Chilblain lupus erythematosus in an adolescent girl

Shikha Bansal, Alka Goel[1]

Department of Dermatology Venereology and Leprology, Safdarjung Hospital, [1]Skin and Laser Clinic, Hari Nagar, New Delhi, India

ABSTRACT

Chilblain Lupus Erythematosus (CHLE) is a rare form of cutaneous lupus erythematosus (LE), more frequently seen in middle aged females. It is characterized by erythematous to violaceous plaques over the acral areas induced by exposure to cold or drop in temperature unlike lesions of lupus erythematosus that worsen with sun exposure. Here, we present a case of chilblain lupus erythematosus in an adolescent girl with few unique features not previously reported.

Key words: Adolescent, anti nuclear antibody, chilblain lupus erythematosus

INTRODUCTION

Chilblain lupus erythematosus is a chronic form of lupus erythematosus seen especially in women, affecting the fingertips, rims of ears, calves, and heels. CHLE lesions may be clinically indistinguishable from simple chilblains or perniosis, which is seen in healthy individuals. Hence, having an understanding of this clinical entity is essential for the clinicians to make a correct diagnosis. Here, we present a case of CHLE in an adolescent girl with few unique features not previously reported.

CASE REPORT

A 13-year-old girl presented with reddish raised asymptomatic lesions over both hands and feet for the past five months. The lesions appeared spontaneously as lentil-sized raised lesions over the right index finger and gradually increased in size and number and coalesced to form larger plaques with appearance of similar lesions over other fingers as well as soles. No personal or family history of oral ulcers, joint pains, photosensitivity, frothing of urine or any systemic complaints in the form of chest pain, abdominal pain, or breathlessness was found. Examination of hands and feet revealed multiple papular lesions, which were erythematous and edematous. They were discoid erythematous scaly atrophic papules to plaques over the ventral and lateral aspect of the fingers of hand and feet as well [Figure 1]. The lesions over the palms and soles were tender to touch. Dorsal aspect of hand had multiple hyperpigmented macules suggestive of postinflammatory hyperpigmentation. Bilateral concha showed violaceous discoid plaques of size 1 × 1 cm [Figure 2]. Nails of both the hands showed ragged cuticle. There was no significant lymphadenopathy and all mucosae were normal.

Hematological investigations were within normal limits except anemia (Hb-10.9g/dL) and raised erythrocyte sedimentation rate (30 mm in first hour). Biochemical tests for hepatic and renal functions, serum proteins, urine examination and chest radiograph were normal. VDRL was non-reactive. Rheumatoid factor was negative. Anti-nuclear antibody test was positive with a speckled pattern of immunofluorescence, and the titer being 1:160. These antibodies were identified as anti-dsDNA, anti-Sm, anti-Ro52, anti-histone and anti-nucleosomal antibodies. Histopathology from the lesion over the finger showed focal basal cell vacuolization of the epidermis and moderate perivascular and periadnexal lymphocytic infiltrate in the dermis [Figure 3]. Direct immunofluorescence (DIF) of lesional skin showed IgM and C3 deposition along the basement membrane zone. Based on the clinical and laboratory findings a diagnosis of CHLE was made and treatment was initiated with potent topical steroids twice daily until complete remission.

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Address for correspondence:
Dr. Shikha Bansal, 962, Sector A, Pocket B and C, Vasant Kunj, Delhi - 110 070, India.
E-mail: contactshikha@rediffmail.com

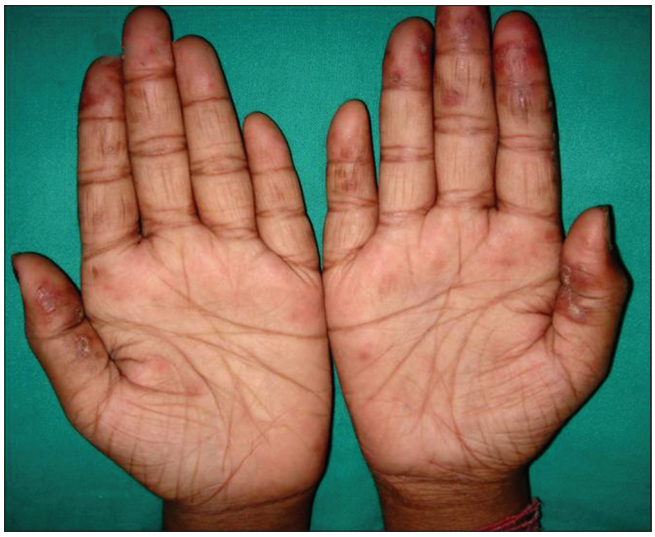

Figure 1: Chilblain lupus erythematosus lesions over the ventral aspect of fingers and palms

