Our Dermatology Online

Case Report

Peutz–Jeghers lentigines revealing epidermodysplasia verruciformis in two siblings

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ABSTRACT

Peutz–Jeghers syndrome (PJS) is an autosomal dominant disease characterized by skin and digestive involvement. Epidermodysplasia verruciformis is a ubiquitous disease characterized by abnormal susceptibility to infection by about twenty human papillomaviruses of group B. Herein, we report two siblings followed for Peutz–Jeghers syndrome with lentiginous skin lesions and pityriasis versicolor-like macules confused with the diagnosis of epidermodysplasia verruciformis. The association of these two rare entities has not yet been reported. However, the cutaneous dyschromic lesions observed in Peutz–Jeghers syndrome are highly characteristic and may be confused with the signs of epidermodysplasia verruciformis, especially for macular lesions in an acral location. This case highlights the value of a skin biopsy in confirming the diagnosis in the face of the appearance of lesions of pityriasis versicolor-like and keratotic papules in a patient with Peutz–Jeghers lentigines.

Key words: Epidermodysplasia verruciformis, Human papillomavirus, Peutz–Jeghers, Morocco

INTRODUCTION

Peutz–Jeghers syndrome (PJS) is an autosomal dominant disorder characterized by polyposis of the gastrointestinal tract [1], pigmentation of the skin and mucous membranes, and a predisposition to oncological diseases. Its association with epidermodysplasia verruciformis (EV) has not been reported in the literature. Generally, EV is an autosomal recessive genodermatosis characterized by the persistent presence of beta group papillomavirus (HPV) in the skin. Recently, a new classification of EV has been proposed distinguishing a classical genetic form, a non-classical genetic form, and an acquired form. Herein, we report the first description of epidermodysplasia verruciformis associated with Peutz–Jeghers syndrome in two siblings.

CASE REPORT

A sixteen-year-old patient, without any notion of consanguinity, followed in pediatrics with his sister for Peutz–Jeghers syndrome with digestive polyposis, admitted to our training for dyschromic lesions on the face, neck, trunk, and upper limbs present since the age of ten years, whose evolution was marked by the appearance of other asymptomatic lesions on the back of the hands and perioral for which he was referred (Figs. 1a – 1c). A clinical examination revealed multiple lentigines on the back of the nose, lips, and palms, hyperpigmented macules on the lips and face, and millimeter-sized papules with keratotic surfaces on the back of the hands associated with a poorly defined pityriasis versicolor-like patch on the back, which was negative on Wood's light (Fig. 1d). We considered the cutaneous manifestations of his pathology or epidermodysplasia verruciformis given the dyschromic patch on the back. A skin biopsy was taken from the dorsal aspect of the hand and confirmed the diagnosis of epidermodysplasia verruciformis. An examination of his sister revealed the same clinical finding (Figs. 2a and 2b). Unfortunately, the patient refused any medical treatment. No genetic studies were performed in either patient.

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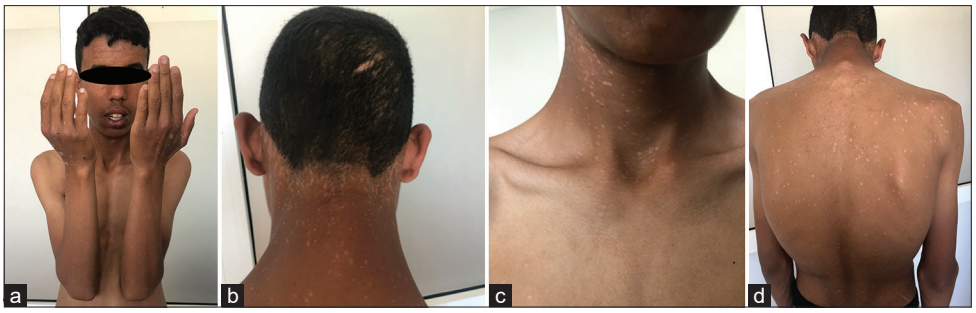
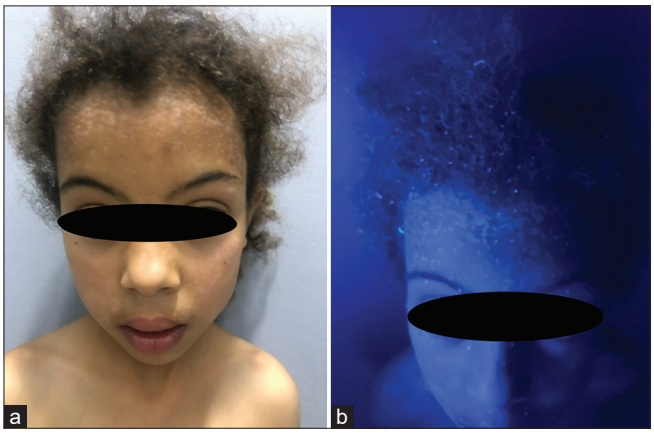


