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Case report

Porokeratosis of Mibelli: an uncommon genodermatosis

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Abstract Porokeratosis of Mibelli is an uncommon, inherited, autosomal dominant disorder characterized by disordered epidermal keratinization and by a predisposition to develop malignant transformation. A case of this rare disorder with classical presentation in a teen aged boy is reported here, which was managed with topical retinoid, superficial chemical peeling and sunscreen lotion.

Key words
Porokeratosis, porokeratosis of Mibelli

Introduction

Porokeratosis is a disorder of epidermal keratinization of uncertain cause, known to be associated with a propensity to develop skin cancer. Five clinical variants of porokeratosis have been described. These include porokeratosis of Mibelli, punctate porokeratosis, linear porokeratosis, porokeratosis palmaris plantaris *et* disseminata, and disseminated superficial porokeratosis.[1] Coexistence of these variants in a single patient has been described only rarely. Disseminated superficial porokeratosis and single plaque porokeratosis of Mibelli have each been documented to occur in association with immunosuppression.[2] The molecular mechanism of this carcinogenesis remains unclear, but p53 has been proposed as a mediator of this process.[3,4] Porokeratosis of Mibelli, first described by Mibelli[5] in 1893, is generally transmitted as an autosomal dominant trait but may occur spontaneously.[1,6] It usually develops in childhood and affects men twice as often as women. The lesions develop as annular, dry plaques surrounded by an elevated keratotic margin and sometimes also a furrow. The centre is often atrophic but may be hyperkeratotic. Lesions appear most often on the limbs and show a tendency to centrifugal spread.[1] The face, genitalia, oral mucosa and cornea may also be affected.[7-9] There may be gradual progression and spontaneous regression leaving mildly atrophic scars. Any diagnostic doubt concerning porokeratosis is erased by cornoid lamella seen on skin biopsy. It is considered to be due to clonal hyperproliferation of atypical keratinocytes. The cornoid lamella consists of a thin column of tightly packed parakeratotic cells within a keratin filled epidermal invagination.[10] There is a risk of malignant transformation, especially after the first five decades, squamous cell carcinoma being the most commonly associated tumour. Bowen's disease and basal cell carcinoma have also been reported.[1,2,11] Medical management includes avoidance of sun exposure, use of sunscreens, topical 5-fluorouracil, topical

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vitamin D-3 analogues (calcipotriol and tacalcitol), topical tretinoin and oral retinoids (isotretinoin, etretinate). Other available modalities are cryotherapy, electrodessication, curettage, dermabrasion and laser therapy.[1,12-14] Surgical excision is most appropriate when malignant degeneration develops. The prognosis is generally good if there is no underlying immunosuppression.

Case report

An 18-year-old boy, resident of district Mianwali, reported with history of progressively enlarging annular plaque involving lips, perioral region, lower part of nose and adjacent areas of the cheeks. It started about seven years ago. He complained of occasional burning and discomfort in the lesion, especially on exposure to sun. He applied various topical medicines over the lesion, but there was no response to treatment. On examination, the lesion was found to be a large annular, atrophic plaque with raised thin, fringe peripheral border (**Figure 1**). The boy was otherwise healthy and did not have any features of immunosuppression. There was no history of such lesions in the family. On the basis of clinical suspicion of porokeratosis, an incisional skin biopsy from the edge of the lesion was performed that confirmed the classical histopathological picture of the disease, with dense hyperkeratosis alternating with a parakeratotic columns of keratinocytes forming cornoid lamellae (**Figure 2**). He was treated with serial weekly facial chemical peeling with 30% salicylic acid solution, daily application of 0.1% tertinoin cream (Retin-A cream) at night along with regular use of physical sunscreen before exposure to sun. After two months of treatment, there was appreciable smoothening of the affected area and he was advised to continue the treatment till 6 months.

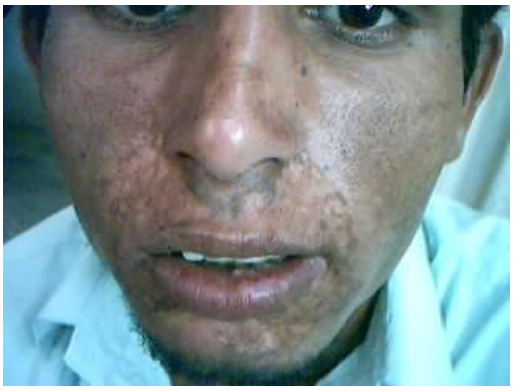

Figure 1 Porokeratosis of Mibelli with characteristic lesion involving central face.

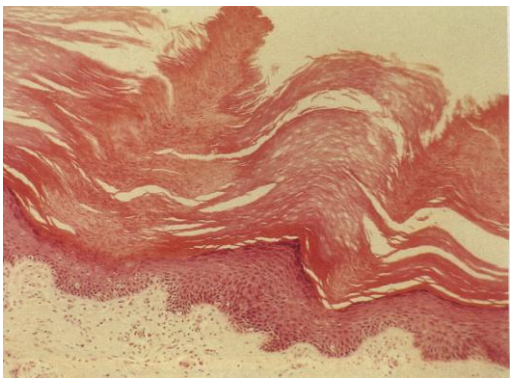

Figure 2 Two cornoid lamellae are seen indenting the underlying epidermis.

Discussion

Epidermal malformations of the skin include a group of heterogeneous developmental defects that result from errors in morphogenesis of the epidermis during intrauterine life. Porokeratosis is one such group of genodermatosis, having many clinical variants, but all are united by the characteristic histologic finding of a cornoid lamella that represents an abnormal clone with abnormal DNA ploidy and mutation.[10] Several risk factors for the development of porokeratosis have been identified; these factors include genetic inheritance, ultraviolet radiation, and immunosuppression.[1,2] An autosomal dominant mode of inheritance has been established for familial cases and spontaneous mutation has also been

described as a possible cause in some cases of porokeratosis.[1,6] In the absence of family history and obvious immunosuppression, spontaneous mutation was probably the cause in our case. The hallmark of histologic diagnosis in porokeratosis of Mibelli and all other forms of the disease is the cornoid lamella, which is formed by hyperproliferative atypical keratinocytes that keratinize rapidly and irregularly and show defective desquamation due to the paucity of intercellular lamellar sheets.[1,10] Although malignancy has been reported previously in various types of porokeratosis, the development of fatal metastatic squamous cell carcinoma in the setting of this disease is a rare event.[11] Porokeratosis of Mibelli has been treated with many topical and surgical modalities in the past; including topical 5-flurouracil, retinoids, cold steel surgical excision, 585 nm pulsed dye laser and CO_2 laser excision.[12-14] We managed our case with more tolerable and safe but still moderately efficacious regimen of chemical peeling with 30% salicylic acid, topical tretinoin cream and sunscreen.

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