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Case Report

Clinical Manifestations of Scleroderma (Systemic Sclerosis): A Case Report

Article History:

Name of Authors & *Affiliation*
Dr. Preeti Nigotia[1], Dr. Vinod Kumar Mourya[2]

[1]Associate Professor, Department of General Medicine, SRVS Medical College, Shivpuri, Madhya Pradesh, India
[2]Senior Resident, Department of General Medicine, SRVS Medical College, Shivpuri, Madhya Pradesh, India

Corresponding Author:
Dr. Preeti Nigotia
Email: *preeti9560@gmail.com*

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Abstract: **Background**: Systemic sclerosis, also known as scleroderma, is a rare chronic autoimmune connective tissue disorder characterized by progressive fibrosis of the skin and internal organs, vascular dysfunction, and immune dysregulation. Clinical presentation is often heterogeneous, with variable cutaneous, musculoskeletal, and visceral involvement. **Case Presentation:** We report a case of a young female who presented with extensive dermatological and musculoskeletal features indicative of systemic sclerosis. The patient exhibited marked skin tightening and thickening over the face and extremities, resulting in sclerodactyly and microstomia. Additional findings included patchy alopecia, hypopigmentation and hyperpigmentation, digital ulcerations, calcinosis over the elbows, and significant nail dystrophy. Oral examination revealed dental crowding and restricted mouth opening. There was no evidence of visceral involvement at the time of evaluation. **Conclusion**: This case highlights the diverse clinical spectrum of systemic sclerosis, particularly emphasizing cutaneous and musculoskeletal manifestations. Early recognition and multidisciplinary management are essential to prevent complications and improve patient outcomes. Comprehensive documentation of such cases enhances clinical understanding and facilitates timely diagnosis, especially in resource-limited settings.

Keywords Systemic sclerosis, Scleroderma, Autoimmune disease, Sclerodactyly, Microstomia.

INTRODUCTION

Systemic sclerosis (SSc), or scleroderma, is a rare but serious systemic autoimmune connective tissue disease characterized by dysregulated immunity, persistent vasculopathy, and progressive fibrosis affecting both skin and internal organs [1,2]. The disease manifests in two main subtypes—limited cutaneous and diffuse cutaneous—differentiated by the extent of skin involvement, which also correlates with prognosis and risk of organ complications [1].

The epidemiology of SSc reveals a global prevalence of approximately 17–19 per 100,000 individuals and an incidence of nearly 1–1.4 per 100,000 person-years, with marked heterogeneity across regions and a striking female predominance [3,4]. Clinical manifestations can range from early skin changes—such as Raynaud's phenomenon, sclerodactyly, skin thickening, pigmentation alterations, telangiectasia, and calcinosis—to life-threatening visceral involvement including interstitial lung disease, pulmonary arterial hypertension, and renal crisis [2,5].

The pathogenesis of SSc is multifaceted, involving a complex interplay of genetic predisposition, environmental triggers, and immune dysregulation leading to endothelial cell injury, chronic inflammation, fibroblast activation, and aberrant extracellular matrix deposition [6,7]. Although considerable advances have been made in understanding these mechanisms, effective disease-modifying therapies remain elusive [1,6].

In this report, we present a case of systemic sclerosis with prominent cutaneous and musculoskeletal features, highlighting the clinical heterogeneity and emphasizing the importance of early recognition and multidisciplinary management.

CASE PRESENTATION

A 42 year female patient came to General Medicine department with cheif complaints of progressive tightening of skin over her face, extremities and trunk for the past 8 years. She also reported intermittent pain and stiffness in her fingers, reduced mouth opening, difficulty in grasping object and recurrent ulceration over pressure points especially at the elbow. At the time of

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presentation in Medicine department her blood pressure was 110/60 mmHg, pulse 82 beats/min regular, RR-20/minute, Afebrile. There was no family history of autoimmune or connective tissue disorders.

