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Case Report

Familial pityriasis rubra pilaris: a case report

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Abstract Pityriasis rubra pilaris (PRP) is a rare papulosquamous disorder of unknown etiology characterized by reddish orange plaques with pityriasiform scaling with follicular keratoses, palmoplantar keratoderma, and occasionally erythroderma. We hereby report a family with five members affected with the condition in three successive generations.

Key words
Pityriasis rubra pilaris, acitretin, familial.

Introduction

Pityriasis rubra pilaris (PRP) is a rare papulosquamous disorder of unknown etiology with an estimated incidence of 1 in 5000 to 1 in 50000 patients.[1] It was first described by Devergie in 1857.[2] It is characterized by reddish orange scaly plaques with pityriasiform scaling showing follicular keratoses, palmoplantar keratoderma, and sometimes erythroderma.[1] The familial subtype, mainly type V PRP is rare and inherited as an autosomal dominant disorder with variable penetrance.[1] Other forms of inheritance as autosomal recessive and X-linked forms have also been described. We hereby report a case of familial PRP, affecting three generations of a family.

Case report

A 30-year-old male patient presented with multiple, reddish scaly lesions distributed over the body along with thickening of skin of palms and soles since childhood. The lesions started initially on dorsum of hands, bilateral elbows, knees and gradually spread to other parts of body. Lesions were asymptomatic except for mild itching. Patient gave history of similar lesions in his father, father's sister, daughter and son (**Figure 1**).

On examination, there were multiple, well-defined, erythematous scaly plaques mainly over trunk, bilateral upper and lower limbs, knees, elbows and nape of neck (**Figure 2a, 2b**). Examination of patient's daughter also showed similar erythematous scaly plaques over knees, elbows and face (**Figure 2c, 2d**). Lesions in son were few and restricted to the bilateral elbows. Islands of normal skin were present in between the plaques. Palms and soles showed waxy diffuse yellowish keratoderma. Hair and mucosae were normal. Finger- and toenails showed nail plate thickening, subungual hyperkeratosis and yellowish discoloration. Systemic examination was within normal limits. Routine investigations including complete blood counts, liver and renal function tests, urinalysis, chest radiographs were within normal limits.

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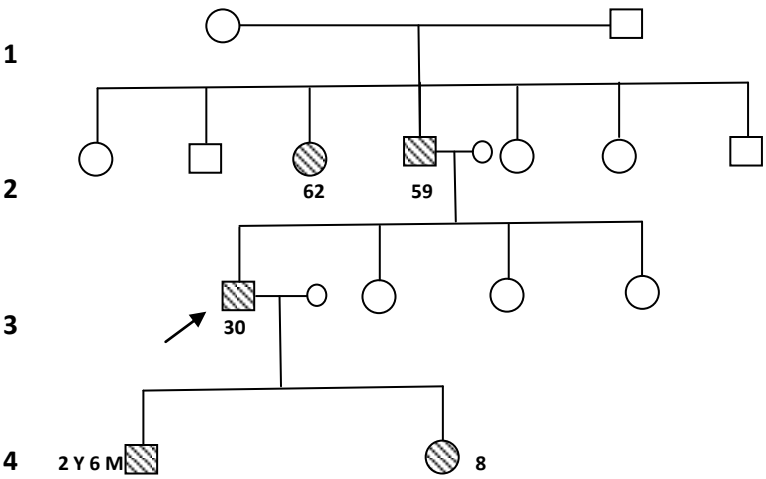

Figure 1 Family pedigree showing five members of the family affected, in three successive generations. The ↗ shows the index case.

