Simultaneous Occurrence of Three Squamous Cell Carcinomas in a Recessive Dystrophic Epidermolysis Bullosa Patient

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Sir,
Squamous cell carcinoma (SCC) is a life-threatening and frequent complication in recessive dystrophic epidermolysis bullosa (RDEB) (1). SCC in RDEB usually develops between the fourth and fifth decades and predominantly on the extremities (2). We have identified three independent SCCs occurring on the extremities of an RDEB patient who had failed to undergo regular dermatological consultations.

CASE REPORT

A 44-year-old Japanese woman who had no history of parental consanguinity or family history of bullous disease consulted our hospital complaining of painful nodules on both her feet (Fig. 1). Her own history revealed that within a few days of birth she developed trauma-induced blisters with scarring. She was soon diagnosed by a dermatologist as having RDEB. Detailed examination later revealed a non-Hallopeau-Siemens RDEB subtype with G1815R and 5818delC mutations in COL7A1 (3). Despite appropriate dermatologic care, she was suffering from continuous widespread erosions over her limbs and back, extensive dystrophic scarring, alopecia, pseudosyndactyl fingers and toes, loose nails, oesophageal stenosis and dental caries. She had stopped regular medical examination spontaneously 20 years previously because her condition failed to improve. Histology of the dome-shaped painful masses on her right foot, the left medial malleolus and the left heel showed all three tumours to be invasive SCCs. They were treated by wide

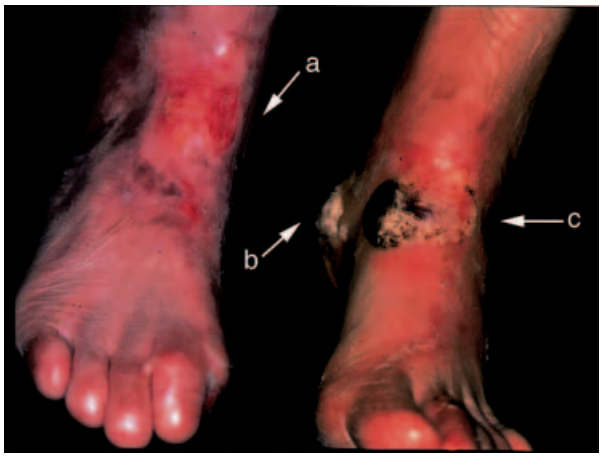

Fig. 1. The patient's three tumours on her feet, all histologically confirmed as well-differentiated, invasive squamous cell carcinomas. Arrows: (*a*) an ill-demarcated hyperkeratotic erythematous plaque, 4 cm in diameter with erosion on the dorsal site of the right foot; (*b*) a well-demarcated hyperkeratotic dome-shaped painful tumour, 3 cm in diameter with black and white necrosis on the left medial malleolus; and (*c*) a well-demarcated hyperkeratotic dome-shaped tumour, 2 cm in diameter with white necrosis and odour on the left heel.

excision followed by skin grafting. The patient has been followed up by a dermatologist for 3 years without signs of metastasis or local recurrences.

DISCUSSION

The most common cause of premature death in RDEB is SCC, which generally behaves aggressively and often metastasizes (1). The National Epidermolysis Bullosa Registry reports that 76.5% of Hallopeau-Siemens (HS) and 27.1% of non-HS RDEB patients experience at least one SCC by the age of 60 (4). Many patients with RDEB are regularly medically examined. As a consequence, early stages of SCC can be identified in the younger patients with RDEB and treated appropriately (5). Therefore, frequent whole body skin inspection with early sampling of suspicious lesions is an essential part of the management in patients with RDEB.

Too much emphasis on the possibility of developing carcinomas may cause patient disapproval and lead to the patients and their families loosing hope for coping with this disabling disease. However, early and on-going patient education raising the awareness of developing SCC from ulcers and scars is necessary for all patients with RDEB. We should at least repeatedly advise them to keep in touch with their dermatologist and not to stop visiting completely.

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REFERENCES

1. Fine JD, Johnson LB, Suchindran C, et al. Premature death and inherited epidermolysis bullosa. In: Fine JD, Bauer EA, McGuire J, Moshell A, eds. Epidermolysis bullosa: clinical, epidemiologic, and laboratory advances and the findings of the National Epidermolysis Bullosa Registry. 1st edn. Baltimore: Johns Hopkins University Press, 1999: 206–224.
2. McGrath JA, Schofield OM, Mayou BJ, McKee PH, Eady RA. Epidermolysis bullosa complicated by squamous cell carcinoma: report of 10 cases. J Cutan Pathol 1992; 19: 116–123.
3. Sato-Matsumura KC, Yasukawa K, Tomita Y, Shimizu H. Toenail dystrophy with COL7A1 glycine substitution mutations segregates as an autosomal dominant trait in 2 families with dystrophic epidermolysis bullosa. Arch Dermatol 2002; 138: 269–271.
4. Fine JD, Johnson LB, Suchindran C, et al. Cancer and inherited epidermolysis bullosa. In: Fine JD, Bauer EA, McGuire J, Moshell A, eds. Epidermolysis bullosa: clinical, epidemiologic, and laboratory advances and the findings of the National Epidermolysis Bullosa Registry. 1st edn. Baltimore: Johns Hopkins University Press, 1999: 175–192.
5. Kawasaki H, Sawamura D, Iwao F, et al. Squamous cell carcinoma developed in a 12-year-old boy with non-Hallopeau-Siemens recessive dystrophic epidermolysis bullosa. Br J Dermatol. In press.