Marfan syndrome presenting with diffuse emphysematous change of the lung

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DESCRIPTION

A 29-year-old never-smoking woman who had been diagnosed with Marfan syndrome at 9 years of age was admitted with persistent exertional dyspnoea. She had previously developed pneumothorax three times and underwent scoliosis surgery and aortic valve replacement. A physical examination revealed a thin, healthy-appearing woman (body mass index 16.4 kg/m²) with pectus excavatum and joint hyperflexibility.

Chest radiography (figure 1) and CT (figure 2) showed severe spinal deformity and diffuse emphysematous change of the lungs. Pulmonary function tests showed a forced vital capacity of 2.7 L (81% predicted), a forced expiratory volume in 1 s (FEV₁) of 1.47 L (49.7% predicted), FEV_{1%} of 53.1% and a diffusing capacity of the lung for carbon monoxide of 48%. The patient has been treated with an inhaled long-acting beta₂-adrenergic agonist and long-acting anticholinergic bronchodilator, and her lung function has been maintained at a plateau level for about 8 years.

Pulmonary involvement is not generally considered a main feature of Marfan syndrome and there are currently no established screening guidelines for Marfan syndrome-associated pulmonary disease. In addition, reports of patients with Marfan syndrome who present with chronic respiratory symptoms are very rare.¹ However, pulmonary function abnormalities are frequently observed in patients with

Figure 2 CT showing severe emphysematous change of the lungs (A, B: axial scanning) and spinal deformity (C: sagittal scanning).

Learning points

- ▶ Patients with Marfan syndrome can present with pulmonary emphysema, even if they are never-smokers.
- ▶ The possibility of coexisting lung disease in patients with Marfan syndrome should be considered, particularly among patients with thoracic cage abnormalities.

Figure 1 Chest radiography showing spinal deformity and overinflation of the lungs.

Marfan syndrome, particularly among patients with significant thoracic cage abnormalities.¹ Thus far, only a small number of reports have shown cases with pulmonary emphysema, pneumothorax and an obstructive pattern and diffusion impairment on pulmonary function tests.^{2 3} The present case suggests the importance of careful evaluation and monitoring for Marfan syndrome-associated pulmonary disease.

Contributors HI researched the topic and wrote the case. RS, YK and HK helped with the research and proofreading of the case report.

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