

Multiple proliferating pilar tumors with porokeratotic adnexal ostial nevus: A rare association



Jalal Maghfour, MS,^a Jennifer Kane, MD,^b Christopher DiMarco, MD,^b
Leslie Robinson-Bostom, MD,^b and Sandy Chai, MD^b
New Orleans, Louisiana, and Providence, Rhode Island

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INTRODUCTION

Porokeratotic adnexal ostial nevus (PAON) is a rare nonhereditary adnexal hamartoma of eccrine and hair follicle components, encompassing porokeratotic eccrine ostial and dermal duct nevus and porokeratotic eccrine and hair follicle nevus. Although PAON has been associated with other syndromes such as palmoplantar syndrome, it has been most commonly associated with a mosaic form of keratitis-ichthyosis deafness syndrome. This is due to mutations in the *GJB2* gene that encodes a gap junctional protein, connexin 26, which plays a role in keratinocyte growth and differentiation. The majority of PAON cases present as solitary lesions. Few reports discuss clinical features associated with PAON, which include psoriasis, alopecia, and focal anhidrosis.¹ To our knowledge, there has been no report on the association of PAON with proliferating pilar tumors, which are rare neoplasms arising from the isthmus of the outer root sheath. Histologically, they are characterized by a mix of squamoid and trichilemmal-type keratinization.²

CASE REPORT

A 33-year-old white woman presented with new-onset, asymptomatic, progressively enlarging nodules. Her medical history was unremarkable except for a PAON diagnosis since birth, with no clinical syndromic association. She denied a family history of similar lesions. Physical examination revealed 2- to 3-mm pink to brown keratotic papules coalescing into Blaschkoid linear plaques localized on the left side of the forehead, left ear, bilateral

Abbreviations used:

PAON: porokeratotic adnexal ostial nevus
SCC: squamous cell carcinoma

aspect of the arms, and lower portion of the legs (Fig 1, A and B). On the bilateral aspect of the palms and wrists, there were punctate pits with central keratotic plugs (Fig 1, C). Additionally, on the chin, left shoulder, right hip, and left buttock, there were solitary asymptomatic nodules ranging from 1 to 3 cm (Fig 2). Punch biopsies of the plaques were performed and histology revealed verrucous epidermal hyperplasia with several tiers of parakeratosis overlying dyskeratosis and hypogranulosis. The cornoid lamella was localized, overlying eccrine ducts and a dilated and distorted follicular infundibulum (Fig 3). Two nodules located on the left buttock and right hip were biopsied, which revealed proliferating pilar tumors with an associated apocrine malformation with ductal metaplasia (Fig 4). These lesions were then surgically excised because of associated discomfort. The remaining 2 nodules on the left shoulder and left shin were neither biopsied nor excised. However, they were clinically similar to the biopsied and excised nodules that were confirmed to be proliferating pilar tumor.

DISCUSSION

PAON may present clinically in 2 different ways. It can appear as comedolike papules with keratotic

From Tulane Medical School, Tulane University, New Orleans^a; and Department of Dermatology, Warren Alpert Medical School of Brown University, Providence.^b

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Correspondence to: Jalal Maghfour, MS, Tulane Medical School, Tulane University, 1430 Tulane Avenue, New Orleans, LA 70130. E-mail: jmaghfou@tulane.edu.

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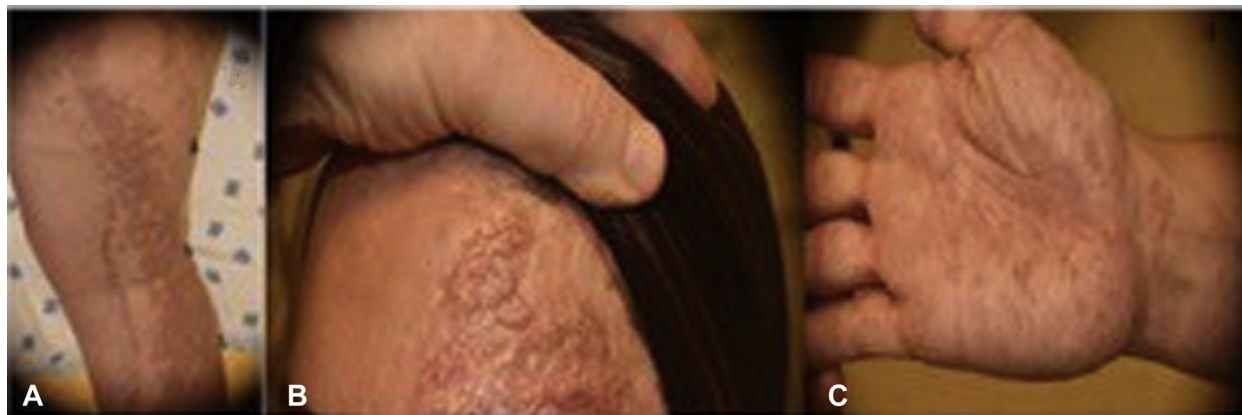


