CHEST

Two Siblings With Interstitial Lung Disease

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A 52-year-old white woman and her 61-year-old white brother separately presented with gradually worsening dyspnea on exertion and cough, and evidence of interstitial lung disease on chest imaging.

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Case Presentations

The woman presented with a several-year history of progressive dyspnea and evidence of interstitial lung disease (ILD) on imaging. She had owned pet cockatiels 3 years prior to the start of her symptoms and had a history of exposure to a moldy environment at work. She had no history of smoking or other relevant respiratory exposures, and had no symptoms to suggest a connective tissue disease or exposure to medications associated with ILD. The patient had a history of hormonally inactive bilateral adrenal hyperplasia and mild splenomegaly, both discovered incidentally on imaging, as well as osteopenia and several foot fractures after minor trauma. Her physical examination was unrevealing. Results of the patient's pulmonary function tests revealed an FVC of 3.09 L (94% of predicted), an FEV_1 of 2.59 L (99% of predicted), an FEV_1 to FVC ratio of 84%, total lung capacity of 4.62 L (93% of predicted), reserve volume of 1.46 L (88% of predicted), and diffusion capacity for carbon monoxide of 11.31 mL/min/mm Hg (49% of predicted), which we interpreted as normal lung volumes with moderately impaired gas transfer.

The woman's brother presented with 1 year of progressive dyspnea on exertion and a cough productive of scant clear sputum. His medical history was notable for two spontaneous pneumothoraces at 29 years of age (for which he underwent doxycycline pleurodesis) and idiopathic thrombocytopenia. He had never smoked cigarettes or used recreational substances but had a history of exposure to asbestos in his 20s during construction work. As with his sister, he had no history that would suggest a connective tissue disease or exposure to medications associated with an ILD. On examination, the patient had crackles over both lower lobes and hypoxia to 84% after walking 450 m on room air. His laboratory studies were notable for a platelet count of 140,000/μL but otherwise normal blood counts. Results of the patient's pulmonary function tests revealed an FVC of 3.77 L (89% of predicted), an FEV_1 of 2.66 L (83% of predicted), an FEV_1 to FVC ratio of 71%, total lung capacity of 5.79 (88% of predicted), reserve volume of 1.88 L (80% of predicted), and diffusion capacity for carbon monoxide of 11.31 mL/min/mm Hg (39% of predicted). Thus, similar to his sister, the patient had normal lung volumes with moderately impaired gas transfer. Results of tests for plasma α_1-antitrypsin level were normal. Both siblings had otherwise normal laboratory studies with no serologic evidence to support autoimmune diseases.

High-resolution chest CT imaging of the female sibling (Fig 1A) showed interlobular septal thickening bilaterally with a craniocaudal gradient, with no honeycombing, ground-glass opacities, nodules,

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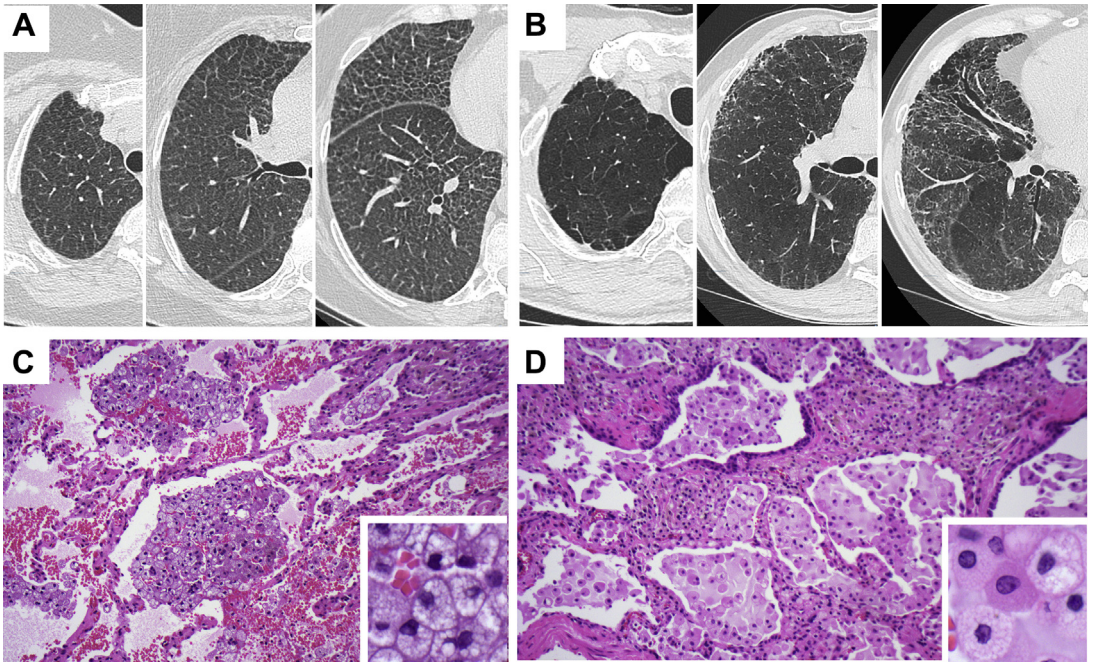

Figure 1 – *Imaging and histologic findings of the (A, C) female and (B, D) male siblings. A, B, representative high-resolution chest CT imaging of upper lobes, level of the carina, and lower lobes (from left to right) of the right lung in both subjects. C, D, hematoxylin and eosin stains of lung histopathology; original magnifications were 100× (main panels) and 400× (insets). The text provides a narrative description of the findings.*

bronchiectasis, lymphadenopathy, or axial gradient. High-resolution chest CT imaging of the male sibling revealed upper lobe-predominant paraseptal emphysema and basilar-predominant reticulation, honeycombing, ground-glass opacities, and traction bronchiectasis, with no nodules, or lymphadenopathy (Fig 1B). In addition, abdominal imaging showed bilateral adrenal enlargement, splenomegaly, and coarse hepatic echotexture.

Lung histopathologic analyses in both siblings revealed marked accumulation of histiocytes containing foamy material within the alveoli (Fig 1C). In the female subject, there was mild fibrosis-associated infiltration of lymphocytes in the interstitium and a small bone marrow pulmonary embolus (not shown). Lung histopathologic analyses from the male sibling showed a similar pattern but with more prominent fibrotic areas associated with a chronic inflammatory infiltrate (Fig 1D).

What is the diagnosis?

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