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A Case of Epidermodysplasia Verruciformis: A First Case Report from Yemen

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1. Abstract

Epidermodysplasia verruciformis (EV) is a rare genodermatosis associated with decreased immunity against certain types of human papillomaviruses that are associated with a high risk for cutaneous dysplasia and skin cancer. Herein, we present the case of a 35-year-old man with seborrheic keratosis-like lesions on his face, plane wart-like lesions on both hands, erythematous macules bilaterally on his neck, and pityriasis versicolor-like lesions on both arms. The onset occurred in the second decade of his life, at which time he reported similar lesions affecting his father and brother. Histopathological findings confirmed the diagnosis of EV. Interestingly, this is the first such case to be reported in Yemen.

2. Significance

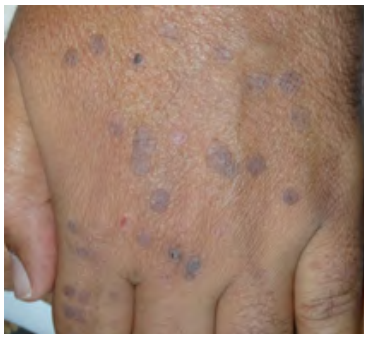
This first reported case of epidermodysplasia verruciformis indicates the prevalence of this unusual and rare skin disease in Yemen and raises awareness about the need for early identification by clinicians and treatment.

3. Introduction

Epidermodysplasia verruciformis (EV) is a rare autosomal recessive genodermatosis caused by a mutation of either EVER1 or EVER2, leading to a specific defect in immunity against certain types of human papillomaviruses (HPV) [1]. It presents with disseminated and persistent HPV infection, giving rise to a typical presentation with three types of lesions: plane wart-like lesions, pityriasis versicolor-like lesions, and reddish plaques [2-4]. Although it is commonly associated with the development of malignant tumors, metastasis is rare [5]. The onset of the highly polymorphic cutaneous lesions of EV commonly occurs during the second decade of life [5]. Approximately 50% of patients develop precancerous lesions and malignant tumors, such as squamous cell carcinomas (SCC) of the skin, subsequently form on sun-exposed sites [6]. The disease is typically characterized by cutaneous polymorphic lesions simulating pityriasis versicolor (PV-like), plane warts (PW-like), or seborrheic keratosis (SK-like).

4. Case Report

A 35-year-old man presented to the dermatology outpatient clinic of the University Hospital in Sana'a, Yemen, with PW-like lesions over the dorsal sides of his hands, SK-like lesions over his face, erythematous macules, and PV-like lesions on both sides of the neck (Figures 1A-D, 2A-D, and 3A). The lesions emerged at 18 years of age. The patient was the oldest of 11 siblings (six brothers and five sisters), but except for the second and ninth brothers, no other siblings had similar lesions. However, when the lesions emerged, his father and his 17-year-old son had similar facial lesions.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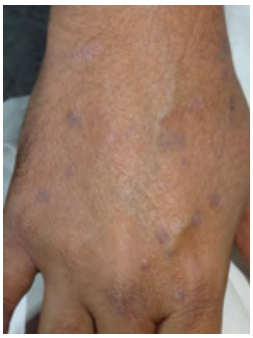
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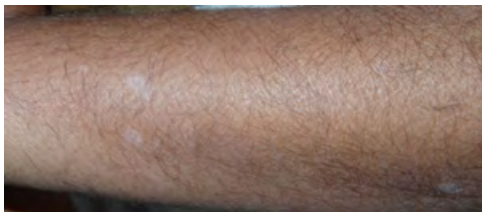
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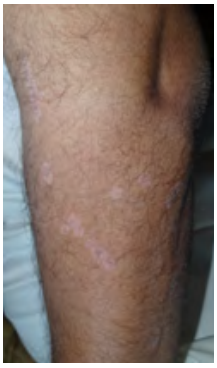
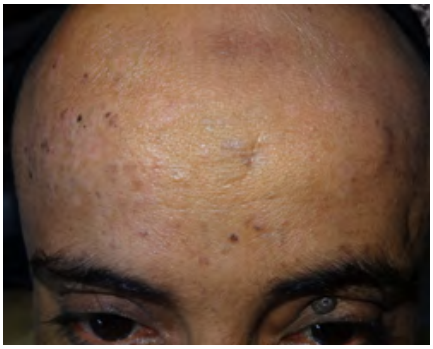


Figure 1D:

Figure 1: A. Brown flat papules resembling plane warts over the dorsum of the left hand. B. Similar lesions over the right hand. C. White hypopigmented macules simulating pityriasis versicolor over the lateral aspect of the left arm. D. Similar lesions over the anterior aspect of the left arm.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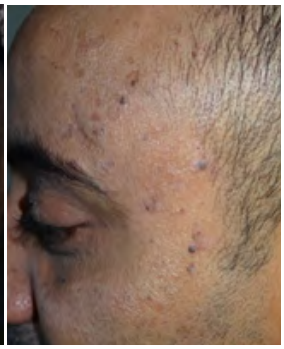
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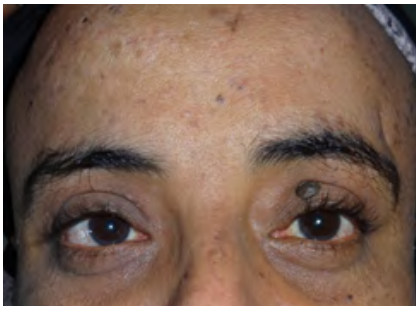
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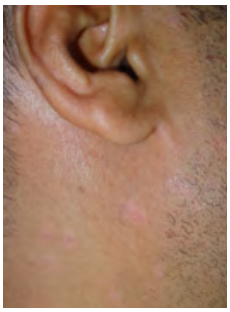


Figure 2D:

Figure 2: A. Flat hypo- and hyperpigmented plaques over the forehead. A compound nevocellular nevus over the left upper lid. B. Flat hyperpigmented plaques over the left temporal and frontal areas. C. Hypo- and hyperpigmented flat plaques over the forehead. Hypopigmented papules over the right upper lid. D. Light red atrophic, pityriasis-like macules over the right side of the neck.

