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Rheumatology

CHILDHOOD ONSET CREST SYNDROME WITH INTERSTITIAL LUNG DISEASE – A RARE CASE REPORT

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ABSTRACT CREST syndrome also known as limited cutaneous systemic sclerosis is a subdivision of systemic sclerosis whose acronym stands for calcinosis , Raynaud's phenomenon , esophageal motility dysfunction, sclerodactyly and telangiectasia .Systemic sclerosis and its subtypes can affect anyone irrespective of age and sex but it has rare occurrence of incidence among pediatric population predominantly affecting female gender (1).Here we report a 10 year old girl with early onset of limited cutaneous systemic sclerosis with strong positivity for anti centeromere antibody. The child presented to outpatient department with complaints of stiffness of joints and tightening of skin . Further examination revealed facial feature suggestive of mouskopf facies , pinched nose , healed palatal ulcer, examination of joints revealed multiple tender joint swellings (figure 1).Systemic examination was normal. Complete blood counts, Renal function and Liver function tests were normal with elevated Erythrocyte sedimentation rate (ESR). Radiographic images of hip joint showed calcium deposits in pubic rami (figure 2). Auto-immune work up was done. ANA profile showed anti centeromere antibody positivity (3 +). On subsequent follow up visit child had complaints of breathing difficulty . Clinical examination revealed fine inspiratory crepitations which provoked the suspicion of Interstitial Lung Disease (ILD) since the child had background of CREST syndrome. Pulmonary Function Test was performed which showed restrictive pattern. Computer tomography of chest was suggestive of Usual Interstitial Lung pattern (UIP) (Figure 3).Patient was started on pulse steroid therapy which was gradually tapered to oral steroids and started on mycophenolate mofetil (MMF) . Child's condition symptomatically improved remission was achieved and is on regular follow up .

KEYWORDS :

INTRODUCTION

Systemic sclerosis is rare connective tissue disorder with complex and unknown pathology.It can be further divided into diffuse cutaneous systemic sclerosis and limited cutaneous systemic sclerosis. Limited cutaneous systemic sclerosis is associated with skin tightening distal to elbow and knees , where as diffuse cutaneous systemic sclerosis is associated with skin tightening proximal to elbow and knees.

Both diffuse and limited cutaneous systemic sclerosis can have multisystem involvement affecting various organ including heart , lungs , kidney and gastro intestinal system. However other system involvement such lung and heart is rare (3). They also have autoantibody positivity – ANA can be positive in 90 % of the cases . anti centeromere , anti-scl 70 and anti RNA polymerase being more specific for systemic sclerosis. There is always a delay in the diagnosis of juvenile systemic sclerosis as well as offers diverse range of treatment modality due its rarity.(6)

The hallmark histopathology change in Ssc is non-inflammatory proliferative/obliterative vasculopathy followed by interstitial/vascular fibrosis. Fibrosis is the predominant pathological finding of systemic sclerosis which can be seen in other organs including salivary gland, thyroid gland , lungs and gastro intestinal system.

Pulmonary involvement in SSc is common. Pulmonary involvement usually associated with inflammatory changes in early stage of the disease , fibrosis and vascular damage occurring later. The pulmonary changes in SSc will be in the form of non specific interstitial pneumonia. Pulmonary artery hypertension occurs due to vasculopathy.

CASE REPORT:

10 Year old single child born out of non consanguineous marriage presented to OPD with complaints joint pain and skin tightening. She also complained of multiple swelling over small joints causing restriction of movements. Local examination of joints revealed multiple hard swelling over the small joints of bilateral hand and elbow joint. Head to toe examination – child had mauskopf facies , healed palatal ulcers , skin tightening distal to elbow joint and knee joint . Systemic examination was normal. Baseline investigation including CBC (complete blood count) and renal function test (RFT) were normal however ESR (erythrocyte sedimentation rate) was elevated .Radiological imaging of joint showed calcium deposits. So in high suspicion of limited cutaneous SSc automune work up – ANA by immunoblotting was done which showed strong positivity for ANA and anti centreomere antibody. A diagnosis of limited cutaneous SSc was done. The child was started on injection methylprednisolone 500 mg intravenous once daily for 3 days following which remission was achieved. Child was started on oral steroids and mycophenolate mofetil after pulse therapy. After 1.5 years during subsequent follow up visit child complained of breathing difficulty on exertion . Examination of respiratory system revealed new onset fine inspiratory crepitations. So to rule out pulmonary involvement PFT (pulmonary function test) was done which showed restrictive pattern suggestive of ILD. Following which a CT CHEST was done to confirm ILD which revealed multiple clusters of thin walled cyst formation with interspersed ground glassing opacity and interlobar septal thickening (reticulation) noted peripherally arranged in superior and posterior basal segment of both lower lobes. Similar changes in apical and anterior segment of both upper lobe noted. The child was started on pulse steroid therapy for 3 days later gradually tapered to oral steroid and mycophenolate mofetil was initiated. Remission was achieved.

