Case Report

A Case Report: Systemic Sclerosis (Scleroderma)

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Abstract:
Scleroderma is a connective tissue disorder, mainly involving skin, blood vessel, and internal organs. Although the pathogenesis of systemic sclerosis remains obscure, it is regarded as an autoimmune disorder because of the presence of Systemic sclerosis associated autoantibodies and various immunological abnormalities. Recent progress in medicine has dramatically improved the treatment of rheumatic diseases; however, the treatment of systemic sclerosis remains disappointing. The risk of mortality in patients with systemic sclerosis is three-to five-times greater than that of an age-and sex-matched population. Therefore, diagnosis in the early disease stage, risk stratification for the development of serious organ involvement and therapeutic intervention with disease-modifying drugs can reduce the maximum degree of fibrosis, leading to improved long-term survival. Recently, new criteria for very early diagnosis of systemic sclerosis have been proposed, which are expected to be useful for regularly following up patients with very early systemic sclerosis, regardless of the absence of skin sclerosis, and for detecting the development of internal organ involvement as early as possible. Here we presented a case of scleroderma who presented with microstomia and tightening of the skin of the whole body. As scleroderma is a clinical diagnosis, higher degree of clinical suspicion is needed to reach diagnosis of this rare and complicated disease & embarking early treatment may reduce the symptoms.

Keywords: autoimmunity, fibrosis, systemic sclerosis, vasculopathy

Introduction:
Systemic Sclerosis (SSc) is a generalized connective tissue disorder characterized by micro-vascular damage and excessive fibrosis of the skin and various internal organs[1,2]. The peak age of onset is the fourth & fifth decades and overall prevalence is 10-20 per 100000 with 4:1 female preponderance[1]. The manifestations are due to diffuse deposition of collagen in the skin and internal organs along with vascular injury and immunologic abnormalities[3]. The skin becomes shiny and taut giving a mask like appearance of the face and claw like appearance of fingers. Orofacial changes are quite characteristic apart from mask like appearance of face and there will be decrease in facial profile and rigidity of tongue. Recent progress in medicine has dramatically improved the treatment of rheumatic diseases; however, the treatment of Systemic sclerosis remains disappointing[3]. The risk of mortality in patients with Systemic sclerosis is three-to five-times greater than that of an age and sex matched population[1-4]. The presence of major organ involvement, including interstitial lung disease (ILD), cardiac involvement, pulmonary arterial hypertension and scleroderma renal crisis, affects the mortality and morbidity in SSc[2,5,6]. Therefore, diagnosis in the early disease stage, risk stratification for the development of serious organ involvement and positive therapeutic intervention with disease modifying drugs will reduce disease severity and improve long-term survival.

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Case Report:
A 40-years old man presented with difficulty in opening the mouth & atrophy of the bilateral fingertips. On query he mentioned that he is suffering from this problem for the last 2 years & it is progressing. He also noticed that his skin of hands & other parts of the body becoming very tight so that he cannot sit for toilet. He also complained that his hands get color change when exposed to cold. On examination the skin of his face reveals smooth, taut, mask like appearance & microstomia. There was pulp atrophy at the fingertips associated with Raynaud's phenomenon. Dysphagia & difficulty in chest expansion was also prominent. On the basis of the history & clinical examination our provisional diagnosis was systemic sclerosis.

Investigations was done accordingly. ESR was high, ANA was positive, Punch biopsy was done from the shin area which showed atrophic epidermis, thickened collagen fibers & perivascular lymphocyte infiltrate.

Considering all the above features we come to final diagnosis of scleroderma. Although it is non-curable disease we treated the patient accordingly to reduce his symptoms.

Discussion:

Scleroderma is an autoimmune disorder involving multiple systems of the body thus making the course of this disease unpredictable. Though the correct nature of the disease is not known, high prevalence of circulating auto antibodies in serological investigation points it toward autoimmune mechanism[7]. The pathogenesis of scleroderma remains unclear but it is characterized by endothelial activation, immune system dysfunction and enhanced fibroblast activity[8]. The endothelium controls the contraction and relaxation of vascular smooth muscle cells, leading to vasospasm and smooth muscle hypertrophy. Eventually it leads to obliteration of the lumen of small arteries and capillaries which leads to ischemia. There is extravasation of inflammatory cells initially predominated by monocytic lineage and later by lymphocytes[8,9]. There is enhanced fibroblast activity which stimulates fibroblast to produce excessive extracellular matrix. Scleroderma clinically presents as two major forms as localized form and systemic form. Localized form affects the skin, without involving the internal organs[10]. There is considerable variation in both the rate of progression and clinical severity of this disease. The most prominent clinical observation is thickened, hidebound skin - especially around the fingers and hands (sclerodactyly). Other features of the disease include - Raynaud's phenomenon, telangiectasia, calcinosis, myositis, arthritis, tenosynovitis, renal failure, esophageal hypomotility, pulmonary fibrosis, and heart failure. A more localized variant of PSS is termed the CREST syndrome (Calcinosis, Raynaud's phenomenon, Esophageal dysmotility, Sclerodactyly and Telangiectasia). Early stages of disease often share clinical and laboratory features of other connective tissue diseases such as systemic lupus erythematosus and rheumatoid arthritis. Raynaud's phenomenon occurs almost universally in SSc and manifests as episodic blanching followed by cyanosis and then rubor. More or less similar frequency of Raynaud's phenomenon was noted in a north Indian study (91%), among the Afro-Caribbean population (93%), and a study from Iraq (100%)[12,13,16]. However, a much lower frequency has been reported in a south Indian study (28%) and a study from Dakar (57%)[14,15]. These discrepancies are probably due to the ethnic variation of the study population and climatic difference. Raynaud's phenomenon often predates other manifestations in the limited subtype and is often found concurrently in diffuse SSc. Our case also showed that Raynaud's phenomenon might either precede or appear simultaneously with the skin tightness. We also observed atrophy of the bilateral fingertips & there was difficulty in opening the mouth in our case. As our patient presented with very typical features consistent with scleroderma we did not do extensive investigations. Although diagnosis of this disease is primarily clinical but many often patient may present with localized presentation when diagnosis become difficult. Once skin thickening has developed, the differential diagnosis includes a wide array of other disorders in which tightening and thickening of the skin prominent features. In diabetic digital sclerosis, vinyl chloride disease, bleomycin induced scleroderma, chronic reflex sympathetic dystrophy, amyloidosis and acrodermatitis are characterized mainly by skin thickening and involvement of digits[9,10]. With the manifestation of Raynaud's phenomenon calcium channel blockers, angiotensin Type II receptor blockers, surgical sympathectomy will be the choice of treatment[10,11]. Cases with digital ulcer, skin fibrosis, arthritis, myositis, drugs like immunosuppressive, non-steroidal anti-inflammatory drugs and low dose of steroids are commonly used. In severe cases where there is renal crisis and pulmonary hypertension angiotensin-converting enzyme inhibitors are used as treatment modalities. We treated our case by calcium channel blocker, angiotensin Type II receptor blockers, non-steroidal anti-inflammatory drugs and by low dose steroid.

