From occupational asthma to pulmonary tuberculosis to systemic sclerosis sine scleroderma: A case report

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Abstract

Systemic sclerosis is a chronic multisystem autoimmune disease of unknown cause that affects the skin and internal organs of the body. It occurs worldwide but has racial and ethnic differences in terms of presentation, blacks tend to have a more diffuse cutaneous presentation. Although genetics plays a significant role in its occurrence, environmental factors including exposure to silica dust, radiation, adulterated rapeseed oil and drugs like bleomycin have also been implicated. It is characterized by progressive fibrosis of the skin and internal organs which is often preceded by inflammation and vasculopathy. We present a case of a 34-year-old poultry farmer who presented with systemic sclerosis sine scleroderma who had initially been misdiagnosed as having occupational asthma and was already on treatment for pulmonary tuberculosis.

Introduction

Systemic sclerosis sine scleroderma, being an uncommon disorder, is a diagnosis that is often missed by many physicians. Although Raynaud's phenomenon is the most common complaint, organs like the esophagus causing dysphagia, and the lungs causing respiratory symptoms are also commonly affected. Involvement of the lungs alone is often misdiagnosed when other features are not overtly expressed.

Case report

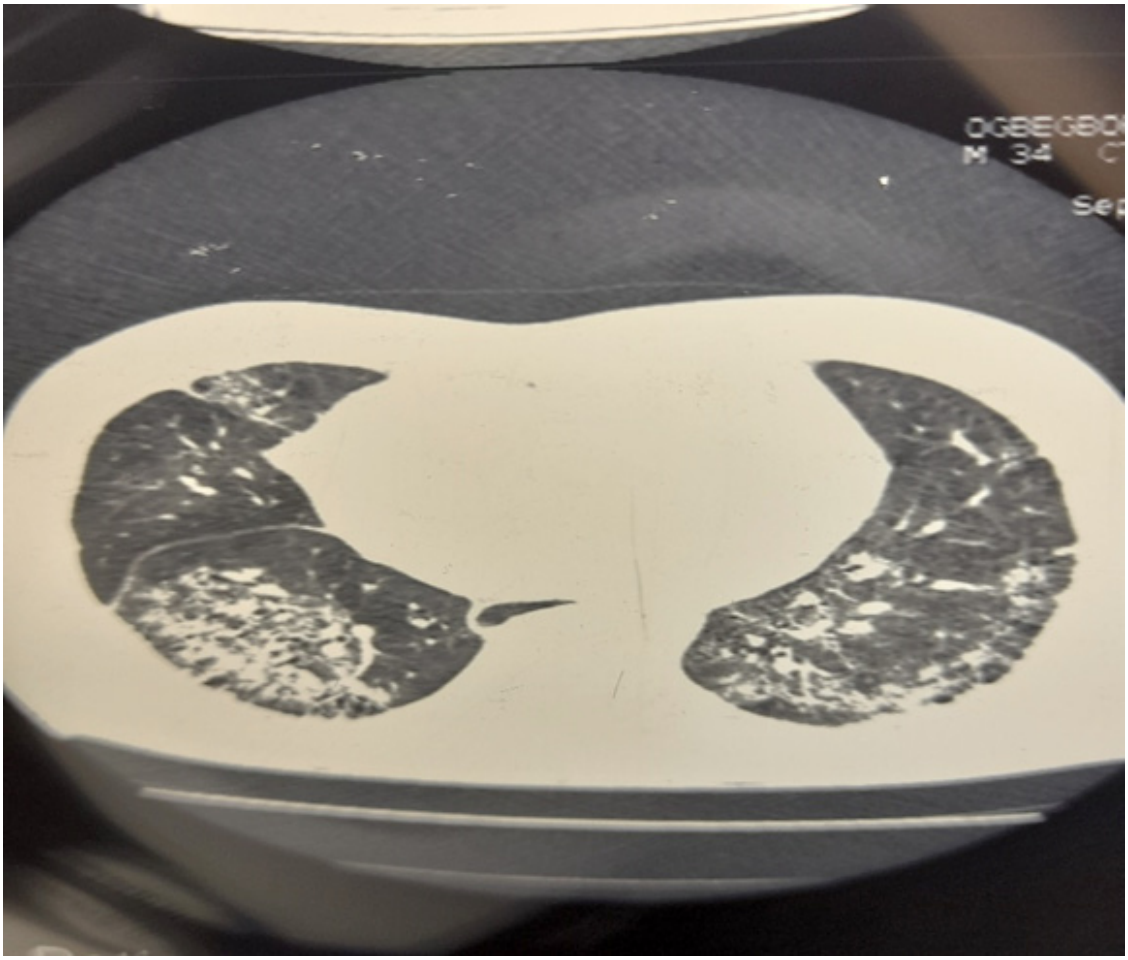
The patient was a 34-year-old male with a 9-year history of progressive difficulty with breathing, recurrent non-productive cough and weight loss. He had been managed by the respiratory unit as case of occupational asthma and was currently being investigated and treated for pulmonary tuberculosis. Rheumatology unit was called on account of complains of polyarthralgia and malaise of 3 years duration.

