ORIGINAL RESEARCH PAPER

Volume-7 | Issue-3 | March-2018 | PRINT ISSN No 2277 - 8179

INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

"A RARE CASE REPORT OF FAMILIAL REACTIVE PERFORATING COLLAGENOSIS"

Dermatology

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ABSTRACT

Reactive perforating collagenosis is a rare form of transepidermal elimination of altered collagen through the epidermis. It can exist in two forms- a rare familial form and a commoner adult form. Very few cases of reactive perforating collagenosis have been reported in literature and here we are presenting a rare case, where two siblings were diagnosed with inherited form of reactive perforating collagenosis with a positive family history.

KEYWORDS

Inherited, familial reactive perforating collagenosis, transepidermal elimination.

Introduction:

Transepidermal elimination is the process in which material from the dermis is extruded to the exeterior with little or no disruption of the surrounding structure.[1] The following conditions can be regarded as primary perforating disorders: Kyrle's disease(KD), Perforating folliculitis(PF), Reactive perforating collagenosis(RPC), and Elastosis perforans serpiginosa(EPS). These disorders might be due to defect in epidermal keratinocytes , hair follicles ,collagen and elastic fibers respectively. Reactive perforating collagenosis is a rare form of transepidermal elimination in which there is an altered collagen extrusion through the epidermis.[2] Less than 50 cases have been reported worldwide in literature so far.[3] Here ,we present a very rare case, wherein both the brothers of a family had inherited form of RPC with positive family history. We are reporting these cases here because of its rarity and classical diagnostic features.

Case Report:

Two patients, a 11 year and a 8 year old siblings presented to the outpatient department with minimally itchy, hard papular eruptions since childhood, with a positive history of grandfather also suffering from the same clinical features with spontaneous resolutions and exacerbations.

On cutaneous examination ,multiple keratotic papules with central adherent keratotic plugs were present over extensor surface of extremities and face[Figure1],[Figure2]. Scarring was seen over the resolved lesions[Figure3]. Koebner's phenomenon was also present.[4] nails, hairs ,teeth and mucosa were normal. On systemic examination complete blood count, blood sugar, liver function test, kidney function test and thyroid profile was normal.

Biopsy from the leg and forearm of the elder brother revealed a typically perforated epidermis with collagen bundles extruding through the epidermis forming a cup shaped epidermal depression, which contained inflammatory cells and keratinous debris. In the papillary dermis, collagen was surrounded by focal epidermal proliferation[Figure4]. Elastic fibers were typically absent. Both the siblings were started on oral isotretinoin, topical steroids and keratolytics once applied daily. The lesions showed appreciable improvement after one month of treatment.

Discussion:

Reactive perforating collagenosis is a rare inherited skin disorder characterized by transepidermal elimination of altered collegen through the epidermis. It is usually precipitated by environmental factors like cold and trauma and associated with intense pruritus. An abnormal response to superficial trauma such as scratching may be implicated in the acquired form. This results in focal damage of collagen, which is then extruded. The acquired form may be associated with diabetes and haemodialysis. It has been reported in association with hypothyroidism and hyperthyroidism, liver disorder, lymphoma and periampullary carcinoma.[5] The inherited form starts in infancy and early childhood as small papules on the extensor surfaces of the hands, elbows and knees following superficial trauma. Each skin coloured papule increases in size over 3-5 weeks to about 6mm and become umbilicated, with a keratinous plug. The lesions regress spontaneously in 6-8 weeks to leave a hypopigmented area or a scar. Kobener's phenomenon may be present.[4] Molluscum, papular urticaria, perforating granuloma annulare, perforating serpiginous elastoma, perforating folliculitis and Kyrle's disesase have to be considered as differentials. The epidermis is typically perforated by disrupted collagen bundles extruding through the epidermis forming a cup shaped epidermal depression, which contains inflammatory cells and keratinous debris. In the papillary dermis, collagen is surrounded by focal epidermal proliferation. On electron microscopy the collagen appears normal ,but gives an abnormal staining pattern with trichrome and phosphotungstic acid hematoxylin.

Lesions are usually self healing without any treatment, but often reoccur. Topical retinoic acid may decrease the number of lesions. Anecdotal reports described successful therapy with isotretinoin, allopurinol, doxycycline, UVB and photochemotherapy.[6,7,8]

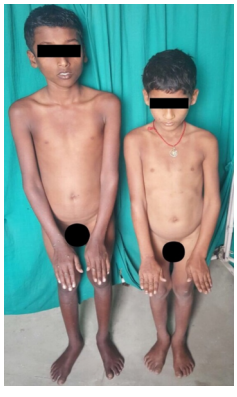

Figure 1. A 11 year and 8 year old siblings showing multiple keratotic papules over extensor surface of extremities.

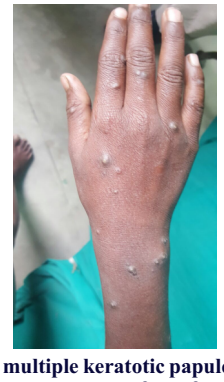

Figure 2. Classic multiple keratotic papules with central adherent keratotic plugs over extensor surface of extremities.

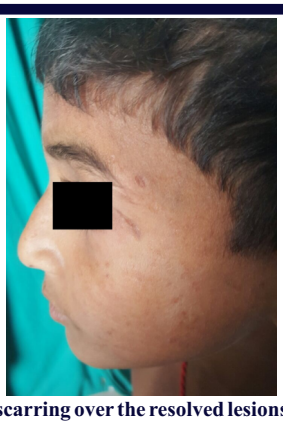

Figure 3. Visible scarring over the resolved lesions of the face.

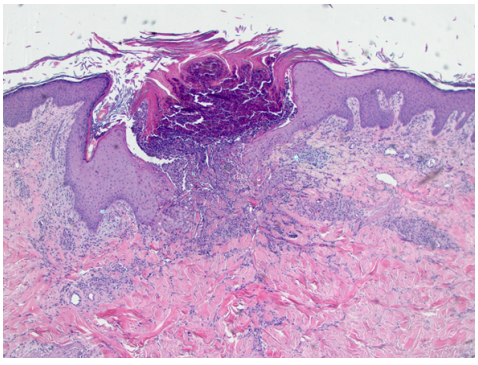

Figure 4. Typically perforated epidermis with the collagen bundles extruding through the epidermis forming a cup shaped epidermal depression which contains inflammatory cells and keratinous debris

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