

LETTER TO THE EDITOR

Atopic dermatitis presenting as generalized poikiloderma with filaggrin gene mutation

Dear Editor,

Poikiloderma is a characteristic skin condition consisting of pigmentation, depigmentation, telangiectasia and atrophy, and is seen in a number of diseases. Here, we report a patient with atopic dermatitis (AD) with filaggrin (FLG) gene mutation, presenting as marked generalized poikiloderma.

A 26-year-old man was referred to us for evaluation of his recalcitrant itchy eruption. In his childhood, the patient had asthma and eczematous lesions, diagnosed as AD, which gradually turned to poikiloderma in his late teens. His AD had been treated with topical corticosteroids, but he denied their extensive usage. On examination, the patient had a poikilodermatous eruption on the chest (Fig. 1a) and back (Fig. 1b), extending to the neck and proximal parts of the limbs. The eruption was a mixture of small pigmentary and depigmentary elements with fine telangiectasia (Fig. 1c). Concomitantly, there were eczematous lesions on the antecubital fossae and dorsal aspects of the hands. He had palmar hyperlinearity (Fig. 1d) and scaly dry skin on the extremities, especially on the shins.

No photosensitivity, nail abnormality or cataracta was found. His SCORAD level was 50.5, and visual analog scale of pruritus was 41.

Laboratory examination revealed high levels of total serum immunoglobulin (IgE (13 207 kU/L; normal, <160) and serum CCL17/TARC (1124 pg/mL; normal, <450). The following values were normal or negative: complete blood counts, leukocyte differentiation, antinuclear and anti-DNA antibodies, complements and soluble interleukin-2 receptor. Histopathologically, a biopsy from poikiloderma showed moderate elongation of rete ridges with melanosis on their tips (Fig. 1e). There was a perivascular infiltrate of lymphocytes intermingled with melanophages in the upper dermis. Cellular atypia of lymphocytes was not observed.

The patient's blood was subjected to *FLG* mutation analysis¹ to detect the eight reported mutations specific to Japanese (R501X, 3321delA, S1695X, Q1701X, S2554X, S2889X, S3296X and K4022X).² Genotyping was performed using polymerase chain reaction and restriction enzyme digest analysis,

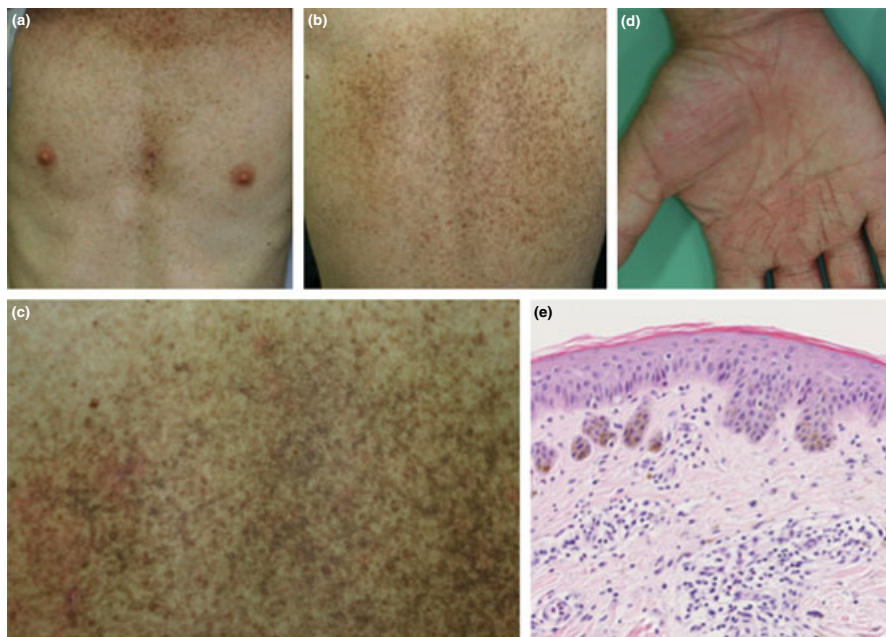


Figure 1. Clinical and histopathological appearances. Poikiloderma-like lesions on the (a) chest and (b) back. (c) Close inspection of poikiloderma on the back. (d) Palmar hyperlinearity. (e) Histopathology, showing melanosis of the tips of elongated rete ridges and infiltration of lymphocytes and melanophages (stain, original magnification $\times 100$, hematoxylin and eosin stain).

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which was approved by our ethics committee.³ We identified S2889X FLG gene heterozygous nonsense mutation in this patient.

The patient was treated with topical tacrolimus and corticosteroids. Currently, his pigmentation has slightly improved.

We diagnosed the patient's poikiloderma as AD because of the childhood-onset long history, concomitance of eczema, presence of asthma, high serum levels of IgE and CCL17, and palmar hyperlinearity with *FLG* mutation. Other poikiloderma-presenting diseases, including mycosis fungoides, dermatomyositis and Rothmund–Thomson disease were excluded. To our knowledge, this is the first report of generalized poikiloderma in association with AD.

Approximately 2% of adult AD patients exhibit rippled, poikiloderma-like lesions on the neck, called “dirty neck”.⁴ Histopathologically, dirty neck shows marked pigmentary incontinence as well as eczematous changes. Deposition of amyloid also may occur. In addition to pigmentary incontinence, our case showed increased melanosis in the epidermal rete ridges. The present case may represent extreme extension of such poikiloderma-like lesions. Various skin lesions are associated with *FLG* mutation, such as xerosis cutis, keratosis pilaris, flexural eczema and dirty neck.⁵ Our case suggests that generalized poikiloderma may be one of the manifestations of AD with *FLG* mutation.

CONFLICT OF INTEREST: None.

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