Case Report

A male with ill-fitting skin with dyspnea

Tushar Pawan Zade[1], Madhuri S Pandharipande[2], Rakhee W Joshi[2], Yugal Nagpure[1]

From [1]Junior Resident, [2]Associate Professor, Department of Internal Medicine, Indira Gandhi Government Medical College and Hospital, Nagpur, Maharashtra, India

ABSTRACT

Scleroderma/systemic sclerosis (SSc) is an orphan disease of unknown etiology, complex pathogenesis, and variable clinical presentations. SSc frequently follows a progressive course and is associated with significant disability and mortality. SSc shows a strong female predominance (4.6:1) as compared to males. The onset of skin manifestation to its progression to systemic features is early among males as compared to their female counterparts. Also, they have rapid clinical and biochemical deterioration. In this case report, we present the case of a 33-year-old male scleroderma with early lung involvement without renal manifestation and deliberate regarding variation of clinical manifestation in male patients in comparison to female counterparts.

Key words: Autoimmune, Interstitial lung disease, Male, Scleroderma

Scleroderma, a connective tissue disorder, is associated with skin thickening and tightening. It is characterized by autoimmunity, inflammation, vasculopathy, and progressive fibrosis of the skin and visceral organs [1-3]. Its estimated annual incidence in the USA is 1.9/100,000, affecting females more than males of the age group of 30–50 years [1,2]. Comparatively in India, the incidence rate is 1.5/100,000 persons per year, which is lower in comparison to the rest of the globe [1-4]. Disease manifestations vary from limited skin involvement with minimal systemic involvement to widespread skin involvement accompanied by systemic involvement or only systemic involvement with no dermal features (scleroderma sine sclerosis).

Scleroderma being rare in the male gender, we describe a case of male scleroderma with early pulmonary involvement without renal dysfunction deviating from its natural course.

CASE PRESENTATION

A 33-year-old male laborer came with complaints of tightening of the skin over the dorsum of both hands, fingers, feet, and toes for the last 2 years, which gradually extended to the midarm and midthigh. He also complained of weight loss, epigastric burning sensation (gastroesophageal reflux disease), bloating sensation, altered bowel habits, exertional dyspnea and easy fatigability, multiple joint pain, stiffness of joint (elbow, distal interphalangeal joint [DIP], proximal interphalangeal joint [PIP], knee, and ankle), and reduced mouth opening. A history suggestive of Raynaud's phenomenon, rash, photosensitivity, and fingertip ulceration was, however, absent.

On examination, the patient had generalized dark pigmentation of the skin, pulse of 80/min afebrile, blood pressure of 120/80 mmHg, and respiratory rate of 16/min. The pallor was evident, the lips were puckered (Fig. 1a), reduced mouth aperture, mouth opening was reduced with opening of one finger width, and salt-and-pepper skin (Fig. 1b) over the neck and chest. Thickening of the skin over bilateral upper and lower limbs, sclerodactyly, and bilateral contractures of PIP and DIP joints could be demonstrated. However, ulceration, telangiectasias, and calcinosis were absent. Respiratory system examination reveled reduced total chest expansion of only 6mm with no adventitious sounds. Other systemic examinations and ophthalmic examination were unremarkable.

A positive anti nuclear antibody (ANA) titre of 1:160 and pulmonary funtion test reveled a restrictive pattern while hign resolution tomography showed changes of ILD. Other investigations are shown in Table 1.

Dianosis and classification of systemic sclerosis (SSc) score was 18 suggesting diagnosis of diffuse cutaneous SSc with ILD. The patient was stared on tablet mycophenolate mofetil 500 mg bd and tablet amlodipine 5 mg od. He was advised to follow-up with a complete blood count and complete metabolic profile, in order to adjust the dosage of medication. However, the patient was lost to follow up of further treatment.

Access this article online	
Received - 09 February 2025 Initial Review - 28 February 2025 Accepted - 26 March 2025	Quick Response code
DOI: 10.32677/ijcr.v11i4.5041	

Correspondence to: Tushar Pawan Zade, Department of Internal Medicine, Indira Gandhi Government Medical College and Hospital, Nagpur, Maharashtra, India. E-mail: tshrzade5@gmail.com

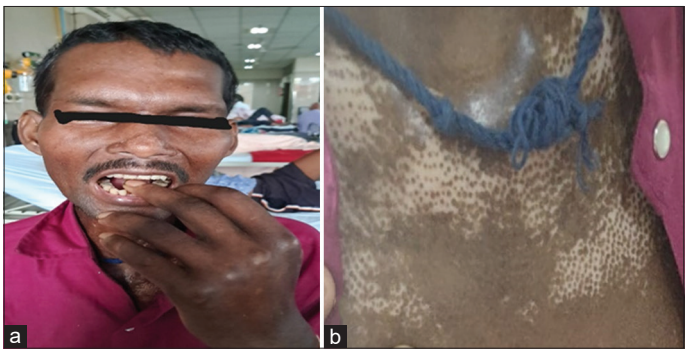

Figure 1: (a) Puckered lips with reduced mouth opening; (b) Salt-and- pepper skin

