



Case Report

Linear Porokeratosis – A rare case report

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ABSTRACT

Linear porokeratosis is a rare disorder of keratinisation resulting from an abnormal clone of epidermal precursors occurring at birth or early adulthood. It is characterized clinically by the presence of sharply demarcated, hyperkeratotic, annular papules with a distinct keratotic edge along the lines of Blaschko. It is common in the distal extremities and is characterized histologically by presence of Cornoid lamella. We present a case of 16 year old female who presented with brownish black, maculo papular, itchy, lesions over anterior aspect of left thigh along lines of blaschko and showed characteristic cornoid lamella on histopathology. Linear porokeratosis has a risk of malignant transformation.

KEYWORDS: Linear porokeratosis, Cornoid lamella, Genodermatoses.

INTRODUCTION

Porokeratosis is a genodermatoses, resulting from a disorder in keratinisation due to an abnormal clone of epidermal precursor presenting with various clinical manifestations but characterised histologically by presence of cornoid lamella. Five types of porokeratosis have been recognized

- a) Classic porokeratosis of Mibelli (PM)
- b) Disseminated superficial actinic porokeratosis (DSAP)
- c) Porokeratosis Palmaris et plantaris disseminata (PPPD)
- d) Linear porokeratosis
- e) Punctate porokeratosis.

Aetiology of various types of porokeratosis is still not known. All except linear porokeratosis show autosomal dominant inheritance pattern. Linear porokeratosis is further classified into localized, Zosteriform, Systematized and generalized. Linear porokeratosis occurs at birth or early childhood and is more common in females. [1]

CASE REPORT

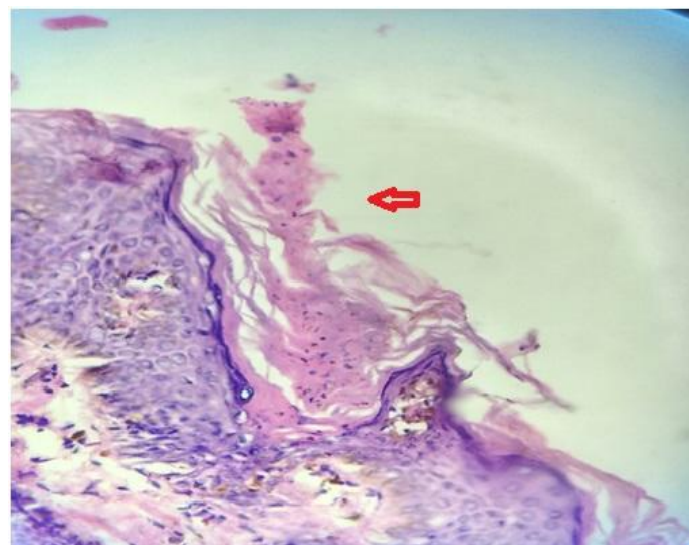
A 16 year old female presented to outpatient department with complaints of an itchy skin lesion over left leg since 3 years. On examination there were numerous linearly arranged multiple discrete to coalescing hyperkeratotic brownish black maculo-papular lesions with central atrophy and raised edges along the Blaschko's line over anterior aspect of left mid thigh extending till knee joint [Figure 1]. No evidence of similar or any other lesion elsewhere on the body. There was no history of similar complaint in the family. Systemic examination was normal.

A biopsy was taken from the peripheral raised edges which on histological examination revealed a keratin filled invagination of the epidermis, center of which had a parakeratotic column containing horny cells which appeared homogenous and contained pyknotic nuclei (cornoid lamella). Granular layer was reduced and few vacuolated keratinocytes were noted. Mixed inflammatory infiltrate was noted in the dermis [Figure 2].

Figure:1 Brownish black maculo-papular lesion with keratotic edge and atrophic center extending linearly from mid thigh to just below knee joint.



