CASE STUDY

100. An Uncommon Presentation of Systemic Sclerosis with Pulmonary Hypertension: A Case Report

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Background: Systemic sclerosis is a rare autoimmune condition that carries high mortality rates due to its varied and often hidden presentations. It is broadly classified into limited and diffuse systemic sclerosis, with the latter associated with a poorer prognosis because of its pulmonary, cardiac, and renal complications. Pulmonary hypertension and interstitial lung disease in diffuse systemic sclerosis (dcSSc) are serious complications that are difficult to diagnose due to their occult and insidious course. These manifestations not only increase morbidity and mortality but also result in significant out-of-pocket healthcare expenditure, especially among individuals with limited access to specialized care. We present a rare case of dcSSc complicated by pulmonary hypertension and interstitial lung disease, emphasizing the need for screening, early detection, and multidisciplinary treatment, especially among high-risk individuals.

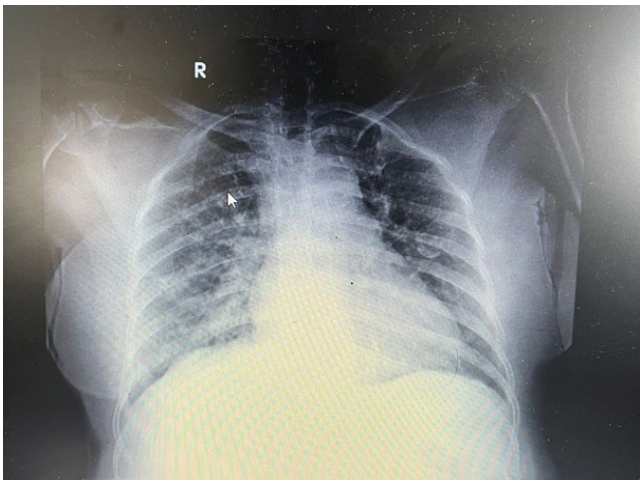
The Case: A 53-year-old Caucasian female presented with a six-month history of progressive dyspnea, dysphagia, Raynaud's phenomenon, and fatigue. She also had proximal interphalangeal joint swelling, unintentional weight loss, and cutaneous changes such as sclerodactyly, calcinosis cutis, and telangiectasias. The skin thickening was diffuse, extending to the trunk, suggesting a diagnosis of diffuse cutaneous systemic sclerosis (dcSSc). Nailfold capillaroscopy revealed dilated capillaries, further supporting the diagnosis of dcSSc.

Laboratory findings were significant for positive ANA with centromere patterns with elevated anti-Scl-70 antibodies. Persistent and progressive dyspnea prompted imaging, which revealed mild interstitial lung disease (ILD) on high-resolution CT and pulmonary hypertension (PH) on echocardiogram, with right ventricular hypertrophy. Pulmonary function tests confirmed reduced diffusing capacity (DLCO). This constellation of findings met the ACR/EULAR 2013 criteria for dcSSc with pulmonary involvement.

A multidisciplinary approach was taken to initiate management in order to address the various systemic manifestations observed. This included immunosuppression with cyclophosphamide (15 mg/kg every 3 weeks) for ILD and bosentan, an endothelin receptor antagonist, for PH. Gastroesophageal reflux and dysphagia were treated with proton pump inhibitors and speech therapy. A common complication of systemic sclerosis is the development of joint contractures, for which physical therapy was initiated.

Conclusion: This case exemplifies the complexity and vigilance required in managing dcSSc complicated by pulmonary hypertension and interstitial lung disease. The insidious onset of symptoms such as fatigue and dysphagia initially suggested gastrointestinal involvement, while pulmonary and cardiac complications remained hidden. Such varied presentations often delay diagnosis, underscoring the importance of heightened clinical suspicion and regular screening in patients at risk. Furthermore, the presence of anti-Scl-70 antibodies signaled a more aggressive disease course. Early recognition of cardiopulmonary complications is thus pivotal, enabling timely initiation of targeted therapies, including endothelin receptor antagonists for pulmonary hypertension and immunosuppressive agents for interstitial lung disease, both of which are critical for improving survival and quality of life. Effective management of dcSSc necessitates a multidisciplinary approach involving rheumatologists, pulmonologists, gastroenterologists, and physical therapists to address its multisystem involvement. A proactive diagnostic and therapeutic strategy, supported by coordinated care, as illustrated in this case, can meaningfully alter the otherwise poor trajectory of diffuse systemic sclerosis.

Figure 1. Chest X-Ray



Legend: Chest X-ray showing bilateral basal reticular opacities suggestive of early interstitial lung disease in a patient with diffuse cutaneous systemic sclerosis.

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