International Journal of Innovative Research and Advanced Studies (IJIRAS)
Volume 5 Issue 1, January 2018

ISSN: 2394-4404

Bullous Ichthyosiform Erythroderma: The Silent Stigma

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Abstract: ***Epidermolytic ichthyosis (EI), formerly known as epidermolytic hyperkeratosis (EHK) or bullous congenital ichthyosiform erythroderma (bullous CIE), is a form of congenital ichthyosis. It is inherited in an autosomal dominant fashion, with about 50% of cases representing spontaneous mutations. Epidermolytic ichthyosis presents at birth with erythroderma, blisters, and erosions and evolves over time into varying degrees of hyperkeratosis. We report 3-year-old girl with her 26 year old father with a generalized hyperkeratosis on the neck, truck, extremities with peeling and superficial erosions.***

Keywords: Ichthyosis form erythroderma, bullous, family trait

I. INTRODUCTION

Epidermolytic ichthyosis (EI), formerly known as epidermolytic hyperkeratosis (EHK) or bullous congenital ichthyosis form erythroderma (bullous CIE), is a form of congenital ichthyosis. It is inherited in an autosomal dominant fashion, with about 50% of cases representing spontaneous mutations so patients may have an affected family member; however, as many as half of reported cases arise as a result of periodic mutations. Rare autosomal recessive cases have also been reported. Epidermolytic ichthyosis is inherited in an autosomal dominant fashion, Epidermolytic ichthyosis presents at birth with erythroderma, blisters, and erosions and progresses over time into variable degrees of hyperkeratosis. Mutations cause defects that conciliation keratin alignment and assembly of intermediate filaments, leading to cellular collapse, blistering, and impaired barrier function. Compensatory hyperproliferation leads to hyperkeratosis.

Defects in genes for keratin 1 (*KRT1*) and 10 (*KRT10*) are the cause of epidermolytic ichthyosis. Epidermolytic ichthyosis is a lifelong condition. Some patients may experience amelioration of symptoms as they age. Risk for morbidity and mortality is highest in the neonatal period, where infants are at increased risk for complications such as sepsis and dehydration because of impaired barrier function.

Usually, these mutations are missense substitutions into the highly-conserved alpha-helical rod and the nonhelical H1 domains of the keratin proteins.

Palmoplantar keratoderma is usually associated with *KRT1* mutations; however, in rare cases, palmoplantar keratoderma may be observed in patients with *KRT10* mutations.

II. CASE REPORT

A 3 yea, old female child with 26 year, old father presented with multiple erosions to the skin OPD

Both were born of full term C section with colloidan body and had thick dark, itchy skin with fluid filled lesions at birth in flexures of both arms and symmetrical all over the body emitting foul odor.

O/e bilaterally symmetrical multiple hyperpigmented to lichenified velvety plaque present around the neck, flexor

ISSN: 2394-4404

aspects of arm forearm bilateral popliteal fossa bilateral groins intergluteal cleft with increased hair volume, fissures on both soles, loss of hyper linearity and thickening of palms and soles.

There was repeated history of fever and cold.

General and systemic examination are normal. Staphylococcal scalded skin syndrome and bullous congenital ichthyosis form erythroderma considered as differential diagnosis.

