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Case Report

Epidermodysplasia Verruciformis: A Case Report and a Brief Review

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Abstract

Epidermodysplasia verruciformis is an uncommon disorder that is transmitted in an autosomal recessive manner. It is characterized by increased susceptibility to human papillomavirus infection, which presents with hypo- or hyperpigmented macular lesions, pityriasis versicolor-like lesions, and an early tendency to transform into skin cancer.

We present a case of a 40-year-old male with complaints of verrucous lesions of the hands and feet. Histopathology was suggestive of EV.

Keywords: Epidermodysplasia verruciformis; Genodermatosis; Human papilloma virus

Introduction

Epidermodyplasia Verruciformis (EV) is a rare, mostly autosomal recessive genodermatosis. It is characterized by an increased susceptibility to Human Papillomavirus (HPV) infection and a defect in the cell-mediated immune response. It occurs in two forms, sporadic or familial [1]. Skin lesions begin in childhood and are mainly observed in sun-exposed sites [2]. We report a case of familial EV with a review of the literature.

Case Presentation

A 40-year-old man, single, born of a first-degree consanguineous marriage, consulted for multiple verrucous lesions of the hands and feet that he had since 4 years old. The patient was a chronic smoker and had a younger brother with the same symptomatology.

The lesions started on the feet and progressively increased in number and size, then appeared on the hands as well. The patient had never previously consulted a physician and, therefore, had not received any treatment.

On physical examination, yellowish and brownish scaly hyperkeratotic lesions were present on the palms and soles (Figure 1 and 2) with subungual hyperkeratosis of the fingernails and toenails. Dermoscopic examination showed a verrucous, cerebriform appearance (Figure 3).

Scalp, oral cavity and genitals were normal. Systemic examination was unremarkable.

A skin biopsy was taken from the lesion of the feet which showed hyperkeratotic and acanthotic epidermis. The keratinocytes had abundant grayish blue basophilic cytoplasm and contained numerous round basophilic cysts.The nuclei were large, round, and empty with a marginal distribution of chromatin and the presence of keratohyaline granules.

Above features were suggestive of EV. Lipid and liver tests normal. A treatment based on oral retinoids was proposed but refused by the patient for the high cost. However, we referred the patient for genetic counseling and he was advised strict photoprotection and regular follow-up.

Figure 1: Clinical aspect of the lesions.

Discussion

Epidermodysplasia Verruciformis is a ubiquitous disease with no sexual, racial or geographical predominance [1]. It is a rare autosomal recessive genodermatosis characterized by an increased risk of cutaneous malignancy. The familial form is more common [3]. However, autosomal dominant inheritance pattern had been reported in two cases [4]. EV can be contracted and is usually seen in immunocompromised patients, such as after renal transplantation, in Hodgkin's disease and systemic lupus erythematosus and with Human Immunodeficiency Virus (HIV) infection[5].

The determinism of this disease is multifactorial, involving genetic factors, extrinsic factors: UV radiation as evidenced by the predominantly affected areas photo-exposed and immunological factors: alteration of cellular immunity [1].

The gene involved is the EVER1 (Epidermine) gene (Epidermodysplasia Verruciformis Enhancing Region) and EVER2

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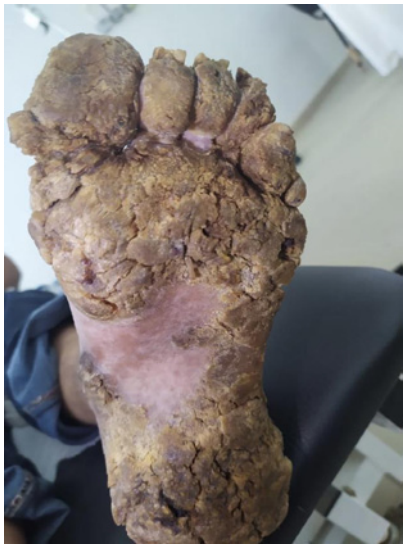

Figure 2: Clinical aspect of the lesions.

