JAMA Dermatology Clinicopathological Challenge

Linear Keratotic Lesions in a Young Woman

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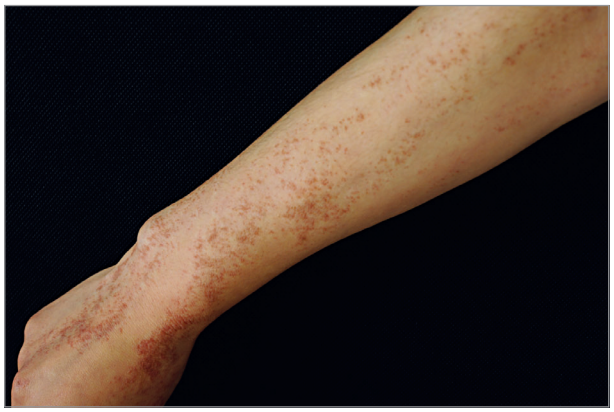

Figure 1. Reddish linear keratotic papules and plaques on the right forearm and dorsal hand distributed in a Blaschkoid pattern.

A woman in her 20s presented with linear hyperkeratotic papules on her right arm. The patient reported that these asymptomatic lesions were present since birth and did not appear to change on sun exposure or with seasons. She had no family history of similar cutaneous feature.

Physical examination revealed that these lesions were distributed in a Blaschkoid pattern. On close inspection, numerous reddish punctate keratotic papules were observed on the patient's right forearm that coalesced into plaques on the dorsum of her right hand (**Figure 1**). No extracutaneous abnormalities were found. A skin biopsy specimen from the patient's right arm was obtained.

WHAT IS YOUR DIAGNOSIS?

A. Epidermal nevus

B. Linear porokeratosis

C. Porokeratotic eccrine ostial and dermal duct nevus

D. Linear Darier disease

Diagnosis

C. Porokeratotic eccrine ostial and dermal duct nevus

Microscopic Findings and Clinical Course

Histologic examination revealed acanthosis, focal hyperkeratosis, and epidermal invagination. The eccrine ducts and ostia were hyperplastic and dilated (**Figure 2**A). The parakeratotic cornoid lamellae were found on the acrosyringium (Figure 2B). A diagnosis of porokeratotic eccrine ostial and dermal duct nevus (PEODDN) was made based on cutaneous and characteristic histopathologic findings. The diagnosis was further confirmed through genetic testing for *GJB2* (OMIM 121011), which was performed using DNA extracted from the affected epidermis and hyperplastic eccrine ducts by laser-captured microdissection. We found a mosaic missense mutation c.134G>A (p.Gly45Glu) in *GJB2*. Cryotherapy was recommended, but the patient refused.

Discussion

First described by Abell and Read[1] in 1980, PEODDN was considered to be an eccrine hamartoma. Clinically, it was featured with early-onset, grouped papules and plaques predominating on the extremities, with comedolike or spiny hyperkeratosis when located on the palms or soles. These lesions are commonly asymptomatic, and the distribution can be bilateral or unilateral with a linear pattern following the Blaschko lines. On histopathologic examination, PEODDN is characterized by cornoid lamellae connected with dilated, hyperplastic eccrine acrosyringium within an epidermal invagination.[2] There is no inflammatory infiltrate in the dermis, whereas hyperplastic eccrine ducts are mostly present together with normal ones.

Easton et al[3] provided the first evidence that mosaic mutations in *GJB2* could cause PEODDN. *GJB2* encodes connexin 26, a member of the gap junction protein family that is crucial for transmembrane communication.

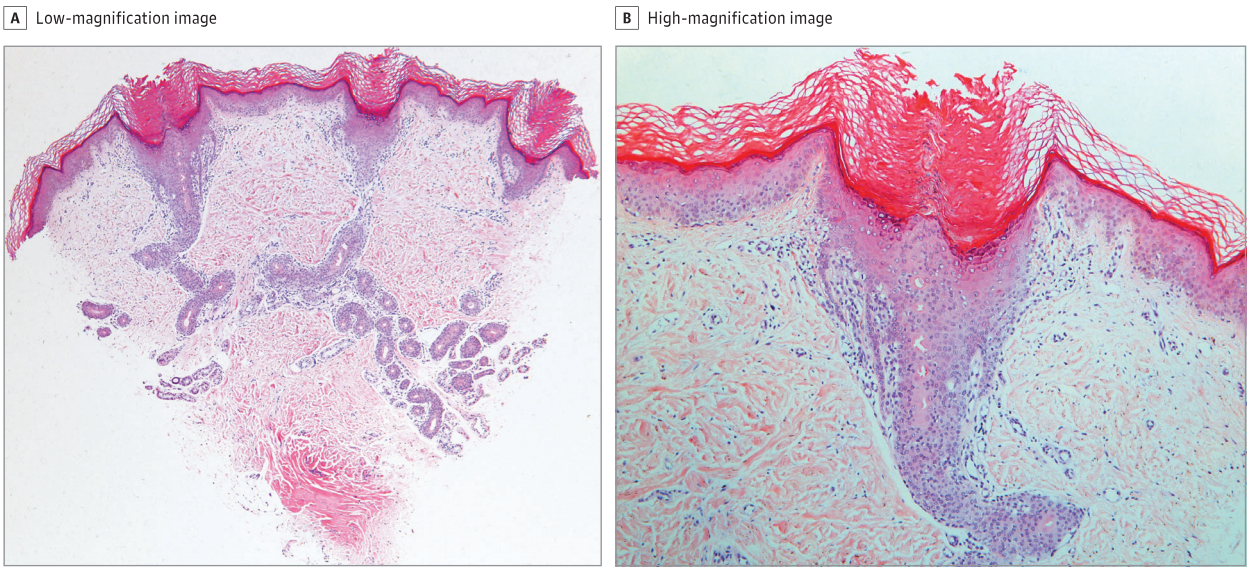

Figure 2. Histologic examination of the skin lesion. A, Hematoxylin-eosin, original magnification ×40. B, Hematoxylin-eosin, original magnification ×100.

Mutations in *GJB2* have been reported to cause keratitis-ichthyosis-deafness (KID) syndrome.[4] Both PEODDN and KID syndrome share a variety of features clinically and histopathologically; thus, PEODDN is considered to be a mosaic form of KID syndrome.

The molecular mechanism of PEODDN remains poorly understood. There is no definitive treatment for PEODDN to date; however, several different therapeutic approaches have been used with limited efficacy, such as topical steroids, anthralin, retinoids, cryotherapy, phototherapy, and surgery.[5]

ARTICLE INFORMATION

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