Porokeratosis of Mibelli With Mutilation: A Case Report

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Porokeratosis is a rare keratinization disorder of the skin characterized by annular plaques with an atrophic center surrounded by a raised keratotic wall that spreads centrifugally. We report a case of porokeratosis of Mibelli with mutilation. A 30-year-old woman presented with atrophic plaques on the index fingers of both hands with a keratotic ridge in some margins of the plaques. There was loss of the distal phalanx of the left index finger. In the right hand, shortening of the right distal phalanx and flexion contracture of the distal interphalangeal joint were noted in the index finger.

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Porokeratosis is a rare keratinization disorder first described by Mibelli[1] and Respighi[2] in 1893. It is characterized by annular plaques with an atrophic center surrounded by a raised keratotic wall that spreads centrifugally. Five clinical types are known: (1) classic porokeratosis of Mibelli, (2) disseminated superficial porokeratosis and disseminated superficial actinic porokeratosis, (3) linear porokeratosis, (4) porokeratosis palmaris et plantaris disseminata, and (5) punctate porokeratosis.[3] We report a case of porokeratosis of Mibelli with involvement of the perioral area and hands that led to destructive changes of the phalanges.

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Case Report

A 30-year-old woman was referred with mutilating lesions on both index fingers that were present since childhood. The lesions started as erythematous scaly plaques on the tips of her index fingers and slowly progressed to the base of these fingers. Both index fingers became sclerotic and shortened after a few years. Similar lesions developed on both thumbs and the perioral region but without mutilation of these areas. The patient reported pain and a burning sensation in the fingers of both hands when exposed to cold weather or cold water. She was a water pipe smoker but had no history of trauma or radiation injury. The family history was noncontributory.

Physical examination revealed well-circumscribed atrophic plaques on the index fingers of both hands. A keratotic ridge could be observed in some margins of the plaques. Well-circumscribed, annular, erythematous plaques with central atrophy also were present on both thumbs, the radial side of the right hand, the ulnar side of the left hand, and the perioral region (Figures 1 and 2). There was nail dystrophy on the right index finger and both thumbs as well as complete loss of the nail of the left index finger. In the right hand, there was limitation of flexion of the proximal interphalangeal and metacarpophalangeal joints, and flexion contracture of the distal interphalangeal joint of the index finger. The tip of the index finger reached only up to the middle of the third finger. In the thumb, there was limitation of flexion of the interphalangeal joint. In the left hand, there was limitation of flexion of the metacarpophalangeal and proximal interphalangeal joints of the index finger and the distal phalanx was absent. There also was limitation of flexion of the interphalangeal joint of the thumb.

A biopsy was taken from the edge of the left hand lesion. Histologic examination showed 2 keratin-filled shallow invaginations of the epidermis with

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Figure 1. Annular erythematous plaques with central atrophy on the radial side of the right hand and the ulnar side of the left hand. Atrophic plaques with some marginal keratotic ridges were present on both index fingers.

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Figure 2. Nail dystrophy of both thumbs with complete loss of the nail of the left index finger. Annular erythematous plaques also were present in the perioral region.

a column of parakeratosis at the center of each invagination. The epidermis was acanthotic and there was a mild, nonspecific, perivascular infiltrate of chronic inflammatory cells in the papillary dermis. Radiography results of the hands showed absence of the distal phalanx, shortening of the middle phalanx, and decreased width of the proximal phalanx of the left index finger (Figure 3). In the right hand, there was marked shortening of the distal phalanx with a compromised distal interphalangeal joint, small erosion of the medial aspect of the middle phalanx with concavity of the cortex, and mild thinning of the proximal phalanx of the index finger.

The patient was asked to complete some hematologic and biochemical tests prior to initiation of treatment but was lost to follow-up.

Comment

Porokeratosis of Mibelli is a rare disease that usually appears during childhood with a male to female ratio of 3 to 1. The mode of inheritance may be autosomal dominant or sporadic.[4]

The etiology of porokeratosis of Mibelli is not known. It has been suggested that a clone of abnormal epidermal cells migrate centrifugally leading to the formation of cornoid lamella. The tendency for development of such abnormal clones is inherited and several triggering factors, such as immunosuppression, irradiation, trauma, and infective agents, lead to phenotypic expression.[5] The tumor suppressor protein p53 is overexpressed in keratinocytes under or adjacent to cornoid lamella in all subtypes of porokeratosis.[6-8]

