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Research Article

DILATED-CARDIOMYOPATHY: UNUSUAL EARLY MANIFESTATION OF CREST SYNDROME

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A B S T R A C T

Systemic Sclerosis (SSc) which is also known as scleroderma is a multisystem autoimmune disease characterised by vasculopathy, organ impairment, immune abnormalitiesand dermal fibrosis. SSc incidence is about 2.7-19.3 new cases per million adults per yearadults with a female to male predominance of 4.6: 1. The presence of cardiac involvement in SSc is often seen in 20% to 25% of patients which is associated with a 70% mortality at 5 years.
A 34 years old female presented with complain ofepisodic bluish discolouration of digits of upper limb and swelling over hands since 1 year. She has dyspnea on exertion since 2 months which was associated with palpitations since 1 month. She was tachypanic with tachycardia & was normotensive and had 99 % saturation on room air. Her face was mask like with loss of wrinkles and expressions with small mouth aperture. On CVS examination, S_1 and S_2 were heard with S_3 and loud P_2 with basal crackles. 2D echo was showing of all cardiac chambers were dilated with ejection fraction of 25%. Her ANA was positive with Anti-Scl70 antibody. Hence, the patient was diagnosis as DCMP (Dilated cardiomyopathy) in CREST syndrome. So, we are reporting this case because of development of cardiovascular complication (DCMP) in very early causes of systemic sclerosis.

INTRODUCTION

Systemic Sclerosis (SSc) which is also known as scleroderma, is a multisystem autoimmune disease characterised by vasculopathy, dermal fibrosis, organ impairment and immune abnormalities. Two forms of Ssc have been described: Diffuse cutaneous systemic sclerosis (*DSSc*) and limited cutaneous systemic sclerosis (*LSSc*). Patients with LSSc display features like Calcinosis cutis, Raynaud's phenomenon, oesophageal dysmotility, sclerodactyly and telangiectasia which together abbreviated as CREST syndrome.Calcinosis occurs due to the calcium deposits in the connective tissues, which are mainly found on the trunks, face, fingers, hands and on the skin above the elbows and knees. Raynaud's phenomenon is a condition in which the small blood vessels of the hands and/or feet contract in response to cold or anxiety.Oesophageal dysfunction is impaired function of the oesophagus that can result in swallowing difficulties. Sclerodactyly is thick and tight skin on the fingers, resulting from deposits of excess collagen within skin layers. Telangiectasia are small red spots on the hands and face that are caused by the swelling of tiny blood vessels. CREST syndrome is an autoimmune disorder, where an immune system stimulates the produce the collagen, which builds up in the skin and internal organs and impairing their function. There is no cure. Aim of the treatment is to relieve the signs and symptoms and preventing complications. Its incidence is about 2.7-19.3 new cases per million adults per yearadults with a female to male predominance of 4.6: 1 [1].

The clinical manifestations are heterogenous and depend on its form and degree of organ involvement. SSc is associated with different systemic involvement present with symptoms like arthralgias, pulmonary hypertension in presence or absence of interstitial fibrosis, primary biliary cirrhosis, renal crises, entrapment neurologic syndromes, risk of malignancies. Cardiac involvement is one of the rare. Patient may present with symptoms likepalpitations, exertional chest pain, dyspnoea, dizziness, and peripheral oedema as a late manifestation. But in our case cardiac symptoms present very early and with a very rapid course. So, we are reporting this case as an unusual and early manifestation of the cardiac symptoms.

Case Report

A 34 years old female presented in Out Patient Department with complainof episodic bluish discolouration of digits of upper limb (UL) since 1 years which was more frequent during winter season, difficulty in holding objects with her both palms with swelling over hands with thickening of skin of palmssince 1 year(fig 1). She also complains of epigastric burning with sour burping and difficulty in swallowing solids

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with difficulty in opening mouth since 6 months and dyspnea on exertion since 2 months which was associated with palpitations since 1 month.

Patient had seeked medical assistance 6 months back for bluish discolouration and stiffening of upper limb fingersfrom a local doctor, but she was not investigated in detail and her symptoms did not get relived (fig 1).

