A Rare Case of Porokeratosis Ptychotropica in A 27- Year-Old Female – A Case Report

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Abstract

Porokeratosis is an uncommon keratinization disorder of the skin that manifests as keratotic papules or annular plaques that spread centrifugally with a raised border. The cornoid lamella is a distinctive diagnostic feature of this disorder. It consists of columns of parakeratosis overlying the epidermis with associated hypogranulosis and scattered dyskeratotic keratinocytes below the corneal layer.

Ptychotropic porokeratosis (verrucous porokeratosis) is a recently classified but rare clinical variant, found in the genitogluteal region, including the natal cleft and buttocks but also seen in the scrotum, penis or vulva. It may be misdiagnosed for several years as it does not have the characteristic clinical features described in classical porokeratosis. However, a biopsy often yields the diagnosis.

In this report, we present a rare case of porokeratosis ptychotropica in a 27-year-old female who presented with recurrent, itchy hyperkeratotic plaques in the inner thighs and perineal area with distinct histological findings, the first case in African literature. The patient was treated with intralesional administration of 5-fluorouracil administered at intervals for 3 months with significant improvement. A literature search revealed less than 50 cases of genitogluteal porokeratosis are documented worldwide, with none reported in Africa.

Keywords: Porokeratosis, Ptychotropica, Verrucous, Cornoid lamellae, Genitogluteal

Un cas rare de Porokératose ptychotrope chez une femme de 27 ans : à propos d'un cas

Résumé

La porokératose est un trouble de kératinisation rare de la peau qui se manifeste par des papules kératosiques ou des plaques annulaires qui s'étendent de manière centrifuge avec un bord surélevé. La lamelle cornoïde est une caractéristique diagnostique distinctive de cette pathologie. Elle se compose de colonnes de parakératose recouvrant l'épiderme avec une hypogranulose associée et des kératinocytes dyskératosiques dispersés sous la couche cornéenne.

La porokératose ptychotrope (porokératose verruqueuse) est une variante clinique récemment classée mais rare, trouvée dans la région génito-fessière, y compris la fente natale et les fesses, mais également observée sur le scrotum, le pénis ou la vulve. Elle peut ne pas être diagnostiquée pendant plusieurs années car ne présentant pas les caractéristiques cliniques décrites dans la porokératose classique. Cependant, une biopsie permet souvent de poser le diagnostic.

Dans cette observation, nous rapportons un cas rare de porokératose ptychotrope chez une femme de 27 ans qui présentait des plaques hyperkératosiques récurrentes et prurigineuses à la face interne des cuisses et dans la région périnéale avec des signes histologiques typiques, le premier cas rapporté dans la littérature africaine. La patiente a été traitée par administration intralésionnelle de 5-fluorouracile administré à intervalles de 3 mois avec une amélioration significative. Une recherche dans la littérature a révélé que moins de 50 cas de porokératose génito-fessière sont documentés dans le monde, aucun n'ayant été signalé en Afrique.

Mots-clés : Porokératose, Ptychotrope, Verruqueux, Lamelles cornoïdes, Génito-fessière

Introduction

Porokeratosis is a primary disorder of keratinization encompassing a heterogeneous group of hereditary and acquired disorders of the clonal proliferation of keratinocytes characterized by dyskeratotic cells and

abnormal keratinocyte maturation.[1] The Austrian dermatologist Isidor Neuman, in 1875, first described the condition. Still, the Italian dermatologist Vittorio Mibelli named it in 1893.[2] It is an uncommon dermatologic disorder that presents with keratotic papules or annular plaques with a raised border that radiates outwards from an indented centre[3] and has been equated to the Great Wall of China with a ridge and moat on its peak.[4]

Porokeratosis has a distinct hallmark of cornoid lamella histologically, a column of tightly fitted parakeratotic cells in the epidermis.[1,5] Usually, genitogluteal porokeratosis involving the genital region, buttocks, perineum, groin and proximal thighs may occur as part of a generalized porokeratosis.[6,7] However, lesions localized to the genitogluteal region are rare; only a few cases are reported in the literature.[4]

