Coexistence of porokeratosis of Mibelli with Gardner's syndrome: A rare case report

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ABSTRACT

Porokeratosis represents a heterogeneous group of disorders characterized clinically by a distinctive ridge-like border and histologically by cornoid lamellae. Gardner's syndrome, a variant of familial adenomatous polyposis (FAP), is an autosomal dominant disease characterized by colorectal polyps, osteomas, epidermoid cysts and soft tissue tumors. Here we report a case of 18 yr old female who presented with porokeratosis of Mibelli with osteoma, multiple epidermoid cytsts, and solitary rectal polyp.

Key words: Cornoid lamella, epidermoid cysts, Gardner's syndrome, osteomas, porokeratosis of Mibelli

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INTRODUCTION

Porokeratosis is a genetic disease transmitted as an autosomal dominant trait featuring abnormal epidermal keratinization, which is histologically characterized by the presence of cornoid lamella.[1,2] The term "porokeratosis" was first described by Vittorio Mibelli in 1893. Since then many variants of porokeratosis have been described, each with differing morphology, distribution, and clinical course.[2] Familial Adenomatous Polyposis (FAP) is characterized by numerous adenomatous colorectal polyps that have an intrinsic tendency to progress to adenocarcinoma. FAP with extra-intestinal lesions such as epidermoid cysts, osteomas, desmoid tumors, dental anomalies is called Gardner's Syndrome.[3] The association of Porokeratosis of Mibelli with Gardner's syndrome is extremely rare without any recorded cases in the literature, prompted us to report this case.

CASE REPORT

An 18-year-old female of low socio-economic status presented to skin OPD with complaints of multiple skin lesions all over the body and swellings on the face, scalp, and extremities of five years duration. Lesions started to develop on the scalp insidiously, and later on left mandible, face, and extremities. Clinical examination of the patient revealed multiple skin plaques present on the face, forearms, back and both lower extremities with elevated margin with a prominent deep furrow and central part showed normal skin [Figure 1]. Fluctuant skin swellings were observed on the scalp and back of trunk, with punctum [Figure 2]. Over the body of left ramus of mandible, there was a bony hard, painless, swelling [Figure 3]. Physical examination of other systems including dental and ophthalmological examination was unremarkable. A clinical diagnosis of Porokeratosis of Mibelli, epidermoid cysts, and osteoma of mandible was made. Family history of the index case revealed father had dysphagia, pain abdomen with episodes of bleeding per rectum, and ultimately died of us suspected gastrointestinal (GIT) origin malignancy. No records of his illness were available. Biopsy of the skin plaques, along with the excision biopsy of cystic swelling over scalp and bony hard swelling of left mandible was done. Because of the family history and clinical presentation (sebaceous cysts and osteoma), a colonoscopy was done which revealed a solitary rectal polyp of 1 × 1.5 cm in size situated 7.0 cm from anal verge. In due course, polypectomy was done and subjected for histological study.

Histopathological examination of the plaque lesions of skin revealed a keratin filled invagination of the epidermis, the center of which showed parakeratotic column (interpreted as cornoid lamella). The horny cells with in the cornoid lamella

