

Epidermolysis Bullosa Simplex-Koebner: A Case Report

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ABSTRACT

Epidermolysis Bullosa Simplex (EBS) has been sub-classified into various subtypes on the basis of clinical features and histopathology predominantly into three divisions: Simplex, Junctional and Dystrophic. EBS is characterised by inter-epidermal blistering with minor internal grossment. It's always inherited in an autosomal dominant fashion and is also the least severe

type of EB. More recently a recessive form of EBS has been described. The Koebner type of EBS (EBS-K), also known as the non-dowling meara or more recently the other generalised form, is usually presented at birth and is one of the rare presentations of EBS. Though always present with an extensive family history EBS-K is very rarely associated with de novo mutation.

Keywords: Blistering of skin, De novo mutation, Newborn, Non-hemorrhagic bullae, Secondary bacterial infection

CASE REPORT

A newborn male child, delivered at home, was presented at the NICU of Sarojini Naidu Medical College, Agra, on very next day after delivery with a complain of blisters arising on various parts of his body since birth [Table/Fig-1]. The child was born to non-consanguineous parents. Gestation period was nine months and newborn was the third birth in order. On examination, he was in distress, afebrile with pulse rate of 128 beats per minute and respiratory rate of 48 per minute. He had a birth weight of 2600 grams and his systemic examinations were unremarkable. The blisters were present on his neck [Table/Fig-2], thorax and abdomen [Table/Fig-3], elbows and knees [Table/Fig-4] and feet [Table/Fig-5]. The blisters were of variable sizes, ranging from 1.5x1.5 cm on neck, hands and feet to 5x5 cm on thorax, abdomen and back. History of similar illness

has been reported in first sibling also, who died at the age of three months due to widespread infection. There was no history of similar illness in either of the parents any other member of their extended families. Mucosal surface was not involved including normal looking nail, palm, soles, scalp and oral cavity. Nikolsky's sign was negative. Gention violet was applied on the wounds to prevent it from infection. The blisters were soft in consistency, were filled with deep yellow coloured fluid [Table/Fig-6]. Skin biopsy was performed. On the basis of biopsy report and clinical features diagnosis of epidermolysis bullosa simplex was confirmed. Non-availability of other diagnostic modalities such as Transmission Electron Microscopy (EM) and genetic analysis studies, restricted the further confirmatory diagnostic investigations. A culture report indicated the signs of staphylococcal infection. Cefotaxime (1gm/10



[Table/Fig-1]: Epidermolysis bullosa simplex showing erosive vesicobullous lesions at sites of friction.

[Table/Fig-2]: A small area of erosion on neck. **[Table/Fig-3]:** A large area of skin sloughing on the abdomen. (images from left to right)



[Table/Fig-4]: Zone of skin sloughing in whole of the right knee. **[Table/Fig-5]:** Small area of skin erosion on left foot involving the great toe. **[Table/Fig-6]:** Epidermolysis bullosa simplex showing blisters, which are filled with fluid and have a deep yellow color. (images from left to right)

mL i.v 12 hrly) and vancomycin (500 mg diluted in 20 mL normal saline i.v 8 hrly) was administered to the patient. Due to widespread infection, despite of medication, the newborn condition deteriorated and he died.

Consent

Baby's parents have given the consent for the publication of images for academic purpose.

DISCUSSION

Epidermolysis Bullosa (EB) is a rare birth disorder typified by a tendency to form non-haemorrhagic bullae even on trivial trauma [1]. According to recent data the prevalence of EB has been estimated to 8-10 per million births with no gender predilection [2]. On the basis of blister formation, EB is further subcategorized predominantly into three divisions: Simplex, Junctional and Dystrophic of which simplex is the least severe. The Koebner is even rarer form of EB, with a prevalence of about two per million [3]. The more severe Epidermolysis Bullosa Simplex (EBS) subtypes include Koebner, Dowling-Meara, and Weber-Cockayne forms. EBS-K usually presents at birth and stages with blisters all over the body except scalp and lacks the typical nail involvement, which is common with other major subtypes of EB [4].

EBS is an assorted group of rare disorders, which are defined by a tendency to develop blisters on the skin and mucosae upon exposure to a trivial trauma [5]. This disease was first to be explained by Von Hebra in 1870 as 'Erblichen Pemphigus', which was later modified to its recent name 'Epidermolysis Bullosa Hereditaria' by Koebner, in 1886. The first classification of this disease was done even much later, in 1962 by Pearson [6].

Centered on the level of separation at the dermo-epidermal junction at the Basement Membrane (BM) zone, EB have been classified into three types - simplex, junctional and dystrophic. The level of separation in

EBS is present at epidermal zone, in junctional EB it is at the level of lamina densa and in dystrophic EB it is found below the basement membrane [7]. EBS has been sub-classified into various subtypes on the basis of clinical features and histopathology. Most cases of EBS are due to mutation in the cytokeratin genes 5 (KRT5) or 14 (KRT14). These genes encode for an intracellular proteins, K5 and K14 respectively, which are responsible for maintaining tissue integrity [7]. K5 and K14 are activated first at the beginning of stratification in the embryonic ectoderm but they are not required for the making and differentiation of epidermis. They largely affect the mechanical integrity of the basal keratinocytes, which causes a substantial trauma to induce a blister [8].

EBS-K is a subtype of generalized EBS and it is characterised by blisters and erosions arising on various parts of the body skin surface and mucosa. These blisters generally don't involve the palms and soles, distinguishing them from patients with localised EBS [4]. The occurrence of milia, scarring, and nail dystrophy is intermediary between that of localised EBS and EBS Dowling Meara, and extra cutaneous findings, other than infrequent blistering in the oral cavity, are rare. In the study subject the blisters spared the palm and soles, unlike the EBS localised the most common subtype of EBS. Milia formation and atrophic scarring was also absent but blisters were induced relatively more easily on pressure or trauma as compared with other type of EBS just like EBS-K. There was almost no extra cutaneous involvement as in the above case and the mucus membrane was generally spared.

The major mode of inheritance is mostly autosomal dominant in EB simplex and autosomal recessive in junctional EB [9] but rarely EBS is also inherited in autosomal recessive fashion. An extensive family history is almost always present with patients with the koebner forms of epidermolysis bullosa simplex, and

the occurrence of sporadic cases is relatively unusual as seen in our case in which two siblings are affected but there is no history of similar illness in either of the parents and their long-drawn-out family. Though there are chances of de novo mutation but such cases are of much severe presentation and are not that frequent [10]. None of the parents have a history of similar disease in their family. The case represents a typical feature of EBS-K but still its inheritance is not that in accordance with that of autosomal dominance. A careful history has revealed no history of blisters formation on pressure or trauma on either of the parents strongly pointing toward the autosomal recessive pattern of inheritance in this case.

CONCLUSION

EBS is one of the most well understood diseases of the EB category, but nevertheless despite knowing its etiology and pathogenesis we still lack the therapeutic options to cure the disease. The disease may be lethal if the lesions are widespread as the infection can spread to internal organs. Genetic counselling is the sole method to prevent the disease. The standard care for EB simplex is generally supportive and preventive, including avoidance of trauma, nutritional support and infection control.

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