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Sero-Negative Systemic Sclerosis: A rare presentation

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ABSTRACT: Systemic sclerosis is an uncommon connective tissue disorder characterised by multi system involvement, heterogeneous clinical manifestation, a chronic and often progressive coarse and significant disability and mortality. s. It is diagnosed by presence of characteristic clinical findings and is supported by specific serologic abnormalities. ANA is positive in case of systemic sclerosis in 90 percent of cases. We report a rare case of this rare disease where patient was ANA, Antitopoisomerase I (anti-Scl-70), Anticenteromere antibody negative.

Keywords: Systemic sclerosis, Antinuclear antibody, Anticenteromere, Antitopoisomerase I

A 42 year old female patient came to the out patient department with history of skin thinkening of both upper limb and lower limb since last 5 months. The skin thickening was insidious in onset and involved both hands and feet. It progressed to involve face and trunks. Patient gave history of stiffening of joints of both hands and leg and was associated with pain and swelling and itching. Patient complained of unable to stand up from sitting position. History of Raynauds phenomenon was present for last 3 months. Patient complained of unable to open mouth fully and was able to open only 2 fingers. There was no history of dysphagia.

On examination, patient was afebrile, pulse was 96/min, blood pressure 100/70 mmHg respiratory rate of 24/min. There was no pallor, cyanosis, clubbing, icterus and lymphadenopathy .Skin thickness was present extending upto elbow joint in upper limb and upto patella in lower limb. Skin thickness was also present on face and trunks. Skin over forearms and abdomen and behind ears showed salt and pepper pigmentation that is depigmentation with perifollicular retention of pigment. Patient had finger tip ulcerations. Rest of the general examination was normal. Cardiovascular, respiratory, abdomen and central nervous system examination was normal. Patient's total score (ACR/ EULAR score for systemic sclerosis) was 13 (greater than 9) – this was consistent with systemic sclerosis.

INVESTIGATION

CBC		**LFT**		**Urine**		
Hb	12	Sr. Bili		Albumin	Nil	
Tlc	6520	Sr Bili (T)	0.6	Sugar	Nil	
Plt	306000	Direct	0.5	Ketones	Nil	
Hct	38.6	Indirect	0.1	M/E	Nad	
Mcv	75	SGOT	49	Esr		35
Urea	27	SGPT	106			
Creat	0.4	ALP	61			

ANA	NEGATIVE			
Scl-70	NEGATIVE(0.50)		**Centromere AB**	Negative(0.09)
		2D		
ECG	WNL	ECHO	NORMAL	NO EVIDENCE OF PULMONARY HYPERTENSION

Anti-RNA polymerase III antibody, anti-CCP antibody, RA factor were negative.

X-ray chest was normal.

Serum protein electrophoresis did not show any monoclonal band.

Skin Biopsy taken from left forearm was consistent with systemic sclerosis. It showed marked thickening of collagen bundles in reticular and papillary dermis with hyalinised appearance.

Discussion

Skin thickening is a characteristic manifestation of SSc, and many physicians recognize this disease by this feature. Systemic sclerosis is associated with positive ANA in 90% of cases. In the EULAR Scleroderma Trials and Research (EUSTAR) database 5390 patients who fulfilled the American College of Rheumatology criteria for systemic sclerosis were enrolled and screened for the absence of ANA. In a study it was found that 92.3% cases were ANA positive, 30.4% cases were anti-centromere antibody positive and 36.4 were Scl-70 positive. In another study of 3249 patients, it was noted that (6.4%) were ANA negative [3]. There are case reports where sero-negative systemic sclerosis is associated with malignancy but these patients were also negative for Raynauds phenomenon [4]. It has been found to be associated as paraneoplastic manifestation of secondary malignancies such as Multiple Myeloma or CA breast. SLE and dermatomyositis may present with similar manifestations but other criteria's are not fulfilled in our case. Our patient did not fulfil any criteria for systemic sclerosis mimic and had no evidence of malignancy.