Porokeratosis is a precancerous lesion, and any of the variants can undergo malignant transformation, mostly into squamous cell carcinoma and rarely basal cell carcinoma.[8] Treatment modalities such as topical, systemic, and surgical therapies have been reported. However, there are no standard guidelines for treatment.[9]

The literature search revealed that by May 2018, less than 50 cases of genitogluteal porokeratosis had been described worldwide,[4] while none had been reported in Africa. Only one case of generalized verrucous porokeratosis was reported from Africa. We report genitogluteal verrucous porokeratosis (porokeratosis ptychotropica) in a 27-year-old female, the only case reported in Africa, and evaluate the diagnostic histopathological features.

Case Report

We report the case of a 27-year-old healthy female who presented with complaints of severe pruritus and several plaques on the inner parts of the thigh and around the perineum. The plaques had increased progressively in size over two years. The plaques appeared hyperkeratotic, verrucous, and scaly, with well-defined borders on clinical presentation (figures 1A, 1B). She had no previous history of skin disorders or other systemic illnesses, and there was no family history of similar lesions. She was treated with topical corticosteroids with no improvement.

We made a clinical impression of viral warts, and the differential was hyperkeratotic lichen planus. Punch biopsies were sent for histologic assessment and showed skin tissue characterized by a hyperkeratotic and acanthotic epidermal layer with prominent hypergranulosis and papillomatosis (figure 2). Many areas show characteristic cornoid lamellae consisting of columns of parakeratosis overlying the epidermis (figure 3) and associated with hypogranulosis and scattered dyskeratotic keratinocytes below the corneal layer (figure 4). Based on the correlation of the histological and clinical findings, a diagnosis of

