An otherwise healthy 13-year-old adolescent boy presented with multiple enlarging asymptomatic lesions on his right forearm that first appeared as small flesh-colored papules when he was 3 years old. The lesions subsequently enlarged centrifugally and, on presentation, appeared as multiple well-circumscribed plaques with a pink atrophic center and a distinct raised border. He denied any symptoms associated with the plaques but noted the lesions got scaly when he was outdoors. He had no other similar lesions elsewhere and no family members were affected.
The Diagnosis: Porokeratosis of Mibelli

In 1893, Mibelli first described porokeratosis in a man with multiple lesions of varying sizes and forms on his arm and hand that first appeared at 2 years of age. The whitish-red lesions were surrounded by a raised border and appeared atrophic in the center, similar to our patient (Figure 1). The patient had a family history of similar lesions. Mibelli called the condition *porokeratosis* because he incorrectly believed that the lesions were caused by hyperkeratosis of the sweat duct pores. Subsequently, the disease was termed *porokeratosis of Mibelli*. Many variants of porokeratosis have been identified since Mibelli’s initial case, including linear porokeratosis and punctate porokeratosis. Disseminated forms include disseminated superficial porokeratosis, disseminated superficial actinic porokeratosis, and porokeratosis plantaris palmaris et disseminata.

The etiology of porokeratosis is unknown, but it is believed to be an autosomal-dominant genodermatosis characterized by a disorder in the epidermal keratinization. Taylor et al believed that the lesion of porokeratosis stemmed primarily from an aberration in the short arm of chromosome 3. Scappaticci et al reported region p12-14 of chromosome 3 specifically was involved in classic porokeratosis. Other physicians have hypothesized that the stimulation of a certain group of cells in the epidermis leads to the development of the porokeratotic lesion. Specifically, these physicians believe the epidermis consists of multiple clones of healthy and mutant cells from birth and that the lesion of porokeratosis results from a group of mutant clonal epidermal cells being overexpressed because of an external factor (eg, a combination of irradiation, infection, trauma, and immunosuppression).

Of all the variants of porokeratosis, disseminated superficial actinic porokeratosis (DSAP) most commonly has been related to frequent sun exposure. As a result, there can be severe exacerbation of DSAP in the summer. Reymond et al described a patient who developed DSAP after extensive psoralen plus UVA therapy. In 1990, Watanabe et al reported that dermal fibroblasts derived from lesions of porokeratosis were hypersensitive to x-irradiation. This finding contributed further to the association between sun exposure and the development of porokeratosis.

Trauma is another possible cause of porokeratosis, which has been specifically noted in patients with previous burn injuries. In addition, the Köbner phenomenon was reported by Savage in 1953 (case presented in 1951) to have a strong correlation in the development of the lesions of porokeratosis in areas of previous trauma.

Immunosuppression has been implicated in the potential development of porokeratosis. Specifically, MacMillan and Roberts hypothesized that adjacent healthy epidermal cells suppress the mutant clones of epidermal cells that have the potential to develop the lesions of porokeratosis. However, immunosuppression may impair the previous mechanism and lead to the development and growth of the lesions of porokeratosis. Consequently, porokeratosis has been reported in patients with active autoimmune diseases, including chronic hepatitis, primary biliary cirrhosis, vitiligo, and cystic fibrosis.

The lesions of punctate porokeratosis typically develop during childhood and are more common in males than females. Punctate porokeratosis most often develops on the extremities and can involve the palms and soles. The initial lesion usually is described as an asymptomatic papule or plaque that slowly increases in size centrifugally. The center of the lesion may appear atrophic and hypopigmented. The border of the lesion is distinctly raised and has a craterlike appearance.

Figure 1. Multiple enlarging asymptomatic lesions on the right forearm.
Histologic evaluation reveals that the sine qua non of porokeratosis is the cornoid lamella, found at the borders of the lesion and composed of a column of parakeratotic cells extending through the orthokeratotic stratum corneum (Figure 2). The granular layer beneath the cornoid lamella either may be absent or reduced in thickness, and the keratinocytes may have a vacuolated appearance. A lymphohistiocytic infiltrate may be present in the subjacent papillary dermis. The epidermis in the central part of the lesion may appear either healthy, thin, hyperkeratotic, or acanthotic.

Porokeratosis has been associated with the development of skin malignancies including Bowen disease, squamous cell carcinoma, and basal cell carcinoma. A retrospective review revealed that a substantial number of patients with porokeratosis developed malignant transformation of their initial lesion. Malignancies were more likely to occur on areas of the body not exposed to the sun, in large porokeratosis lesions, and in patients with previous ionizing radiation exposure. Also, malignant transformation was more likely to occur in linear porokeratosis than in disseminated forms. Gray et al demonstrated that the staining pattern of keratinocytes in the center of the lesion was similar to actinic keratosis and squamous cell carcinoma. Also, Arranz-Salas et al proposed that an alteration in p53 expression has led to the increased incidence of malignancies arising from the lesions of porokeratosis.

Although multiple treatments for porokeratosis have been reported, the therapeutic response usually is poor. Localized lesions can be removed by surgical excision, cryotherapy, CO2 laser, or dermabrasion. Topical and intralesional corticosteroids of varying potencies and topical tretinoin all have shown varying degrees of success in treating this disorder. Systemic etretinate also has been noted to be an effective treatment. However, recurrence is likely following cessation of therapy. Sunscreen commonly is prescribed to prevent potential solar-induced irritation.

REFERENCES