Cutaneous examination revealed diffuse skin thickening, hyperpigmentation and hypopigmentation in a mottled pattern over the face and extremities, consistent with salt-and-pepper dyspigmentation. The scalp showed evidence of **patchy non-scarring alopecia**, with visible dermal atrophy and tight adherent skin. The facial skin was taut, with limited expression, microstomia, and perioral radial furrowing. The oral cavity examination demonstrated dental crowding and reduced oral aperture, approximately 24mm on maximal opening.

Musculoskeletal examination revealed flexion contractures of the fingers with digital clawing. The fingers were tapered with **sclerodactyly**, and there was marked **nail dystrophy** with subungual hyperkeratosis. Multiple digital ulcers and **calcinosis cutis** were noted over the extensor surfaces of both elbows. Joint mobility in the hands and wrists was restricted due to fibrotic changes.

Radiographic evaluation (X-ray of both hands) demonstrated **acro-osteolysis** of distal phalanges, joint space narrowing, and soft tissue contractures. These findings supported the clinical diagnosis of systemic sclerosis with significant skeletal involvement.

Systemic evaluation including respiratory, renal, and gastrointestinal assessment did not reveal any overt signs of visceral involvement at the time of presentation. Laboratory investigations showed CBC- 10.2 gm/dL, TWBC 7,400/mm3, platelet count 158,000/mm3, ESR-24; LFT, RFT, RBS-within normal limit, CRP levels High (1.1), ANA test by ELISA positive (28 unit), Anti DS DNA high (120).

The patient was clinically diagnosed with **diffuse cutaneous systemic sclerosis (dcSSc)** based on ACR/EULAR 2013 classification criteria [8] and was referred to a multidisciplinary care team for immunosuppressive therapy initiation, physical rehabilitation, dental consultation, and regular follow-up.