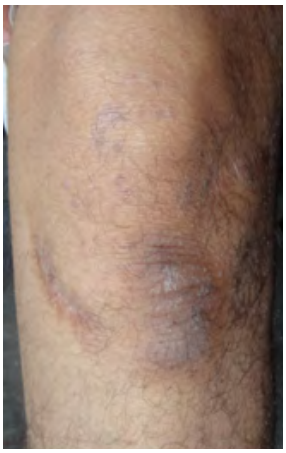
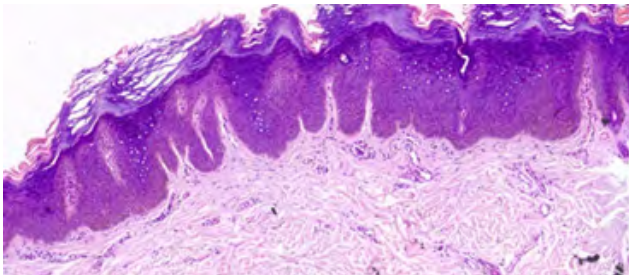


Figure 3A: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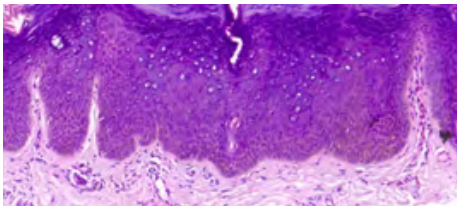
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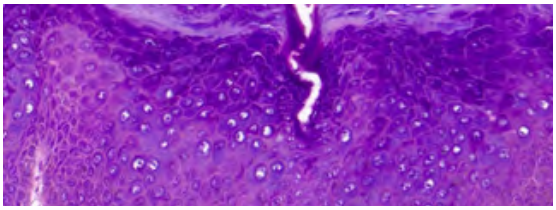
Figure 3D:

Figure 3: A. Hyperkeratotic plaques over the left knee. B. Overview of biopsies. Note the pale-stained cytoplasm of the large "dysplastic" keratinocytes visible within the stratum basale and stratum granulosum of the epidermis, a pathognomonic histological feature of EV (H & E ×40). C. Close-up view of B (H & E ×100). D. Close-up view of C (H & E ×400).

5. Discussion

EV is an autosomal recessive genodermatosis induced by the mutation of either EVER1 or EVER2. It presents with disseminated HPV infection, a predisposition to the development of malignant tumors, and cellular immunological abnormalities [1]. EV was first described by Lewandowsky and Lutz in 1922 [7]. Six cases of autosomal dominant inheritance and five of X-linked inheritance have been reported [8-11]. EV is very rare, with only 500 cases reported up to 2017 [12]. The congenital type of EV may be either familial or sporadic due to de novo mutation. The acquired type tends to appear in immunosuppressed individuals, such as human immunodeficiency virus (HIV) patients or transplant recipients. Many different types of HPV have been implicated in the development of cutaneous lesions in patients with EV, although it has been regarded as a model of cutaneous HPV oncogenesis and an example of genetic predisposition to viral-induced SCC [13]. Its prevalence does not differ based on race or geographical area [8]. PW-like lesions are the first to appear, mainly on the dorsal sides of the hands and fingers. This is generally followed by characteristic EV PV-like lesions a few years later. HPV5 and HPV8 are the most commonly identified HPV types found in EV-related cancers (up to 90%) [14]. Some PW-like HPV types (HPV3, HPV10) are associated with EV, while others are more specific to EV, including types 5, 8, 9, 12, 14, 15, and 17. Molecular techniques used to identify the type of HPV lesions in individual patients with EV who frequently express several HPV types can influence the most commonly recognized HPV types. Approximately 30–60% of patients with EV will develop SCC in sun-exposed areas; this frequently begins in the third and fourth decades of life [15]. HPV types 5 and 8 are the most commonly found in EV-associated SCCs, followed by HPV types 14 and 47 [14]. A lower level of interleukin-10 predisposes patients with EV to the development of SCC [15]. Malignant transformation of cutaneous epidermal lesions in EV is generally seen in connection with HPV types 5, 8, 17, 20, and 47 [15]. These lesions usually develop when patients are in their 30s, decades after the initial presentation. SCCs developing in patients with EV rarely metastasize, although they can be locally destructive [15]. Bowen disease, actinic keratoses, and basal cell carcinomas are common complications of EV. Malignant tumors rarely develop extracutaneously. Defective cell-mediated immunity is observed in patients with EV and in various immunosuppressed states, including HIV. EV is caused by an autosomal recessive mutation in either TMC6/EVER1 or TMC8/EVER2 on chromosome 17. These genes belong to the novel transmembrane channel-like (TMC) gene family and encode integral membrane proteins of the endoplasmic reticulum that regulate zinc metabolism. The gene defect of a second susceptibility locus has been mapped to chromosome 2, but the exact gene remains unknown. The existence of X-linked recessive transmission has been suggested based on reports from several other pedigree species with classical EV. Few cases of familial EV have been reported, although sporadic cases are the most commonly reported. Fourteen members of an EV pedigree among 41 individuals were reported from India, across several generations, with second-degree consanguinity.

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