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Case Report

Giant squamous cell carcinoma in a patient with epidermodysplasia verruciformis

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ABSTRACT

Epidermodysplasia verruciformis is an autosomal recessive skin disease usually presenting as multiple flat warts and pityriasis versicolor-like macules in early youth, possessing a great risk of developing skin cancer due to a lack of defense against beta HPV. Herein, we report the case of a 29-year-old female, a known case of EV, who presented with a verrucous growth on the forehead persistent for the previous one year. While clinical and dermoscopic examinations led to the suspicion of squamous cell carcinoma, it was confirmed by histopathological examination following a skin biopsy.

Key words: Epidermodysplasia verruciformis; SCC; Dermoscopy

INTRODUCTION

Epidermodysplasia verruciformis (EV) is a rare autosomal recessive genodermatosis that usually presents in early childhood as verrucous papules and plaques resembling pityriasis versicolor, verruca plana, or seborrheic keratosis, most commonly on the skin of the head, neck, and upper extremities, characterized by widespread infection with specific strains of human papillomavirus (beta HPV) [1]. There is a lack of defense against beta HPV in these individuals, which increases the likelihood of developing non-melanoma skin cancers, most commonly squamous cell carcinoma in these individuals [2]. These patients, therefore, serve as models for studying susceptibility to beta HPV and its carcinogenesis.

CASE REPORT

A 29-year-old female patient (Fitzpatrick skin type IV), normotensive, non-diabetic, belonging to a rural area, a diagnosed case of epidermodysplasia verruciformis with consanguinity in the parents' marriage (first degree) and the absence of such a condition in other family members, reported with a verrucous growth (5 × 6 cm) on the center of the forehead and multiple crusted plaques and ulcerations bilaterally on the forehead present for the last one year, which began as a small plaque on the pre-existing lesions of epidermodysplasia verruciformis one year previously and progressed to involve the central part of the forehead, with a rapid increase in size and pus discharge (Figs. 1a and 1b). The swelling was well-defined, firm, and not attached to the underlying structures. Diffuse swelling with crusted plaques was observed around the bilateral periorbital areas and the nasal bridge. An examination of other body areas revealed multiple seborrheic keratosis-like lesions on the face and neck. A general physical and system examinations were normal. The cervical lymph nodes were uninvolved. Hematological and biochemical investigations were within the normal limits.

Dermoscopy of the growth revealed multiple structureless, milky-white areas, yellowish, homogeneous areas, hemorrhages, and erosions on background erythema with short, linear, and polymorphic vessels (Fig. 2). An incisional biopsy taken from the lesion revealed poorly differentiated squamous cell carcinoma. The lesion was surgically excised followed by radiotherapy.

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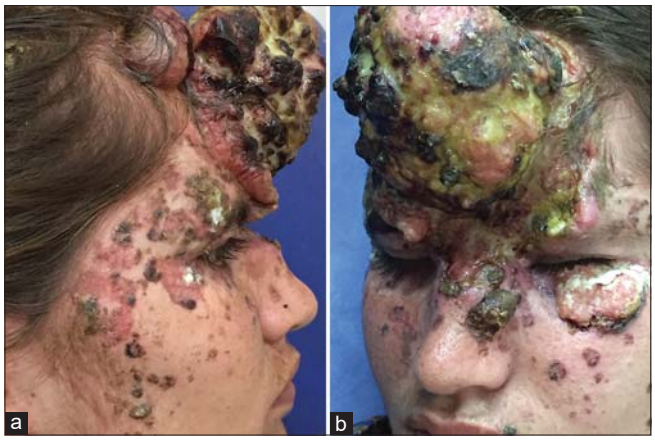

Figure 1: (a and b) Clinical image revealing a verrucous growth in the center and multiple crusted plaques and ulcerations bilaterally on the forehead in a patient with EV.

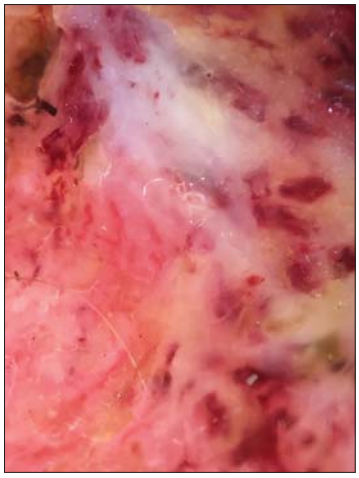

Figure 2: Dermoscopic image revealing multiple structureless, milky-white areas, yellowish, homogeneous areas, hemorrhages, and erosions on background erythema with short, linear, and polymorphic vessels, suggestive of SCC.

DISCUSSION

The clinical manifestations of EV begin in childhood, and up to 60% of patients with EV develop non-melanoma skin cancer, mainly squamous cell carcinoma (SCC) [3,4]. Such a skin cancer occurs usually in the fourth or fifth decade of life and is localized mainly in sun-exposed areas, indicating an important role of environmental factors, notably UV irradiation [5,6].

Beta HPV has a potential role in developing skin cancer in immunocompromised patients yet causes mainly unapparent skin infections in immunocompetent individuals, with types 5 and 8 being particularly more common forms in EV [7,8]. The inherited form of EV, which is caused by a mutation in TMC6/EVER1 or TMC8/EVER2 has a defect in the ability to mount an immune response to certain HPV types in keratinocytes [9]. However, there are normal immune capabilities against other infectious pathogens. The beta HPV types identified in patients with EV who develop skin malignancies are found throughout the general population. In persons without the EVER mutations or EV, these HPV types have not been shown to produce dysplasia or malignancy [10]. Patients with EV cannot appropriately control beta HPV replication and, therefore, have a strong antibody response against a broad variety of beta HPV types [11].

CONCLUSION

Patients with epidermodysplasia verruciformis are at a high risk of developing non-melanoma skin cancers, thus proper counseling and follow-up are needed for the timely management of dysplastic changes in any existing lesions. HPV is the viral agent clearly associated with the malignant transformation of cells.

CONSENT

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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