Scleroderma- A case report

Abstract-Scleroderma or systemic sclerosis is a chronic multisystem disease characterized by excess deposition of connective tissue in skin and internal organs, associated with microvasculature changes and immunologic abnormalities.
We hereby report a case of scleroderma in a 52 year old female with classical clinical and histopathological findings.

Introduction-Scleroderma is a rare connective tissue disorder with unknown and complex pathogenesis. Scleroderma can be divided into two forms, localized Scleroderma (morphea, linear scleroderma, and scleroderma en coup de sabre), or Systemic sclerosis, which can further be classified as either limited systemic sclerosis (formerly known as the CREST syndrome) or diffuse systemic sclerosis based on clinical and serological criteria.[1]
The pathogenesis of systemic sclerosis is complex and includes vascular alterations, immunological dysregulation, and extensive tissue fibrosis.[2] However, the trigger of this early putative vascular injury remains elusive. Ineffective angiogenesis, which can be assessed using nailfold capillaroscopy, is clearly apparent in scleroderma. A critical imbalance between factors promoting vasoconstriction (e.g., endothelin) and vasodilation (e.g., nitric oxide)is a key factor promoting vascular changes.Subsequent to vascular injury and immune disturbances, activated fibroblasts result in the excess deposition of extracellular matrix, which includes collagen, resulting in organ dysfunction and tissue fibrosis.[3]
Limited cutaneous systemic sclerosis, formerly known as the CREST syndrome, is associated with skin thickening distal to the elbows, distal to the knees, and/or face without trunk involvement. Diffuse cutaneous systemic sclerosis is associated with skin thickening that may involve skin proximal to the elbows, proximal to the knees, face, and/or trunk. Antinuclear antibodies (ANA) may be present in more than 90% of cases of systemic sclerosis, and at least one of the more specific autoantibodies (anti-centromere, anti-SCL70, and anti-RNA polymerase III) is present in up to 70% of the cases. The organs most frequently affected by scleroderma are the skin, gastrointestinal tract, lungs, kidneys, skeletal muscle, and pericardium.[4,5,6]

Case report-

A 52 year old female, who is a farmer hailing from Thiruporur, presented to our OPD with complaints of thickening of skin over the hands and tightness of skin over the face for 6 months.

She was apparently normal 6 months back following which she developed thickening of skin, first over the distal part of her fingers, which were initially pruritic and edematous, then gradually extending proximal to the metacarpophalengeal joints. It was associated with difficulty in joint movement.

She also complained of skin tightness over the face, which was of gradual onset initially involving the perioral area, which was associated with difficulty in opening her mouth.There was history of occasional episodes of skin discolouration associated with pain on exposure to cold temperatures over the fingers, suggestive of Raynaud's phenomenon. Patient gives a history of painful ulcers and whitish hard raised lesions present over fingers. She also claims to have decreased sweating over hands. Occasional episodes of myalgia were observed. No history of fever. No history of breathlessness, dry cough, chest pain or palpitations. No history of nausea, constipation, diarrhoea, regurgitation, abdominal bloating or discomfort. No history of oliguria, swelling of feet, swelling around eyelids or frothy urine.No history of headache, visual disturbances or seizures. No history of grittiness in eyes, tingling or numbness over extremities. No history of photosensitivity, rash over face or oral ulcers. No history of drug intake or previous treatments for present condition.She is not a known case of diabetes, hypertension, tuberculosis,bronchial asthma. No history of any drug allergy. No past surgical history.No similar complaints in the family.

