An unusual spiculated presentation of follicular porokeratosis

Paulina M Young BS, Joan Leavens MD, Sara Gaspard MD, Gene Kim MD, April W Armstrong MD MPH

Affiliations: Department of Dermatology, Keck School of Medicine of the University of Southern California, Los Angeles, California, USA

Corresponding Author: April W. Armstrong MD, MPH, 1975 Zonal Avenue, KAM 510, Los Angeles, California 90089, Email: armstrongpublication@gmail.com

Abstract

Follicular porokeratosis is a rare variant of porokeratosis in which the coronoid lamellae are confined to hair follicles. It classically presents with annular plaques with a surrounding keratotic ridge. However, the clinical presentation has shown significant variation in reported cases. We present the case of a 26-year-old man with chronic diffuse follicular spicules associated with alopecia. Clinical findings were suggestive of trichodysplasia spinulosa, but multiple biopsies showed findings consistent with follicular porokeratosis. Diffusely spiculated presentation as present in our patient has not been previously reported in the literature. It is important to recognize the necessity of histopathologic confirmation in a diagnostically challenging condition such as follicular porokeratosis.

Keywords: follicular porokeratosis, porokeratosis, follicular spicules, trichodysplasia spinulosa

Introduction

Porokeratosis is a disease of abnormal keratinization that presents clinically with annular plaques with a surrounding keratotic ridge [1]. It was first described by Vittorio Mirabelli in 1893 [2]. Several different subtypes of porokeratosis exist, all sharing the common histologic finding of cornoid lamellae [3].

In recent years, a new variant in which cornoid lamellae are confined to follicles, termed follicular porokeratosis, has been described. The diagnosis of follicular porokeratosis is based on the histologic presence and distribution of the cornoid lamella. However, the clinical presentation has not been as definitive, making diagnosis a challenge in the clinical setting.

We report the unusual case of a young man with an 8-year history of diffuse follicular spicules associated with alopecia, considered initially by clinical examination to be trichodysplasia spinulosa but with histopathologic features consistent with follicular porokeratosis.

Case Synopsis

A 26-year-old man with Sturge-Weber syndrome and hidradenitis suppurativa presented to the dermatology clinic for management of diffuse follicular spicules on the scalp, face, and trunk associated with alopecia. The follicular spicules had been present stably for approximately 8 years and were associated with xerosis and intermittent pruritus.

Physical examination of the scalp revealed large geometric alopecia with follicular spicules over the frontal region and vertex (**Figure 1**). There was a violaceous patch located in the left V1 distribution associated with mild left-sided facial hypertrophy, consistent in appearance with a port-wine stain. The

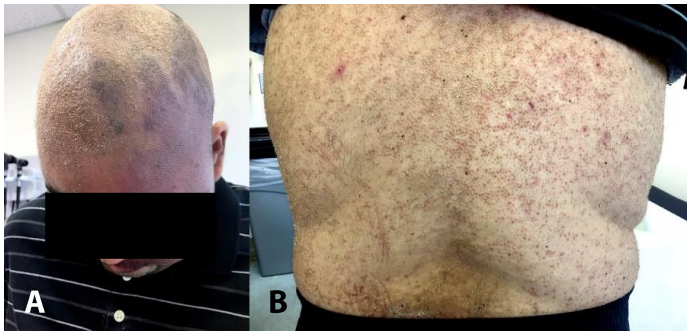

Figure 1. ***A)*** *Scalp with follicular spicules and alopecia.* ***B)*** *Trunk with diffuse follicular spicules.*

nose, chin, and ears had numerous spicules located at the follicular orifices. Follicular prominence with spicules were located diffusely over the trunk, arms, and lower legs (**Figure 1**).

