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Prospective Cross-Sectional Clinical Study of Autoimmune Connective Tissue Diseases in Pediatric Age Group in Tertiary Care Hospital

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Abstract: ***Introduction: Autoimmune Connective Tissue Diseases(AICTD) may occur during childhood, each with variable clinical presentations; amongst the more common are lupus erythematosus (LE), morphea and juvenile dermatomyositis (JDM).[(1)]Juvenile onset systemic sclerosis(SSc) and mixed connective tissue disease(MCTD) are rare. Aim and objective: To assess clinical presentation, course of disease and complications of paediatric onset autoimmune connective tissue diseases. Material and method: Prospective, cross-sectional study of 36 paediatric patients diagnosed as autoimmune connective tissue disease from Jan-2011 to Aug-2019 was done. Results: Out of total 36 patients, 22 patients had lupus erythematosus, 9 patients had morphea, 3 patients had juvenile onset systemic sclerosis, 1 patient had mixed connective tissue disease and 1 had juvenile dermatomyositis. Conclusion: Autoimmune connective tissue diseases are not uncommon in paediatric age group and presenting complaints and diagnostic methods may differ in paediatric age group.***

Keywords: Pediatric onset autoimmune connective tissue disorders

1. Introduction

Autoimmune Connective Tissue Diseases(AICTD) may occur during childhood, each with variable clinical presentations; amongst the more common are lupus erythematosus (LE), morphea, and juvenile dermatomyositis (JDM).[(1)]Juvenile onset systemic sclerosis (SSc) and mixed connective tissue disease(MCTD) are rare.

Pediatric onset of these disorders carries a diagnostic dilemma for the clinicians. Dermatologists have the unique opportunity to diagnose these autoimmune disorders based on cutaneous findings which may predate systemic symptoms.

Aim and Objective

To analyse the clinical presentation, course of disease and complications of paediatric onset AICTD

2. Material and Method

Prospective cross-sectional study from Jan-2011 to Aug-2019 of 36pediatric patients (below 18 years of age) presentingwith autoimmune connective tissue disease was done. Detailed history was taken and thorough examination was done for all the patients. All routine investigations including ANA titre and ANA profile, histopathologic examination and ultrasonographic examination were noted. Special investigations like pulmonary function test, 24 hour urinary protein and radiological scans etc were performed as and when required and recorded to rule out systemic association.

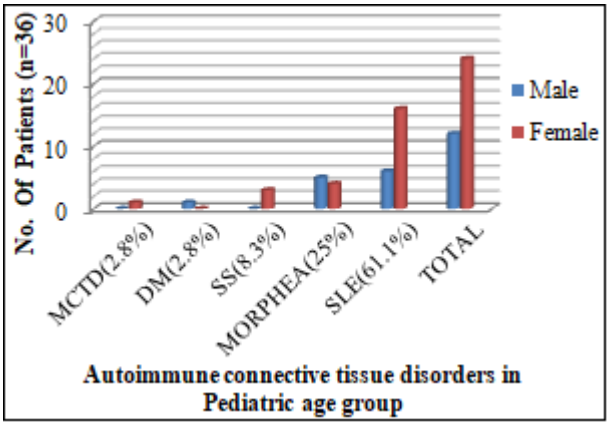

Chart 1: Autoimmune connective tissue disorders in pediatric age group

3. Results

Out of 36 total patients, 12 were male and 24 were female (M:F =1:2). Age ranged from 1.5 year to 17 year. Out of 36 patients of AICTD, 22 patients LE (61.1%), 9 patients had morphea (25%), 3 patients hadSSc (8.3%), 1 patient had MCTD (2.8%) and 1 JDM (2.8%).