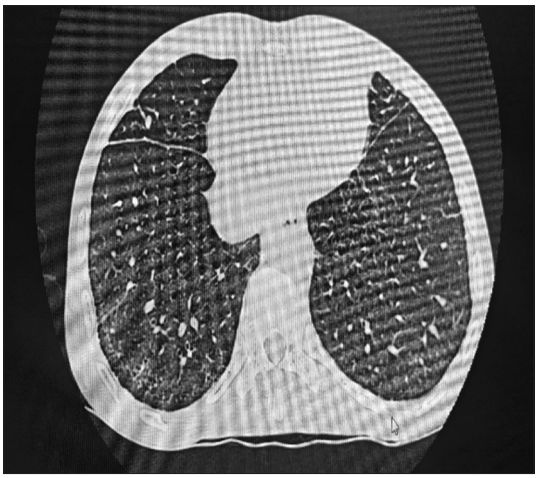

Figure 2: High-resolution computed tomography suggestive of early features of interstitial lung disease [5]

Table 1: Laboratory investigation

Investigations	Results	Interpretation
Complete blood count	HB-7.7 G/dL MCV-54.3 fL WBC-9.8×10^9/L PLT-648×10^9/L	Microcytic anemia
C-reactive protein	7.6 MG/DL	Moderate elevation
Kidney function test	creatinine-0.6 urea-34	Normal range
ANA BLOT	SSA- + RO + SCL-70 +++ Other antibodies Negative	Strong positive titer suggestive of autoimmune disease (systemic sclerosis)
Pulmonary function test	fvc-1.64 (53%) fev1-1.18 (44%) fev1/fvc-72.0 (89%)	Restrictive lung disease (ILD)

HB: Hemoglobin, MCV: Mean corpuscular volume, WBC: White blood cell, PLT: Platelet count, ANA: Antinuclear antibody, FVC: Forced vital capacity, FEV 1: Forced expiratory volume

DISCUSSION

SSc is less common in males but tends to present with more diffuse disease, elevated acute phase reactant, pulmonary, and muscular involvement in the initial phase of the disease [5,6]. They have an increased risk of cardiovascular and renal crisis, and the duration from the diagnosis to the development of pulmonary hypertension is shorter with increased severity of digital vasculopathy [1,6-9].

Men than in females are more likely to carry anti-scl-70 or anti-RNA polymerase III antibodies than anticentromer antibodies, which is supported by the findings of Taouch *et al*. [5,8,10-12]. A case series showed antinuclear antibody positivity in 70% of cases, of which 40% had anti-scl-70 antibody-positive reports [10], although a higher probability of being antinuclear antibody negative was demonstrated by several studies [12,13]. As per a study done by Hussein *et al*. [9] and several other studies [1,2,6,7,9], diffuse SSc has a survival of 40–60% at 10 years onset of non raynauds features phenomenon signs and symptoms than limited SSc which has a survival of 70–75% at 10 years. Scleroderma sine scleroderma- a rare form has a poor prognosis, attributed to an increased risk of malignancy.

ILD can develop in any patient with SSc, including those with pulmonary hypertension, but the risk is increased in those with diffuse (rather than limited) cutaneous SSc, those with anti-Scl-70/anti-topoisomerase I antibody, those with absence of anticentromere antibody [11,12]. While it can occur at any time, the risk of developing ILD is maximum during the initial phase of SSc, so patients should be monitored closely in the first few years after diagnosis. An increased extent of lung fibrosis on HRCT and a low forced vital capacity are predictors of early mortality; it was also evident in the present case.

Most SSc patients' deaths are due to pulmonary involvement or recurrent infection [11]. When compared, males have a higher rate of premature deaths attributed to early manifestation of systemic features [6,7,11].

In the present case, the patient on presentation had skin thickening of gradual onset with progressive course without any systemic complaints; thus, our initial differentials included diffuse cutaneous SSc, scleredema diabeticorum, hyperkeratosis, and paraneoplastic syndrome. However, after clinical evaluation, EULAR criteria for SSc were met, leading to a diagnosis of SSc. Nonetheless, in this male patient with SSc had preserved renal funtion and early pulmonary manifestation which is not in congruence with findings in other case reports/series [1-3,11,12].

CONCLUSION

Although scleroderma is rare in males, it tends to manifest earlier with pulmonary and renal complications, necessitating prompt diagnosis and treatment. With advancements in treatment modalities, survival rates have significantly improved, further emphasizing the importance of early diagnosis and intervention.

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Funding: Nil; Conflicts of interest: Nil.

How to cite this article: Zade TP, Pandharipande MS, Joshi RW, Nagpure Y. A male with ill-fitting skin with dyspnea. Indian J Case Reports. 2025; 11(4):172-174.