Laboratory studies included a complete blood count remarkable for WBC 11.2. Basic metabolic panel was unremarkable. An SPEP ordered previously was within normal limits. Vitamin A levels were also within the normal range. Clinical findings are suggestive of trichodysplasia spinulosa. However, the characteristic viral changes were absent on electron microscopy. Multiple punch biopsies were performed over several years, each revealing follicular hyperkeratosis with perifolliculitis and narrow columns of parakeratosis associated with dyskeratotic keratinocytes, consistent with cornoid lamellae (**Figure 2**). The patient was diagnosed with follicular porokeratosis based on the histologic findings.

Multiple punch biopsies were performed over several years, each revealing follicular hyperkeratosis with perifolliculitis (**Figure 2**). The patient was diagnosed with follicular porokeratosis based on the histologic findings.

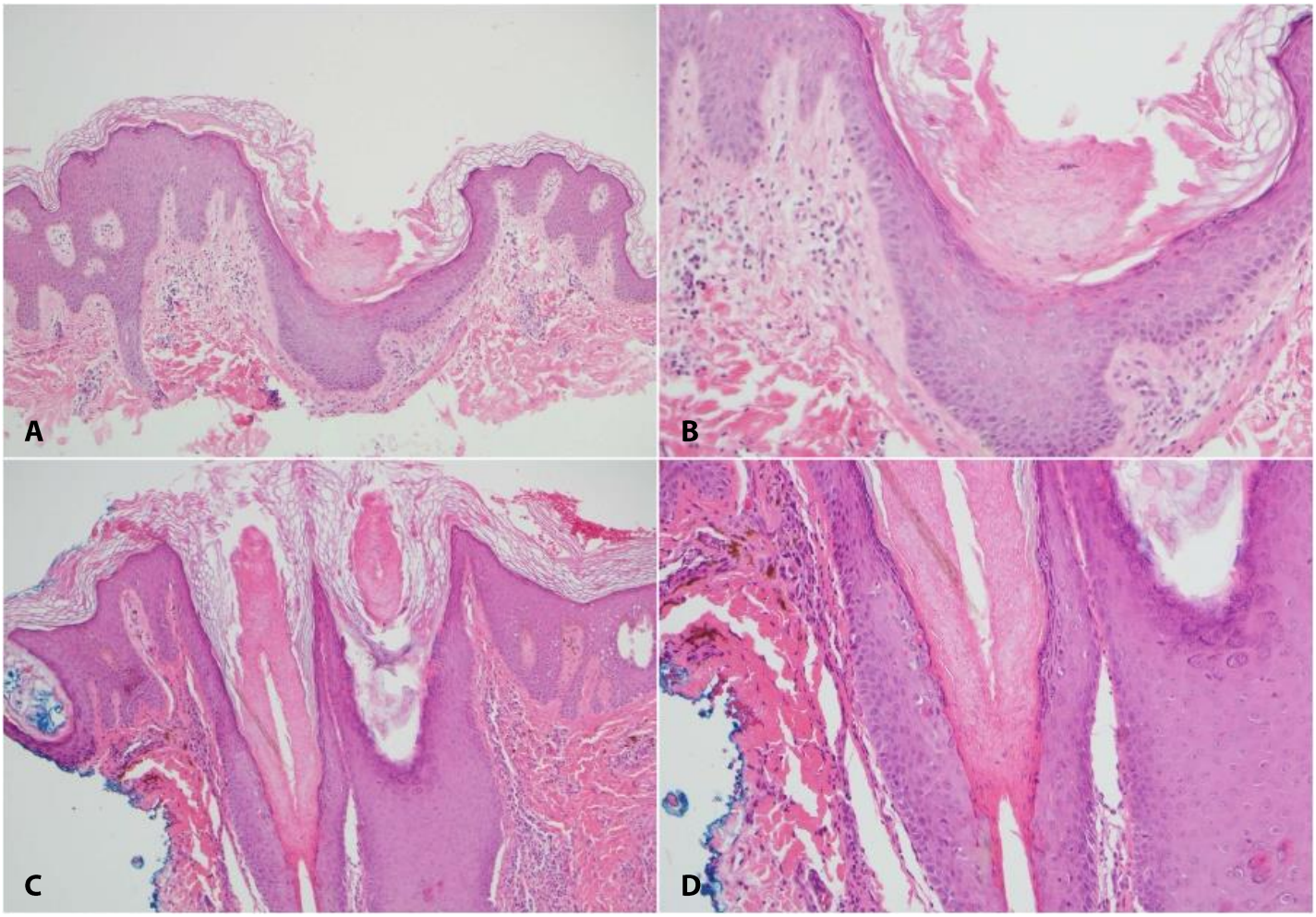

Figure 2. *Two separate biopsy specimens both showing a tier of parakeratosis in the follicular infundibulum with associated hypogranulosis and dyskeratosis.* ***A)*** *Erythematous papule with hyperkeratotic spicule, left flank H&E, 100×;* ***B)*** *200×;* ***C)*** *Right scalp 100×,* ***D)*** *200×.*

Past treatments were overall unsatisfactory and included tretinoin cream, which was drying for the patient. Amlactin cream and ketoconazole shampoo were also tried without significant improvement. He was then restarted on acitretin, which the patient had previously tried and discontinued because of skin abscesses and spiculization. The patient again could not tolerate the acitretin, as it worsened his xerosis and pruritus. Since discontinuing acitretin, the patient has been on a regimen of ketoconazole shampoo for the scalp and gentle moisturization for the body. Another topical medication will likely be added following resolution of the active dryness and irritation.

Case Discussion

Porokeratosis is a disorder of keratinization presenting clinically as annular plaques surrounded by a keratotic ridge [4]. The histopathologic hallmark of this disease is the presence of cornoid lamella [2]. This can be seen in biopsy samples as a narrow column of parakeratosis with underlying dyskeratosis and loss of the granular layer [3]. Several subtypes of porokeratosis have been recognized to date. These include porokeratosis of Mibelli, disseminated superficial actinic porokeratosis, superficial disseminated porokeratosis, porokeratosis plantaris palmaris et disseminata, and linear porokeratosis [1]. Rarer variants of porokeratosis also exist, including porokeratosis ptychotropica and giant porokeratosis [5, 6].