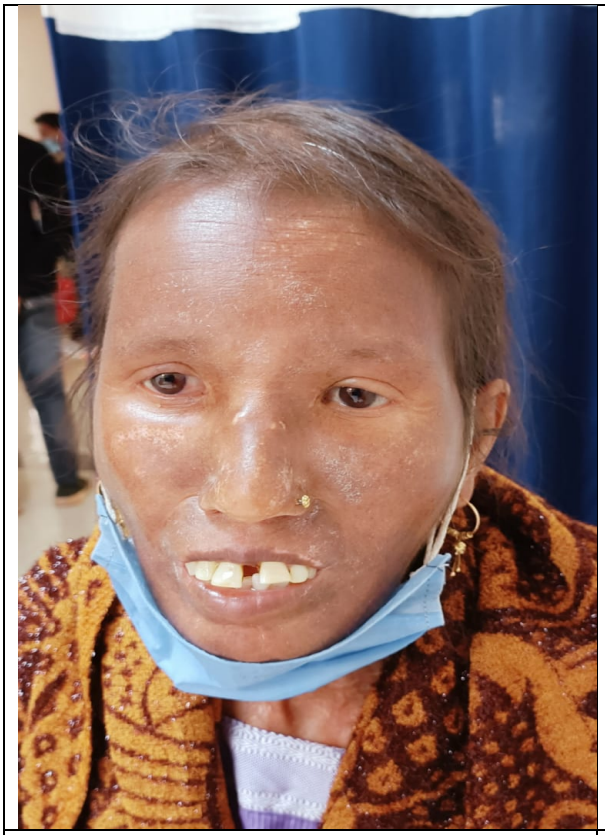

Image 1: Mask like (Expressionless) face and nasal alar resorption

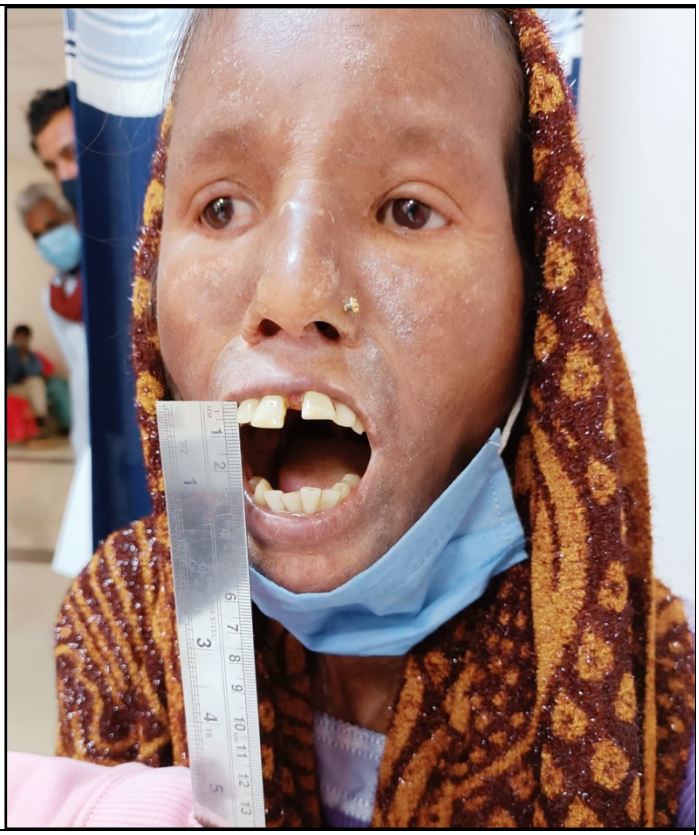

Image 2: Patient's maximum mouth opening

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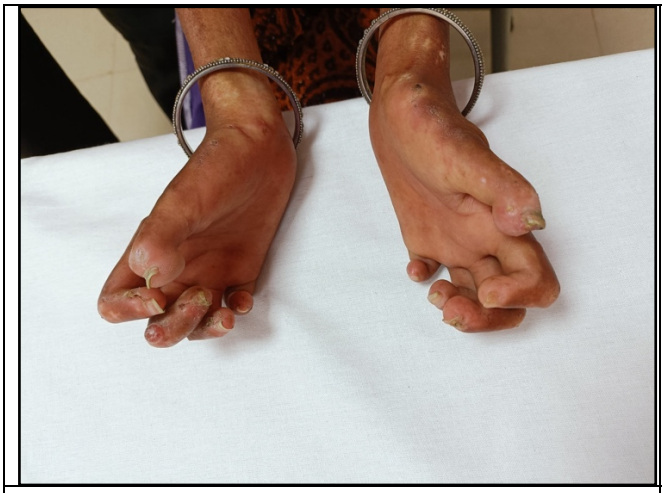

Image 3: Sclerodactyly, clawlike position of fingers, loss of digital pulp with erosion.

