CASE REPORT

Porokeratotic eccrine ostial and dermal duct nevus: A medical case

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ABSTRACT

Porokeratotic eccrine ostial and dermal duct nevus is a rare adnexal hamartoma, characterized by keratotic papules that exhibit comedo-like openings presenting in a linear disposition. It's a rare condition, fewer than 70 cases have been reported. Nowadays its considered in the adnexal ostium nevi group. Its importance lays on the somatic mutation found on the nevi tissue, which is similar to the keratitis-ichthyosis-deafness (KID) syndrome, meaning that there is a possibility this mutation can be inherited or passed down to the offspring, with the consequent risk of developing KID syndrome.

INTRODUCTION

The Porokeratotic eccrine ostial and dermal duct nevus (PEODDN) is an adnexal hamartoma with eccrine differentiation, that generally follows the Blaschko lines.[1] Since it was first described in 1979, approximately 70 cases have been reported.[2] It is considered a mosaic form of keratitis ichthyosis deafness (KID) syndrome due to a somatic mutation in GJB2 gene, which codes for the gap junction protein connexin 26.[3]

The objective of this work is to report a case of his rare condition, describe the characteristics of the dermatoscopic findings and show the importance of its' diagnosis.

CASE REPORT

A 9 years old female, without previous conditions, presents a lesion in the external region of her right thumb since birth that slowly increased in size. She denies any other local symptoms (fig1A).

The dermatoscopy study showed bright hypochromic pitted kerarotic papules in a linear pattern in the external area of her right thumb (fig1A).

The dermoscopy study features comedo-like openings in a linear disposition over a keratotic white yellowish base (fig1B).

A differential diagnosis was performed to discard lichen striatus, verrucous epidermal nevus and verruca vulgaris.

A biopsy was required for a histopathological analysis which revealed dilatation of intraepidermal eccrine ducts associated to a corneal and columnar porokeratotic obstruction. Also a moderate focal and superficial lymphocytic infiltrate in dermis. The histopathological diagnosis is compatible to Porokeratotic eccrine ostial and dermal duct nevus (fig2).

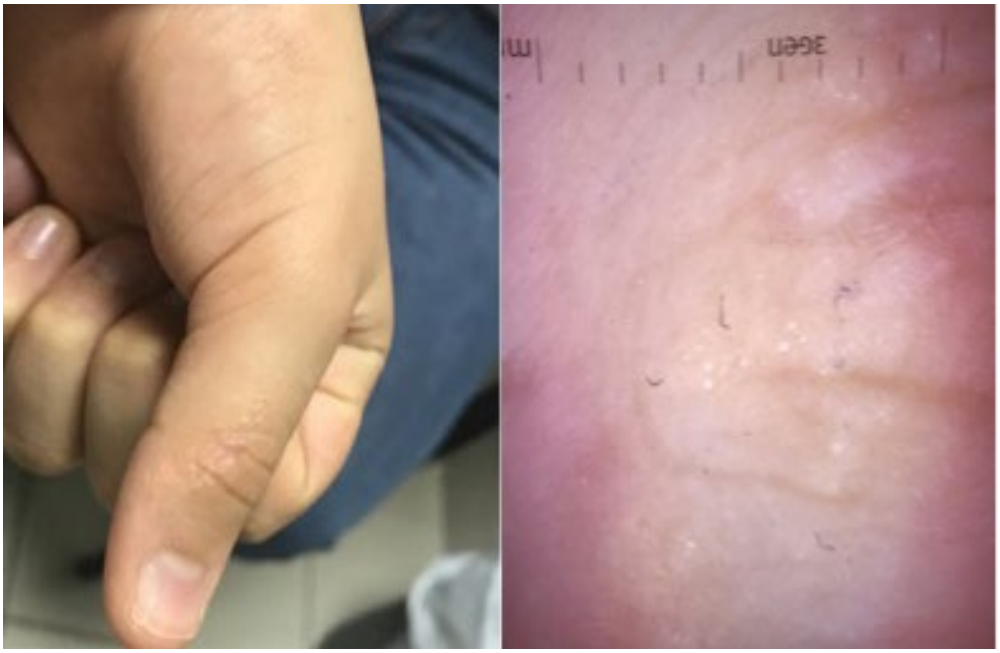

Fig1A. Small bright flesh colored keratotic papules, grouped in a linear patern on external side of right thumb. B. dermoscopy: comedo-like openings in a linear disposition over a keratotic base.