Another rare variant of porokeratosis, in which the cornoid lamellae are exclusively limited to the follicular infundibula, was formally termed follicular porokeratosis in 2009 [7]. There have been fewer than 20 reported cases of follicular porokeratosis in the existing literature [1, 2]. Factors involved in pathogenesis may include UV radiation and genetic predisposition [2]. Other proposed associations include infection (i.e. Hepatitis **C)**, certain drugs, and immunosuppression [7]. There is a slight male-to-female predominance in reported cases. The age range of patients is broad, spanning from pediatric to the elderly [1, 2]. Symptoms can include pruritus, but there have been several asymptomatic cases as well [1, 2].

No treatment is known to be effective, as several methods has been tried with variable degrees of success. These include topical medications such as imiquimod 5% cream, keratolytics, and 5-FU [8]. Excision as well as curettage and cautery have been tried in patients with fewer lesions. In the case reported by Trikha et al., the lesions diagnosed as follicular porokeratosis were treated with imiquimod 5% cream daily for two weeks with little improvement noted, possibly because the medication was unable to penetrate the cornoid lamella located deep within the follicle [9].

The clinical appearance of follicular porokeratosis was first described as multiple, small (<10mm), persistent or static, follicle-centered lesions in either a photo- or non-photodistributed area [7]. Another common description is of erythematous or brown annular plaques with a peripheral keratotic ridge [8]. However, within these broad parameters, there have been a wide variety of presentations. Sud et al. presented four different cases of follicular porokeratosis with shared histologic findings but contrasting clinical presentations. For example, one case was similar in appearance to a solar lentigo, whereas another was suspected to be a facial basal cell carcinoma [8]. Based on existing reports, there does not seem to be a coherent clinical phenotype to the histologic diagnosis of follicular porokeratosis.

