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Epidermolytic Hyperkeratosis: A Case Report

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Abstract:
Epidermolytic hyperkeratosis is a rare dermatosis of autosomal dominant inheritance that affects both sexes equally. It is also called as Bullous congenital ichthyosiform erythroderma of Brocq. We report a case of 3 year old boy presenting with dark thick skin around neck, forearms, knees, hips since a year. The clinical examination and histopathological analysis confirmed the diagnosis. The discussion highlights the clinical manifestations, histopathology and management options in a case of Epidermolytic Hyperkeratoses.
Keywords: *Epidermolytic hyperkeratosis; genodermatoses;*

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I. Introduction:

Epidermolytic hyperkeratosis is a rare dermatosis of autosomal dominant inheritance that affects both sexes equally, with a prevalence of 1:100,000 to 1:300,000. The name was first introduced by frost and Van scott. It is also called as Bullous congenital ichthyosiform erythroderma of Brocq. It presents at birth with generalized erythema, blisters and erosions. In the months after birth, erythema and blistering decrease, while hyperkeratotic scaling that is especially prominent over the joints, neck and dorsum of hands and feet increases. The condition is caused by mutations in either keratin 1 or 10 gene, located on chromosome 12 or 17 respectively.

II. Case Synopsis:

We report the case of a 3 year old boy born to a non-consanguineous marriage who presented to our OPD with dark and thick skin around his neck, forearms, knees, hips and buttocks since 1 year. He had a history of generalized erythema and blistering at the time of birth. The patient is otherwise healthy with no other medical illnesses. On examination, the patient was a healthy looking young boy who had attained normal developmental milestones. The boy was non-toxic with multiple hyperkeratotic, hyperpigmented scaly plaques which were present around the neck, cubital fossa, elbows, popliteal fossa, knees, pelvis and gluteal region with prominent cobblestone pattern over the joints (Fig 1a & 1b). The lesions increased in winters with improvement under mositurizers and keratolytics like salicylic acid 12% creams. No similar lesions were present among the family members. Histopathologic findings confirmed the diagnosis(Fig 2). The patient was advised to apply moisturizers thrice daily, salicylic acid 12% and a combination cream containing urea 12%, lactic acid 6%, glycerine 3% and ammonium chloride 0.5% at night over the thick scales. The patient was advised to have regular follow-up in the OPD.

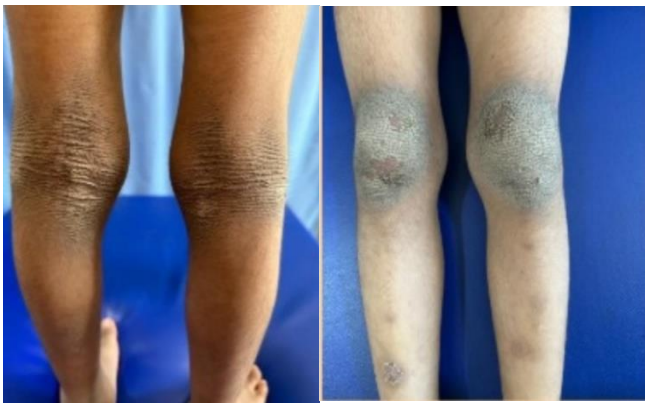

Fig 1a & 1b. Multiple hyperkeratotic, hyperpigmented scaly plaques present over B/L knees and B/L popliteal fossa.

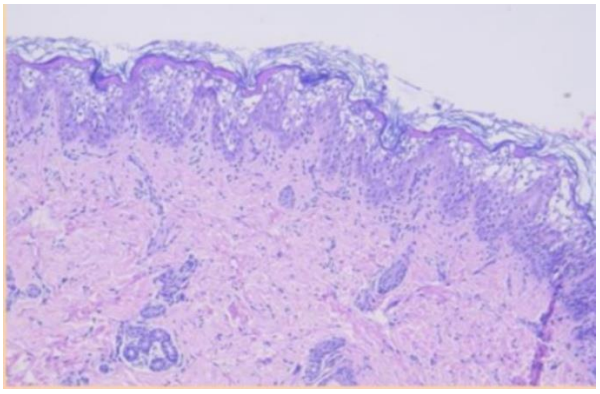

Fig 2. Histopathology shows 1) Hyperkeratosis, papillomatosis and acanthosis.
2) Basket weave epidermis with granular and vacoular degeneration.
3) Sparse perivascular and interstitial lymphocytic infiltrate in upper dermis.
4) Tonofilaments are lost- promote blistering.

III. Discussion:

Epidermolytic hyperkeratosis is rare genodermatosis with a prevalence of 1 in 100,000 to 300,000. It was first described by Brocq in 1902. It is an autosomal dominant condition with a 100% penetrance, although approximately 50% of all cases represent new spontaneous mutations. KRT 1 AND KRT 10 gene mutations represent the underlying cause. At birth the infant may show erythema, blistering and erosions resulting in a burnt child appearance. As the patient gets older, erythema and blistering becomes less apparent and later the disease is characterized by hyperkeratosis with cobblestone pattern (most prominent over joints) with pronounced ridging of the flexures. In approximately 60% of patients, palmoplantar hyperkeratosis develops and may result in recurrent painful fissures, contractures and sclerodactyly. Some cases are complicated by sepsis, fluid loss and electrolyte imbalance. Severely affected children may be of short stature, although may catch up in adolescence. The condition is associated with markedly increased epidermopoiesis. Keratin 1 mutation is associated with severe palmoplantar hyperkeratosis while keratin 10 is not. The patients are managed conservatively with the use of topical emollients, appropriate antibiotics and antiseptic creams when required. Topical calcipotriol helps in some cases. Oral retinoids may be required in severe cases, it reduces skin fragility and blistering. High concentration keratolytics can also be used.

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