Histopathologic observations of all variants of porokeratosis typically show the characteristic cornoid lamella, which is a column of parakeratotic cells in the center of the keratin-filled invaginations of the epidermis. The lower epidermis exhibits a loss of the granular layer and several dyskeratotic and vacuolated cells. There is a perivascular infiltrate of chronic inflammatory cells in the papillary dermis.[9]

The lesions of porokeratosis of Mibelli may be solitary or a few and generally begin as asymptomatic papules that enlarge centrifugally, leaving the central area atrophic, hairless, and hypopigmented. The lesions are more common on the extremities, especially the dorsal surface of the hands, feet, and ankles.[3,4,10] There is a high incidence of skin malignancies, including squamous cell carcinoma, basal cell carcinoma, and Bowen disease, in the lesions of porokeratosis.[4,11,12] Malignant changes are more frequent in nonexposed skin, in large porokeratotic lesions, in lesions with long duration, and in patients with prior exposure to ionizing radiation. It also is more common in the linear form of porokeratosis of Mibelli.[11-14]

Treatment of porokeratosis is difficult, though various treatment regimens have been described. Localized lesions can be removed by cryotherapy, electrocautery, dermabrasion,[15] CO_2 laser therapy,[16]

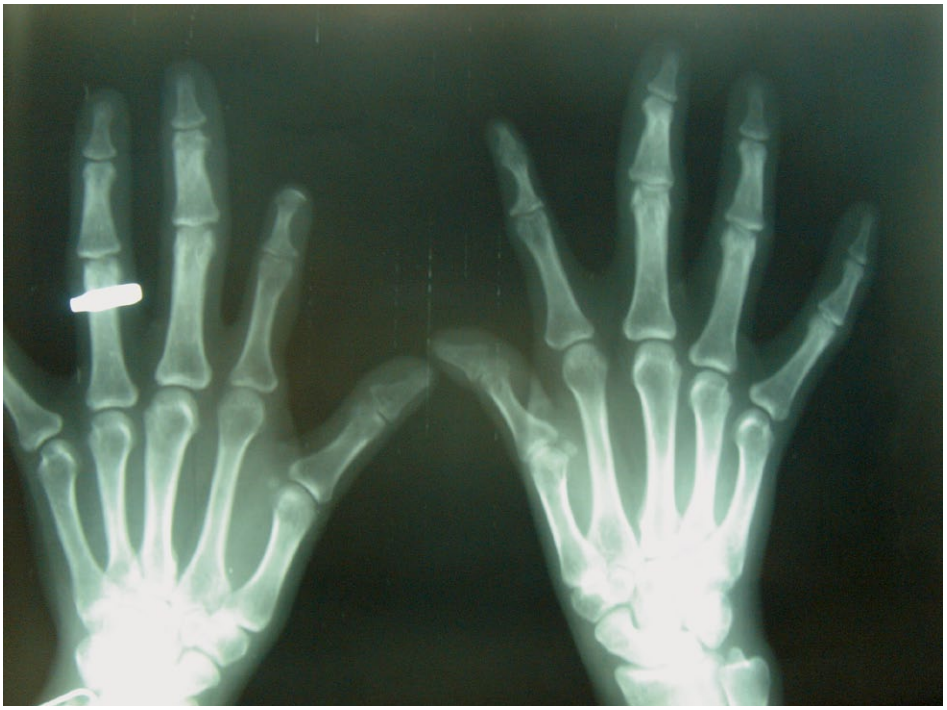

Figure 3. Radiography of the hands showed absence of the distal phalanx of the left index finger with marked shortening of the distal phalanx of the right index finger.

and surgical excision. Other treatments include keratolytics, topical 5-fluorouracil,[17] topical or intralesional cortisone,[18] topical tretinoin,[19] and imiquimod cream 5%.[20] Oral etretinate may be effective,[21] but it led to worsening of the disease in one patient.[22]

Destructive changes in porokeratosis are rare. Rahbari et al[23] reported 2 cases of facial porokeratosis of Mibelli with extensive destructive changes. Bhutani et al[24] reported a patient with porokeratosis of Mibelli and porokeratosis palmaris et plantaris disseminata who had nail dystrophy, bony resorption of distal phalanges of the fingers and toes, and flexion contracture of the interphalangeal joints. Another case reported by Handa et al[25] showed similar features with loss of distal phalanges of the hand and flexion deformities. Ramesh et al[26] described a patient with generalized porokeratosis of Mibelli who had pseudoainhum in the right second and third toes resulting from a porokeratotic lesion of the right foot.

Our case report emphasizes the importance of considering porokeratosis of Mibelli among the differential diagnoses of patients with cutaneous lesions associated with mutilation.

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