Similarly, our patient's presentation differed significantly from the classic lesions of follicular porokeratosis. He presented with pronounced follicles, each with a prominent spicule and associated alopecia (**Figure 1**). These were located diffusely over the trunk, face, and limbs. The spiculated lesions more closely resemble those of trichodysplasia spinulosa, which presents with thousands of folliculocentric, monomorphous papules with fine keratotic spicules and progressive alopecia [10]. Other conditions with a similarly spiculated presentation were also considered but were ruled out based on the clinical history (**Table 1**), [11, 12]. On histology, the biopsied lesions showed follicular changes consistent with follicular porokeratosis while lacking the characteristic trichohyalin granules of trichodysplasia spinulosa (**Figure 2**). Additionally, no polyoma viral particles

were observed under electron microscopy. Ultimately, the diagnosis of follicular porokeratosis rested on the histopathologic findings.

We considered the possibility that there could be an association between Sturge-Weber Syndrome and follicular porokeratosis. However, upon a review of the existing literature, we could not find any genetic, epidemiologic, or pathophysiologic associations between the two conditions. Reported cutaneous manifestations of Sturge-Weber are all vascular in nature, with a facial port-wine stain being the syndrome's dermatologic hallmark [13-15]. It is likely that our case is an isolated incident of concurrent Sturge-Weber Syndrome and follicular porokeratosis, but there remains a possibility that there is a previously unreported correlation between these disease processes.

Conclusion

Our patient is one of the few reported cases of porokeratosis with exclusive follicular involvement. Ours' is also one of the presentations that diverges from the classically described phenotype of follicular porokeratosis, that of erythematous plaques with a keratotic ridge [8]. Our patient's unusual spiculated presentation has not been previously reported in the literature. There is a standing question as to whether follicular porokeratosis is a distinct clinical entity or rather a histologic variant of porokeratosis [2]. Along with the cases presented by Sud et al., our case suggests that there may not be a consistent clinical phenotype for follicular porokeratosis. However, further studies are needed, given that understanding of the topic remains limited. The presented case contributes to the existing knowledge base available on follicular porokeratosis. It reflects the diagnostic challenges of this rare condition in the clinical setting and emphasizes the necessity of histopathologic confirmation.

Potential conflicts of interest

The authors declare no conflicts of interest.

References

1. Zhao M, Sanusi T, Zhao Y, et al. Porokeratosis with follicular involvement: report of three cases and review of literatures. *Int J Clin Exp Pathol*. 2015;8:4248-52. [PMID: 26097620].
2. Sun R, Chen H, Lian S, Zhu W. Follicular porokeratosis: a case study and literature review. *Eur J Dermatol*. 2017;27:332-4. [PMID: 28677581].
3. Kim J, Wood BA, Harvey NT. Follicular porokeratosis of the nose: two further cases of an emerging variant of porokeratosis. *Pathology*. 2015;47:482-5. [PMID: 26126043].
4. de Almeida HL, Jr., Guarenti IM, de Castro LA, Rocha NM. Follicular involvement in porokeratosis. *J Eur Acad Dermatol Venereol*. 2007;21:109-11. [PMID: 17207181].
5. Yeo J, Winhoven S, Tallon B. Porokeratosis ptychotropica: a rare and evolving variant of porokeratosis. *J Cutan Pathol*. 2013;40:1042-7. [PMID: 24274427].
6. Avhad G, Jerajani H. Porokeratosis of Mibelli: Giant variant. *Indian Dermatol Online J*. 2013;4:262-3. [PMID: 23984259].
7. Pongpudpunth M, Farber J, Mahalingam M. Follicular porokeratosis: distinct clinical entity or histologic variant? *J Cutan Pathol*. 2009;36:1195-9. [PMID: 19519877].
8. Sud A, Shipman AR, Odeke M, et al. Follicular porokeratosis: four new cases. *Clin Exp Dermatol*. 2017;42:881-6. [PMID: 28748571].
9. Trikha R, Wile A, King J, et al. Punctate follicular porokeratosis: clinical and pathologic features. *Am J Dermatopathol*. 2015;37:e134-6. [PMID: 26485244].
10. Kirchhof MG, Shojania K, Hull MW, et al. Trichodysplasia spinulosa: rare presentation of polyomavirus infection in immunocompromised patients. *J Cutan Med Surg*. 2014;18:430-5. [PMID: 25348766].
11. Caccetta TP, Dessauvagie B, McCallum D, Kumarasinghe SP. Multiple minute digitate hyperkeratosis: a proposed algorithm for the digitate keratoses. *J Am Acad Dermatol*. 2012;67:e49-55. [PMID: 21050621].
12. Hwang S, Schwartz RA. Keratosis pilaris: a common follicular hyperkeratosis. *Cutis*. 2008;82:177-80. [PMID: 18856156].
13. Chernoff KA, Schaffer JV. Cutaneous and ocular manifestations of neurocutaneous syndromes. *Clin Dermatol*. 2016;34:183-204. [PMID: 26903185].
14. Klar N, Cohen B, Lin DDM. Neurocutaneous syndromes. *Handb Clin Neurol*. 2016;135:565-89. [PMID: 27432683].
15. Purkait R, Samanta T, Thakur S, Dhar S. Neurocutaneous syndrome: a prospective study. *Indian J Dermatol*. 2011;56:375-9. [PMID: 21965842].

