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Atypical manifestations of hemangiomas in epidermolysis bullosa

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Epidermolysis bullosa (EB) is a group of inherited bullous diseases that are classified into four subtypes according to the level of skin cleavage: EB simplex, junctional EB (JEB), dystrophic EB (DEB), and Kindler EB (1). Despite their clinical variability, all EB subtypes have a more or less high risk of the patient developing cutaneous squamous cell carcinoma (cSCC), which can be life-threatening (2). JEB and recessive dystrophic EB (RDEB) are particularly associated with cSCC, with incidences ranging from 6.8% to 16.2% for JEB and 35.4% to 70% for RDEB (2). So, it is crucial to monitor for cSCC when following up EB patients over the long term. There have been a few reports on benign cutaneous tumors in EB. Here, we introduce two Japanese EB patients who developed hemangiomas in the skin.

Case reports

Patient 1

A 67-year-old female with RDEB presented with a keratotic nodule on her right forearm (Fig. 1a, b). She was compound heterozygous for c.5443G >A (p.Gly1815Arg) and c.5819del (p.Pro1940Argfs*65) in *COL7A1* (3). She had a recurrent history of cSCC. The 1cm-wide nodule was erosive on its surface and had been present for about a year (Fig. 1a, b). Because it had been slowly enlarging, the nodule was resected for possible cSCC. Histopathology showed epidermal hyperkeratosis, various degrees of acanthosis, and ectasia of dermal thin-walled vessels surrounded by epidermal ridges (Fig. 1c). Epidermal erosions and fibrin deposits in the dermis were seen. No cell atypia was observed in the epidermis. Based on these findings, we diagnosed solitary angiokeratoma.

Patient 2

A 38-year-old male with JEB presented multiple red-colored papules on the scalp (Fig. 2a, b). He was compound heterozygous for c.1179del (p.Ala394Leufs*9) and c.4159C>T (p.Gln1387*) in *COL17A1* (4). The papules were 5 to 8 mm in diameter. Dermoscopy showed a red background, clustered red lacunae, and a white surface. A skin biopsy specimen from the papule revealed numerous capillaries with narrow lumina and prominent endothelial cells arranged in a lobular fashion in the subpapillary dermis (Fig. 2c). We diagnosed multiple cherry hemangiomas.

Discussion

The clinical manifestations of the angiokeratoma in Patient 1 were atypical, due to its erosive surface, possibly associated with skin fragility. The development of an angiokeratoma has been reported in an RDEB patient (5), and it is possible that recurrent wound healing in EB skin might result in angiokeratoma formation. Angiokeratoma can be a differential diagnosis of cSCC, especially in EB patients. Although the dermoscopic findings of cherry hemangioma in Patient 2 were typical, the relatively large papules and the scalp lesions were uncommon for an ordinary cherry hemangioma. To the best of our knowledge, cherry hemangiomas in JEB have not been reported. These cases point out that hemangiomas can show atypical manifestations in EB patients and that histopathological evaluation is essential for EB tumors.

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Figure legends

Figure 1. Clinical features of Patient 1.

(a, b) A 10 x 10-mm keratotic red nodule with an erosive surface on the outer right forearm.

(c) Histopathology of the nodule. Scale bar: 200 um.

Figure 2. Clinical features of Patient 2.

(a) Multiple red papules on the scalp (yellow arrowheads).

(b) Dermoscopic features of the area enclosed by the yellow rectangle (a).

(c) Histopathology of the papule. Scale bar: 200 um.

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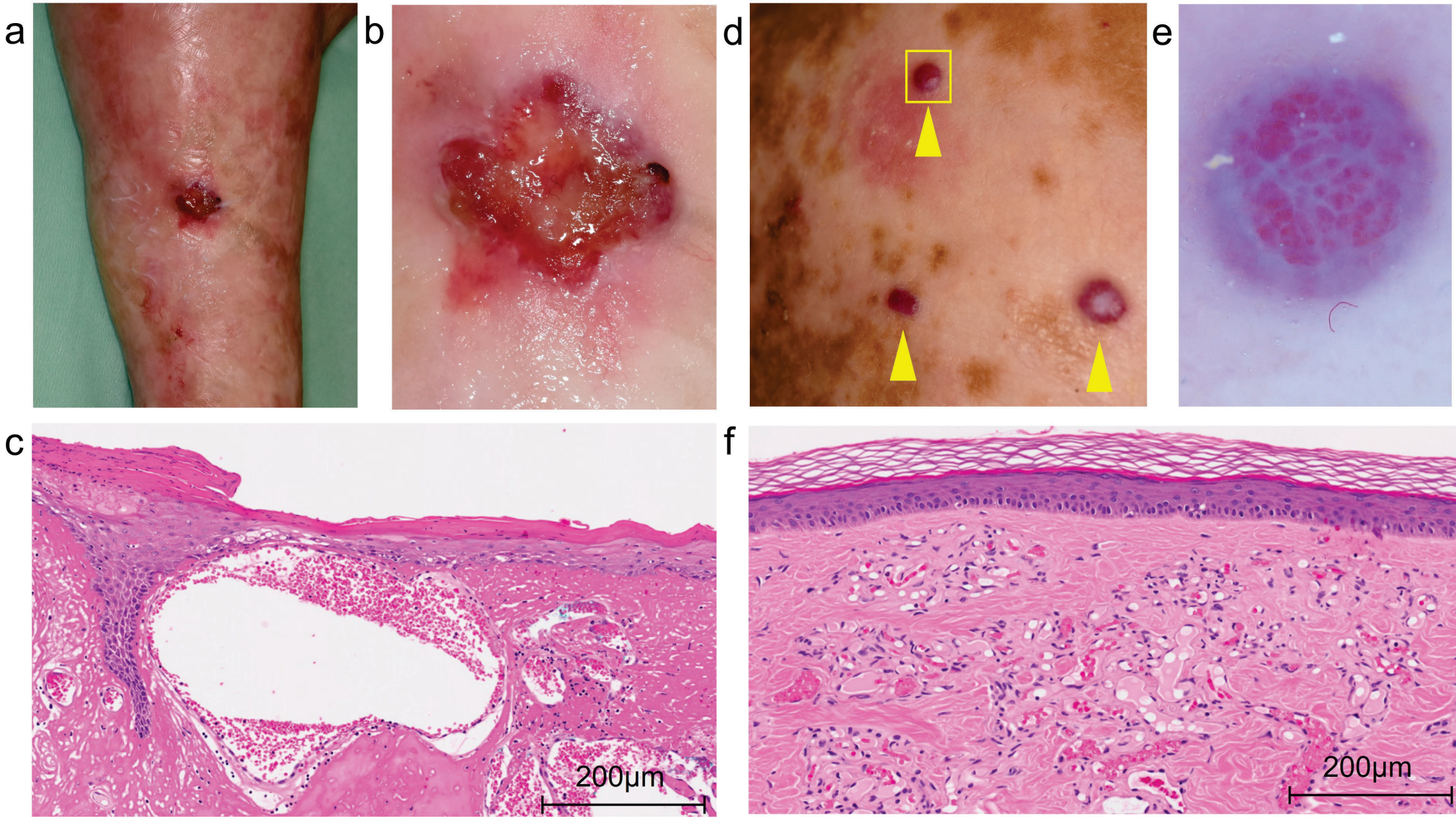
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