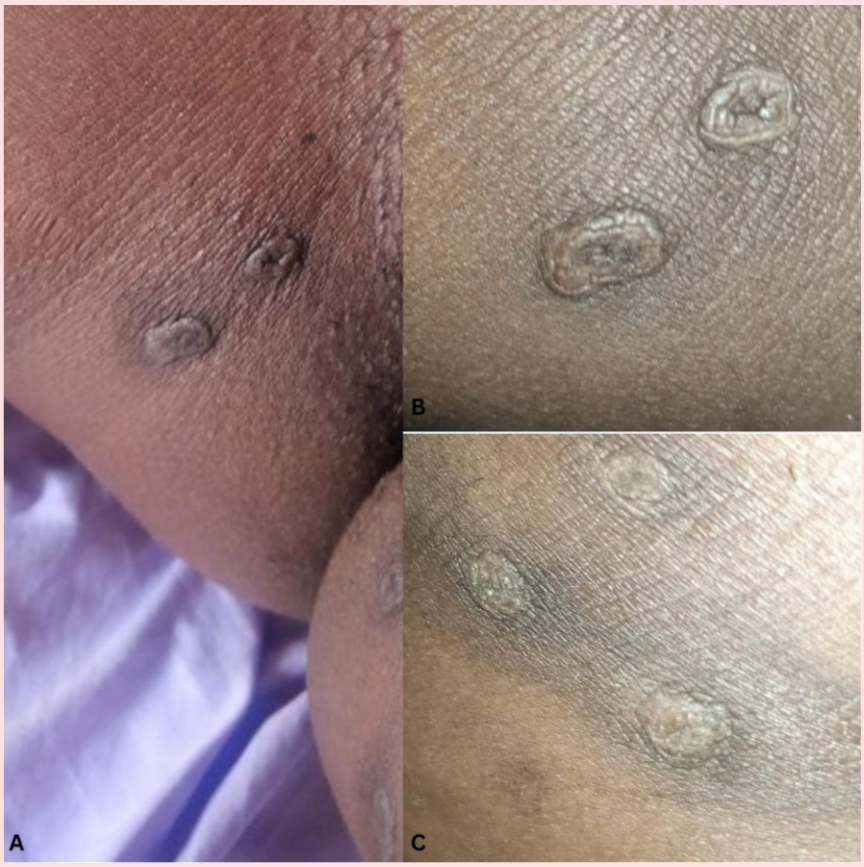

Figure 1: (A) Clinical image showing multiple annular, hyperkeratotic, scaly and verrucous plaques in the perineum. **(B)** A close-up view of the plagues shows central dimpling. **(C)** After one month of treatment with intralesional 5-fluorouracil, the surfaces are more flat and less verrucous.

verrucous porokeratosis (porokeratosis ptychotropica) was made. The patient was treated with intralesional 5-fluorouracil, administered every 2 weeks for 3 months. The itching significantly improved after a month of administration, and the lesions flattened out (figure 1C). Subsequently, she stopped attending the follow-up clinic.

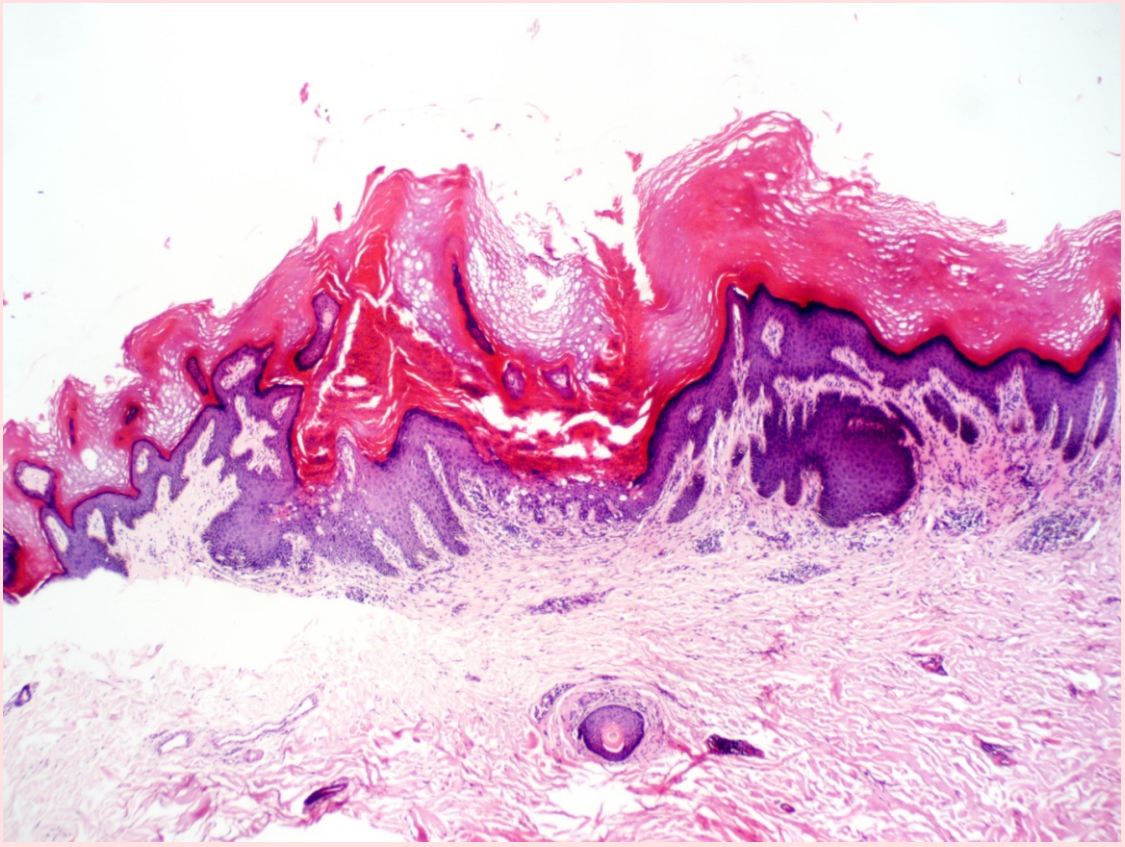

Figure 2: Histologic section at low power shows hyperkeratotic, acanthotic, and papillomatous epidermal layers with areas of prominent hypergranulosis. H&E x40