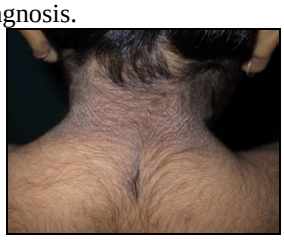

Figure 1: Neck of Daughter

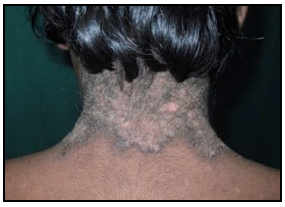

Figure 2: Neck of Father

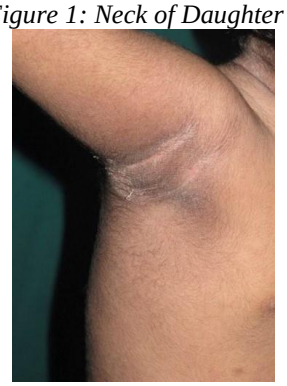

Figure 3: Axilla Of Daughter

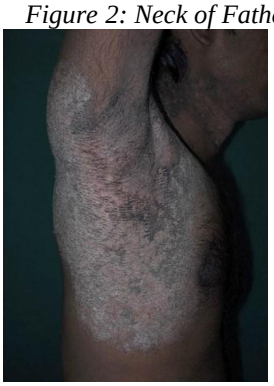

Figure 4: Axilla of Father

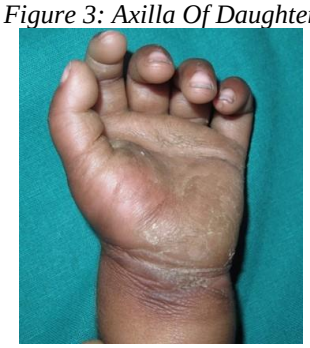

Figure 5: Palm Of Daughter

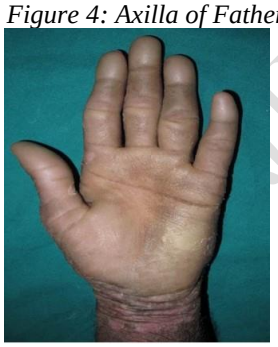

Figure 6: Palm Of Father

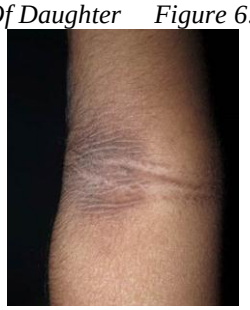

Figure 7: Popliteal Fossa of Daughter

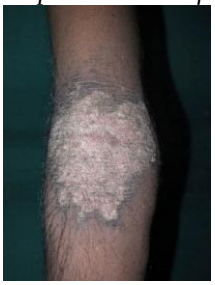

Figure 8: Popliteal Fossa of Daughter

III. INVESTIGATIONS

Both patients had normal blood chemistry. X-ray & USG studies, blood and wound cultures were negative.

IV. BIOPSY FINDING

Epidermis showing keratinized stratified squamous epithelium features of hyperkeratosis, koilocytic changes and elongated ridges. Dermis is fibro collagenous showing perivascular and interstitial inflammatory infiltrate.

Based on this finding we confirmed diagnosis as bullous congenital ichthyosis form erythroderma.

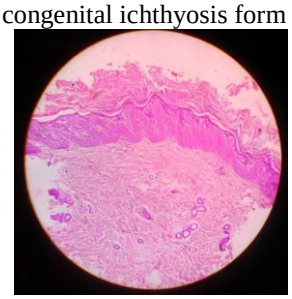

Figure 9: Biopsy of Daughter

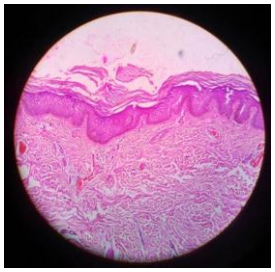

Figure 10: Biopsy of Father

V. DISCUSSION

From birth the skin is noted to be fragile with blisters and peeling. Often there is no evidence of ichthyosis at birth and the skin appears red with superficial erosions. During the first few years of life it may be difficult to distinguish non-bullous from bullous ichthyosis from erythroderma. Patients with generalized epidermolytic ichthyosis may be born to parents with epidermolytic epidermal nevi (mosaic epidermolytic ichthyosis) However, the histological changes in later are diagnostic, with vacuolation and cavitation in the upper layer of the epidermis and the disorder is sometimes to be referred to as epidermolytic hyperkeratosis. Once blisters have developed, there is usually no diagnostic difficulty, but they may be minor feature of disorder and not noticed by the patient [16]. The hyperkeratotic lesion in the feature in the flexure, often have a characteristically moist, rather, yellow appearance. Bullous ichthyosis form erythroderma, is inherited as autosomal dominant trait. Novel mutations in both genes continue to be reported.

If the mutation also involves gonadal cells, which is thought to be more likely in patients with more extensive cutaneous involvement, affected individuals can have offspring with generalized epidermolytic ichthyosis. From early childhood, the skin becomes scalier and the redness and blistering less obvious. The skin thickening can affect any pad of the body but is most prominent on the scalp, around the neck and in the skin creases of the armpits, elbows and knees. Many patients with this condition develop thickening of the skin of the palms and soles. Older children and adults suffer from repeated skin infections especially in the skin folds.

It is possible that one of the parents may have a dark warty birth mark usually in a line, which may be the only expression of this disorder.

Accurate diagnosis of epidermolytic ichthyosis (EI) is important in order to properly inform and counsel parents. Genetic counseling and prenatal diagnosis also can be offered [18] Newborns with epidermolytic ichthyosis who have denuded skin are at increased risk for infection, secondary sepsis, and electrolyte imbalance. These newborns should be transferred to the neonatal ICU to monitored and treated as needed. They should be handled gently to avoid further trauma to the skin.

Wound care for blistering and moisturization/emollients are important in the newborn period. In older children, topical emollients and topical keratolytic are generally the mainstays of treatment. The accumulation of scale predisposes to overgrowth of bacteria, in particular with *Staphylococcus aureus*, which is often associated with odor. Patients may benefit from the use of mild antibacterial soaps or dilute bleach baths. Some patients may also benefit from therapy with oral or topical retinoids.

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