DIFFERNTIAL DIAGNOSIS

1) SCLERODERMA

The term scleroderma is commonly used to describe a group of localized skin disorders. These occur more commonly in children than in adults. In contrast to SSc, localized scleroderma is rarely complicated by Raynaud's phenomenon or significant internal organ involvement. Morphea presents as solitary or multiple circular patches of thickened skin or, rarely, as widespread induration (generalized orpansclerotic morphea); the fingers are spared. Linear scleroderma—streaks of thickened skin, typically in one or both lower extremities—may affect the subcutaneous tissues, leading to fibrosis and atrophy of supporting structures, muscle, and bone.

2) EOSINOPHILIC FASCIITIS

Eosinophilic fasciitis is a rare idiopathic disorder associated within duration of the skin that generally develops rapidly. Adults are primarily affected. The skin has a coarse cobblestone "peau d'orange" appearance. In contrast to SSc, internal organ involvement is rare, and Raynaud's phenomenon and SSc-associated autoantibodies are absent. Furthermore, skin involvement spares the fingers. Full-thickness excisional biopsy of the lesional skin reveals fibrosis of the subcutaneous fascia and is generally required for diagnosis

3) Drug Induced

Bleomycin, carbidopa, pentazocine, cocaine, penicillamine, vitamin k has been documented to cause systemic sclerosis but our patient had no history of any such drug intake

4) Endocrine disorders

Diabetes mellitus and myxoedema can be associated with skin thickening. Can be associated with POEMS (Polyneuropathy, Organomegaly, Endocrinopathy, Monoclonal gammopathy and Skin changes) syndrome

5) Scleromyxoedema

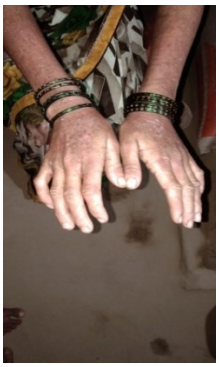
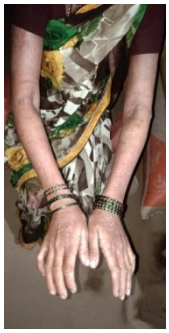
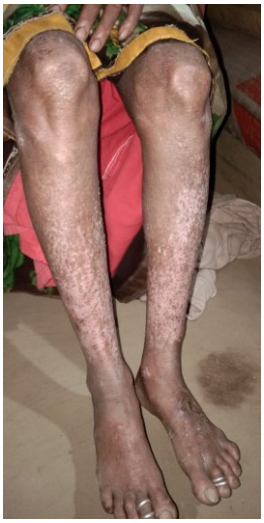
Scleromyxoedema is characterized by yellow and waxy papules arranged in a linear fashion. When the papules coalesce, induration similar to sclerodermatous changes may be noted. This is also common with paraproteinemias

Other conditions may mimic systemic sclerosis by causing hardening of the skin. Diagnostic hints that another disorder is responsible include the absence of Raynaud's phenomenon, a lack of abnormalities in the skin on the hands, a lack of internal organ involvement, and a normal antinuclear antibodies test result.

Meticulous history taking, examination and thorough diagnostic evaluation is important before labelling patient as sero-negative systemic sclerosis

CONCLUSION

Thorough clinical evaluation of patients is needed to rule out mimics of Systemic Sclerosis. This is a rare presentation of relatively rare disease where there was no evidence of underlying malignancy and patient had negative Anti-Nuclear Antibodies.

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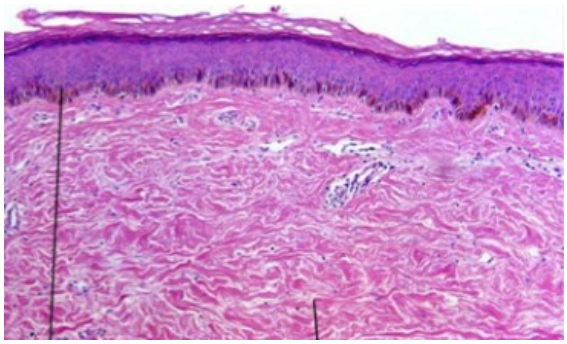

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