

Squamous cell carcinoma arising from mucosal porokeratosis

Prateek Maharana¹, Subrat Kumar Patel²

From ¹Assistant Professor, Department of Dermatology, Veer Surendra Sai Institute of Medical Sciences and Research, Burla,

²Consultant Dermatologist, Department of Dermatology, District Headquarter Hospital, Bargarh, Odisha, India

A 60-year-old male presented to the dermatology outpatient department with complaints of multiple annular plaques over photo-exposed areas and a progressively enlarging growth over the lower lip. The lesions over the extremities had been present for nearly 2 years, while the lip lesion had developed over the preceding three months with a gradual increase in size and occasional bleeding. The patient was immunocompetent, with no history of tobacco use, alcohol intake, or prior radiation exposure. His vital parameters at presentation were stable with a blood pressure of 128/78 mmHg, pulse rate of 78 beats/min, respiratory rate of 18 breaths/min, and temperature of 36.7°C. Cutaneous examination revealed multiple well-defined annular plaques with raised hyperkeratotic hyperpigmented borders and central atrophy over photo-exposed areas of the bilateral upper limbs (Fig. 1). The lesions were asymptomatic but slowly progressive. In addition, ulceroproliferative growth measuring approximately 2.5 × 2 cm with irregular margins, crusting, and areas of necrosis was noted over the lower lip (Fig. 2), arising from a pre-existing plaque. The surrounding mucosa showed features suggestive of porokeratosis. No regional lymphadenopathy was detected.

Histopathological examination from a representative annular plaque demonstrated a coronoid lamella with underlying loss of the granular layer and focal dyskeratotic keratinocytes, accompanied by a superficial dermal lymphocytic infiltrate, confirming the diagnosis of porokeratosis. A biopsy obtained from the ulceroproliferative lip lesion showed invasive nests and sheets of atypical squamous cells extending into the dermis, exhibiting nuclear pleomorphism, hyperchromasia, increased mitotic activity, and individual cell keratinization. Prominent keratin pearl formation was noted, confirming a well-differentiated squamous cell carcinoma (SCC). The patient was referred to the surgical oncology department, where a wide local excision of the lesion with appropriate margins was performed. The patient is currently under follow-up for 3 months; no evidence of local recurrence or metastasis was noted.

Porokeratosis encompasses a group of disorders of keratinization, including disseminated superficial actinic porokeratosis, linear porokeratosis, and punctate variants, all characterized by the presence of a coronoid lamella histologically. Clinically, these lesions typically appear as annular (ring-shaped) plaques with a raised, thread-like keratotic border and a relatively depressed or atrophic center. The condition is thought to arise from an abnormal clone of keratinocytes, leading to disordered epidermal differentiation. Lesions most commonly occur on sun-exposed areas and may remain stable for years; however, in some cases, they can gradually enlarge or develop complications. Malignant transformation has been reported in approximately 7.5–11% of cases, most commonly into SCC [1,2]. Risk factors associated with malignant transformation include long-standing lesions, chronic ultraviolet exposure, immunosuppression, and large or linear variants of porokeratosis.

Malignancy arising in mucosal porokeratosis is exceedingly rare, with fewer than ten cases documented to date. In previously reported cases, malignant transformation was attributed to cumulative ultraviolet radiation, genetic instability within the abnormal keratinocyte clone, and chronic inflammation leading to DNA damage [3]. Although the patient lacked conventional risk factors, malignant transformation may be attributed to intrinsic clonal instability of keratinocytes within the coronoid lamella. Chronic ultraviolet exposure to the lower lip and repeated microtrauma may have contributed to cumulative DNA damage, facilitating carcinogenesis. The pathogenesis involves the expansion of an abnormal keratinocyte clone forming the coronoid lamella. Genetic instability, ultraviolet-induced mutations, and impaired epidermal differentiation contribute to malignant transformation [4].

Similar cases have been sporadically described in the literature. Kanitakis reported that cutaneous malignancies arising in porokeratosis are predominantly SCC, although basal cell carcinoma and Bowen's disease have also been documented [1]. Sertznig *et al.* emphasized that long-standing lesions located on sun-exposed areas

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Correspondence to: Dr. Prateek Maharana, Department of Dermatology, Veer Surendra Sai Institute of Medical Sciences and Research, Burla, Odisha, India. E-mail: drprateekmaha@gmail.com

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Figure 1: Cutaneous examination revealed multiple well-defined annular plaques with raised hyperkeratotic hyperpigmented borders and central atrophy over photo-exposed areas of the bilateral upper limbs



Figure 2: Ulceroproliferative growth measuring approximately 2.5×2 cm with irregular margins, crusting, and areas of necrosis was noted over the lower lip

are particularly susceptible to malignant transformation.² Compared with previously reported cases, our patient demonstrated the rare occurrence of SCC arising from mucosal involvement of porokeratosis on the lower lip, highlighting the need for vigilance in monitoring such lesions.

This case underscores the importance of regular surveillance of porokeratosis lesions, particularly those located on photo-exposed or mucosal surfaces, to detect early malignant transformation. Patients with porokeratosis should be counseled regarding the risk of malignant transformation and advised to undergo regular follow-up. Any lesion showing rapid growth, ulceration, or bleeding should undergo prompt biopsy. Photoprotection and avoidance of chronic irritation are recommended.

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