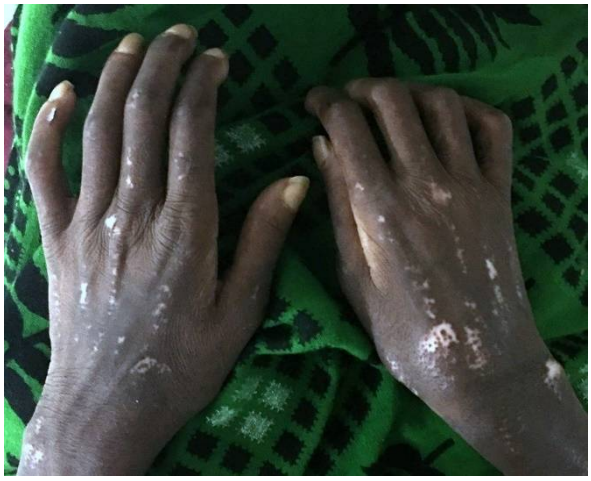

Fig 1 Sclerodactyly

Patient was tachypnic with respiratory rate of 22 per minute & had tachycardia of 110 beats per minute, regularly regular rhythm. Although she was normotensive and had 99 % O_2 saturation on room air. Jugular venous pressure was slightly elevated & hepatic jugular reflex was present.

On local examination, patient's face was mask (mauskopf) like with loss of wrinkles and expressions with small mouth aperture (microstomia) (fig 2). Skin over both hands was shiny with fixed flexion contracture at proximal interphalengeal joint with redness over nail fold. There was a localised swelling over palm with a chalky prominence which was further found to be calcinosis cutis (fig 3). Telangiectasia was present over upper torso (fig 4). On cardiac auscultation S_1 and S_2 were heard with S_3 with loud P_2. On respiratory auscultation Velpro crackles were audible bilaterally with reduced air entry on both sides.

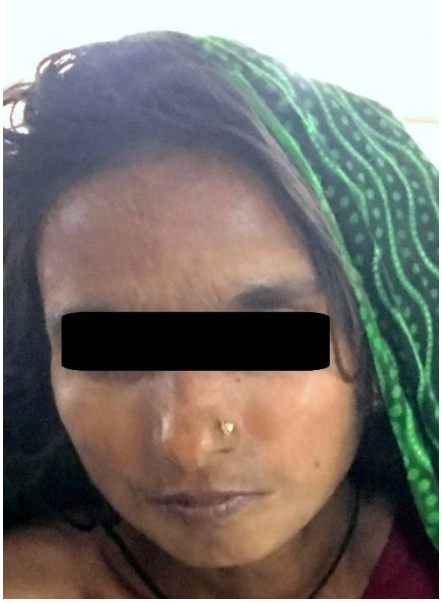

Fig 2 Mask like face with loss of wrinkles and creases

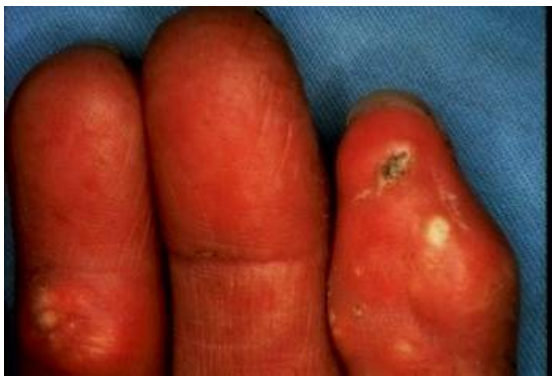

Fig 3 Calcinosis cutis

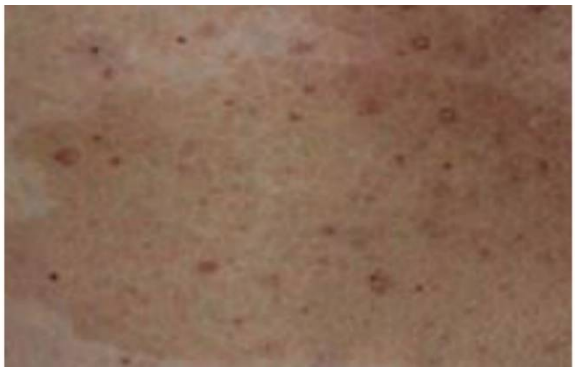

Fig 4 Telangiectasia

On investigating, her hemogram and ESR were normal. Her ECG was within normal limits. Chest roentgenography was donewhich was suggestive of enlargement of cardiac silhouette with prominent vasculature with kerley B line (fig 5).

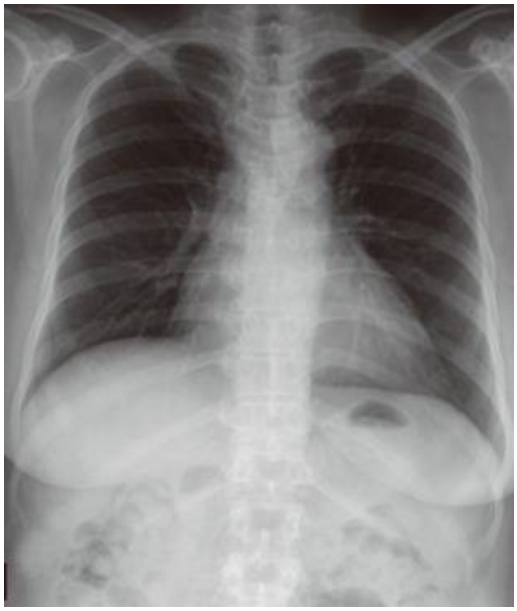

Fig 5 Suggestive of cardiomegaly.