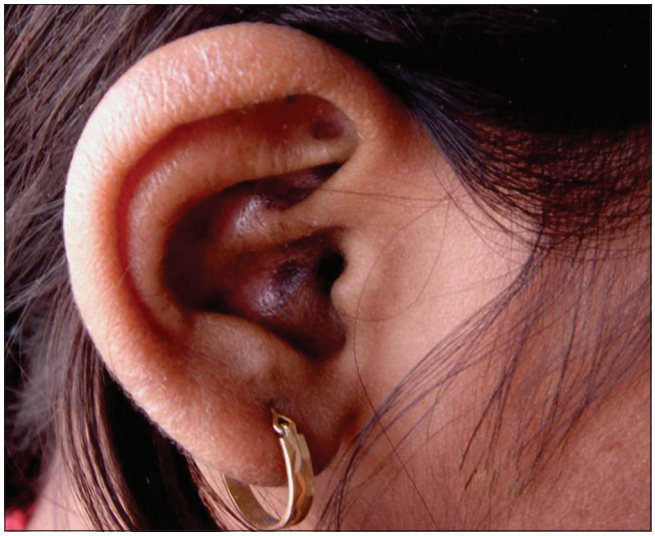

Figure 2: Typical discoid lupus erythematosus lesions over the concha

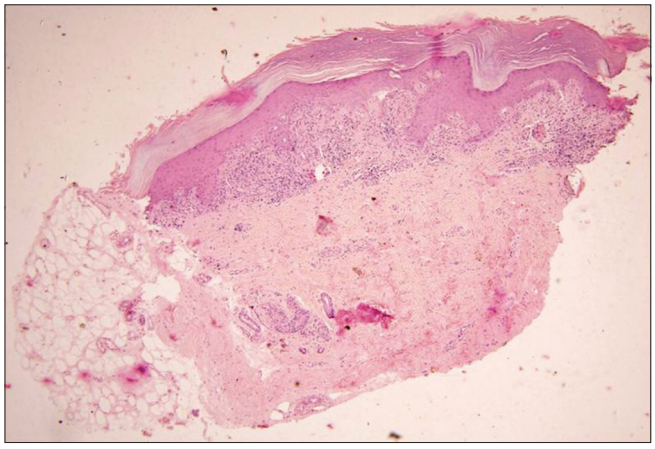

Figure 3: Histopathology showing vacuolization of the basal cell layer and a perivascular and periadnexal lymphocytic infiltrate (H and E, ×10)

DISCUSSION

Chilblain lupus erythematosus is a rare manifestation of chronic cutaneous lupus erythematosus, first reported by Hutchinson in 1888[1]. According to a review published in 2008[2], about 70 cases have been reported in the literature, with only few in the adolescent age group. The two forms of CHLE are described - 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 CHLE remains unknown, though vasoconstriction or microvascular injury secondary to cold and possible hyperviscosity and stasis due to immunological abnormalities are usually implicated. An association of CHLE with anorexia and intestinal lymphoma has also been reported but not conclusively proven. Pregnancy induced chilblain lupus has been described in literature.[4]

CHLE is clinically characterized by symmetrically distributed circumscribed, occasionally infiltrated erythematous to violaceous pruriginous or painful plaques over dorsal and lateral aspects of hands and feet, appearing during cold, damp weather conditions. Involvement of ears, nose or trunk is uncommon. CHLE may be associated with lesions of discoid lupus erythematosus (DLE) or other forms of cutaneous lupus erythematosus and may progress to systemic lupus erythematosus (SLE) in up to 18% of patients. It can clinically simulate lupus pernio, however, it is essential to make a correct diagnosis; although clinically the two entities are similar and have in common their association with systemic disease (lupus erythematosus or sarcoidosis), the prognosis and treatment differ considerably.[5] Verrucous form of chilblain lupus has also been described in literature.[6] In most patients, immunological abnormalities are frequently observed in the form of polyclonal hypergammaglobulinemia, raised rheumatoid factor, antinuclear antibodies. Anti-Ro/SsA and antiphospholipid antibodies have also been found to be positive in a few studies.[7] Su *et al.,*[8] proposed the Mayo Clinic Diagnostic criteria for a confirmative diagnosis of CHLE. Major criteria include skin lesions in acral locations induced by exposure to cold or a drop in temperature, and evidence of lupus erythematosus in the skin lesions by histopathologic examination or DIF. Minor criteria are the coexistence of SLE or other skin lesions of DLE; response to anti-lupus erythematosus therapy; and negative results of testing for cryoglobulin and cold agglutinins. Diagnosis of CHLE is confirmed if patient fulfills both the major and any one of the minor criteria. Our patient presented with erythematous, scaly lesions over both the hands and feet that started during the cold month of December. The lesions initially appeared to be simple perniosis, but on detailed cutaneous examination, typical DLE lesions were observed over bilateral concha though the patient or her family members had not noticed them. The diagnosis of CHLE was further confirmed by positive antinuclear antibodies as well as by skin biopsy and direct immunofluorescence. Thus,

our patient fulfilled both major as well as two minor criteria as suggested by Su *et al*.[8]

The treatment includes protection from cold by physical measures and topical or oral antibiotics, if the lesions are infected. Topical steroids and calcium channel blockers have been found to be effective.[9] In recalcitrant cases, local immunosuppressive agents such as tacrolimus, pimecrolimus or systemic agents such as systemic steroids and mycophenolate mofetil[10] may be tried. Antimalarial drugs (chloroquine or hydroxychloroquine) have shown to have good effects on SLE typical symptoms but not on chilblain themselves. The patient was advised protection from cold and application of potent topical steroids twice daily over DLE as well as chilblain lesions. She showed clearance of lesions in two months. She is being followed up for the appearance of new lesions in next winter. We propose to start her on calcium channel blockers in winters if the lesions become symptomatic during that period.

Our patient showed unique features in the form of early age of onset, concentration of chilblain lesions over the ventral aspect compared to the dorsal aspect of the hands and feet and the presence of anti-Ro 52, anti-histone, anti-Smith, ds DNA and nucleosomal antibodies. A few case reports have reported the presence of Anti Ro/SS-A in CHLE but none has reported the presence of multiple subtypes of antinuclear antibodies. Although rare, it is an important entity that the physician must have an understanding of, as it can be confused with perniosis.

The risk of developing SLE in such cases of CHLE is estimated to be about 18%, hence we plan to follow up the patient for clinical/laboratory evidence of progression to SLE.

To conclude, CHLE lesions may be clinically indistinguishable from simple chilblains or perniosis seen in healthy individuals, hence detailed examination as well as investigations must be done in suspicious cases.

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