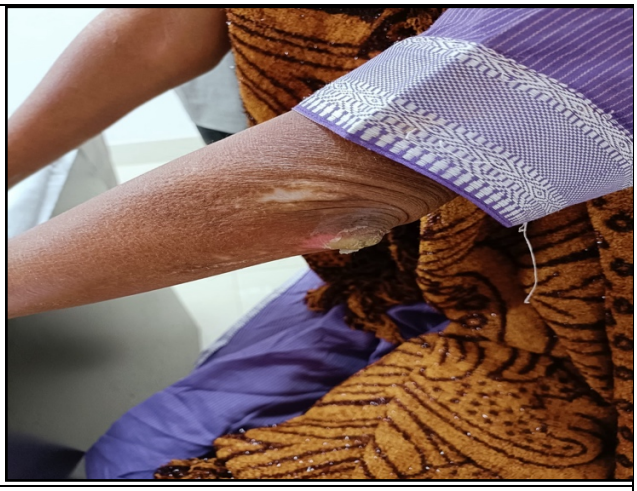

Image 4: Calcinosis and ulceration at pressure point like elbow

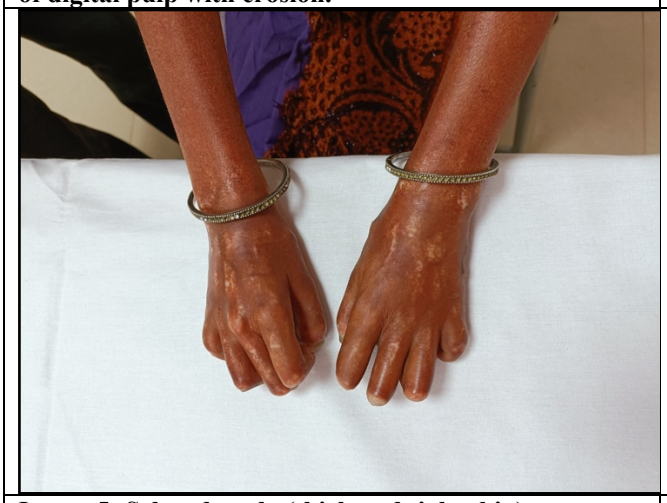

Image 5: Sclerodactyly (thick and tight skin)

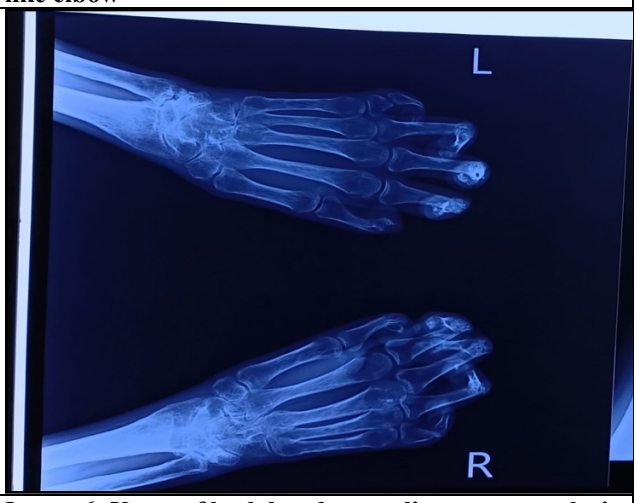

Image 6: X-ray of both hands revealing acro-osteolysis of distal phalanges with soft tissue contractures.

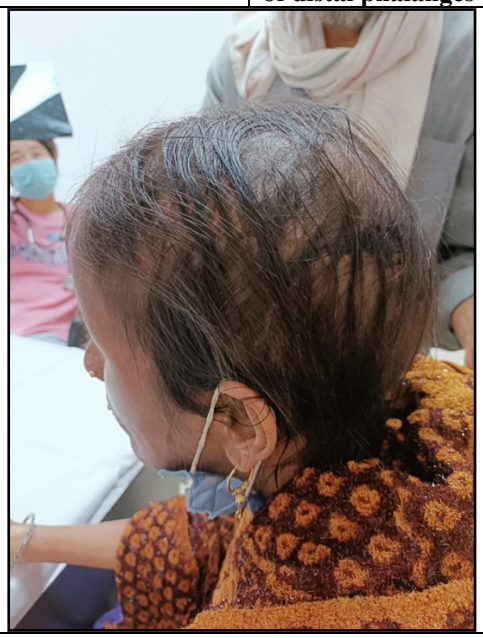

Image 7: Scalp image revealing non-scarring alopecia

DISCUSSION

The presence of acro-osteolysis in our patient's hand radiographs is consistent with well-recognized musculoskeletal manifestations of systemic sclerosis (SSc), where distal phalangeal bone resorption is reported in approximately 20–25% of cases. This process is believed to result from chronic vascular ischemia, progressive skin tightening, and impaired angiogenesis [9]. Such skeletal changes are indicative of the underlying vasculopathy and fibrotic cascade that are central to the disease pathogenesis.

Calcinosis cutis, as observed in our patient over pressure-bearing areas such as the elbows, represents another common systemic complication of SSc. It is associated with a significant clinical burden—pain, limitation of mobility, cosmetic deformity, and psychological distress—with many patients experiencing a marked reduction in quality of life [10]. Current treatment options are limited; pharmacological agents such as calcium channel blockers, bisphosphonates, warfarin, and minocycline have been trialed, but their therapeutic benefit remains inconsistent. Surgical or procedural removal is typically reserved for severe or symptomatic cases [11].

