a crown of thorns in dermoscopy, corresponded to increased and dilated dermal capillaries, a new finding that was confirmed by histologic analysis and, to our knowledge, not described previously. In conclusion, as with the clinical and pathologic presentations, the RCM features associated with cutaneous lesions of Degos disease seem to exhibit very distinctive RCM features.

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Sibling Cases of Hailey-Hailey Disease Showing Atypical Clinical Features and Unique Disease Course

Hailey-Hailey disease (HHD), a well-characterized autosomal dominant hereditary disease, is caused by mutation in ATP2C1 gene and clinically shows characteristic erosive lesions predominantly on the intertriginous areas. We herein report sibling cases of HHD with novel mutations in the ATP2C1 gene that showed unique and atypical clinical phenotypes mimicking seborrheic dermatitis, pemphigus vulgaris, or pemphigus foliaceous as well as considerable alterations during the disease course.

Report of Cases | Case 1. A 61-year-old Japanese man with intractable pulmonary arterial hypertension, pulmonary fibrosis, and emphysema was receiving oxygen supply from a nasal...
tube at the time of presentation. Physical examination revealed seborrheic dermatitis–like diffuse scaly erythemas on the face and anterior neck, where the nasal tube of the oxygen supply contacted the skin (Figure 1A). In addition, extensive erosive erythemas with severe pain were seen on the abdomen, and a few pemphigus vulgaris–like flaccid bullae were also observed (Figure 1B). No skin lesions were seen in any intertriginous areas, including axillae and groin. No mucosal membranes were involved. Detailed medical history indicated that, beginning at age 19 years, the patient had occasional mild eczematous lesions on the axillae, groin, popliteal fossa, and head.

Case 2. A 57-year-old younger sister of patient 1 reported that, beginning at age 20 years, she had developed dermatitis-like skin lesions on the genital region during menstrual periods. Subsequently, itchy erosive lesions appeared on the axillae and groin, which worsened in the summer every year. The skin lesions on the axillae and groin gradually decreased, while skin lesions on the face, neck, and chest continued to develop.

Physical examination revealed erythemas with small erosions scattered on the forehead, neck, chest, and upper back (Figure 1C) but not on the intertriginous areas, except for minimum lesions on the left axilla. There were no apparent vesicles or bullae.

Comment | Histopathologic analysis of the seborrheic dermatitis–like lesion and pemphigus vulgaris–like lesion in case 1 and pemphigus foliaceus–like lesion in case 2 revealed acantholysis without any dyskeratotic keratinocytes in the lower epidermis (Figure 1D and E). Indirect immunofluorescence studies showed IgG antibodies to epithelia of monkey esophagus and rat bladder in case 1 (Figure 2A and B), but no conclusive pattern was seen in case 2. Genetic study revealed a novel splice-site mutation in the ATP2C1 gene in both cases (Figure 2C).

The 2 cases were diagnosed as HHD by identification of a novel mutation in the ATP2C1 gene. However, while both patients showed more typical HHD features when they were
younger, as they aged into their 60s, they manifested atypical clinical features and unique disease courses. Intriguingly, the clinical features in the 2 cases were significantly different from each other, in spite of the presence of the same genomic mutation. Taking the changes in clinical features during the disease courses of the 2 cases together, we speculate that some environmental conditions, in addition to the genetic defect, may affect the development of skin lesions of HHD.

A study of 58 patients with HHD in the United Kingdom indicated that predilection sites were the flexure areas, including the groin, axilla, perineum, inframammary region, umbilicus, and retroauricular region, in descending order, as well as neck/nape, shoulders, chest, arms, and back. The abdomen was not involved in any cases. Only a few cases showed lesions on the popliteal fossae, head, and face, which were frequently associated with eczematous changes but rarely showed bullae or pustules.

Two clinically unique and atypical HHD cases have been reported, although genetic study was not performed in either case. One case showed seborrheic dermatitis-like lesions on the scalp as well as other typical HHD lesions, while the other case showed generalized vesiculobullous lesions with occasional pustules. To our knowledge, our patients represent the first sibling cases with seborrheic dermatitis-like, pemphigus vulgaris-like, or pemphigus foliaceus-like atypical clinical and histopathologic features of HHD diagnosed by genetic analysis.

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