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Juvenile Systemic Sclerosis (JSS): A Rare Case from Berhampur, Odisha, India

Authors
Dr Sunil Kumar Agarwalla[1], Dr Kali Prasad Mishra[2], Dr Nithya B.G[3] Dr Dibya Ranjan Panda[4]
[1]Associate professor, Department of Pediatrics, M.K.C.G Medical College, Berhampur, Ganjam, Odisha, 760004, India
[2,3,4]Junior Resident, Department of Pediatrics, , M.K.C.G Medical College, Berhampur, Ganjam, Odisha, 760004, India

Abstract
Juvenile systemic sclerosis (JSS) is a multisystem connective tissue disease characterized by skin fibrosis and internal organ involvement. It has a low prevalence (1/1,00,000) even in a tertiary facility setting. Reynaud phenomenon, Proximal sclerosis, sclerodactyly are the predominant skin manifestation. Respiratory involvement occurred in two thirds of patients, and it manifested as dyspnea as well as abnormal imaging and/or pulmonary function tests; Pulmonary hypertension is a poor prognostic sign. Dysphagia was the commonest gastrointestinal symptom. The most frequent musculoskeletal symptom was arthralgia. Reynaud phenomenon heralds the beginning of the disease. Capilaroscopy 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 more rare. Cardiopulmonary disease is the most frequent visceral involvement, leading to significant morbidity. Herewith we are going to present a 11 year old female child presented with JSS.
Keywords: *Juvenile systemic sclerosis (JSS), Connective tissue disorder, Capilaroscopy.*

Introduction

Juvenile scleroderma encompasses a range of conditions unified by the presence of fibrosis of the skin. Juvenile scleroderma is divided into 2 major categories, Juvenile localized scleroderma (JLS also known as morphea), limited largely to skin, and Juvenile systemic sclerosis (JSS), with multisystem organ involvement. JLS is more common in pediatric population with many sub groups (JLS: JSS =10:1), linear scleroderma is the commonest subtype. JSS have a more severe morbidity & mortality, two varieties found- diffuse and limited. Diffuse subtype is more common in children. Etiology being obscure till now, some triggering factors like trauma, infection, subclinical graft vs host reaction from persistent maternal cells, vascular endothelial cell injury are considered. Mechanism of disease appears to be combination of vasculopathy, autoimmunity, immune activation and fibrosis.(1)

Case Presentation

A 11year old female child from Rayagada district of odisha from a lower socio economic status born out of non consanguineous marriage presented to our hospital with complains of progressive

thickening of the skin, restriction of movement across all the small and large joints and dyspnoea on exertion for last 6 months. These symptoms are not associated with fever, rash, joint swelling or deformity, oral ulcer, swelling of face or legs. There is no similar attack in past. There is no history of blood transfusion, hospitalization or contact TB in past. Child was neuro developmentally normal and immunized as per National immunization schedule (NIS). The family history reveals one maternal uncle died at the age of 12 with same type of complaints. O/E child was conscious, oriented, afebrile. HR-110/min, RR-30/min, SpO2-96% in room air, BP-100/70mm Hg. There is some pallor, no icterus, clubbing, cyanosis, lymphadenopathy or edema. On head to toe examination there is generalized tightness of the skin (loss of elasticity), restriction of range of movement around all joints, fish mouth and puckering of skin of distal part of all fingers. Systemic examination reveals no abnormality.

On investigation complete blood count within normal limits, ASO, CRP, Sickling are negative, raised ESR, RFT and Electrolytes within normal limit. 2 D ECHO reveals mild PAH and TR, normal LV systolic function and normal valves. Anti Scl-70 came as 115. 2 units (>80 unit is strongly positive).

