Print ISSN: 2321-6379
Online ISSN: 2321-595X

Case report

Familial Disseminated Superficial Actinic Porokeratosis

Archana Uthayakumar[1], Muthukumaran Rajaram[2], Eswaramurthi Balasubramanian[3], Madhavan Ramamoorthy[3]

[1]Post Graduate Student, Department of Dermatology, Venereology, and Leprology, Coimbatore Medical College Hospital, Coimbatore, Tamil Nadu, India, [2]Professor, Department of Dermatology, Venereology, and Leprology, Coimbatore Medical College Hospital, Coimbatore, Tamil Nadu, India, [3]Assistant Professor, Department of Dermatology, Venereology, and Leprology, Coimbatore Medical College Hospital, Coimbatore, Tamil Nadu, India

Abstract

Porokeratosis is a disorder of keratinization resulting from abnormal clonal expansion of keratinocytes. It may be sporadic or familial with autosomal dominant pattern of inheritance with variable penetrance. Lesions start as papules which develop into annular plaques with thin, thread like elevated rim. Diagnosis is confirmed by histopathological examination of pathognomonic Coronoid lamella. Here, we are reporting a familial case of disseminated superficial actinic porokeratosis in a mother and daughter.

Key words: Coronoid lamella, Familial, Parakeratosis, Porokeratosis

INTRODUCTION

Porokeratosis is a rare disorder of keratinization resulting from clonal expansion of keratinocytes characterized by well-defined plaque with hyperkeratotic ridges. It may be sporadic or familial with autosomal dominant pattern of inheritance with variable penetrance.[1] Exact pathogenesis is unclear, possible theories of pathogenesis include premature apoptosis of keratinocytes, reduced keratinocytic expression of filaggrin, and loricrin. In disseminated superficial actinic porokeratosis mutations in the SART3 and MeValonate Kinase genes on chromosome 12q24 normally play a role in keratinocyte differentiation and protect keratinocytes from apoptosis in response to damage from ultraviolet radiation (UV).[1,2] Risk factors include genetic inheritance, UV radiation and immunosuppression. Lesions start as papules or plaques which develop into annular lesions with thin, thread like elevated rim. Diagnosis is confirmed by histopathological examination of lesion which shows the pathognomonic Coronoid lamella. Malignant change into squamous cell carcinoma may occur.

Access this article online

www.ijss-sn.com

Month of Submission : 11-2020
Month of Peer Review : 11-2020
Month of Acceptance : 12-2020
Month of Publishing : 01-2021

CASE REPORT

A 50-year-old female presented with complaints of asymptomatic skin lesions over face, chest, back, and right forearm for past 35 years. Initially, lesions started over face which gradually progressed to involve upper chest, right forearm, and back. History of similar lesions was present in her two brothers, sister, and her daughter. On examination, multiple well-defined annular plaques of varying size ranging from 1 × 1 cm to 2 × 2 cm present over face, upper chest [Figure 1a-c], right forearm, back [Figure 1d-f] with central atrophy and peripheral raised hyperpigmented furrowed margins. Oral mucosa, palms, and soles and genitals were normal.

Her 27-year-old daughter, presented with complaints of asymptomatic skin lesions over face for past 10 years. She is a known case of hypothyroidism on treatment. On examination multiple well-defined annular plaques of sizes ranging from 0.5 × 0.5 mm to 1 × 1 cm with central atrophy and peripheral raised hyperpigmented furrowed margins were present over face, scattered over forehead, and around eyes and cheeks [Figure 2a-c].

Corresponding Author: Dr. Muthukumaran Rajaram, Department of Dermatology, Venereology, and Leprology, Coimbatore Medical College Hospital, Coimbatore, Tamil Nadu, India.

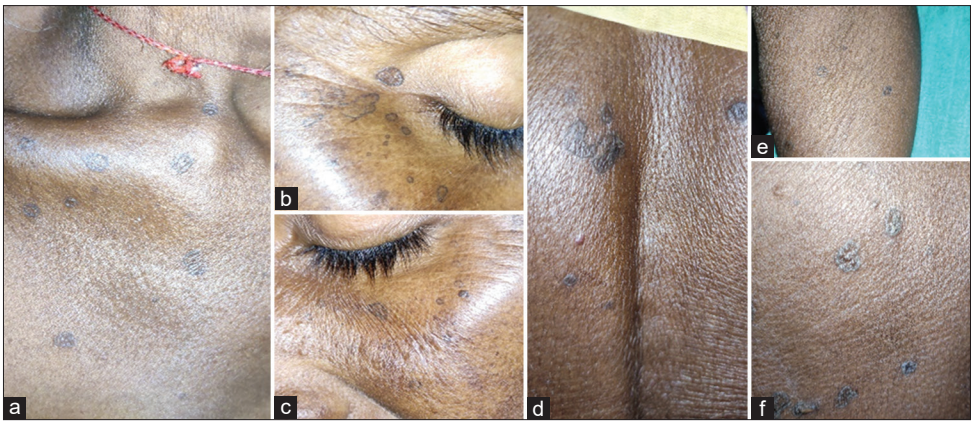

Figure 1: (a-c) Multiple plaques with central atrophy and peripheral hyperpigmented raised furrowed margin in the upper chest, right and left side of face in mother, (d-f) Multiple plaques with central atrophy and peripheral hyperpigmented raised furrowed margin in back and left forearm in mother

