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Case Report

Early onset chilblain lupus erythematosus with atypical presentation in an Indian girl

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ABSTRACT

Chilblain lupus erythematosus (Chilblain LE) is a rare form of lupus erythematosus with only 70 cases reported worldwide with most of the patients in middle age and very few cases in adolescent age group. Herein, we are reporting a 16 years old girl presented with an early age of onset at 10 years with atypical presentation who fulfilled Mayo clinic diagnostic criteria for Chilblain LE.

Keywords: Chilblain lupus, Atypical, Early onset

INTRODUCTION

Chilblain lupus erythematosus (Chilblain LE) is a chronic form of lupus erythematosus (LE) reported mostly in middle-aged female patients. It may either precede the development of systemic lupus erythematosus (SLE) or occur after the diagnosis of SLE is established. It can be an isolated form also.[1] The study was done to present a case of chilblain LE in an Indian adolescent girl who had early onset and atypical presentation.

CASE REPORT

A 16 years old school going girl presented to us in the department of dermatology with complaints of recurrent red elevated lesions over palms and soles which initially appeared during winters and subsequently persisted throughout the year for five to six years with aggravation during winters, associated with mild itching and pain on and off. History of self resolution of some lesions in summer was there. She also gave history of erythema over nose and cheeks during winters without any raised lesion or other symptoms. She also complained of diffuse hair loss. There was no history of photosensitivity, joint pains, oral ulcers, other systemic symptoms, renal complaints or frothing of urine. There was no related positive family history.

On examination, well to ill defined erythematous non tender plaques of size ranging from 5 mm to 2 cm were seen bilaterally over palms. The lesions were blanchable on pressure. No lesion in any other part of the body was noted at the time of presentation (Figure 1).

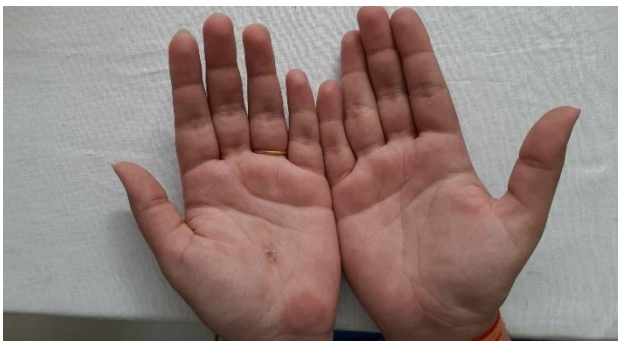

Figure 1: Well to ill defined erythematous plaques over palms.

Due to the long duration of recurrent and persisting lesions, a clinical possibility of chilblain lupus erythematosus was kept and further investigations done. Antinuclear antibody titre and Rheumatoid arthritis factor were highly positive with values of 182.34 units and 231 IU/ml respectively. Erythrocyte sedimentation rate was 80mm. Cryoglobulins or cold agglutinins were absent. Other routine blood and urine investigations were within normal range. Punch biopsy was taken from a lesion over the left palmar region. Histopathological examination with Hematoxylin and Eosin (10X magnification) stain showed interface pathology, basal layer vacuolization, occasional apoptotic keratinocyte, upper to deep dermal perivascular lymphocytic infiltrate. Mild edema and few extravasated erythrocytes were observed in papillary dermis. PAS stain showed mildly thickened basement membrane. Alcian blue stain showed mild interstitial mucin deposition in reticular dermis (Figure 2 and 3).

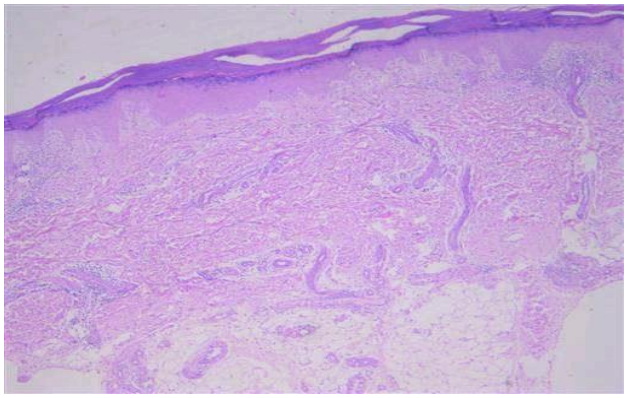

Figure 2: Histopathology (H & E stain, 10X) showing interface pathology, basal layer vacuolization, dermal perivascular lymphocytic infiltrate, thickened basement membrane.

