CASE REPORT
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SKIN BLISTERING ASSOCIATED WITH SEVERE SCARRING AND PHOTOSENSITIVITY AFFECTING TWO SIBLINGS - KINDLER SYNDROME OR DYSTROPHIC EPIDERMOLYSIS BULLOSA? – A CASE REPORT

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Abstract

Kindler syndrome is an autosomalrecessive inherited condition characterized by acral bullae, progressive poikiloderma, photosensitivity along with mucosal involvement. Kindler syndrome and dystrophic epidermolysis bullosa, both can have similar clinical presentation and it may be difficult to differentiate them from each other, especially in neonatal period. We are reporting here the case oftwo siblings having photosensitivity with poikilodermatous changes, acral blistering and mucosal involvement, features clinically consistent with Kindler syndrome.

Key words: Skin blistering,Photosensitivity,Dystrophic epidermolysis bullosa,Kindler syndrome.

Introduction

Dystrophic Epidermolysis Bullosa (DEB) and Kindler syndrome are genetic blistering disorders which are characterized by traumatic blistering, which usually starts at birth or in infancy. The fragility of skin results in blisters appearing at trauma prone sites and these lesions usually heal with secondary changes like scarring, milia formation and nail changes. Dystrophic epidermolysis bullosa has both dominant and recessive pattern of inheritance and is characterized by a defect in collagen 7 protein which causes defect in the sublaminadensa, while Kindler Syndrome has recessive pattern of inheritance and is characterized by defect in Kindlin 1. In addition to the fragile skin, teeth and nail changes seen in DEB, Kindler syndrome has progressive poikiloderma, photosensitivity and mucosal inflammation also.[1,2]

Case Report

A 23-year-old female borne out of non-consanguineous marriage presented to our out-patient department with history of clear fluid filled lesions rupturing to leave behind crusted raw areas all over the bodysincethe age of 3 months, more over areas prone to trauma, like knees, elbows and dorsa of feet and hands. She also gave history of photosensitivity with burning sensation on exposure to sun and fluid filled lesions developing at the sites of sun exposure. There was also a history of similar complaints in her younger sister. On examination, patient had multiple well defined irregular shaped erosions with overlying crust present over both shoulders, elbows and knees. [Figure 1] There were multiple areas of atrophic scarring over both elbows, knees, dorsa of hands and feet with mild poikilodermatous changes.

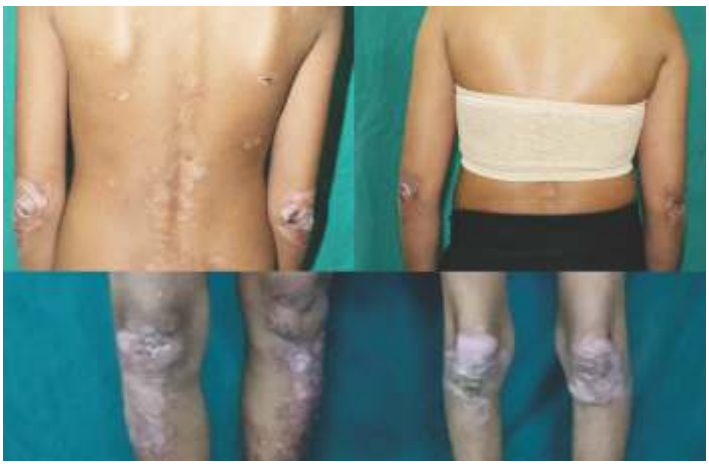

Figure 1 : Multiple well defined irregular shaped erosions with overlying crust present over bilateral elbow and knee

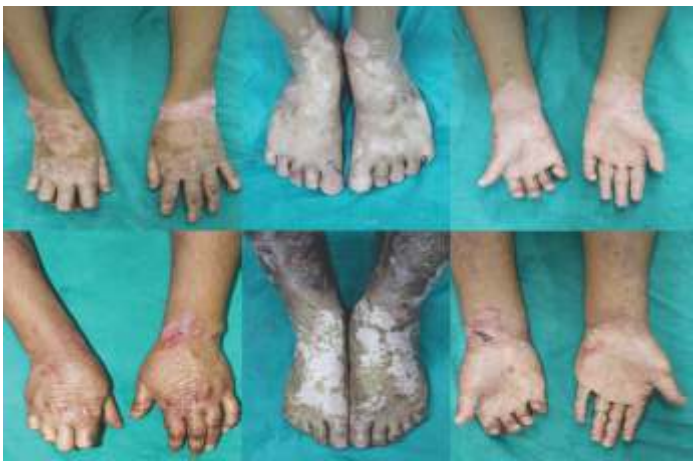

Figure 2 : Atrophic scarring over both elbows, knees, dorsa of hands and feetwith mild poikilodermatous changes, clawing of both hands and anonychia over bilateral fingers and toes

