JRD Journal of Rheumatic Diseases Vol. 22, No. 4, August, 2015
http://dx.doi.org/10.4078/jrd.2015.22.4.269

Clinical Image

Childhood Chilblain Lupus Erythematosus

Eui Hyun Oh[1], Jae-Bum Jun[2], Joo Yeon Ko[1]
Departments of [1]Dermatology and [2]Internal Medicine, Hanyang University College of Medicine, Seoul, Korea

Chilblain lupus erythematosus (CLE) is a rare form of cutaneous lupus erythematosus (LE) and diagnosed by chilblain like skin lesions in acral locations induced by cold exposure and characteristic histopathologic findings similar to LE [1]. Sometimes, CLE can coexist with other cutaneous LE and have one of the other American College of Rheumatology criteria for systemic LE [2]. Patients with CLE should avoid cold exposure and could usually be controlled by anti-LE therapy such as topical or systemic steroid and antimalarial agents [1,2].

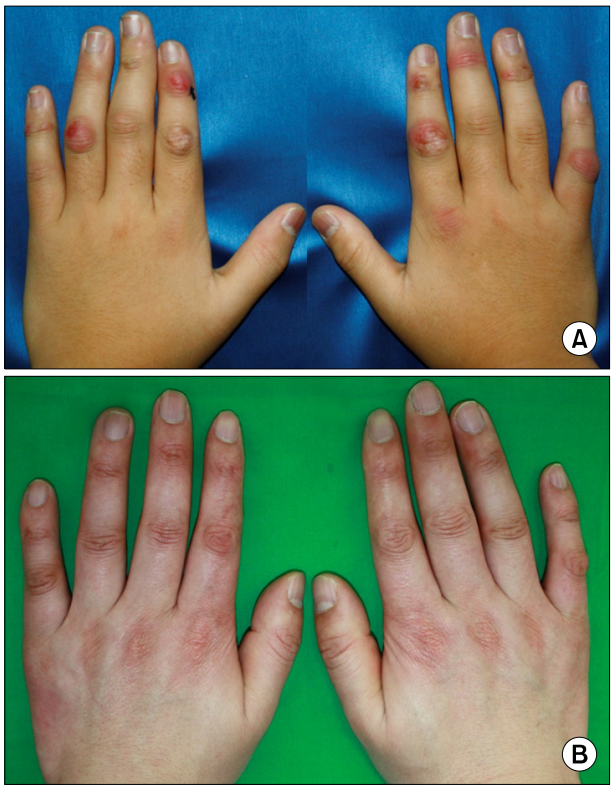

Figure 1. (A) Multiple erythematosus scaly plaques were observed on dorsal aspects of patient's hands. (B) Similarily, his father showed erytheamtous skin lesions on dorsum of hands.

We describe a 14-year-old boy with a history of painful, cold-induced inflammatory lesions on his fingers, toes, and ears since infancy. Recurrent episodes of these skin changes in every winter led to postinflammatory hyperpigmentation and skin hardening on the multiple knuckles of the hands and both ears (Figures 1 and 2). His father had suffered from similar lesions (Figures 1 and 2) in winter since childhood but symptoms had ameliorated with age. Histopathologic and direct immunofluorescent findings from biopsied skin were compatible with LE (Figure 3). After treatment with systemic steroid and hydroxychloroquine for diagnosed CLE, significant improvement was observed.

Unlike sporadic CLE whose pathogenesis remains unknown, two missense mutations in TREX1 were described in several cases of familial CLE [3-5]. Although sequencing of TREX1 was not carried out in this case, patient's family history and early onset of disease could lead to his diagnosis of childhood CLE with a familial trait.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

REFERENCES

1. Hedrich CM, Fiebig B, Hauck FH, Sallmann S, Hahn G, Pfeiffer C, et al. Chilblain lupus erythematosus: a review of literature. Clin Rheumatol 2008;27:949-54.
2. Viguier M, Pinquier L, Cavelier-Balloy B, de la Salmonière P, Cordoliani F, Flageul B, et al. Clinical and histopathologic

Received : April 7, 2015, **Revised :** April 28, 2015, **Accepted :** May 13, 2015

Corresponding to : Joo Yeon Ko, Department of Dermatology, Hanyang University College of Medicine, 222 Wangsimni-ro, Seongdong-gu, Seoul 04763, Korea. E-mail : drko0303@hanyang.ac.kr