In LE patients (n=22) common age-group was 11-17 years (M:F =1:2.66). Youngest age incidence was seen in one male patient presenting at 18 months of age with joint pain,fever,maculopapular lesions , global milestone delay and positive IgG Ab to DsDNA , Ro and Sm/RNP autoantibodies s/o subacute cutaneous lupus

erythematosus(SCLE).12 patients presented with malar rash and 2 had Raynaud's phenomenon at the time of presentation.Renal involvement was seen in 6 patients of LE (27.3%).Out of 22 only 2 (9.1%) had neuropsychiatric illness.Most common type was acute cutaneous LE (59.1%) while 3 patients had subcutaneous LE (13.6%) and 6 had chronic cutaneous LE(27.3%) while 1 of them had biopsy proven diagnosis of bullous LE. ANA titre and profile were consistent with LE in 18 patients (81.81%).

Figure 1, 2: A 18 month old patientof SCLE with maculopapular lesions with oral ulceration.

In morphea (n=9), common age group was 4-18 years,with slight male predominance. Linear morphea was most common type.Out of 9 pateints of morphea 3 patients had head and neck involvement ,one had Parry Romberg syndrome and one had En coup de sabre. None had any ocular or dental complications. 2 patients of linear morphea had contracture as a complication.one patient had generalised morphea.

Figure 3: A 13 year old female patient of en coup de sabre, figure 4: A 17 years old female patient of parry Romberg syndrome

Three Patients of SSc were from 16-18 years of age group and all 3 were female patients. Two had proximal skin induration, sclerodactyly, digital tip ulcers, salt and paper pigmentation, internal organ involvement suggested by abnormal pulmonary function test and changes on CECT. One patient had CREST syndrome (calcinosis, Raynaud's phenomenon, esophageal dysmotility, sclerodactyly and telangiectasia). All patients had positive ANA titre with positiveScl-70,RNA,Ss-a/Ro52 autoantibodies in two of them.

JDM was seen in 17year old male who presented with heliotrope rash,gottron papules, calcinosis cutis and periungual changes along with positive ANA titre and profile. No muscle involvement was found.MCTD was seen in 15 year old female. Patient presented with raynauds phenomenon, malar rash, muscle weakness, diffuse hairloss, pancytopenia, pulmonary involvement and positive anti U1 RNP.

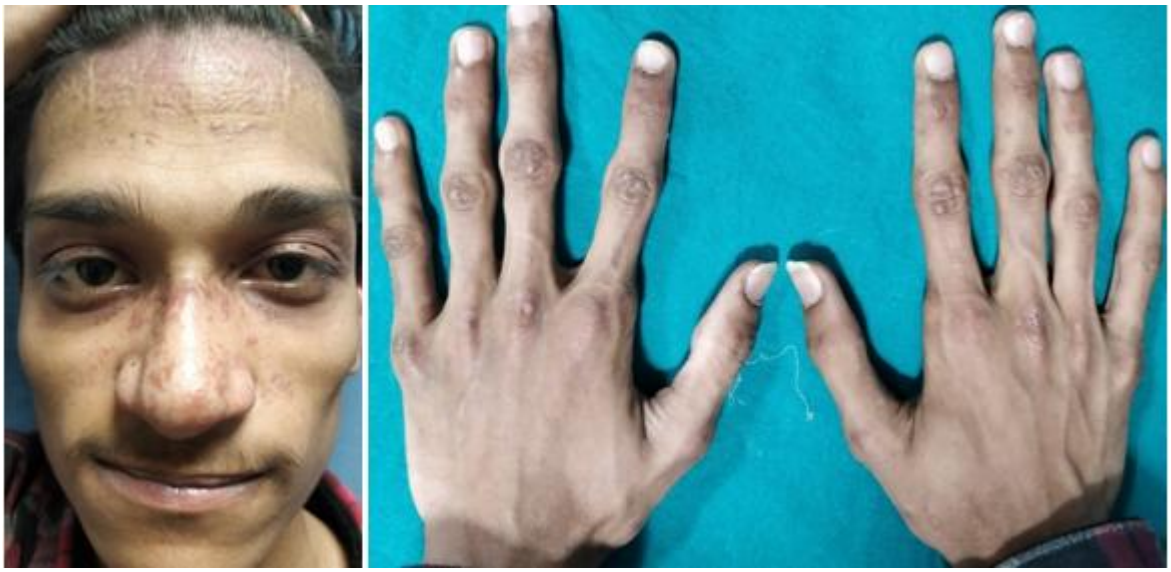

Figure 5, 6: A 17 year old patient of JDM with heliotrope rash and gottron papules

Out of total 36 patients, 4 patients (3 of LE and 1 of SS) died during the course of the disease during study period. All other patients are under regular follow up and have short term remission withon and off exacerbation. Male patients with LE have particularly poor response to therapy and relentless progression of disease requiring more number of hospital admissions.