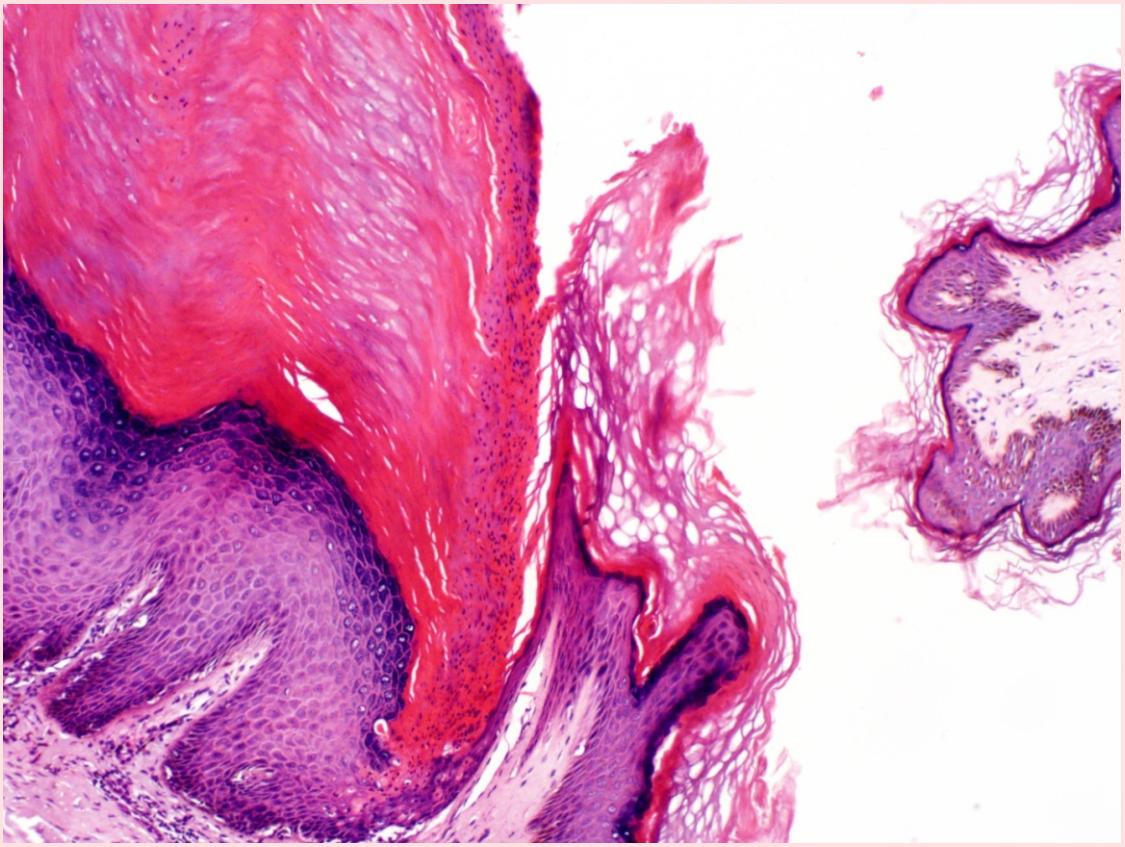

Figure 3: Histologic section at medium power showing an angulated cornoid lamella composed of a tall column of parakeratosis. The underlying epithelium shows loss of the granular layer. H&E x100