Table 1. *Differential diagnosis of diffuse cutaneous spicules.*

	Pathogenesis	Distribution	Clinical Presentation	Histopathology	Associations
Trichodysplasia spinulosa [10][11]	Trichodysplasia spinulosa polyomavirus (TSPyV)	Mainly ears and face, can involve trunk and limbs	Folliculocentric papules with keratotic spiny projections, often with alopecia	Dilated hair follicles with proliferation of inner root sheath cells containing large trichohyaline granules Electron microscopy (EM): intracellular polyomavirus particles	Immunosuppression (usually due to therapies for solid organ transplant or hematologic malignancies)
Multiple minute digitate hyperkeratosis [11]	Unknown	Trunk and limbs, sparing face and palmoplantar surfaces	White, yellow, brown, or skin-colored spicules; sometimes flat-topped, dome-shaped, or crateriform papules	Focal columns of orthokeratotic hyperkeratosis arising from tented epidermis EM: reduced keratohyaline granules and normal or increased Odland bodies	N/A
Lichen spinulosus [11]	Unknown	Mainly trunk and limbs	Follicular keratotic papules with horny spines grouped into plaques	Follicular keratin plugs with dense lymphocytic perifollicular infiltrate	N/A
Hyperkeratotic spicules [11]	Immunoglobulin deposition	Mainly face (especially nose), but may involve scalp, trunk, and limbs	Follicular or non-follicular hyperkeratotic spicules	Focal columns of orthokeratotic or parakeratotic hyperkeratosis with homogenous compact eosinophilic inclusions	Paraproteinemia, multiple myeloma, cryoglobulinemia
Keratosis pilaris [12]	Unknown	Mainly extensor surfaces of upper arms, thighs, buttocks	Grouped folliculocentric keratotic papules with a variable degree of perifollicular erythema	Orthokeratotic keratin plugs inside dilated hair follicles May be mild perivascular lymphocytic infiltrate in upper dermis	Atopic dermatitis, ichthyosis vulgaris
Phrynoderma [11]	Various nutritional deficiencies (often Vitamin A)	Posterolateral upper arms and anterolateral thighs, can spread to elbows, knees, and neck.	Hyperkeratotic follicular papules	Hyperkeratosis with prominent follicular plugging	N/A
Spiny keratoderma [11]	Unknown	Palms and/or soles	Discrete keratotic plugs	Focal columns of orthokeratotic or parakeratotic hyperkeratosis overlying hypogranular epidermis EM: reduced keratohyaline granules and normal Odland bodies	Malignancy