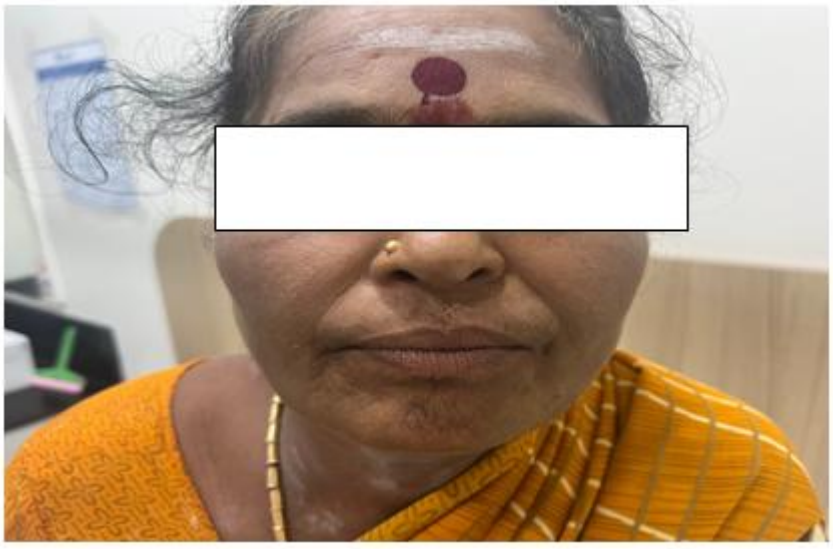

Fig-1: Taut facial skin with induration

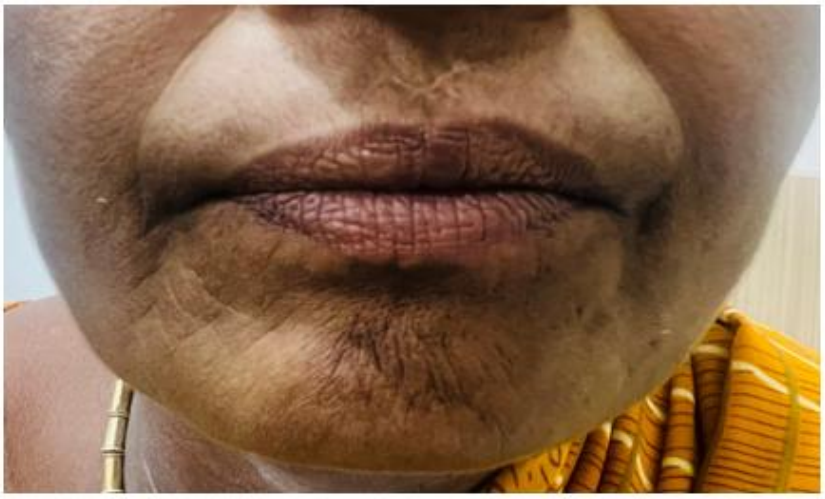

Fig-2: Perioral rhagades giving a purse string appearance

Fig-3

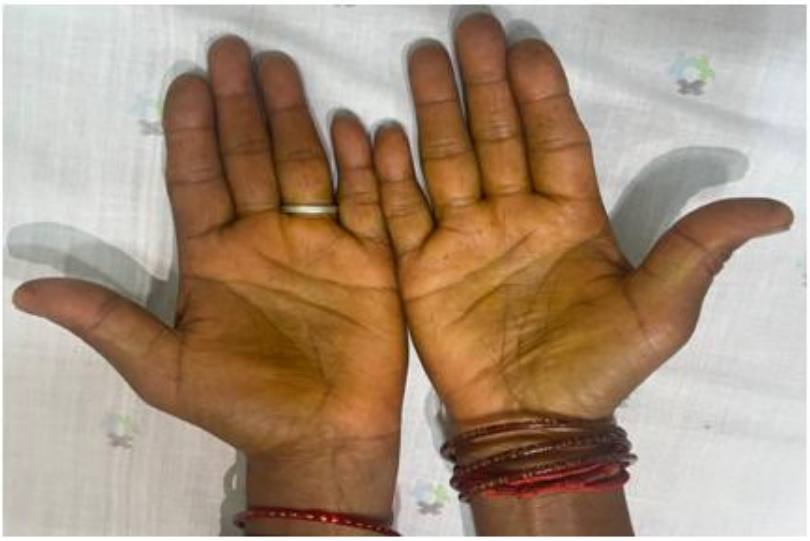

Fig-4

Fig.3 and 4 showing puffy fingers extending proximal to the metacarpophalengeal joint

