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Case Report

Scleroderma: a case report

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ABSTRACT

Scleroderma is systemic multi organ autoimmune disorder characterized by hardening of skin. Also known as systemic sclerosis. Estimated annual incidences of approximately 19 cases per million persons. The limited skin disease has a 10-year survival rate of 71%, whereas those with diffuse skin disease have a 10-year survival rate of just 21%. Risk is higher in women than men and peak in individuals aged 30-50 years. It has no definitive treatment. It may be limited or diffuse depending upon manifestations of symptoms or signs affecting internal organs especially lungs, heart, or kidney. We report a case of scleroderma with pulmonary hypertension and interstitial lung disease in our hospital who presented with tightening of skin, joint pain, dysphagia, and breathlessness. On examination skin appeared dark, shiny, and tight, with loss of hair, paraesthesia and digital ulceration. Patient also has history of Raynaud's phenomenon. On investigation, Scl-70 and ANA (antinuclear antibodies) by enzyme immunoassay came positive. HRCT thorax was suggestive of interstitial fibrosis and PFT revealed moderate restriction. On 2D echocardiography, mild pulmonary hypertension was present while barium swallow showed motility disorder involving oesophagus. On view of extensive systemic involvement like skin, respiratory system, gastrointestinal system and heart, we would like to present this rare disorder.

Keywords: Scleroderma (SSc), Pulmonary hypertension (PAH), Raynaud's phenomenon, Interstitial lung disease (ILD)

INTRODUCTION

Systemic sclerosis is a chronic autoimmune disease characterised by thickening and fibrosis of the skin. Internal organs like heart, lungs, kidneys and gastrointestinal tracts are often involved. It is connective tissue disorder of unknown etiology, heterogeneous clinical manifestations and chronic and often progressive course.[1] Patients with scleroderma may develop either a localized or a systemic (body-wide) form of the disease.[2] Gintrac in 1847 introduced the term "scleroderma" as skin is most obvious organ involved.[1]

Localized scleroderma usually affects only the skin on the hands and face. Its course is very slow, and it rarely, if ever, spreads throughout the body (becomes systemic) or causes serious complications. There are two main forms of localized scleroderma: morphea (patches of hard skin) and linear (band of hard skin involving bone) scleroderma.[2]

Systemic scleroderma is also called systemic sclerosis. This form of the disease may affect the organs of the body, large areas of the skin, or both. This form of scleroderma has two main types: limited and diffuse scleroderma.[2] In limited disease [formerly called

CREST (Calcinosis, Raynaud's phenomenon, oesophageal dysmotility, sclerodactyly, and telangiectasia's) syndrome], skin tightening is confined to the fingers, hands, and forearms distal to the elbows, with or without tightening of skin of the feet and of the legs distal to the knees. In diffuse disease or diffuse cutaneous systemic sclerosis (dcSSc), the skin of the proximal extremities and trunk is also involved. Both dcSSc and lcSSc are associated with internal organ involvement; however, patients with dcSSc are at greater risk for clinically significant major organ dysfunction. These patients are at risk for early pulmonary fibrosis and acute renal involvement.

Systemic sclerosis sine scleroderma is a rare disorder in which patients develop vascular and fibrotic damage to internal organs (phenotypically similar to that in limited scleroderma), in the absence of cutaneous sclerosis.[3]

Pulmonary Arterial Hypertension (PAH) in SSc can be an isolated precapillary pulmonary artery hypertension or secondary in association with Interstitial Lung Disease (ILD). PAH developing in SSc is particularly severe and one year survival following diagnosis is 55%.[4]

Pathogenesis

Three cardinal features of the disease are vasculopathy, cellular and humoral auto immunity, and progressive visceral and vascular fibrosis in multiple organs. Autoimmunity and altered vascular reactivity may be the earliest manifestations of systemic sclerosis. The pathophysiology involves several cell lines, such as endothelium, fibroblasts, lymphocytes and their mediators. The vascular phase begins in the endothelium of small vessels throughout the body, although the primary event that triggers endothelial damage is unknown. Tissue hypoxia is one of the primary events that modify vascular tone.[4]

The endothelium contributes to the regulation of the contraction and relaxation of vascular smooth muscle cells through the production and release of endothelium-derived vasoactive substances including prostacyclin [prostaglandin (PG)I_2], endothelium-derived relaxing factor (EDRF or nitric oxide), and endothelin. Endothelin-1 plays an important role in the pathogenesis of scleroderma. Elevated plasma endothelin is seen in scleroderma-associated pulmonary hypertension and Raynaud's phenomenon. Other key growth factors that have also been implicated in the pathogenesis are Connective Tissue Growth Factor (CTGF), and Platelet-Derived Growth Factor (PDGF).[3]

Thus we report such a case of diffuse scleroderma presenting with diffuse skin hardening, dysphagia and breathlessness having Raynaud's phenomenon, pulmonary hypertension and early pulmonary fibrosis.