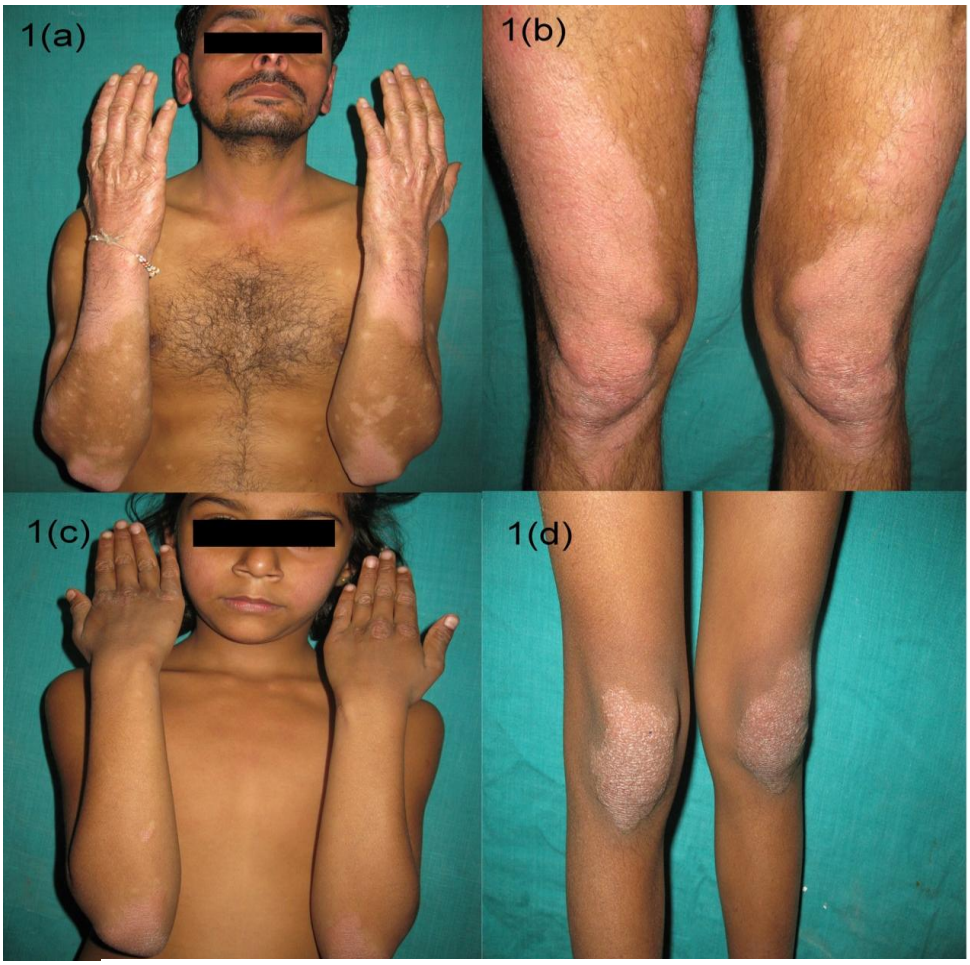

Figure 2(a) Well-defined erythematous plaques over bilateral elbows, dorsum of hand, forearm and face in a 30-year-old male. **2(b)** Well-defined erythematous scaly plaques over bilateral knees extending up to thighs in the same patient. **2(c)** Well-defined erythematous scaly plaques over bilateral cheek, knuckles and elbow in 8-year-old-daughter of the patient. **2(d)** Similar kind of erythematous plaques over bilateral knees in the daughter.

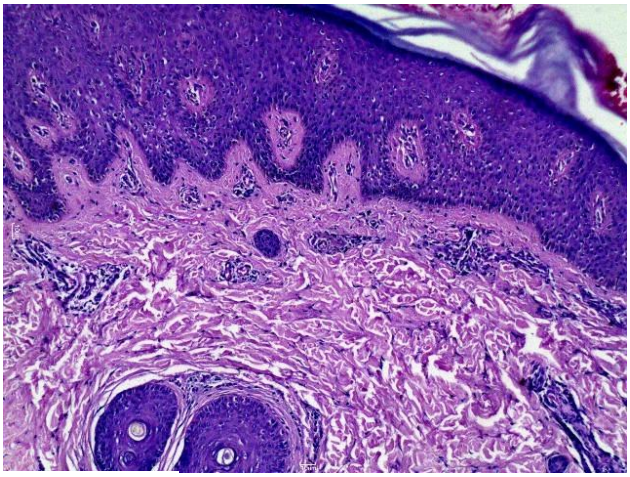

Figure 2 Sparse superficial perivascular lymphohistiocytic infiltrate with irregular psoriasiform hyperplasia of the epidermis showing alternate zones of parakeratosis and orthokeratosis in stratum corneum along with hypergranulosis (Hematoxylin and eosin, x400).

