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].

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

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 periappendageal, 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).