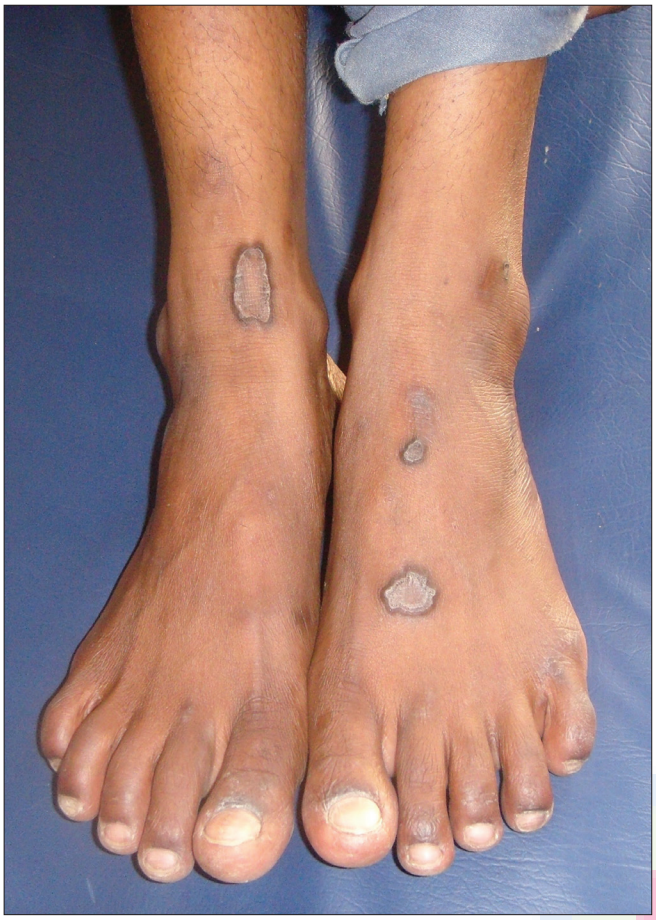

Figure 1: Porokeratosis of Mibelli

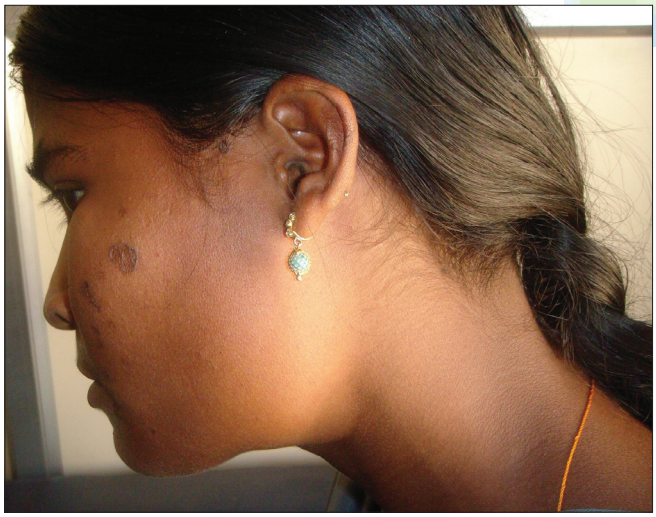

Figure 3: Osteoma of mandible

appeared homogenous with pyknotic nuclei. Granular cell layer of epidermis was absent under the cornoid lamella. Superficial dermis revealed focal chronic nonspecific inflammatory infiltrate composed of lympho-mononuclear leucocytes. There was no evidence of malignancy or dysplasia. Diagnosis of porokeratosis of Mibelli was made [Figure 4]. Cystic swellings revealed the histopathology of epidermoid cyst. Bone lesion showed the

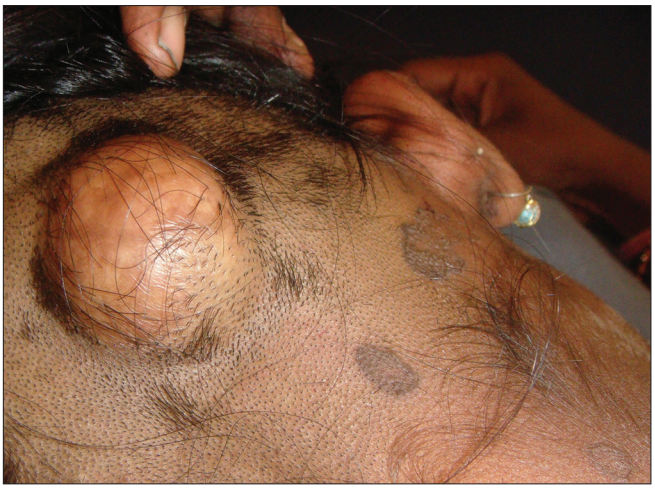

Figure 2: Epidermoid cysts

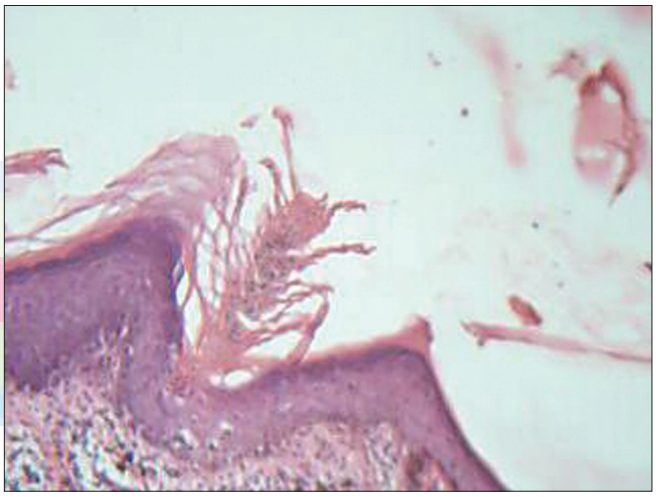

Figure 4: Parakertotic column (Cornoid lamella) in porokeratosis Mibelli, (H and E, ×100)

histopathology of a benign osteoma showing mature osteocytes [Figure 5]. Histopathological examination of rectal polyp showed hyperplastic polyp of rectal mucosa with focal adenomatous change, mild nonspecific inflammation, cystic change, congested vessels with no evidence of malignancy [Figure 6].