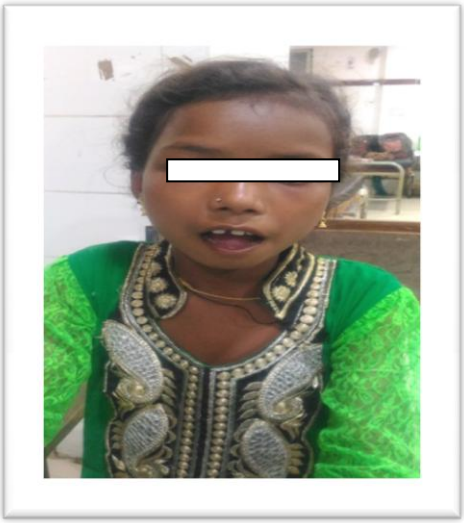

Figure-1- shows fish mouth like appearance

Figure-2 shows non pinchability of skin

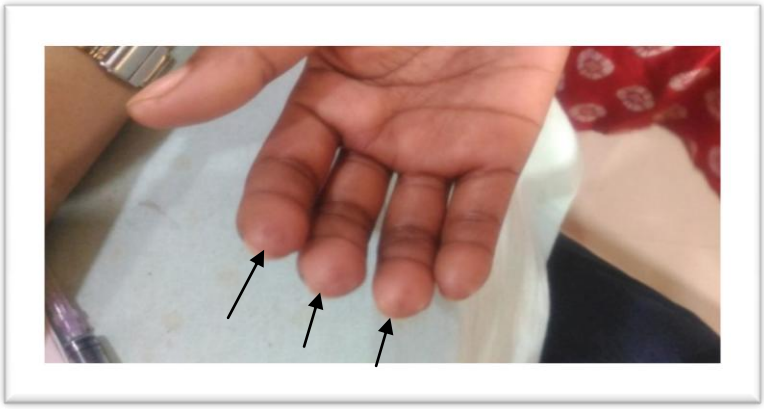

Figure-3 shows puckering of tip of fingers

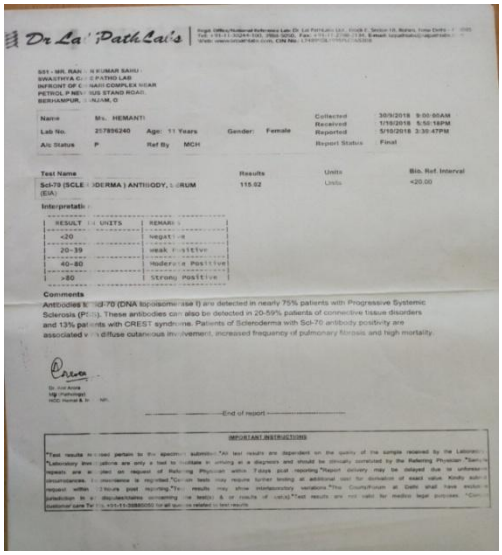

Figure-4 shows report of antiscl-70

Discussion

Juvenile systemic sclerosis (JSS) is a low prevalence autoimmune multisystem connective tissue disease of obscure etiology which is characterized by skin fibrosis and internal organ involvement[2,3]. JSS also has an insidious onset with a prolonged course charecterized by periods of remission and exacerbation, ending in either remission or, more commonly, chronic disability and death[1]. The skin manifestation of JSS include an early phase of edema that spreads proximally from the dorsum of hands and fingers & includes the face, that followed by induration and fibrosis of skin resulting in loss of subcutaneous fat, sweat gland & hair follicles. Progressively flexon contracture around joints. In face it results in decreased mouth apperture. Raynaud phenomenon, sclerodactyly, acroosteolysis are prominent cutaneous manifestation[1,2]. Cardiopulmonary disease is the most frequent visceral involvement, leading to significant morbidity and progressive cardiopulmonary complications are known to be the main causes of death in these patients[3-4]. Further, patients with JSS often suffer from gastrointestinal tract disorders that may progress to significant dysmotility causing complications like oropharyngeal dysphagia, esophageal dysphagia, gastroesophageal reflux, gastroparesis, pseudo-obstruction, bacterial overgrowth, intestinal malabsorption, constipation, diarrhea, fecal incontinence, malabsorption, weight loss and severe malnutrition[5,6]. Additionally, renal disease is also considered an important cause of morbidity and mortality in JSS. The spectrum of renal complications includes scleroderma renal crisis, normotensive renal crisis, antineutrophil cytoplasmic antibodies-associated glomerulone-phritis, penicillamine-associated renal disease, and reduced renal functional reserves manifested by proteinuria, microalbuminuria, or isolated reduction in glomerular filtration rate[7]. According the newly proposed pediatric criteria [8] the patient has to fulfill one major and two minor criteria, with an age of onset of the disease before the age of 16 years. The major criterion is proximal skin sclerosis/induration of the skin. The minor criterion can be a defined scleroderma specific organ involvement of the involved organs, listed in the classification, and specific serologic findings, defined in the classification paper too. This criterion is prospectively not validated yet, but hopefully it will enable to make the diagnosis of JSS earlier in the disease course and create a larger clearly defined patient population. An advantage of the JSS criteria compared to the ACR criteria is, that not only pulmonary involvement as an internal organ involvement, can define the disease, but other scleroderma specific organ involvements or antibody profile can be used for the classification purposes.

Patients with JSS may have anemia, leukocytosis, eosinophilia & autoantibodies (ANA & anti scl 70) positive. High resolution CT, pulmonary function test, echocardiography & manometry are useful tools for diagnosing & monitoring visceral involvement in JSS. Treatment for JSS target specific disease manifestation. Cold avoidance, calcium channel blockers(nifedipine,amlodipine), losartan, prazosine , bosentan, sildenafil are used for raynaud phenomenon. ACE inhibitors are indicated for hypertension associated with renal diseases. Methotrexate or Mycophenolate mofetil may be benificial for skin manifestation. Cyclophosphamide & mycophenolate mofetil are used to treat pulmonary alviolitis & prevent fibrosis. Use of high dose cyclophosphamide , anti thymocyte globulin(ATG) & autologous stem cell transplantation gives good prognostic results in majority of patients[1]. In our case diagnosis was made by clinical examination, classification criteria & positive anti scl 70. The CBC, electrolytes, RFT came normal. 2D echo reveals mild PAH and TR. She was given Tab Cyclophosphamide 50mg OD for 6 months & Nifedipine 10mg OD for 6 months and adviced to come for follow up.

Conclusion

Out of all rheumatological disorders in children JIA, SLE tops the list. But any child coming with

hardness of skin particularly acral end of extremities, trismus, chewing difficulty, tiredness, one has to keep juvenile systemic sclerosis in mind. Mostly it is a clinical entity but will be confirmed by anti scl 70 assey. Early diagnosis and early intervention can have a prolonged quality of life. Such rarity of this case prompted us to go for publication.

Conflict of interest: No.

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