ORIGINAL RESEARCH PAPER

Internal Medicine

BEYOND THE SKIN: A DEEPER LOOK AT SYSTEMIC SCLEROSIS

KEY WORDS: Systemic sclerosis, Interstitial lung disease, Chronic kidney disease

Dr. Deval Ghelani* Junior Resident, Department of General Medicine, Dr D.Y. Patil Medical College & Hospital, Nerul, Navi Mumbai. *Corresponding Author

Dr. Prashant Purkar Professor, General Medicine, Dr D.Y. Patil Medical College & Hospital, Nerul, Navi Mumbai.

ABSTRACT

Systemic sclerosis (SSc) is a rare autoimmune connective tissue disease characterized by fibrosis, vasculopathy, and multisystemic involvement. We report a case of a 64-year-old female with diffuse cutaneous systemic sclerosis (dcSSc) presenting with joint pain, dyspnea, and signs of interstitial lung disease (ILD) and chronic kidney disease (CKD). Clinical findings included sclerodactyly, indurated skin, typical facial features and bilateral pedal edema. Investigations revealed anemia, raised inflammatory markers, anti-Scl-70 positivity, NSIP-pattern ILD, and renal involvement without hypertensive crisis. This case highlights the progressive multisystemic nature of SSc, with pulmonary and renal manifestations as primary morbidity contributors. Early recognition and multidisciplinary management can significantly improve outcomes in such patients.

INTRODUCTION

Systemic sclerosis (SSc) is a chronic autoimmune connective tissue disorder marked by widespread vasculopathy, immune dysregulation, and progressive fibrosis affecting the skin and various organs. The disease shows a broad spectrum of clinical phenotypes. Studies show that up to 80% of SSc patients develop interstitial lung disease, and renal involvement is detected in 50% over the disease course, particularly in diffuse cutaneous forms. [1–3]

CASE REPORT

A 64-year-old female presented with history of pain in multiple small joints over 3 years and a 1-month history of dry cough and dyspnea.

On Examination

General examination: Pallor, bilateral pedal edema, sclerodactyly, fish-mouth facies, indurated skin. Respiratory: Bilateral basal crepitations. Cardiovascular, abdominal, neurological: No abnormalities.

Musculoskeletal signs were suggestive of inflammatory synovitis.

INVESTIGATIONS

Hemoglobin: 8.8 g/dL
Serum creatinine: 3.25 mg/dL
24-hour proteinuria: 282 mg/24 hours
ESR: 110 mm/hr, CRP: 11.1 mg/L
ANA (IF): Positive (+++), mixed pattern
ANA blot: Anti-Scl-70 positive
HRCT Thorax: NSIP pattern of ILD
2D Echo: Trivial TR, EF 55%

Renal ultrasound: Small left kidney with bilateral loss of cortico-medullary differentiation.

Renal imaging and lab findings indicated chronic involvement. The absence of malignant hypertension ruled out scleroderma renal crisis, aligning with findings that CKD without crisis is common in longstanding disease. [4] Thus the final diagnosis of Diffuse cutaneous systemic sclerosis with ILD and chronic kidney disease was made.

MANAGEMENT

Patient was managed with multidisciplinary management involving pulmonology, nephrology, rheumatology. Main pharmacological management was Disease-Modifying Antirheumatic Drug (DMARD) for arthritis and Immunosupressives for ILD. Supportive: Pulmonary rehabilitation and dietary counseling.

DISCUSSION

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. Virtually every organ can be affected.

SSc is an acquired sporadic disease with a worldwide distribution and affecting all races. SSc shows a strong female predominance (4:1). An additional risk factor for SSc is having an affected first-degree family member, which increases disease risk 13-fold. Although SSc can present at any age, the peak age of onset in women with both limited and diffuse cutaneous forms is 65–74 years. [5–7]

Multisystemic Involvement Includes:
Skin: Dryness,Increased tightness, mask facies, pursed lip, puckered mouth, Raynaud's phenomenon

Pulmonary: ILD and pulmonary arterial hypertension (PAH)

Renal: CKD and scleroderma renal crisis

Cardiac: Pericardial effusion, arrhythmias, myocardial fibrosis

Gastrointestinal: Esophageal dysmotility, GERD, malabsorption

Musculoskeletal: Arthritis, myalgia, tendon friction rubs

Neurological: Peripheral neuropathy, autonomic dysfunction

Pulmonary complications particularly interstitial lung disease (ILD) and pulmonary arterial hypertension (PAH) are leading causes of morbidity and mortality. Renal crisis, though less frequent remains a life-threatening complication.

Early diagnosis is essential for timely intervention and monitoring. Autoantibodies, such as anti-topoisomerase I (Scl-70), are frequently associated with dcSSc and correlate with severe organ involvement. Management focuses on symptom relief, slowing disease progression, and addressing organ-specific complications, with immunosuppressive therapies are showing some efficacy in ILD. [8,9]

Given its complexity and systemic nature dcSSc requires multidisciplinary care and ongoing research to better

understand its pathogenesis and identify targeted therapies.

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