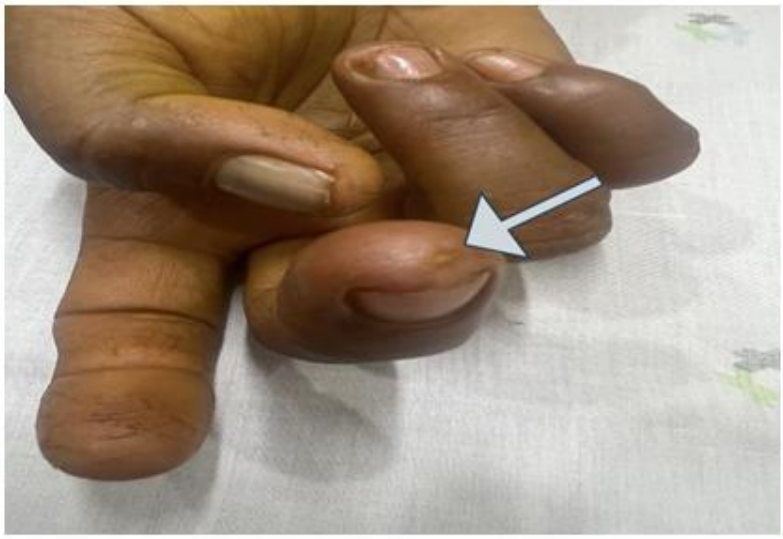

Fig-5: Pitted scar over tip of the right middle finger

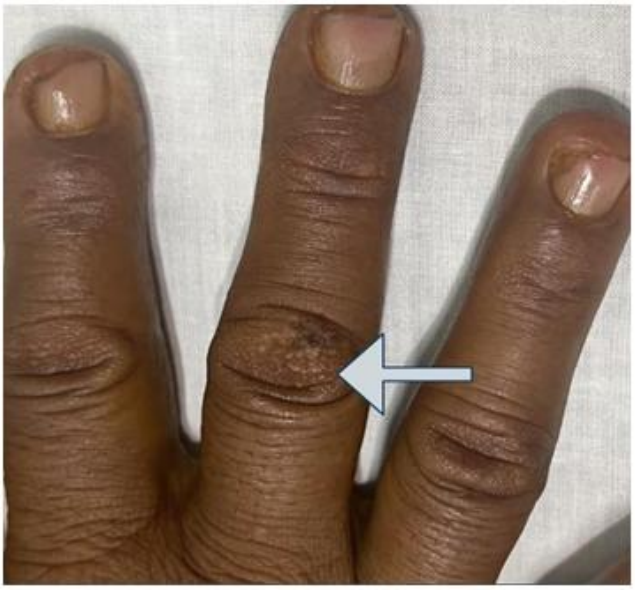

Fig-6: White chalky deposits over the dorsum of right middle finger

On examination, patient was conscious and well oriented. Vitals were stable. General examination was normal. No significant findings on systemic assessment. On cutaneous examination, the patient's facial skin was taut with induration(Fig-1). Ingram sign was positive. Perioral rhagades were present, giving a purse string appearance(Fig-2). On examination of the hands,Sclerodactyly extending proximal to metacarpophalangeal joints on both hands present(Fig-3 and 4). Pitted scar was present at the tip of right middle finger(Fig-5). Loss of hair over all fingers was noted. Calcinosis cutis was present over right middle finger(Fig-6) and left index finger. No flexion deformities or loss of pulp of finger. No significant nail fold changes were observed. Tendon friction rubs were absent. Raynaud's phenomenon was clinically elicited. Onycholysis, fissuring over palms and soles and dental caries present on further examination. Routine heatological tests were normal,chest Xray had no significant findings.RFT and glucose profile were normal.

