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Case Report

Juvenile Systemic Sclerosis- A Case Report

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Abstract

Juvenile systemic sclerosis (JSS) is a multisystem disorder as well as rare disease of childhood, and the amount of published data is limited. It appears that its clinical presentation differs from adult disease and the limited form affects only very few children. The organ involvement pattern differs also from the adult form. Prognosis seems to be better with a 5-yr survival of 95% of the JSS patients. Though the incidence is very rare but we describe a 10-yr-old boy who presented with typical features of JSS.

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Introduction

Progressive systemic sclerosis (PSS) is a rare connective tissue disease of unknown etiology characterized by increased collagen deposition leading to fibrosis and subsequent degeneration of the skin and internal organs. Approximately 12 new cases of PSS per million of population are diagnosed in the United States each year, with females being affected three to four times more frequently than males. There is no racial predilection. It most commonly occurs between 30 and 50 years of age.[1] PSS in children is very rare. One and one-half per cent of all patients diagnosed are younger than 10 years old, and 7% of cases occur in patients between 10 and 19 years old.[2] Childhood PSS also has a female predominance of more than 75% and usually begins before puberty.[2,3]

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. [1,4-6] A more localized variant of PSS is termed the CREST syndrome (calcinosis, Raynaud's phenomenon, esophageal dysmotility, sclerodactyly, and telangiectasia).[5]

The precise etiology of PSS is unknown; however several factors seem to play a key role. Immunologic studies suggest that the pathogenesis of the disease may be an autoimmune response directed against endothelium. Furthermore, the early stages of the disease also may be characterized by the presence of an inflammatory cell infiltrate in the dermis. Others have shown coagulation abnormalities that result in thrombosis, occlusion, and extensive ischemic tissue damage.[7, 8] Fibroblast tissue culture studies

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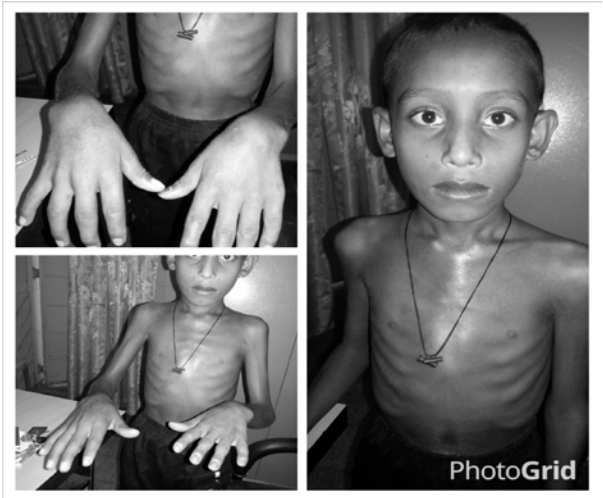
have confirmed increased secretion of collagen proteins. In vitro studies showed that increased collagen production is due to an overexpression at the transcriptional level. The fibroblasts in these patients exhibit growth abnormality.[7, 9]

Case report

A 10-yr-old boy, born of non-consangiuneous marriage, presented with complaints of pain in the multiple small and large joints of both extremities with restriction of movements for 8 months. Over same period of time he noticed skin tightening of both hands gradually involving forearm, arm, face and body as a whole. Simultaneously, patient experienced bluish discoloration of finger tips on exposure to cold (Raynauds Phenomenon) along with difficulties of swallowing both solids as well as liquids. There was no history of cough, epigastric or chest pain, dyspnoea on exertion, passage of dark colored urine and alteration of bowel habit. Family, personal and past histories were noncontributory.

Clinical examination revealed a thin built pale child showed hidebound skin in hands, forearms and trunk. The face was tense and taut with thin lips and beaking of nose. Mouth opening was restricted. Musculoskeletal system revealed generalized wasting of muscles with sclerodactyly in both hands. Flexion deformity as well as restriction of movements was in hips, knees, ankles, wrists and interphalangeal joints.



Investigations revealed normocromic normocytic anemia with high ESR (Hb% 9.30, ESR-125), CRP, creatinine and urine analysis, CXR were normal. Serology revealed RA-negative but ANA were positive, due to lack of facility Anti-centromere and anti topoisomerase was not done. Lung function was restrictive. Biopsy was not done due to noncooperation.

Discussion

Although JSS is a rare disease resembling adult progressive systemic sclerosis (PSS), has a number of distinguishing features.[12,13] Similar to adult disease, it is characterized by Raynaud's phenomenon (90-95%), diffuse skin involvement and a microvasculopathy leading to progressive dysfunction of the esophagus, lungs, heart and kidney. This patient presented with typical cutaneous features of PSS.

Pulmonary involvement, though invariable in PSS, may remain asymptomatic or may present with dyspnoea and cough. Although our patients were asymptomatic, and showed a restrictive type of lung disease on pulmonary function tests.

Esophageal dysfunction occurs in most patients, is the main cause of gastrointestinal morbidity in children with JSS although only half will complain of dysphagia, [11] our patients had dysphagia.

Bodemer et al have reported that cardiac abnormalities like pericarditis, left or biventricular failure or arrhythmias are common in pediatric systemic sclerosis, [14] but in our patient didn't have such complaint.

Renal involvement is seen in 40-60% patients in the form of malignant hypertension, proteinuria, and azotemia. It generally occurs rapidly within the first three years of PSS and indicates a guarded prognosis, [11] our patients did not have so far developed any clinical or laboratory abnormalities suggestive of renal dysfunction.

Antinuclear antibodies are reported in 70% to 90% of patients. This patient showed ANA positivity with high titre.

Our patients and his family were counseled regarding the slow progression of this disorder a supportive measures like protection from the cold, etc. The prognosis in childhood scleroderma depends on functional impairment and the course of visceral disease is difficult to predict.[15] However, Foeldvari et al have reported that most patients of juvenile systemic sclerosis show a favorable outcome and a significantly better survival rate as compared to their adult counterparts.[16] Thus the prefix 'progressive' is not uniformly applicable and should be avoided.

Conclusion

JSS is a low-prevalence disease and incidental cases are very sporadic, even in a tertiary level. Raynaud's phenomenon heralds the beginning of the disease. Capillaroscopy is a major adjuvant in the diagnosis, since autoantibody determination may not offer sensitive and specific markers. Skin and vascular manifestations are the most common clinical features, while internal organ involvement is rarer. However, cardiopulmonary disease is the most frequent visceral involvement, leading to significant morbidity, and potentially death.

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