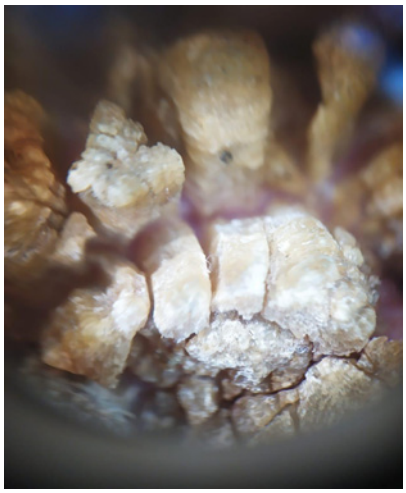

Figure 3: Dermoscopic aspect.

on the q-region of chromosome 17, which is also probably involved in certain familial forms of psoriasis [2]. Indeed, these patients are potential reservoirs of HPV5 and the two conditions would be due to two different mutations of the same gene [1]. Other associations with certain genetic diseases such as chondrodysplasia, neurofibromatosis type 1 and congenital bone dysplasia have been described.

The onset of the disease is usually between 4 and 8 years of age, most often before the age of 20 years, but exceptionally it may appear later [6]. In EV lesions, papillomavirus types 5 and 8 are found, viruses that are found in 80% of subjects in a normal asymptomatic population [6]. Types 5, 8 and 47 are highly oncogenic, found in more than 90% of epidermolysplasia verruciformis carcinomas. Types 14, 20, 21 and 25 have low oncogenic potential and are most often detected in benign lesions. HPV 3 and 10 are not specific for epidermolysplasia verruciformis, they are also associated with flat warts in the general population [1].

EV results in the appearance of scaly macules and papules of sometimes exuberant, pseudotumor evolution, mainly on the hands and feet. The primary skin lesions are multiform, usually verrucous, lichenoid, hyperkeratotic, and flat-topped [3]. Over time, they increase in number and tend to develop into papules that resemble flat warts, pink to brownish in color, a few millimeters in size, with a smooth surface [3]. It may also manifest as a macular rash similar to that seen in pityriasis versicolor; associated with scaly wart-like papules [6].

EV has pathognomonic histological features. The keratin layer is loose with a basket-weave-like appearance. The most characteristic findings are the presence of clear cells in the granular and spinous layers with occasional enlarged, hyperchromatic, atypical nuclei. The nucleoplasm is clear, and keratohyaline granules of various sizes and shapes are present. In premalignant tumors, the normal keratinocyte maturation is preserved, while in the malignant lesions, the normal surface maturation of keratinocytes is lost. The premalignant lesions display features similar to actinic keratoses with prominent atypical, dyskeratotic cells [7].

The prognosis of EV is related to the oncogenic potential of certain HPVs inducing carcinomas in photo-exposed areas in 30 to 60% of cases [1]. Dysplastic lesions and skin cancers occur after 30 years of age, most often in photo exposed areas, in the form of actinic keratoses and Bowen's disease. Squamous cell carcinomas develop on one or more lesions in about 20% of cases [1].

At present, there is no specific and effective treatment for EV. The goal for the management of EV would be to prevent the progression of benign lesions to malignancy through preventive measures such as genetic counseling, photoprotection, and symptom monitoring for early detection of pre-malignant and malignant lesions [3]. Pharmacological treatments include oral and topical retinoids, interferon, immunotherapy, imiquimod and cimetidine. A surgical approach, including electrosurgical ablation and cryotherapy, is also used to manage benign and premalignant lesions [8]. Malignant lesions have also been treated surgically [8].

Conclusion

Epidermodysplasia verruciformis is a rare disease with a high potential for transformation.

It is imperative to educate the patient and to do a rigorous regular follow up in order to diagnose early precancerous lesions.

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