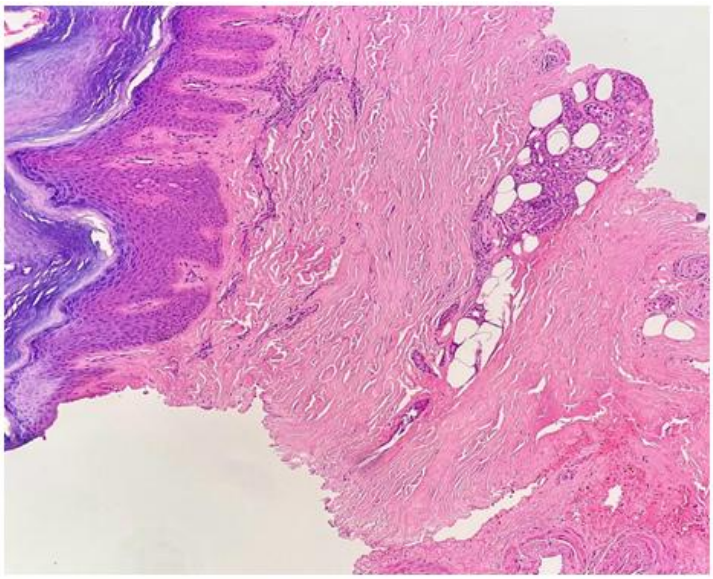

Fig-7: H&E staining showing hyperkerstosis, hypergranulosis and acanthosis of epidermis. Thick, compact, homogenous and hyalinised collagen bundles in the dermis

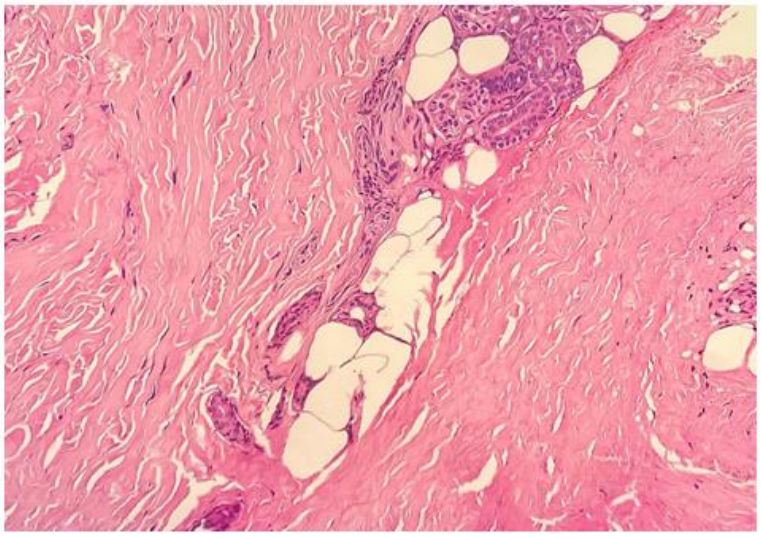

Fig-8: H&E staining showing collagen formation below subcutaneous tissue

Skin biopsy sample was obtained from the right middle finger which on examination revealed, hyperkeratosis,hypergranulosis and acanthosis of the epidermis. Dermis showed thickened compact homogenous hyalinised collagen with absent eccrine glands. Collagen bundles were present below the sub cutaneous tissue. Based on the above findings, a diagnosis of limited

scleroderma was made. Patient was started on Sildenafil, NSAIDS and Vitamin E supplements. Patient is under regular follow up.

Conclusion-Systemic sclerosis is still undoubtedly a challenge for clinicians, because of the variety of symptoms. Scleroderma course and prognosis depend on the clinical picture and character of organ involvement, especially kidney, heart and lungs.As the pathogenesis of systemic sclerosis is still unclear, the treatment is based on disease-modifying and organ-specific drugs. Therapeutic decisions should be made after appropriate assessment of symptoms, disease duration, activity and complications.Although early diagnosis and new therapeutic options significantly improve systemic sclerosis prognosis, it is still characterized by a severe course and high risk of early death. Studies are needed to define effective treatments in these diseases, which cause great morbidity.

References-

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