DISCUSSION

The clinical picture, family history and multiple lesions diagnosed histologically in the index patient, we contemplated the possibility of Porokeratosis of Mibelli with an evolving Gardner's Syndrome. Gardner's syndrome start with a few polyps in the 10-20 year age group and develop into its full blown clinical picture by the fourth decade of the patient, with an intrinsic tendency to progress to adenocarcinoma. It is caused by a germline mutation in the adenomatous polyposis coli (APC) gene located on the long arm of chromosome 5.[3]

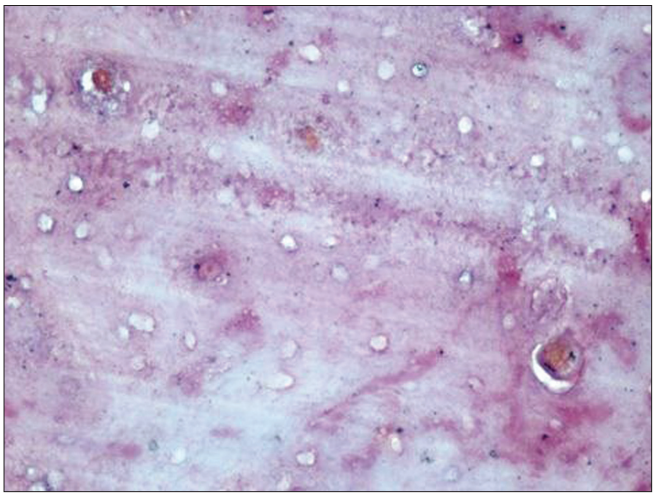

Figure 5: Osteoma with mature osteocytes (H and E, ×200)

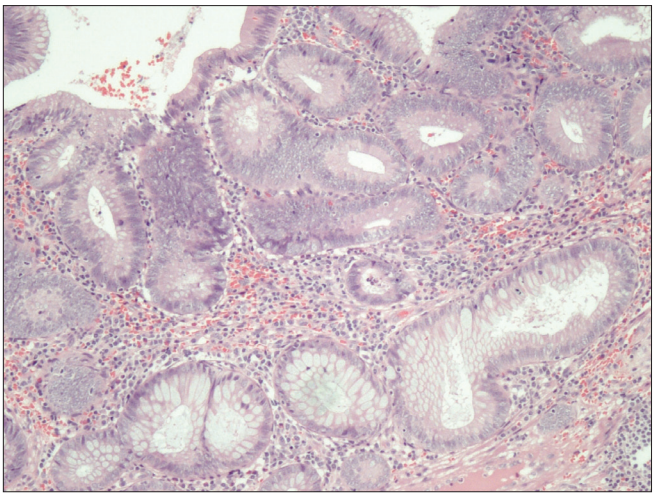

Figure 6: Hyperplastic polyp of rectal mucosa with focal adenomatous change (H and E, ×100)

Presence of osteomas is required to make the diagnosis of Gardner's syndrome. The mandible is the most common location; however, osteomas may occur in the skull and the long bone. Osteomas precede the clinical and radiographic evidence of colonic polyposis, therefore, they may be sensitive markers for the disease.[4] Porokeratosis is a heterogeneous group of disorders characterized by distinct clinical findings of a keratotic ridge with a central groove that corresponds to the cornoid lamellae on histology.[5] Etiopathogenesis of porokeratosis is unknown but genetic inheritance as well as acquired form has been reported. Heredity, immunosuppression, infection, and ultraviolet rays have been found to play a role in the causation of porokeratosis. Autosomal dominant mode of inheritance has been reported in all forms of familial porokeratosis. Linear porokeratosis is said to represent the type 2 segmental form of the disease. No specific candidate gene has been found. However, genetic loci of disseminated superficial actinic porokeratosis have been detected in the region 12q23.2-24.1 (DSAP 1) and 15q25.1-26.1 (DSAP 2). Recently, another loci has been found within an 8.2 cM or 11.9 Mb region between markers D1S438 and D1S464 (DSAP 3). No gene or genetic locus has been found specifically for Mibelli's type.[6] Malignancy, although rare, has been reported in almost all forms of porokeratosis. Larger lesions have higher malignant potentiality, particularly, the giant variety that is most frequently reported to show malignant transformation. Malignancies reported in porokeratosis are squamous cell carcinoma, Bowen's disease, basal cell carcinoma, diffuse large B-cell lymphoma, etc.[6] Porokeratosis in association with lichen planus, diabetes mellitus, CAP syndrome (craniosynostosis, anal anomalies, and porokeratosis), Bloom's syndrome, cystic fibrosis etc, have been reported.[7-11] To the best of our knowledge this is the first case reported of porokeratosis of Mibelli with Gardener's syndrome.

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