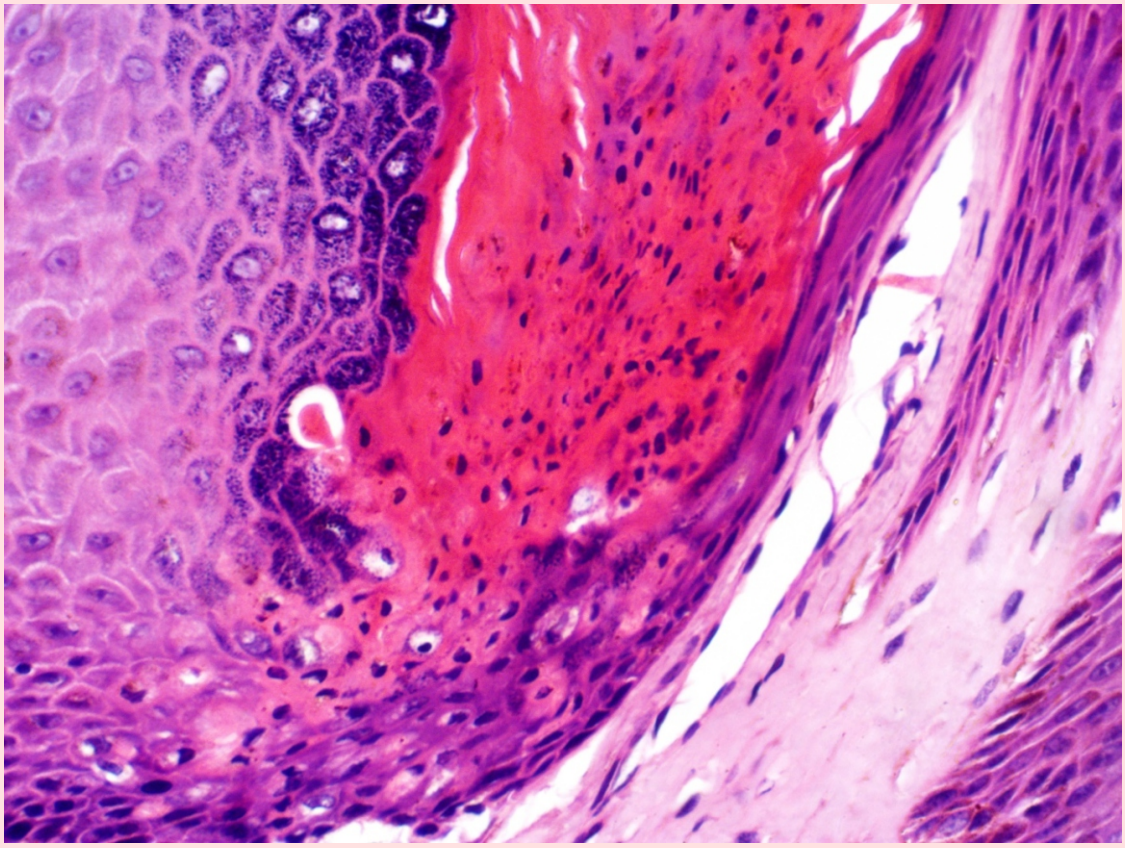

Figure 4: The high-power section of Figure 2 shows the base of the parakeratotic column, characterized by hypogranulosis and composed of vacuolated dyskeratotic cells. H&E x400

Discussion

Genitogluteal porokeratosis was first described in 1985; it is a rare variant of porokeratosis characterized by the development of verrucous plaques in the genital region, buttocks, perineum, groin, and proximal thighs.[10] This case report presents a 27-year-old female with porokeratosis ptychotropica, a specific subtype of genitogluteal porokeratosis.