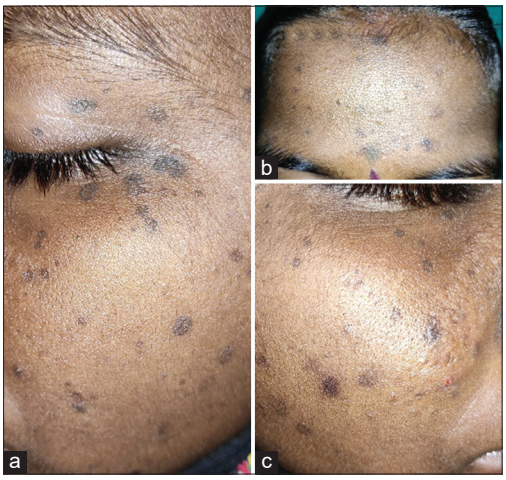

Figure 2: (a-c) Multiple plaques with central atrophy and peripheral hyperpigmented raised furrowed margin in forehead, bilateral cheeks in daughter

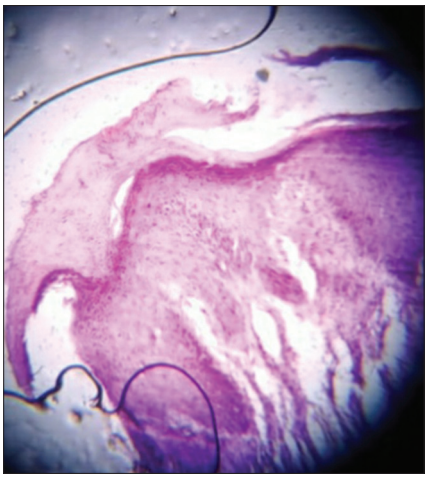

Figure 3: Hyperkeratosis, epidermal invagination filled with parakeratotic keratinocytes (coronoid lamella)

Histopathological examination of margin of lesions in both cases shows hyperkeratosis, epidermal invagination filled with thin column of tightly packed parakeratotic keratinocytes called coronoid lamella [Figure 3].

On the basis of history, clinical and histopathological examination, diagnosis of disseminated superficial actinic porokeratosis in mother and daughter was made.

DISCUSSION

Mibelli described classical porokeratosis in 1893. The lesions of porokeratosis results from peripheral expansion of abnormal, mutant clone of epidermal keratinocytes. Clinically, it is classified into localized and disseminated forms. Localized forms include porokeratosis of Mibelli, linear porokeratosis, punctate palmoplantar porokeratosis, perianal and genital porokeratosis. Disseminated forms include disseminated superficial porokeratosis, disseminated superficial actinic porokeratosis, systematized linear porokeratosis, and disseminated palmoplantar porokeratosis. Other unusual and rare forms include porokeratosis developing in gluteal region – porokeratosis ptychotropica, reticular and follicular variants. It may sometimes resemble punctate keratoderma, Darier disease, Cowden disease, and Arsenical keratosis.[3]

Malignant change is rare, if occurs squamous cell carcinoma is common, followed by Bowen's disease, basal cell carcinoma, and diffuse large B cell lymphoma.[4]

Histologically, it is characterized by coronoid lamella-thin column of tightly packed parakeratotic keratinocytes. Underlying granular layer is absent or attenuated. Dyskeratotic keratinocytes are present at the base of coronoid lamella.

Treatment options depend on the size and site of the lesions and include cryotherapy, topical 5-fluorouracil, topical imiquimod, topical Vitamin D analog, curettage, cautery, photodynamic therapy, and carbon dioxide laser.[5] Oral retinoids such as isotretinoin and acitretin were tried with variable success.

Familial disseminated superficial actinic porokeratosis is reported less in Indian literature and hence we report this case.

REFERENCES

1. Le C, Bedocs PM. Disseminated superficial actinic porokeratosis. In: Stat Pearls. Treasure Island, FL: Stat Pearls Publishing; 2020.
2. Chu AC, Teixeira F. Acquired disorders of epidermal keratinization. In: Rooks Textbook of Dermatology. 9th ed., Vol. 3. Hoboken: Wiley Blackwell; 2016. p. 87.
3. Requena L, Requena C, Cockerel CJ. Benign epidermal tumours and proliferations. In: Dermatology. 3rd ed., Vol. 2. New York: Elsevier, Saunders; 2017. p. 1801.
4. Sarma N, Boler AK, Bhattacharya SR. Familial disseminated plaque type porokeratosis with multiple horns and squamous cell carcinoma involving anal skin. Indian J Dermatol Venereol Leprol 2009;75:551.
5. Inakanti Y. Familial porokeratosis of Mibelli: A peerless entity. J Pak Assoc Dermatol 2015;15:144-7.

How to cite this article: Uthayakumar A, Rajaram M, Balasubramanian E, Ramamoorthy M. Familial Disseminated Superficial Actinic Porokeratosis. Int J Sci Stud 2020;8(10):4-6.

Source of Support: Nil, **Conflicts of Interest:** None declared.