Fibrotic involvement of soft tissues, including the face and perioral region, can contribute to microstomia, which is reported in around 7% of patients with SSc. This reduction in mouth opening (<30 mm) is strongly correlated with disease severity and systemic organ involvement, and it can impair nutrition, speech, and self-care [12]. Other craniofacial features in SSc include xerostomia from salivary gland fibrosis, telangiectasia, and tongue rigidity, all of which can increase susceptibility to mucosal injury, infections, and nutritional compromise [13].

Beyond the musculoskeletal and cutaneous domains, SSc involves multiple organ systems, including the lungs, heart, gastrointestinal tract, and kidneys, driven by the same vasculopathic and fibrotic mechanisms. While organ-specific management is essential, supportive measures targeting functional impairment also play a role. Recent interventional approaches, such as pulsed CO_2 laser therapy for perioral fibrosis, have shown measurable functional improvement (mean mouth opening gain of 8.5 mm at 12 months) with minimal adverse effects [14]. Similarly, structured physiotherapy programs have demonstrated potential in preserving tissue mobility and halting disease-related functional decline, although larger controlled trials are needed to confirm sustained benefit [15,16].

The cornerstone of SSc management is to halt disease progression and minimize complications. In the present case, treatment included methotrexate (15 mg/week) and low-dose methylprednisolone (4 mg/day). Given the increased risk of renal crisis in SSc, glucocorticoids—particularly prednisone—should be administered cautiously at doses below 10 mg/day. The pathogenesis of SSc involves a triad of inflammation, vasculopathy, and fibrosis; therefore, therapeutic regimens are tailored to address these mechanisms through immunosuppressive agents, vascular-targeted therapies, and antifibrotic strategies. Commonly used immunosuppressants include mycophenolate mofetil, cyclophosphamide, corticosteroids, methotrexate, and selected biologic agents [17,18].

In this case, amlodipine 10 mg/day, a calcium channel blocker, was initiated to address Raynaud's phenomenon and associated vasculopathy. Standard vascular therapies in SSc include calcium channel blockers, angiotensin-converting enzyme (ACE) inhibitors, angiotensin II receptor blockers (ARBs), and prostacyclin analogs [19].

Cutaneous manifestations were managed with topical corticosteroids and regular emollient application. Chronic inflammation in SSc promotes atrophy of the skin, adnexal structures, and glands, leading to xerosis, reduced sebum production, and increased vulnerability to trauma; hence, daily use of moisturizers is recommended [20]. Fibrotic changes in the hands may progress to joint contractures, emphasizing the importance of patient education regarding Raynaud's phenomenon, including avoidance of precipitating factors such as nicotine, emotional stress, sympathomimetic drugs, and cold exposure [21].

Potential complications of SSc encompass sclerodactyly, digital ulceration, gangrene necessitating amputation, interstitial lung disease, pulmonary hypertension, inflammatory myocarditis, arrhythmias, conduction defects, restrictive cardiomyopathy, and acute or chronic renal failure [22]. Gastrointestinal tract involvement occurs in over 60% of patients, presenting as dysphagia, peptic strictures, peptic ulcer disease, pseudo-obstruction, and malabsorption. Prognosis is largely determined by the extent of internal organ involvement, with pulmonary complications representing the leading cause of mortality. Reported survival rates are approximately 74.9% at 5 years and 62.5% at 10 years [23].

CONCLUSION

This case underscores the multifaceted burden of systemic sclerosis, featuring digital bone resorption, peripheral calcinosis with ulceration, and orofacial dysfunction from microstomia—each reflecting the disease's hallmark vascular fibrosis. Incorporation of combined therapeutic approaches—ranging from laser therapy, tailored rehabilitation, to symptomatic surgical interventions—demonstrates the imperative of a multidisciplinary management strategy. Early detection and integrative care pathways are vital to preserve function, reduce morbidity, and enhance quality of life in SSc patients.

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