Her BNP (Brain Natriuretic Peptide) was 422, echocardiography had shown of dilated cardiac chamberswith severe global hypokinesia with ejection fraction was 25%, with severe diastolic dysfunction, grade II Mitral regurgitation and mild pulmonary arterial hypertension suggestive of dilated cardiomyopathy (fig 6). Barrium meal was also done which was suggestive of abnormal dilatation of oesophagus (fig 7). Her capillary nailfold examination revealed distorted widened and irregular loops with areas of vascular dropouts.

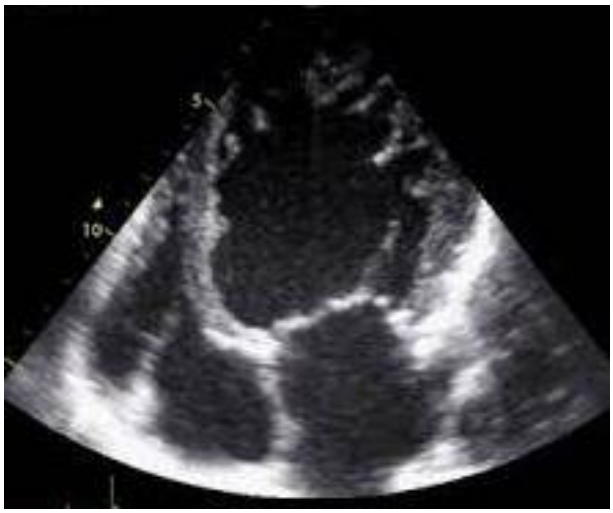

Fig 6 All the camber are dilated in 2D echo.

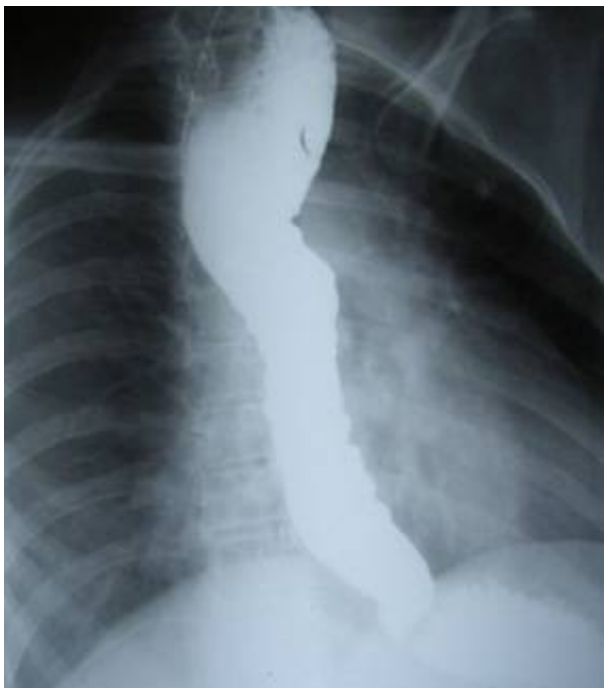

Fig 7 Dilated Oesophagus

ANA was positive with centromeric pattern and Anti-Scl70 (topoisomerase I) antibody was found positive with 2.39 titre. Hence all above clinical finding which were confirmed by investigation lead us to the diagnosis of this patient as CREST with DCMP

Patient was managed with immunosuppressants, short term steroids, antiplatelet, diuretics and other symptomatic management. Over a course of few days patient was symptomatically better and was discharged from ward.

DISCUSSION

CREST which is also known as limited scleroderma usually has an onset of approximately 30-65 years of age. In 1910, Thibierge and Weissenbach described the first case report, called as CRST (calcinosis cutis, Raynaud phenomenon, sclerodactyly, and telangiectasia). Velayos *et al* in 1979 [2] reviewed 13 patients, with CREST and CRST syndromes, which are syndromes equivalent.

The pathologic hallmark of scleroderma is progressive fibrosis of tissues. Collagen (types I, III, IV, & VII), fibronectin, glycosaminoglycans, and proteoglycans, deposited in the interstitium and in the intima of small arteries [3]. Fibrosis is found in clinically evident in affected tissues.

Some researchers suspect that other factors increase the risk to have the disease, including g*enetic factors*, s*ex* (female predominance), *race* (affects blacks more than whites) & some *environmental* factors such as exposures to, such as polyvinyl chloride, benzene, silica may trigger the disease.