Fig 1. Shown are 2- to 3-mm pink to brown keratotic papules coalescing into Blaschkoid linear plaques localized on the lower leg (A) and left side of the forehead (B). On the bilateral aspect of the palms and wrists, there were punctate pits with central keratotic plugs (C).



Fig 2. Asymptomatic well-demarcated nodules ranging from 1 to 3 cm, involving the left shoulder (A), right hip (B), and chin (C).

plugs and central pits in palmoplantar distribution. It can also manifest as papules and plaques resembling linear verrucous epidermal nevi.³

The majority of PAON cases tend to be unilateral. Bilateral involvement is rare and it is most commonly observed in association with other syndromes. Other than keratitis-ichthyosis deafness, bilateral involvement in PAON has been observed with syndromes that contain a mutated variant of connexin 26. These include palmoplantar keratoderma, Vohwinkel syndrome, and Bart-Pumphrey syndrome.³

PAON remains a rare variant form of porokeratosis, which is a known risk factor for squamous cell carcinoma (SCC) and Bowen disease. Linear porokeratosis has the highest risk of developing into SCCs.⁴

Transformation of all forms of porokeratosis, including PAON, to SCC seems to be due to abnormal expression of p53, a tumor suppressor protein. Abnormal p53 is overexpressed in the majority of porokeratosis cases, with the highest concentration at the base of the cornoid lamella.^{5,6}

Additionally, the presence of cornoid lamella within SCC suggests the possibility that the

carcinoma develops secondary to abnormal proliferation of epithelial cells.⁷

We report a unique case of a patient who had extensive PAON with the subsequent development of numerous proliferating pilar tumors. The latter arise from pilar cysts as a result of trauma, irritation, or chronic inflammation; however, the mechanism remains unknown. A histologic hallmark of both proliferating pilar tumors and pilar cysts is trichilemmal keratinization. Proliferating pilar tumors are common in women older than 50 years. The tumors often present as solitary nodules with a predilection for the scalp.⁸

On histology, proliferating pilar tumors may exhibit cytologic atypia. In our case, no atypia was appreciated, with a background apocrine malformation.

Rarely, proliferating pilar tumors may undergo malignant transformation. Lesions arising in locations other than the scalp have a higher risk of becoming malignant, with possible lymph node metastases. Characteristics of malignant proliferating pilar tumors include rapid growth, diameter greater than 5 cm, increased mitotic activity, and atypia on histology.⁸

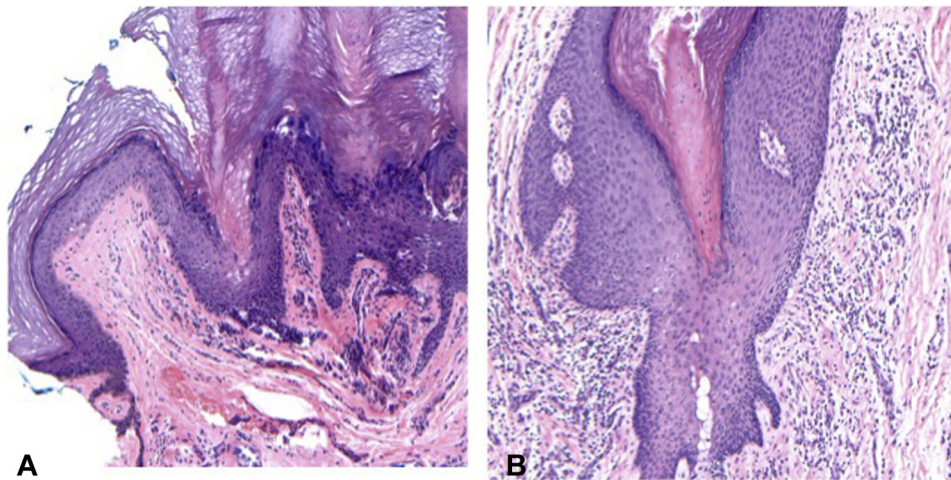


Fig 3. Histopathology of a keratotic papule (shown at low power) revealing verrucous epidermal hyperplasia with several tiers of parakeratosis overlying dyskeratosis and loss of granular layer. Cornoid lamella overlies the eccrine duct (**A**). Histopathology of another keratotic papule revealing cornoid lamella overlying a dilated and distorted hair follicle (**B**).