Further review revealed a history of Raynaud's phenomenon, digital scars at the tips of his fingers, dysphagia, weight loss and recurrent bilateral pedal swelling. He denied a history of hair loss, malar or photosensitive rash, mouth ulcers, joint swelling, skin tightness and/or proximal muscle weakness. There was no history suggestive of renal involvement. He had no history of smoking, past chronic exposure to ionizing radiation or silica dust.

Examination had revealed a young man who was chronically ill looking, and bilateral pitting leg edema. He was in respiratory distress with a respiratory rate of 36 cycles/min with coarse crepitations in the lung base bilaterally, a pulse rate of 112 beats/min, a left parasternal heave, and a loud pulmonary component of the second heart sound. Skin examination had confirmed the presence of digital scars at tips of his fingers but there was no skin thickening or sclerodactyly.

Initial X-ray done had suggested pulmonary tuberculosis however, gene Xpert and sputum AAFB were both negative. Chest CT scan done showed features of interstitial lung disease with non-specific interstitial pneumonitis pattern of involvement (Figure 1) and features of right ventricular enlargement. Echocardiogram and lung function test showed features in keeping with moderate pulmonary hypertension and restrictive pattern respectively. ANA done was positive with a titer >1:5120 [pattern] and positive anti-Scl 70 antibody. On account of the above, a diagnosis of systemic sclerosis sine scleroderma associated interstitial lung disease with pulmonary hypertension and cor-pulmonale was made.

Figure 1: Chest CT scan of patient



He was commenced on mycophenolate mofetil, hydroxychloroquine, low dose methylprednisolone, tadalafil/sildenafil, bosentan, pirfenidone, spironolactone, and torsemide. He was also commenced on long-term oxygen therapy at 3 liters/min for 15 hours daily using an oxygen concentrator.

Discussion

Systemic Sclerosis (SSc) is a chronic multisystem autoimmune disease of unknown cause which is characterized by progressive fibrosis of the skin and internal organs. The pathogenesis of SSc comprises of inflammation, vasculopathy and fibrosis[1]. It is more common in females with a ratio of 3F:1M and African – Americans have a higher incidence than Caucasians and also tend to present with the diffuse cutaneous subset. The prevalence of SSc in Nigeria and Africa is not known, however there are publications of systematic reviews and hospital-based studies are available. A sex ratio of 5.4F:1M was obtained from a recent systematic review[2,3]. A retrospective study done by Adelowo *et al*[4] found 14 patients in a private hospital in Lagos Nigeria.

Systemic Sclerosis Sine Scleroderma (ssSSc), first described by Rodnan and Fennell, is a subset of systemic sclerosis without or with minimal cutaneous features. The hallmark of ssSSc is the involvement of internal organs including the lungs, the hearts and/or the gastrointestinal tract.

A prevalence of 2% was found in Mexico, while in a study from 1808 patients from the Italian Systemic sclerosis PRogressive INvestIGation registry 3.4% of these patients were classified as having ssSSc[5, 6]. While Raynaud's phenomenon was the commonest finding (90% of patients), the esophagus was the most commonly affected organ in the Mexican study. This was followed by the lungs (interstitial lung disease and pulmonary hypertension) and the heart.

The patient fulfilled the 2013 ACR/EULAR criteria for systemic sclerosis without the skin thickening[7]. He had fingertip scars (3 points), interstitial lung disease and pulmonary hypertension (2 points), Raynaud's phenomenon (3 points) and positive scleroderma related autoantibodies (3 points). These made a total score of 11. He also fulfilled all the criteria for ssSSc proposed by Poormoghim *et al*[8]. This criterion requires the absence of skin thickening, the presence of Raynaud's phenomenon or peripheral vascular involvement, positive antinuclear antibody, involvement of visceral organs typical of systemic sclerosis, and the absence of another defined connective tissue disease.

The time lapse from onset of symptoms to diagnosis of our patient is typical for patients with ssSSc. Being a poultry farmer, he had previously been managed for occupational asthma and had also been commenced on anti-tuberculosis medication before a rheumatology consult was suggested on account of a history of arthralgia. Management for tuberculosis is not uncommon for patients presenting with similar complaints due to its prevalence in our environment. He was commenced on mycophenolate mofetil, methylprednisolone, tadalafil/sildenafil, bosentan, pirfenidone, spironolactone and oxygen therapy. He has since improved clinically as evidenced by reduced episodes of dyspnea, easy fatigability and need for oxygen therapy.

Challenges faced during management of this patient included high cost of investigations and medications as well as the unavailability of laboratories for serological tests. Samples for this patient were analyzed outside the country and had a turn-around-time of two weeks. The purpose this case report serves is, in addition to increasing awareness of this disease, to increase the awareness and suspicion in patients who are suspected of having pulmonary tuberculosis but lack bacteriologic evidence.

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