Histopathological examination from a lesion over back showed irregular psoriasiform hyperplasia of the epidermis. Stratum corneum showed alternating zones of parakeratosis and orthokeratosis. Papillary dermis showed sparse superficial perivascular lymphohistiocytic infiltrate and mild edema (**Figure 2**). Histopathological examination of a biopsy from knee of his daughter showed similar findings.

Based on history, clinical examination and investigations, the diagnosis of PRP was confirmed. Patient was prescribed oral acitretin 25 mg twice a day along with antihistamines. The lesions showed marked improvement in form of reduction in erythema and scaling after 4 weeks of treatment. As the disease of patient's father, daughter and son was mild so they were prescribed topical mid-potent corticosteroids only along with antihistamines.

Discussion

PRP is a disorder of keratinization of unknown cause that progresses sometimes to erythroderma along with disabling keratoderma. The disease is subclassified into six types, including both hereditary and acquired forms.[1] Although most cases of PRP are sporadic, familial transmission consistent with autosomal dominant inheritance has been reported.[2,3,4,5] The underlying etiopathogenesis of PRP has not been fully elucidated. Several familial occurrences have led investigators to consider the genetic basis for this disorder. The derangement of vitamin A metabolism or deficiency of vitamin A as cause of PRP has been an area of debate. Good response of PRP patients to systemic or topical retinoids suggested a possible role of vitamin A metabolism in etiology of PRP.[4] Diagnosis is established by characteristic clinical presentation aided by histopathological examination.

Griffiths divided PRP into 5 categories based on clinical features, age of onset, and prognosis: classic adult type, atypical adult type, classic juvenile type, circumscribed juvenile type, and atypical juvenile type. More recently, an HIV-associated type was added to this classification system. The classic adult type (type I) accounts for 50% of cases and has the best prognosis, with 80% clearing by 3 years, whereas the familial form of the disease may persist throughout life. PRP is characterized by small follicular papules that coalesce into orange, scaly plaques, palmoplantar keratoderma, diffuse scaling of the scalp, and frequent progression to exfoliative erythroderma with characteristic islands of normal skin.[1] Histopathology is characterized by light microscopy findings of alternating orthokeratosis and parakeratosis, focal or confluent hypergranulosis, follicular plugging, broad rete ridges and sparse superficial dermal lymphocytic perivascular infiltration. Electron microscopic features of PRP include decreased numbers of keratin filaments and desmosomes, enlarged intercellular spaces, parakeratosis with lipid like vacuoles and large number of lamellar

granules, and a focal split of the basal lamina at dermo-epidermal junction with lymphoid cells in the dermis.[6,7] PRP has been associated with various cutaneous and systemic disorders including vitiligo, lichen planus, alopecia universalis, Kaposi varicelliform eruption, seronegative arthritis, myositis, myasthenia gravis, hypothyroidism, celiac sprue and HIV.[1] Rarely, internal malignancies have been recorded in adult onset PRP. Systemic retinoids are first-line therapy in the management, although the results are variable. Other treatment modalities that have been tried include photochemotherapy, methotrexate, topical calcipotriol, keratolytics, cyclosporine, azathioprine, fumaric acid esters, extracorporeal photochemotherapy and tumor necrosis factor alpha antagonists i.e. infliximab, etanercept.[4,8] This family represents a case of familial PRP with features of autosomal dominant inheritance. The treatment of PRP should depend upon various factors like age of the patient, gender and severity of the disease.

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