CASE REPORT

65 year old female presented to hospital with complains of darkening and tightening of skin all over body which was gradual in onset over 10 months (Figure 1). Patient also has history of discolouration of skin on exposure to cold which begin over 7 months. Over 2 months, patient started complaining of dysphagia and breathlessness. Gradually patient complained loss of wrinkling over face and limitation of movement at knee joint due to skin tightening.

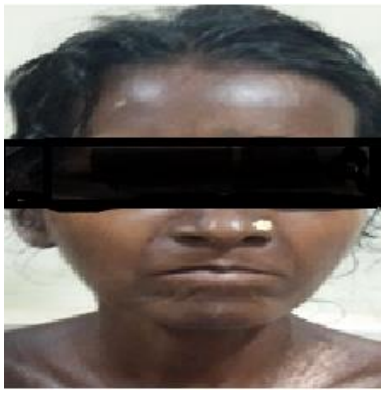

Figure 1: Pinched nose.

On inspection, skin appeared shiny, tight and loss of hair over face, fingers, knees, and toes which was diffuse. Swelling over distal interphalangeal joints with pitting scars over finger tips was present (Figure 2 & 3). Hyperpigmentation of skin over trunk is seen (Figure 4).

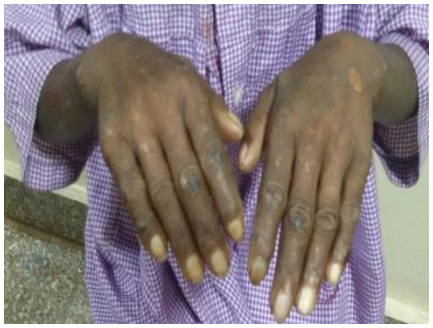

Figure 2: Pigmentation and tightening of skin.

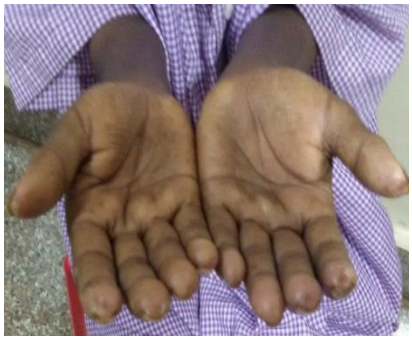

Figure 3: Pitting scar over fingertips.

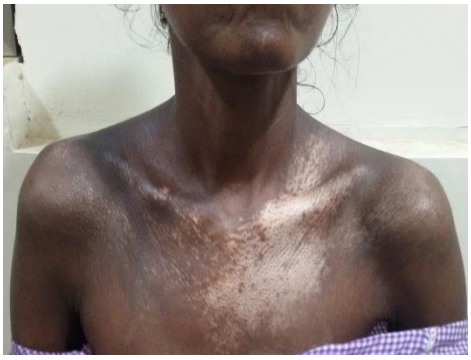

Figure 4: Pigmentation and tightening of skin.

On further evaluation and blood investigation, anti-nuclear antibody was positive (2.7, reference range up to 1.2) while anti SCL - 70 antibody was positive (198, reference range up to 20 RU/ML) which confirmed diagnosis. Later barium swallow was done which was suggestive of motility disorder or connective tissue disorder in lower 2/3rd of oesophagus. High Resolution Computed Tomography (HRCT) of chest was done which revealed interstitial thickening of bilateral interlobular areas of lungs suggests early interstitial fibrosis. Pulmonary function test showed moderate restriction. 2D echocardiography showed pulmonary hypertension of more than 40 mmHg with left ventricular diastolic dysfunction. Rest other blood parameters appeared normal with no renal involvement.

Thus above findings gave us diagnosis of diffuse scleroderma with multiorgan involvement.

DISCUSSION

Systemic sclerosis is a connective tissue disorder of unknown etiology. Raynaud's phenomenon, defined as episodic vasoconstriction of small arteries of fingers and toes is the first symptom in 95% of cases. In dcSSc, interval between Raynaud's phenomenon and appearance of other manifestations is brief over weeks or months while in lcSSc, it follows over years. Involvement of skin leads to firm, thickened and eventually tightly bound skin to underlying subcutaneous tissue. Involvement of face results in thinning of lips, loss of skin wrinkles and facial expressions. The nose takes on a pinched or beaklike appearance. In oesophagus there is hypotonia or atony with hypokinesia or aperistalsis in lower two-third of oesophagus. More than 50% of patients with scleroderma have musculoskeletal features, majority of which are seen in hands. Involvement of fingers leads to sclerodactyly, atrophy and resorption of soft tissue at finger tips with soft tissue calcification. Pulmonary fibrosis and secondary pulmonary hypertension seen in diffuse variety is a cause for the mortality and presents with exertional dyspnoea, fatigue, palpitation, chest pain, syncope and cough. It is thus necessary for regular check-up and evaluation of any organ involvement for definitive management in decreasing early progression of disease.

Although there is no definitive cure for scleroderma, early diagnosis followed by addition of calcium channel blocker or ACE inhibitors and symptomatic relief with physiotherapy and psychotherapy involving proper nursing care to avoid tissue injury plays an important role to reduce mortality and modify quality of life.

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