An Unusual Case Report of Dystrophica Epidermolysis Bullosa in a Child

Sneh Kumar[1], Amar Taksande[2], Shruti Chaudhary[3], Revat Meshram[4], Sachin Damke[5]

[1, 2, 3, 4, 5] Department of Paediatrics, Jawaharlal Nehru Medical College, Sawangi, Wardha, Maharashtra, India.

INTRODUCTION

Epidermolysis bullosa (EB) is a genetically inherited severe skin disease involving dermal-epidermal junction. Based on the appearance and involvement, it is grouped into simplex, junctional & dystrophic forms. These disorders represent heterogeneous phenotypes and are correlated with a variable range of complications, from localized skin fragility to neonatal death. Genetic testing had made a precise diagnosis and it requires only supportive and symptomatic therapy. Here we report an atypical case of dystrophica epidermolysis bullosa in a 6-year-old male child.

Epidermolysis bullosa (EB) is a general term used for heterogeneous group of congenital, genetic blistering disorders. It has a wide spectrum of clinical presentations.[1] It is characterized by induction of blisters by trauma, exacerbation of blistering in warm weather and healing with scarring. EB can be categorized under three major groups - epidermolysis bullosa simplex (EBS), junctional epidermolysis bullosa (JEB) and dystrophic epidermolysis bullosa (DEB). EB simplex has an incidence and a prevalence rate of 10.75 and 4.65, 2.04 and 0.44 of junctional EBs and 2.86 and 0.99 of dystrophic EBs and recessive dystrophic EB 2.04 and 0.92 respectively.[2] The dystrophic forms are characterized by deformities of the skin including coalescence of the fingers, nail changes and milia formation.[3] This case report highlights the rare presentation of recurrent episodes of blisters and limb deformities in 6 - year - old male children.

PRESENTATION OF CASE

A 6-year-old male child, born out of non-consanguineous marriage presented with a complaint of skin lesions all over the body since birth. There was no history of fever, cough, vomiting, ear discharge, trauma, convulsion, headache, unconsciousness or weakness in the limbs. At birth, the child had blisters on the left lower limb which was progressed to the whole body within a month. The child's developmental milestone was normal. Parents stated that the child could not walk because of blisters in both legs. They also complained of the frequent occurrence of oral ulcers.

Grossly, the child looked emaciated and thin built. On anthropometric examination, his weight was 7 kgs and height 73 cm which was suggestive of severe stunting and severe wasting as per WHO classification. His vitals were temperature of 38.5°C, a pulse of 92 beats / minute, and blood pressure of 96 / 60 mmHg, respiratory rate of 20 breaths / minute, and oxygen saturation of 98 % in room air.

Corresponding Author:
Dr. Amar Taksande,
Department of Paediatrics,
Jawaharlal Nehru Medical College,
Sawngi, Wardha, Maharashtra, India.
E-mail: amar.taksande@gmail.com

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On general examination, there was mild pallor, no icterus, and cyanosis or clubbing. There was no cervical lymphadenopathy. Both hands and feet showed mitten deformity with pseudosyndactyly. Hands showed flexion contracture of all fingers (Figure 1), adduction contracture of thumb and anonychia. Atrophic scarring was seen over the abdomen, bilateral upper limbs and lower limbs. Multiple superficial erosions were seen over the medial aspect of the right arm, abdomen, and chest with areas of hypo and hyperpigmentation in between. Pseudosyndactyly along with multiple ulcers over the medial aspect of the left knee, shin of the tibia and left foot with sloping edges associated with the erythematous base and purulent discharges were present (Figure 2). All deciduous teeth were present with the oral cavity. Bilateral corneal haziness was present on ophthalmic examination. Normal heart sounds were auscultated on cardiovascular examination and air entry was bilaterally equal on respiratory examination. His abdomen was soft without hepatosplenomegaly. Neurological examination was normal.