The term "genitogluteal porokeratosis" was first introduced in 1985 to describe cases of porokeratosis affecting the genital region. Subsequently, the term "porokeratosis ptychotropica" was coined in 1995 to refer to the flexural location of this condition specifically.[11]

Porokeratosis ptychotropica typically presents as slowly growing, pruritic, verrucous plaques in the natal cleft, buttocks, penis, scrotum, vulva, and thighs.[12] It is a relatively uncommon condition resulting from the disordered progression of epidermal cells,[13] with fewer than 50 reported cases worldwide. This present patient is a young female; the age at presentation is consistent with some previously reported cases; however, most studies suggest a male predominance, with 90% of cases occurring in males aged 27 to 84 years. The gluteus is the most commonly affected area, followed by the genitogluteal region and the gluteal region involving the extremities.[14] Although it is mainly restricted to these regions, ptychotropic porokeratosis has been described with lesions in other body parts.[15]

While the exact pathogenesis of this lesion remains unclear, sun exposure and repeated minor frictional trauma due to tight clothing are implicated as potential risk factors.[4] In this case, the patient presented with itchy, verrucous plaques, leading to a clinical suspicion of viral warts and a differential diagnosis of hypertrophic lichen planus. However, the presence of cornoid lamellae on histopathological examination was crucial for the correct diagnosis. Porokeratosis ptychotropica is often misdiagnosed as psoriasis, chronic eczema, common warts, lichen simplex chronicus, dermatophytosis, or candidiasis due to its lack of the classic peripheral ridge and moat appearance seen in other types of porokeratosis.[4]

The dermoscopic examination can be helpful; in porokeratosis ptychotropica, dermoscopy often reveals sharply demarcated annular lesions with a thick peripheral brown rim and a regular dotted vessel

pattern. These findings differ from those in psoriasis, dermatophytosis, and lichen simplex chronicus.[16] In contrast, the dermoscopy of verruca vulgaris shows papillomatosis and dilation of the capillaries of the dermal papillae, and hyperkeratosis may occur depending on the clinical presentation of the lesion.[17] However, in porokeratosis ptychotropica, only hyperkeratosis is observed, and the other components of the viral warts are absent.

Histopathological examination is essential for the definitive diagnosis of porokeratosis ptychotropica.[12,14,15] The key diagnostic feature is the presence of multiple cornoid lamellae within the epidermis, as observed in this case. Other histological findings may include acanthosis, hypergranulosis, and scattered dyskeratotic keratinocytes. The limited number of reported cases of genitogluteal porokeratosis in this part of the world may be attributed to misdiagnosis and restricted access to healthcare facilities.

Porokeratosis is a premalignant condition with a risk of malignant transformation, primarily into squamous cell carcinoma. Molecular studies show the overexpression of p53 associated with this disorder, and occasionally, clonal expansion of abnormal epidermal keratinocytes can be seen.[9] The risk of malignant transformation is estimated to be between 6.9% and 30%,[5,9] with certain factors, such as older age, history of radiation therapy, chronic lesions, lesions on extremities, and the linear variant, increasing the risk.[18,19] Less frequently, malignant transformation into basal cell carcinoma may occur. If the lesion has not undergone a malignant transformation, excision is curative.[20] Of all the variants of porokeratosis, the linear variant and giant porokeratosis are the most susceptible to malignant transformation. Malignant transformation is rare in disseminated superficial actinic porokeratosis.[21]

The treatment of porokeratosis ptychotropica can be challenging, and the response to various treatments is often poor.[4,8] In this case, intralesional 5-fluorouracil effectively improved symptoms and reduced the lesions' size. Other options include cryotherapy, imiquimod, calcipotriol, topical steroids, antifungals, and lasers. Treatment choice depends on the severity of the condition and the patient's preferences.[10]

In conclusion, Porokeratosis ptychotropica is a rare variant of porokeratosis, with fewer than 50 cases described in the literature and none in Africa. Pruritic, verrucous plaques in the genitogluteal region characterize it. This case highlights the importance of considering porokeratosis ptychotropica as a differential diagnosis in patients with pruritic, verrucous plaques in the genitogluteal region, especially when conventional treatments fail.

Histopathological examination is essential for diagnosis, and there is a risk of malignant transformation. While treatment options are limited, intralesional 5-fluorouracil may sometimes be effective.

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