DISCUSSION :

Juvenile onset of systemic sclerosis has an extremely rare incidence based on epidemiological study and incidence of case reports being very less across the world(1), and has a female preponderance (2) . the amount of data published on juvenile systemic sclerosis is limited. Presentation and organ involvement pattern differs when compared to adult population. Hence many questions arise about the evaluation of the juvenile systemic sclerosis , it progression and treatment.

It is very important to follow up with patients even though limited form is more common there is a higher chance of the disease progressing with other system involvement. Hence follow-up will ensure in early identification of disease progression which helps in early intervention which inturn will improve the quality of life.

It still remains a big hurdle for the physician in treating Juvenile

systemic sclerosis. In our case the child was treated with pulse steroids during flare up and was maintained with oral steroids and mmf during remission after consulting with rheumatologist . DMARDS are used widespread but no study has been proven beneficial. Some studies have showed biologicals to be helpful especially disease which progress rapidly with baseline treatment strategy(5). Although studies focusing on treating juvenile SSc with MMF is limited , MMF has proven effective in treating adult population with interstitial lung disease .(3)

CONCLUSION :

Calcinosis cutis commonly occurs in patients with systemic sclerosis, especially the limited form (CREST). Twenty-five percent to 40% of patients with limited systemic sclerosis will develop calcinosis cutis ten years after the onset of disease. Control of Juvenile systemic sclerosis is of utmost priority as it can have a negative impact on growth , development and scholastic activity of the child as well as physically disabling.

Management approach for juvenile systemic sclerosis should be tailored made considering the possible adverse effect of medication considering the presentation of disease , organ involvement and adverse effect of medication. In this case 10 year old who initially presented as calcinosis cutis -complete work revealed juvenile systemic sclerosis. The incidence of juvenile systemic sclerosis is 0.05 per 100,000 children which makes a rare presentation. Methotrexate helps in management in skin sore related to diffuse sclerosis(6).Digital vasculopathy being a common clinical presentation of systemic sclerosis -anti platelet therapy as platlet aggregation and activation is increased in raynauds phenomenon but 2 studies have concluded use of anti platelet have no significant benefit (7). The child was started on steroids during flare up once remission was achieved mmf was started . Tolerance for MMF was observed patient general condition improved and patient was discharged after achieving remission. Patient is currently under regular follow up.

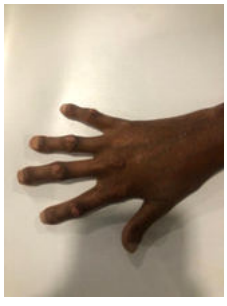

Figure 1 : deposistion of calcium over joint spaces

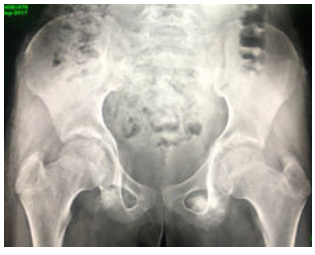

Figure 2: calcium depoisits in bilateral pubic rami

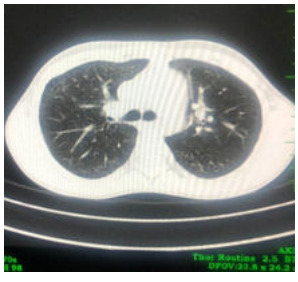

Figure 3 : CT showing reticular pattern over bilateral lung field.

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