In 2004, Nadashkevich *et al* proposed the classification criteria (1) autoantibodies to centromere proteins, Scl-70 (topo I) and fibrillarin; (2) bibasilar pulmonary fibrosis; (3) contractures of the digital joints or the prayer sign; (4) dermal thickening proximal to the wrists; (5) calcinosis cutis; (6) Raynaud phenomenon (at least a 2-phase color change); (7) esophageal distal hypomotility or reflux esophagitis; (8) sclerodactyly or nonpitting digital edema; and (9) telangiectasias, which can be remembered by the abbreviation ABCDCREST. Fulfilling 3 or more criteria, definitely indicatessystemic scleroderma with sensitivity as 99% and specificity as 100%[4]. In my patient, 5 criteria is fulfilling for the diagnosis of the systemic scleroderma.

CREST can be difficult to diagnose. Signs and symptoms vary widely and often resemble those of other connective tissue disorders hence clinical findings can be confirmed by investigations. Calcinosis is the pathologic calcification of soft tissues which can be subclinical. It can be tender, painful and can even ulcerate, drain a white chalky substance, when it became secondarily infected. While the entire intestine can be involved in scleroderma, esophageal involvement is most commonly found. Barrett esophagitis has been found in scleroderma patients. Esophageal adenocarcinoma, a malignant transformation of Barrett esophagitis, has also been documented in scleroderma patients. Telangiectases are lesions formed by collections of dilated blood vessels on the face, upper trunk, and hands. In ourPatient all the sign of CREST was present after properly evaluating the patient.

In the typical course of limited scleroderma, the patient first notices Raynaud phenomenon. Over the years, fingers become puffy, then the skin thickens slowly known as sclerodactyly and internal organ manifestations are delayed. Our patient also noticed features suggestive of Raynaud phenomenon as first symptoms.

Other systemic manifestations as described in literature are arthralgias, pulmonary hypertension in presence or absence of interstitial fibrosis, primary biliary cirrhosis, renal crises, entrapment neurologic syndromes, risk of malignancies and rarely myocardial involvement.[5]

The presence of cardiac involvement in SSc is often underestimated due to the occult nature. More recent studies suggest that clinical evidence of myocardial disease may be seen in 20% to 25% of patients with SSc [6]. Cardiac disease occursdue to myocardial fibrotic change or secondary to PAH. It may be asymptomatic until well developed. Patient presentation with chest pain, dyspnoea, palpitations, dizziness, and peripheral oedema and can be detected through echocardiogram monitoring. Potential cardiac problems are

due to[7,8], Microvascular coronary artery disease (resulting in myocardial ischaemia), Accelerated atherosclerosis with subsequent coronary heart disease, Myocardial fibrosis, Left ventricular (LV) systolic dysfunction, LV diastolic dysfunction, Pericarditis or pericardial effusion; these may cause cardiac impairment or congestive cardiac failure, Arrhythmias and conduction defects (including bradyarrhythmias and tachyarrhythmias), Endocarditis.

The presence of clinical cardiac involvement in SSc is a harbinger of a poor prognosis. Medsger and Masi[9] showed that clinical cardiac disease in SSc was associated with a 70% mortality at 5 years and the presence of pulmonary arterial hypertension is a poor prognostic sign with a higher mortality rate in patients with SSc. In our case report Patient had developed serious cardiac complication as dilated cardiomyopathy. The critical point to note here is that very early occurrence of such complication and its rapid course. Cardiac complication is known to occur in very late stage of the disease, but in our case, this dreaded complication occurred within 6 months of development of classical symptoms and patient was quite symptomatic also.

Unfortunately, CREST has no known cure. The condition carries both physical and psychological consequences, so holistic approachto management should be taken. Although some immunosuppressive agents like cyclophosphamide, methotrexate and mycophenolate mofetil has resulted in durable disease remission with improved skin induration but their role in prevention of development and progression of complication is not a well-studied. Antifibrotic agent like D Penicillamine has also been under trials to prevent progression. Vascular therapy for Raynaud phenomena and myocardial perfusion with ACE inhibitors and ARB's and Calcium channel blockershave also been tried and proven in improving perfusion. Our patient was treated for Congestive cardiac failure in dilated cardiomyopathy along with immunosuppressive. Our patient improved and was successfully discharged from the ward.

CONCLUSION

Hence CREST syndrome with typical symptoms (calcinosis, Raynaud's phenomenon, oesophageal dysmotility, sclerodactyly and telangiectasia) with early major organ impairment is rare presentation and patient usually not screened for the same. Our case report highlights the need for thorough clinical examination to diagnose such serious complication in very early stage, so that prompt management can be given initiated early and further fatal complication can be prevented.

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