4. Discussion

LE is rare in paediatric age group, with an incidence of 10–20 in 100,000 Caucasian children [2]with a male:female ratio of 1:4.5 between the ages of 6 and 18 years.[3] Also in our study, it is 3.7 times more common in female than male.SLE can occur at any age, although it is rare before five years of age. Here we report one case which presented at 18 months of age which is very rare.Central nervous system manifestations are a major cause of morbidity and mortality in juvenile SLE.[4]In childhood, SCLE is exceedingly rare, with only 15 cases of childhood-onset SCLE reported to date. Compared with the adult SCLE population, children had a higher rate of progression to SLE (83 %) and were more likely to have lesions below the waist than adults with SCLE [5].

Juvenile onset systemic sclerosis (SSc) represents a minorityof cases of systemic sclerosis in children as morphea (localized form of the systemic sclerosis) occurs at least 10 times more commonly than systemic sclerosis in children.[6]childhood -onset scleroderma is rare. Less than 3% of all cases are childhood onset.[7]In juvenile onset SSc, the limited cutaneous variant is exceedingly rare, contrasting with its predominance among adults with SSc. The term lcSSc is preferable to CREST syndrome (calcinosis, Raynaud phenomenon,oesophageal motility disorders, sclerodactyly and telangiectasis).[8] Here we reported one case of CREST syndrome in pediatric age group.The gastrointestinal tract is the most commonly involved internal organ in juvenile onset SSc, as in adult disease.Pulmonary disease is now the major cause of death in SSc.Renal disease, although no longer the major cause of death in systemic sclerosis but it is an important complication. In our study, pulmonary involvement was seen in all 3 patient and 1 patient died due to cardio- respiratory failure within 2 years of disease presentation.

Morphea is a connective tissue disorder of unknown aetiology. The mean age of onset in children is between 7 and 10 years.As with many other connective tissue diseases morphoea predominantly affects females, with a female : male ratio of 2 – 3 : 1 [9]. In childhood, linear variant is the most common subtype.[10] In our study also most common type presented was linear morphea. Linear morphoea of the head is termed ‘en coup de sabre’.Parry – Romberg syndrome is characterized by hemifacialatrophy, mainly affecting the subcutaneous tissue, muscles and bones. We have reported 1 case of Parry-Romberg syndrome in female patient and 1 case of en coup de sabre in male patient.

Juvenile dermatomyositis (JDM) is usually considered in the differential diagnosis either when erythematous rashes arise on the face or extremities or when acquired symmetrical muscle weakness is present.[11]The rash can precede or follow muscle weakness. Gastrointestinal, cardioresoiratory, genitourinary and neurological involvement can be seen in children with JDM

Mixed connective tissue disease (MCTD) [12], describes the clinical situation in which symptoms of a number of connective tissue diseases (CTDs) occur in one patient. There may be presence of anti-U1 -ribonucleoprotein (anti-U1 -RNP) in Sharp’s original description of MCTD in 1972 [13]. The condition can occur in childhood and may be more severe in children.In our study we reported one case of MCTD.It is unpredictable to give any prognostic ideato parents as autoimmune connective tissue diseases escape control at any time or new symptom or new organ involvement may be seen at any time.

5. Conclusion

This study suggest that autoimmune connective tissue diseases are not uncommon in paediatric age group and presenting complaints and diagnostic methods may differ in paediatric age group.So its detailed knowledge can aid dermatologist in prompt diagnosis and early intervention to prevent permanent complications and sequelae in paediatric age group.Clinical Studies focusing on paediatric onset autoimmune connective tissue disease are scanty in literature.We hope to curb the disease at the earliest in all pediatric cases to make their childhood suffering free.

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