Figure :2 Histopathological section of the skin biopsy showing the characteristic Cornoid lamella with reduced granular layer and vacuolated keratinocytes beneath cornoid lamella. (H & E 40x)



DISCUSSION

Porokeratosis was first described by Vittorio Mibelli in 1893 who coined the term ‘porokeratosis’ from the Greek word ‘poros’ (pore of sweat gland) and ‘keratosis’ (horny thickening) [2]. Linear porokeratosis is listed as a rare disease by Office of Rare Diseases (ORD) of the National Institute of Health, meaning thereby that linear porokeratosis affects fewer than 20,000 people in the US . [3] Linear porokeratosis accounts for 3.5–16.7 % of all forms of porokeratosis. [1]

Etiology is still unknown but has been postulated to be due to immune suppression, UV radiation. Occasionally there may be a family history or association with other forms of porokeratosis [4]. Linear porokeratosis has also been seen in monozygotic twins. [5]

Linear porokeratosis usually occurs at birth or early childhood and has a slight predilection for females. Associations with other medical conditions like Crohns disease [6], burns [7], and recipients of organ transplantation [8] and end stage liver disease [9] has also been observed. It has been sub classified into unilateral, zosteriform, systematized and generalized forms.

Linear porokeratosis presents as sharply demarcated brownish black, hyperkeratotic, annular, with a distinct keratotic edge along the lines of Blaschko surrounding a central zone of atrophy. The lesions of linear porokeratosis which vary in size (0.5–1.0 cm), depth (≥ 1 mm), number (few to many) are grouped and linearly arranged on extremities affecting the distal portion more than the proximal areas and on the trunk. In our case, lesions were

distributed more over proximal than distal areas of limb. They are asymptomatic, but can be associated with pruritus.

Biopsy should be taken from the keratotic edge of the lesion to demonstrate the characteristic histopathology finding of cornoid lamella. Histological evaluation reveals a keratin filled invagination of the epidermis, center of which has a parakeratotic column containing horny cells which appear homogenous and contain pyknotic nuclei. This is the characteristic finding of Linear Porokeratosis and is referred to as cornoid lamella [10]. This classic finding is believed to be a peripheral clonal expansion of aberrant keratinocytes [11] which is supported by the presence of helper T cells, suppressor T cells, and Langerhans cells approximating the cornoid lamella, perhaps providing a stimulatory signal[11]. Granular layer may be reduced or absent. Differential diagnosis includes linear verrucous epidermal nevus, linear lichen planus, incontinentia pigmenti (stage II), and lichen striatus.

Studies have demonstrated the presence of an abnormal DNA ploidy and abnormalities in keratinocyte maturation beneath the parakeratotic column [12]. Cultured fibroblasts taken from the underlying dermis has also been found to have instability of the short arm of chromosome 3. [12] Over expression of p53 in the nuclei of keratinocytes in the basal layers of the epidermis beneath the cornoid lamella has also been demonstrated. [11]

Linear porokeratosis has the highest risk of malignant transformation and metastasis among all forms of porokeratosis seen in 7.5 - 11 % of cases which is most commonly Squamous cell carcinoma followed by Basal cell carcinoma.[3,13] Malignant transformation results from

allelic loss or loss of heterozygosity.[14] The development of a nodule or sore within a porokeratosis lesion warrants biopsy evaluation. This warrants the need for a close follow-up of the patient. In a survey conducted in Britain between the periods 1964- 1994 of Porokeratosis, Sasson and Krain found that large lesions, those of long-standing duration and the linear type were at greatest risk for malignant transformation. [15] Aside from malignant transformation, it has been observed that linear porokeratosis may rarely cause nail and bone dystrophy. [16, 17]

Treatment options include destruction using cryotherapy, electrodesiccation, CO2 laser [18] dermabrasion [19] or surgical excision. Successful treatment also has been observed by inducing a cell-mediated/cytotoxic response using 5-fluorouracil and imiquimod cream 5%. Retinoid therapy has yielded inconsistent results.

CONCLUSION

This case of linear porokeratosis is being reported because of the rarity of the lesion, unusual distribution of the lesion, to highlight its characteristic histopathological findings and to stress the need for close follow up of the case in view of the risk of malignant transformation.

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