Figure 1: (a) Multiple papules with a keratotic surface on the forearms and dorsal surface of the hands associated with pigmented macules on the lips. (b and c) Multiple hypopigmented and pigmented macules on the neck.(d) Dyschromic pityriasis versicolor-like plaques on the back.



Figures 2: (a and b) Hypochromic macules on the face not taking contrast in Wood's light on the forehead in the sister.

DISCUSSION

SPJ is a rare autosomal dominant disease defined by the combination of periorificial lentiginosis-like skin involvement and digestive, pulmonary, and reproductive organ involvement [1]. The prevalence is estimated at 1 in 200,000, with no sex or race predominance, and the average age of diagnosis was 22 years in a review of 75 cases [2]. In 1921, Jan Peutz, [3], a Dutch physician, reported a family with gastrointestinal polyposis and mucocutaneous lentiginosis. In 1949, Harold Joseph Jeghers [4] published a detailed description of patients with intestinal polyposis and skin pigmentation, [1]. In a literature review of seventy cases, skin signs appeared earlier in boys (5–10 years) than in girls (10–15 years), which was not the case in our two patients [3]. These skin manifestations are a revealing sign of the disease, yet they are not the first to appear. In all cases, they are lentigines, brown- or buff-colored macules, oval, and generally less than 5 mm in diameter. They are most often found on the nose, lips, and oral mucosa, as was the case in our patient. The nasal mucosa, periorbital areas, elbows, dorsal surface of the fingers, toes, and plamo-plantar areas may also be affected [5], as in our patient.

This condition poses a problem in differential diagnosis, especially with epidermodysplasia verruciformis, a rare skin condition of genetic origin characterized by abnormal sensitivity of the skin surface to papillomavirus. Clinically, EV manifests itself in childhood between the fourth and eighth years of age, most often before the age of twenty, and lasts throughout life with flat, wart-like lesions and a macular rash similar to that known in pityriasis versicolor (pityriasis versicolor-like), disseminated and persistent [6] as was the case in our patient. The lesions usually begin on the back of the hands and forehead, progressively spreading to the limbs, neck and trunk, in rare cases reaching generalized forms, with the mucous membranes usually respected [7].

EV is associated with mutations in the EVER1/TMC6 and EVER2/TMC8 genes. Recently, a new classification of VE has been proposed distinguishing between a classical genetic form, a non-classical genetic form, and an acquired form. Recent discoveries of other genes involved in EV, including RHOH, MST-1 and CORO1A, have complicated the classification of EV and EV-like syndromes. In our patient, we assume that it was a classical form due to the presence of a familial case, as well as the age of onset of the lesions and the absence of an infection or immune deficiency [8].

In general, both conditions have a high risk of malignant transformation in both children and adults, with a lower prevalence in children [3], requiring regular and long-term follow-up.

There is no standardized treatment for mucocutaneous pigmentation, which is present in the majority of patients. Treatment is mainly cosmetic for the dermatological manifestations of Peutz–Jeghers and involves cryosurgery, dermabrasion, and Q-switched laser [4]. While for EV, complete regression of the

lesions has never been observed, as they are refractory to conventional wart treatments, preventive treatments for complications and symptomatic treatments of the lesions may be proposed, including alpha and beta interferons or imiquimod, yet their efficacy is inconsistent and temporary [9]. Cosmetically intrusive, benign tumors are treated with oral retinoids, yet results are inconsistent with a relapse on dose reduction or discontinuation. Photodynamic therapy and cryotherapy appear to be effective on viral lesions. [10]. Given the limited economic means of our two patients, only depigmenting and keratolytic creams and an early, permanent, and rigorous photoprotection were proposed.

CONCLUSION

This case was interesting because of the association between the two rare entities as well as the interest of a skin biopsy in confirming epidermodysplasia verruciformis in front of the appearance of pityriasis versicolor-like and keratotic papules in a patient with Peutz–Jeghers lentigines.

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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