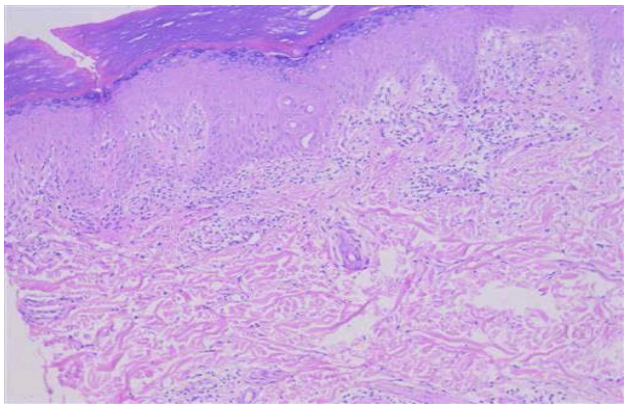

Figure 3: Magnified view (40X) of histopathology.

A diagnosis of chilblain LE was made with clinical examination and laboratory investigations. She was started on potent topical steroid (clobetasol propionate 0.05% cream) twice a day along with pentoxyphylline and hydroxychloroquine and was advised cold protection. After two months of therapy, she showed good improvement and is still under regular follow up.

DISCUSSION

Chilblain lupus, a rare cutaneous form of systemic lupus erythematosus, was first described by Jonathan Hutchinson in 1888 and was alternatively named after him as “Hutchinson lupus”.[2] It is derived from the Anglo-Saxon terms “chill” and “blegen”, a synonym for “sore”.

Two forms of Chilblain LE are described namely familial and sporadic. Familial form of chilblain lupus manifests in early childhood and is caused by a heterozygous mutation in the TREX1 gene that encodes a 3’-5’ DNA exonuclease.[3] The pathogenesis of sporadic chilblain LE remains unknown, though vasoconstriction or microvascular injury secondary to cold and possible hyperviscosity and stasis due to immunological abnormalities are usually implicated. The rheological phenomena discussed above may be aggravated by immunological anomalies. Sporadic Chilblain LE usually affects middle-aged females whilst familial chilblain LE manifests in early childhood. Generally, lesions occur at first during cold or damp periods and do not remit completely. Usually, they are located on the dorsum of the hands and fingers.

A set of diagnostic criteria have been proposed by the Mayo Clinic after studying a small number of patients with chilblain lupus erythematosus. This includes two major criteria and four minor criteria. Major criteria include acral skin lesions associated with cold temperature and evidence of lupus erythematosus in skin lesions on histopathology or direct immunofluorescence. Minor criteria include coexistence of systemic lupus erythematosus or other skin lesion of discoid lupus erythematosus or response to antilupus erythematosus therapy or negative cryoglobulin/cold agglutinin studies. For diagnosis, 2 major and 1 minor criteria needed.[4]

Patients may also display hypergammaglobulinaemia (over 2/3), positive rheumatoid factor (in half), antinuclear antibody, antiphospholipid or anti-Ro antibodies. They are usually negative for antidouble-stranded DNA antibodies.[5]

Most patients respond well to symptomatic therapy and cold protection. Antimalarial agents like chloroquine or hydroxychloroquine are useful. Many cases respond well to topical steroids. Secondary infections can be treated with antibiotics. In recalcitrant cases, local immunosuppressive agents like tacrolimus or pimecrolimus may be an option. Other drugs reported to be useful in the past include pentoxyphylline, etretinate, and gold. Boehm et al reported successful treatment with mycophenolate mofetil (MMF), which may be considered if other treatment regimens are insufficient.[6] A therapeutic study conducted in dermatology department of Al-Yarmouk Teaching Hospital, Baghdad, Iraq during four winter seasons between 2010 and 2014 concluded

that pentoxyphylline is an effective and safe drug for treatment of primary perniosis.[7]

A study which compared idiopathic chilblain patients and patients with chilblains associated with LE concluded that feminine sex and persistence of lesions beyond the cold season were significantly related to chilblain LE.[8] Chilblain LE may be associated with lesions of discoid LE or other forms of cutaneous LE and may progress to SLE in up to 18% of patients. Hence, it is critical to differentiate it from simple chilblains whenever there is any suspicion.

CONCLUSION

In our case, age of onset of lesions was around 10 years which is very earlier as chilblain LE is reported in around 70 cases till 2008 according to a review of literature and it was more common in middle aged with very few cases in adolescent group.[5] The lesions usually occur over the dorsum of hands whereas our patient had over ventral aspect. She fulfilled two major and one minor criteria of Mayo clinic diagnostic criteria. Hence, we are reporting for the rarity of the condition and also for the atypical presentation in our case.

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