pISSN: 2093-940X, eISSN: 2233-4718

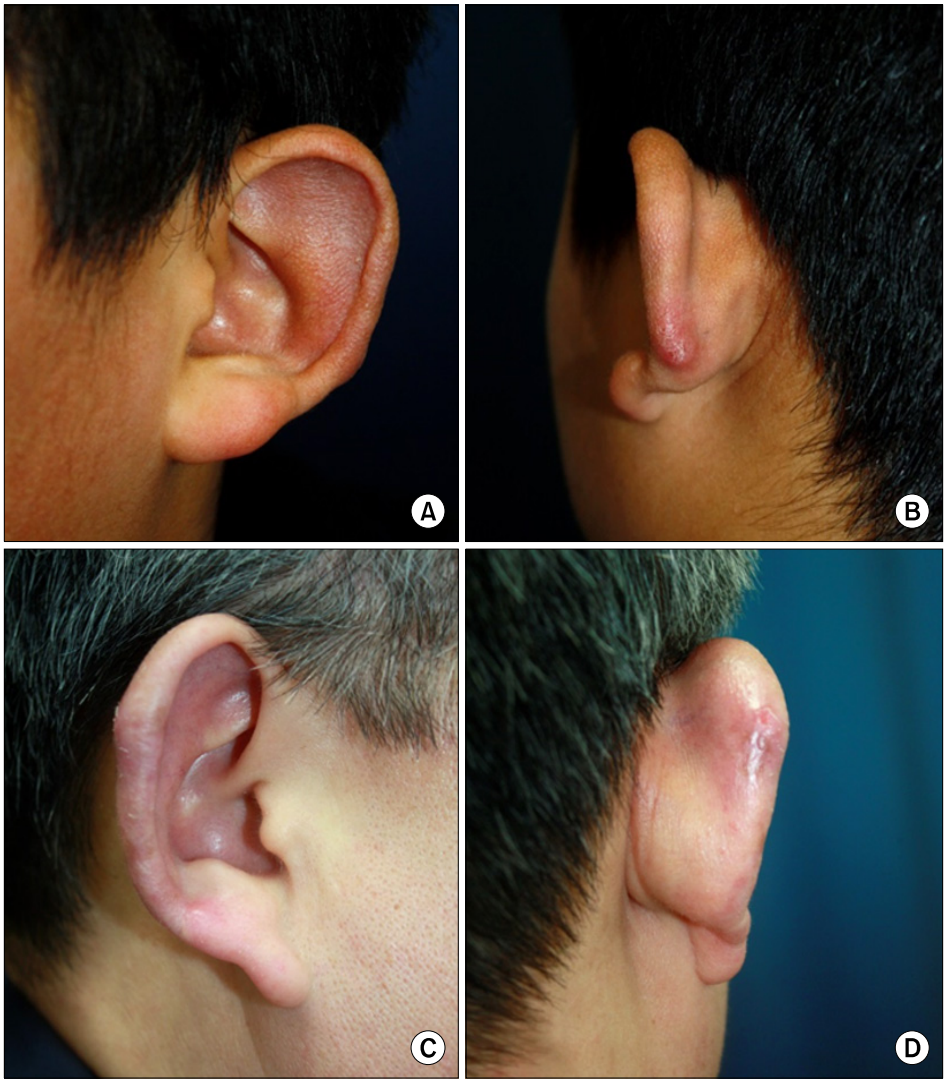

Figure 2. Erythematous to purpuric swollen scaly patches were noted on the ear of the patient (A, B) and his father (C, D).

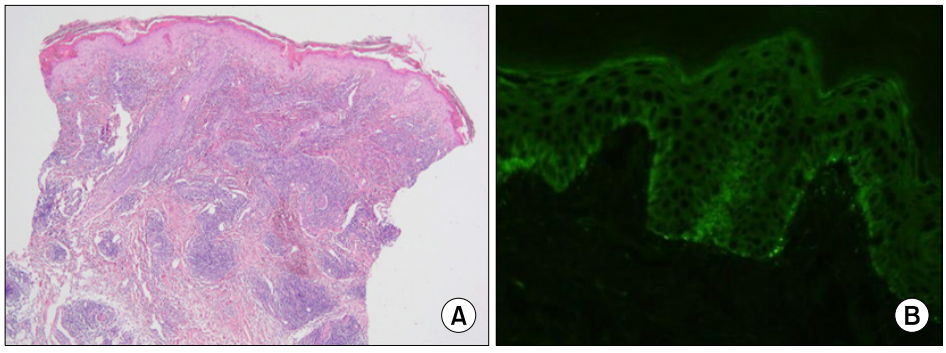

Figure 3. (A) Histopathologic examination revealed interface dermatitis and prominent periappendigeal, perivascular and perineural lymphohistiocytic infiltrates throughout the entire dermis (H&E, ×40). (B) Lesional direct immunofluorescence results showed granular deposits of immunoglobulin M in the basement membrane zone (×200).

features and immunologic variables in patients with severe chilblains. A study of the relationship to lupus erythematosus. Medicine (Baltimore) 2001;80:180-8.
3. Tüngler V, Silver RM, Walkenhorst H, Günther C, Lee-Kirsch MA. Inherited or de novo mutation affecting aspartate 18 of TREX1 results in either familial chilblain lupus or Aicardi-Goutières syndrome. Br J Dermatol 2012;167:212-4.
4. Abe J, Izawa K, Nishikomori R, Awaya T, Kawai T, Yasumi T, et al. Heterozygous TREX1 p.Asp18Asn mutation can cause variable neurological symptoms in a family with Aicardi-Goutieres syndrome/familial chilblain lupus. Rheumatology (Oxford) 2013;52:406-8.
5. Günther C, Hillebrand M, Brunk J, Lee-Kirsch MA. Systemic involvement in TREX1-associated familial chilblain lupus. J Am Acad Dermatol 2013;69:e179-81.