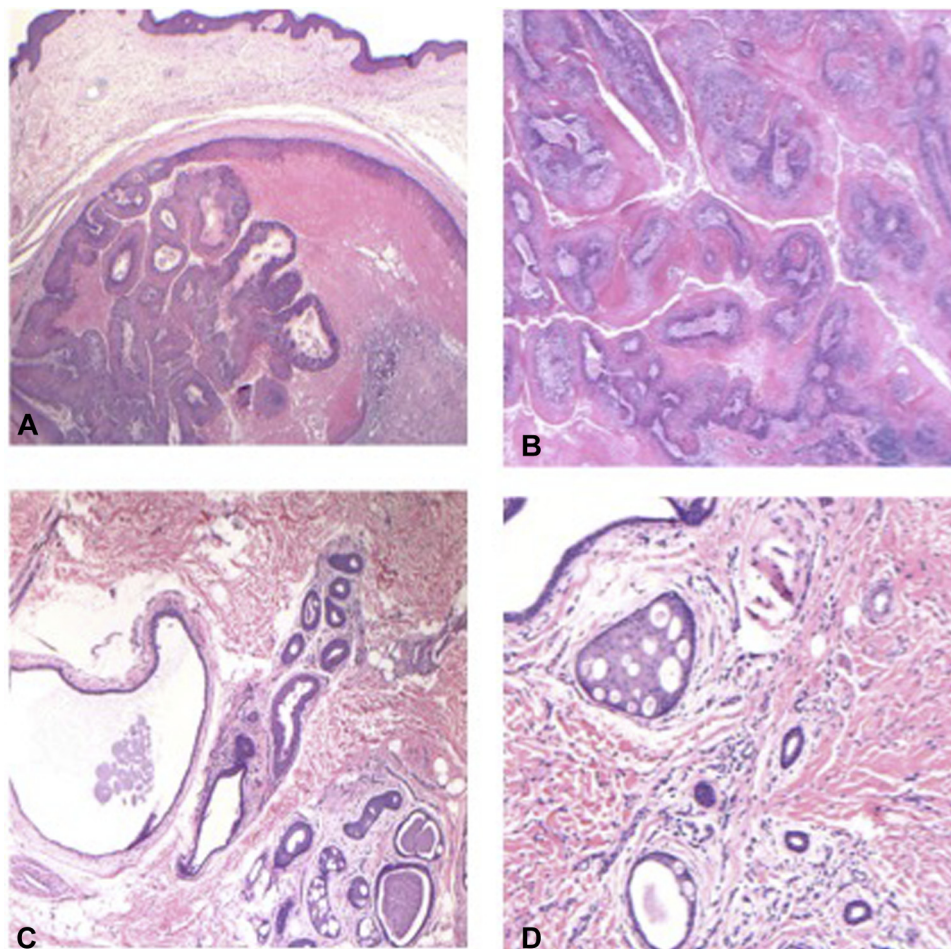


Fig 4. Scanning magnification of proliferating pilar tumor (**A**). Histopathology of 1 of the nodules revealed a well-circumscribed nodule within the deep dermis. The nodule was composed of squamous epithelium without atypia, forming interlacing lobules with abrupt keratinization and without a granular layer (**A** and **B**). Another tumor was biopsied and showed similar features with associated apocrine malformation with ductal metaplasia (**C** and **D**). (**A**, Original magnification: $\times 2$.)

Because these lesions can be aggressive, with higher recurrence rate and metastatic potential, Mohs micrographic surgery appears to be a therapeutic option.⁹

In summary, we describe a rare case of widespread, bilateral PAON associated with multiple proliferating pilar tumors. Although this may have been a coincidental finding, given the development of multiple proliferating pilar tumors in association with PAON, an unknown syndromic association is possible.

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