper and middle lobes, or 6) unclassifiable IIP. Based on clinical behaviour the patient was classified as having progressive, irreversible disease with the potential for stabilisation, according to the classification by Travis et al. 2013 (1).

Management and follow-up

The patient was started on corticosteroids and advised to avoid his usual work environment but continued to deteriorate. Several months later he presented with a productive cough with further progressive deterioration in blood gases and a DL_{CO} of 19% predicted. PET/CT failed to provide any further useful information and his diagnosis remained one of unclassifiable IIP. However, due to the fact that the patient had two first-degree relatives with documented ILD, the criteria for familial pulmonary fibrosis were fulfilled and the patient underwent testing for genetic predisposition. As is described in the literature, both the radiological and pathological data of familial pulmonary fibrosis are often unclassifiable (2). While the results were pending, the patient and his family were informed about lung transplantation and agreed to undergo pre-transplant evaluation. The patient is now enrolled in a lung transplantation waiting list.

Conclusion

This case highlights a difficult diagnosis in a very young man with severe disease. The multidisciplinary team identified six potential diagnoses, but given the inconclusive nature of the patient's clinical, radiological and histopathological information, the case was initially categorised as unclassifiable IIP and ultimately as familial pulmonary fibrosis. The challenges that are still associated with accurate classification and the absence of guidance available to direct treatment in unclassifiable patients are under-

scored in this case. While clinical classification according to disease behaviour can be a useful adjunct to IIP classification, in this case the prognosis was exceptionally poor. The patient's condition did not stabilise after treatment with corticosteroids and avoidance of work environment.

References

1. Travis WD, Costabel U, Hansell DM, et al.; ATS/ERS Committee on Idiopathic Interstitial Pneumonias. An official American Thoracic Society/European Respiratory Society statement: Update of the international multidisciplinary classification of the idiopathic interstitial pneumonias. Am J Respir Crit Care Med 2013; 188 (6): 733-48.
2. Borie R, Kannengiesser C, Nathan N, Tabèze L, Pradère P, Crestani B. Familial pulmonary fibrosis. Rev Mal Respir 2015; 32 (4): 413-34.

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