Figure 3 : Oral cavity showing multiple erosions with overlying slough over the hard palate and cheilitis

There was contracture with resultant immobile clawing of both hands and anonychia over bilateral fingers and toes. [Figure 2] In the oralcavity, there were multiple small erosions with overlying white slough over the hard palate and bilateral buccal mucosa; along with cheilitis. [Figure 3] She also had multiple, well defined, and flat topped, dark brown to violaceous atrophic papules and some plaques, predominantly over the forehead, upper chest and back and milia were present over both helix of ear. [Figure 4] The teeth were normal and there were no lesions on the scalp.

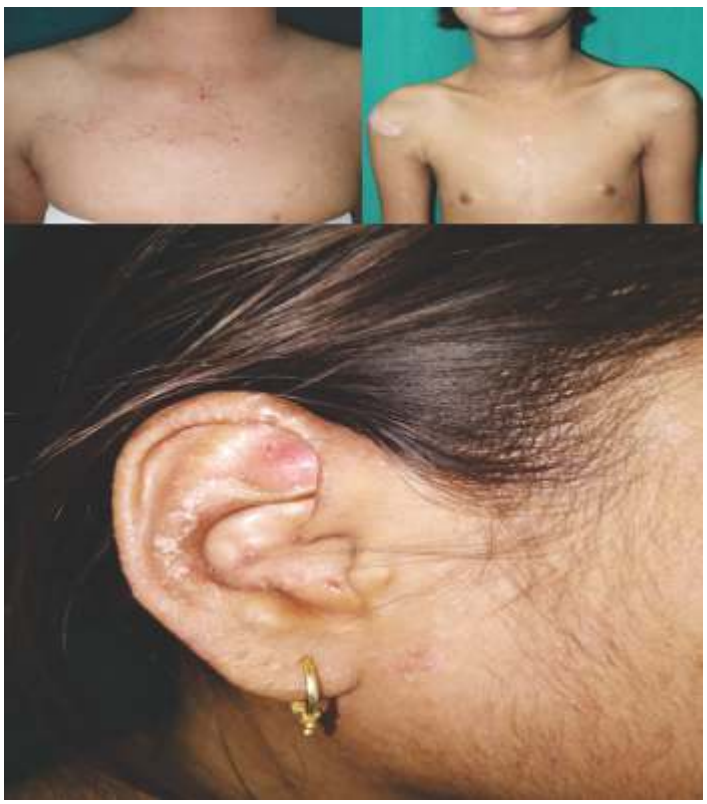

Figure 4 : 4Well defined dark brown to violaceous atrophic papules and plaques, over upper chest and back; milia over helix of ear

Routine hematological investigations were normal and a shave biopsy was sent from one of the lesions over her chest. Histopathological examination showed basal cell degeneration and split formation at multiple level at dermo epidermal junction along with melanophages in the dermis on hematoxylin and eosin stain (H&E). [Figure 5] Electron microscopy and genetic analysis could not be done because of resource constraints.