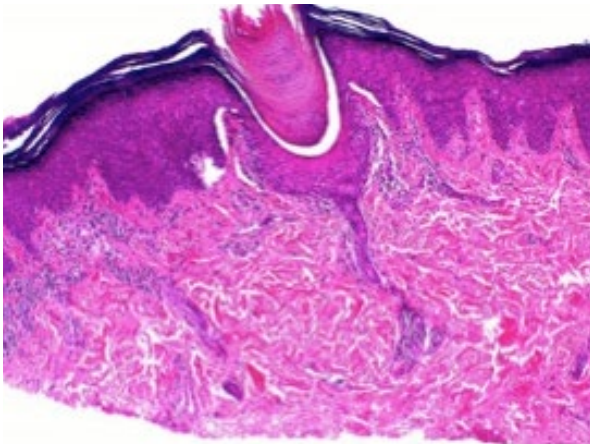

Fig2. Fragment of skin reveals the presence of a cornoid lamella over a dilated eccrine duct. Mild focal lymphocytic infiltrate.

The treatment began with one session of cryotherapy, but the patient did not return to the consult, so it was impossible to evaluate the final results.

DISCUSSION

Porokeratotic eccrine ostial and dermal duct nevus is a congenital hamartoma.[5] It was first described in 1979 as a nevus of the sudoriferous duct by R.A. Marsden et al.[7] Is a very rare condition, less than 70 reported cases exist at the moment.[2]

Its name has been debated and since 2009 the term porokeratotic adnexal ostial nevus was introduced in other to understand the Porokeratotic eccrine ostial and dermal duct nevus, the eccrine nevus and the Porokeratotic eccrine hair follicle nevus.[4,8]

Clinically it is characterized by multiple keratotic or atrophic papules grouped in plaques that have small cavities filled with keratin, giving the aspect of comedomes. They have a linear distribution following the Blaschko lines. These lesions are predominately located on the palms and soles, although they have been described on head, neck and chest associate to anhidrosis and alopecia. The lesions present since birth or during childhood. They are similar to the comedome nevus, with the difference that the porokeratotic nevus presents on palms and soles where hair follicles are absent.[5,6]

The histopathological hallmark is the presence of a cornoid lamella with subjacent acrosyringium. Acanthosis, hyperkeratosis and papillomatosis can be found in the epidermis.[4,6] Similar to the findings on the medical case previously mentioned.

It should be differentiated clinically from linear porokeratosis, linear lichen plaus, linear verrucous epidermal nevus and linear Darier's disease,[5,6] since these conditions can be very similar and difficult to distinguish clinically, having both erythematous papules with a keratotic or verrucous surface grouped

in plaques in a linear pattern, that could follow the blaschko lines. It should be distinguished a specific characteristic for each of these conditions; for example linear verrucous epidermal nevus has a chronic, progressive, inflammatory and pruritic evolution, or in lichen planus the polygonal, violaceous papules with fine white scales (Wickham´s striae) could help for the correct diagnosis.

The importance of this nevus is centered that only recently somatic mutations of GJB2 were discovered in affected tissue by Porokeratotic eccrine ostial and dermal duct nevus that were not found on normal healthy skin of the mother of a patient suffering from KID syndrome. Mutations of autosomal dominant inheritance of GJB2 could cause KID syndrome, individuals with somatic mosaicism have a risk of transmitting systemic diseases to their children and individuals diagnosed with acral porokeratotic nevus should be advised about the higher risk for their children to develop KID syndrome.[9]

CONCLUSION

Being that Porokeratotic eccrine ostial and dermal duct nevus is a rare condition, we have reported a new clinical case with its specific dermoscopic characteristics that have not been described in the literature. We have pointed the importance of its diagnosis so the patients know the possibility of their children inheriting this mutation as well.

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