Figure 1.
Flexion Contracture of All Fingers along with Hypo and Hyper Pigmentation Over Chest and Abdomen

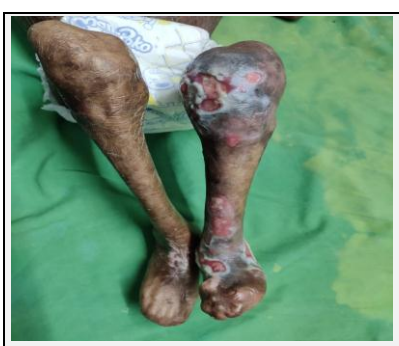

Figure 2.
Superficial Erosions Over Left Knee and Ankle along with Bilateral Pseudosyndactyly

Laboratory investigations revealed haemoglobin 9g / dl, WBC 6,200 / mm^3, platelets 1.6 lakh / mm^3. He had normal serum electrolytes and renal function tests. Parents were not willing for the neuro-imaging study. A diagnosis of dystrophic epidermolysis bullosa was made and the local application of fusidic acid and physiological hypoallergenic lotion was started.

DISCUSSION

EB is a heterogeneous group of rare diseases characterized by skin and mucosal blistering when exposed to mechanical stresses.[4] In 1870 Von Hebra first listed it as "erblichen pemphigus". In 1886 Koebner[5] introduced the present term 'epidermolysis bullosa hereditaria'. While further classification was first done by Pearson in 1962.[6] This disease includes inherited conditions of the skin's fragility that are marked by skin and mucosa blistering in reaction to little or no noticeable trauma. These conditions are heterogeneous and have various complexes spanning from localized skin fragility to neonatal death.[2]

According to the extent of dermo-epidermal separation in the basement membrane region, three main classes - simplex, junctional and dystrophic were described.[7,8]

The separation in the EB simplex (EBS) is intraepidermal. It is at the thick lamina of the junctional EB (JEB) and under the membrane in dystrophic EB (DEB).[2-5] EBS is a none scarring, autosomal dominant or recessive disorder. The defect is in keratin 5 or 14. The filaments of basal keratinocytes are malformed, leading to intraepidermal bullae formation. It is majorly classified further into generalised and local forms based on the area of involvement. Bullae heal with minimal or without scar formation. The localized form commonly involves hands and feet.

JEB can be Herlitz or Non Herlitz type. The Herlitz is fatal involving most of the body parts right from birth. The mucosa of the systemic epithelium is affected and healing is delayed. Septicaemia is a common cause of death. The children fail to thrive, have teeth loss, infection, and anaemia. In Non –Herlitz type, the blisters are present from the neonatal period but the associated condition is milder. On microscopy, a cleavage plane in lamina lucida is observed in the basal cells.

DEB results from a mutation in collagen which is a major supportive anchoring fibril of the basement membrane, keeping epidermis and dermis together. The dominant variant is the most common type with variable presentation. Blisters may be present at birth mostly found over bony prominences with good healing. A recessive form of DEB is severe and generalised associated with severe involvement of all body parts. The blisters are frequent, scarring and milia formation is pre-dominant. A similar finding was present in our case also. The stricture formation of mucosa and squamous cell carcinoma is common. Scarring and fibrosis cause pseudosyndactly. Septicaemia is the most common cause of death.

The treatment is mainly supportive. A definitive diagnosis of inherited EB is made with the monoclonal antibody assay and with transmission electron microscopy (TEM) and immunofluorescence (IF) antigen imaging. The skin needs to be properly biopsied, as stated by Intong and Murell, to acquire a correct diagnosis.[9] Treatment is usually symptomatic and supportive which includes oral iron therapy, oral hygiene and regular dressings.

CONCLUSIONS

Epidermolysis bullosa is a poor prognostic, genetically based disease with skin fragility. The diagnosis was based on clinical results. Thus, this is an uncommon report of a male child of six years of age of dystrophic epidermolysis bullosa.

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