Conclusion:

Scleroderma is a multisystem disorder with oral, cutaneous manifestation and organ involvement. Autoantibody profile will help in identifying disease severity and organ involvement, differentiating scleroderma from other diseases. Physicians and patients should be more attentive to the potential risk factors for organ damage, particularly very early in the disease, even when the patients may not be symptomatic. Treatment should be initiated as soon as problems are identified.

References:

1. Ralston SH, Mclnnes IB. Rheumatology & Bone disease. In: Boon NA, Colledge NR, Walker BR, editors, Hunter JAA, international editor. Davidson's Principles and Practice of Medicine, 22nd ed. Edinburgh: Churchill Livingstone; 2014. p 1112-3.

2. Simeón CP, Armadans L, Fonollosa V, Solans R, Selva A, Villar M, et al. Mortality and prognostic factors in Spanish patients with systemic sclerosis. Rheumatology (Oxford). 2003; 42(1): 71–5.

3. Jacobsen S, Halberg P, Ullman S. Mortality and causes of death of 344 Danish patients with systemic sclerosis (scleroderma). Br J Rheumatol. 1998; 37(7): 750–5.

4. Abu-Shakra M, Lee P. Mortality in systemic sclerosis: a comparison with the general population. J Rheumatol. 1995; 22(11): 2100–2.

5. Bryan C, Howard Y, Brennan P, Black C, Silman A. Survival following the onset of scleroderma: results from a retrospective inception cohort study of the UK patient population. Br J Rheumatol. 1996; 35(11): 1122–6.

6. Tyndall AJ, Bannert B, Vonk M, Airò P, Cozzi F, Carreira PE, et al. Causes and risk factors for death in systemic sclerosis: a study from the EULAR Scleroderma Trials and Research (EUSTAR) database. Ann Rheum Dis. 2010; 69(10): 1809–15.

7. Ioannidis JP, Vlachoyiannopoulos PG, Haidich AB, Medsger TA Jr, Lucas M, Michet CJ, et al. Mortality in systemic sclerosis: an international meta-analysis of individual patient data. Am J Med. 2005; 118(1): 2–10.

8. Takehara K, Sato S. Localized scleroderma is an autoimmune disorder. Rheumatology (Oxford). 2005; 44(3): 274-9.

9. Yamamoto T. Scleroderma – Pathophysiology. Eur J Dermatol. 2009; 19(1): 14-24.

10. Marmary Y, Glaiss R, Pisanty S. Scleroderma: oral manifestations. Oral Surg Oral Med Oral Pathol. 1981; 52(1): 32-7.

11. Barnett AJ, Miller M, Littlejohn GO. The diagnosis and classification of scleroderma (systemic sclerosis). Postgrad Med J. 1988; 64(748): 121-5.

12. Sharma VK, Trilokraj T, Khaitan BK, Krishna SM. Profile of systemic sclerosis in a tertiary care center in North India. Indian J Dermatol Venereol Leprol. 2006; 72(6): 416–20.

13. Flower C, Nwankwo C. Systemic sclerosis in an Afro-Caribbean population: A review of demographic and clinical features. West Indian Med J. 2008; 57(2): 118-21.

14. Dia D, Dieng MT, Sy TN, Diallo M, Fall S, Ndongo S, et al. Systemic scleroderma: 92 cases in Dakar. Dakar Med. 2003; 48(2): 82–6.

15. Krishnamurthy V, Porkodi R, Ramakrishnan S, Rajendran CP, Madhavan R, Achuthan K, et al. Progressive systemic sclerosis in south India. J Assoc Physicians India. 1991; 39(3): 254–7.

16. Al-Adhadh RN, Al-Sayed TA. Clinical features of systemic sclerosis. Saudi Med J. 2001; 22(4): 333–6.