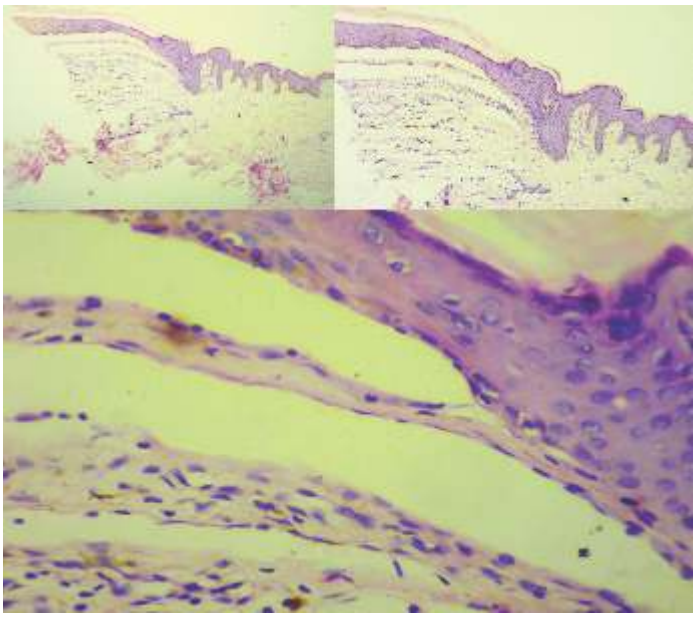

Figure 5 : Basal cell degeneration with split at multiple level at dermo epidermal junction along with melanophages in the dermis. (H&E stain 40x,100x,400x)

Discussion

Kindler syndrome was first described in 1954 by Theresa Kindler in a child with acral blistering, pigmentary changes and photosensitivity. The KIND1 gene encodes the protein Kindlin 1 which connects the actin cytoskeleton to the extracellular matrix. Recently, a novel mutation in FERMT1 gene has also been discovered.[3]Both dystrophic epidermolysis bullosa and Kindler syndrome can have similar clinical presentation and it may be difficult to differentiate them from each other, especially in neonatal period. There are many features which may help in differentiation, but the definite diagnosis can however be made after molecular studies only [Table 1].[4]

In Kindler syndrome, photosensitivity and acral blistering decrease with age, and appearance of poikiloderma and cutaneous atrophy tend to gradually worsen. Atrophic changes which typically appear as cigarette paper like wrinkled skin are most prominent over the sun exposed areas, most commonly site being the dorsal aspect of the hands and feet but may become generalized by adolescence. Mucosal involvement is common in both and may present as ectropion, corneal erosions, gingival inflammation, periodontal disease and scarring of the external urethral meatus. In Kindler syndrome, additional oral mucosal

findings include advanced periodontal bone loss, and leukokeratosis of buccal mucosa, trismus and a form of desquamative gingivitis.[5] Other less common features include ichthyosis, palmoplantar hyperkeratosis, light colored hair, pseudoainhum, nail changes including dystrophy and long and thick cuticle of nail,and an increased susceptibility to the development of squamous cell carcinoma.[6-9] The hands may develop pseudosyndactyly, similar to some cases of dystrophic EB. Chronic colitis may complicate the cases of Kindler syndrome sometimes.

The initial diagnosis relies upon careful clinical examination, family history and establishing the level of blister formation. Traditionally, transmission electron microscopy and immunofluorescence microscopy using a panel of antibodies against the candidate proteins implicated in EB are the preferred methods. The main objectives of skin biopsy are first to establish the level of blistering or tissue separation and, second, to search for other clues that may be indicative of the underlying disorder and therefore helpful in the diagnosis. Histopathologic examination alone cannot distinguish between dystrophic epidermolysis bullosa and Kindler syndrome. Kindler syndrome, shows variable plane of cleavage or duplicated lamina densa as compared to specific cleavage planes in epidermolysis bullosa. In addition, there may be epidermal atrophy, basal layer vacuolization, variable epidermal melanin content, dermal melanophages and capillary dilatation, which is more prominent in older patients when the poikiloderma sets in.[10] Immunostaining with anti-kindlin 1 antibody shows decreased staining of the epidermis in KS.[11]

Diagnostic criteria have been proposed by Fischer et al. and the presence of four major criteria makes a diagnosis of KS. The major criteria include:[12]

1. Acral blistering beginning in infancy

2. Progressive poikiloderma,

3. Cutaneous atrophy

4. Photosensitivity,

5. Fragility and/or swelling of gums.

The minor criteria include syndactyly and involvement of other mucosae. Additional features including palmoplantar keratoderma, ectropion, pseudoainhum, oral leucokeratosis, squamous cell carcinoma, onychodystrophy, skeletal abnormalities, and dental problems may be seen. The diagnosis is "confirmed" if four major criteria are fulfilled, as seen in our patient. The presence of three major and two minor criteria makes a "probable" diagnosis, while presence of two major and two minor/additional features makes the diagnosis "possible".

We have presented this case because of considerable overlap between these two conditions, and rarity of Kindler syndrome; it is important to diagnose Kindler syndrome because photo protectionto prevent any cutaneous malignancy needs to be advised to the patient along with management akin to dystrophic EB.

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