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CASE REPORT

Epidermolysis Bullosa Simplex

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ABSTRACT

Epidermolysis bullosa (EB) is a rare group of inherited skin disorders that manifests as blistering of the skin in the varying degrees of severity. The severity can range from a mild, localized disease to a generalized, devastating process. The three major types of EB include simplex, junctional and dystrophic epidermolysis bullosa. EB simplex (EBS) is the most common and dominantly inherited disease. In EBS, the blisters are usually present at birth or appear during the neonatal period [1],[2]. Secondary infection is the primary complication. A newborn with extensive blistering of the skin, minimal oral lesions and secondary infection with a staphylococcus is described.

Key words: Epidermolysis bullosa, newborn, secondary bacterial infection

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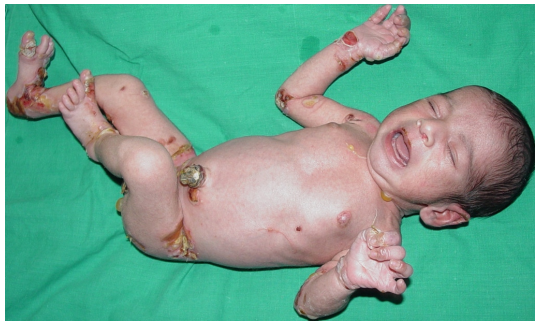
Introduction

Epidermolysis bullosa is a family of inherited skin disorders that is characterized by blister formation in response to the little or no apparent trauma. Epidermolysis bullosa simplex, junctional epidermolysis bullosa and dystrophic epidermolysis bullosa are the three major types. EBS is the most common among them [1],[2]. EBS may manifest either at birth or during the neonatal period. We describe a male neonate with blistering of the skin during the neonatal period.

Case Report

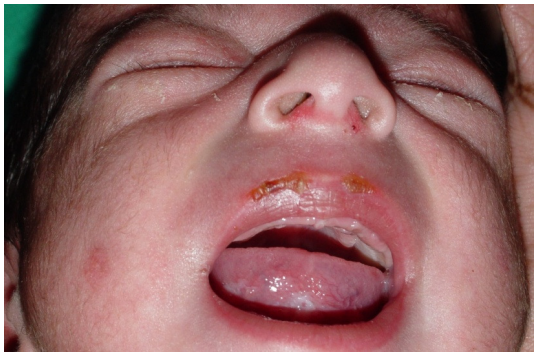
A 9-day old, male newborn was referred to us with a history of blistering of the skin since day 2 of life. He was born vaginally at 38 weeks of gestation to a 3rd gravida mother. Parentage was non-consanguineous. Antenatal period was uneventful. He cried soon after birth. His birth weight was 2740g. On the 2nd day of life, his mother noticed bullous lesions over the back which gradually progressed to involve the neck, trunk and the extremities. Minimal trauma elicited fresh bullae. Within 2-3 days, bullae ruptured with serious discharge and healed without scarring. At admission, neonate had various stages of multiple clear fluid filled vesiculobullous lesions over the neck, trunk and extremities. The size of the lesions varied from 1×1cm to 4×4 cm [Table/Fig 1]. Ruptured bullae and newly erupted bullae were seen at various places. There were raw areas at the site of ruptured bullae.

[Table/Fig 1]. Multiple blisters and skin sloughing in a newborn



Nikolky sign was negative. There were minimal perioral lesions [Table/Fig 2]. Oral cavity, conjunctiva, cornea, nails, scalp and genitalia were normal. Systemic examination was normal. There was no family history of bullous skin lesions. Few lesions had crusting.

[Table/Fig 2]. Minimal lesions on the lips



A diagnosis of EBS was considered. Dermatologists confirmed the diagnosis. Biopsy was not considered in view of characteristic lesions and their distribution. Gram stain of the serious discharge from the lesions showed few pus cells. Culture grew Staphylococcus aureus. With erythromycin therapy and local mupirocin, discharge from the lesions had decreased and general condition of the neonate had improved.

Discussion

Epidermolysis bullosa (EB) is a group of inherited skin disorders that presents as blistering of the skin. The severity varies [1],[2],[3],[4]. Most cases are inherited as autosomal dominant or recessive [1],[2], [5]. Based on the precise structural level at which the split responsible for blistering occurs the condition has been categorized into three main types: simplex, junctional and dystrophic.

EB simplex (EBS), a dominantly inherited disorder, is the most common. The dermal-epidermal junction of the newborn skin is a vital area of attachment. Any defects in the components that compromise this junction can lead to fragility of the skin. Blisters are usually present at birth or appear during the neonatal period. Sites of predilection are the hands, feet, elbows, knees, legs and scalp. Intraoral lesions are minimal. Nails rarely become dystrophic and usually regrow even when they are shed. The dentition is usually normal. EBS blisters typically heal with minimal to no scar or milia formation. The extent and severity of disease improves with age [1].

The defect in epidermolysis bullosa simplex is in the cytoskeleton structural proteins, keratin 5 or 14, which makes up intermediate filaments of the basal keratinocytes. The intraepidermal bullae result from cytolysis of the basal cells. Disease severity correlates with the location within the keratin gene mutation. Mutations in highly conserved genetic sequences lead to severe disease, whereas mutations in less critical keratin gene regions manifest as milder, more localized symptoms. In Koebner variant, there is generalized blistering but no mucosal or nail changes. In the Weber-Cockayne form, blistering is confined primarily to the easily traumatized palms and soles. Patients with the most severe subtype, Dowling-Meara EBS, have generalized blistering, prominent nail thickening and sloughing, and significant mucosal erosions. Our case had generalized blisters with no involvement of mucosa or nails and hence satisfies as Koebner type [1],[2],[4].

Treatment involves supportive care and prevention of complications. Wound management, nutritional support, and infection control are important aspects of care [1],[2],[4]. The skin should be protected from trauma, including tapes and adhesives. The blister should be punctured with a sterile needle or a blade by leaving the roof of the blister intact. This may prevent the accumulation of fluid and pressure and may thus prevent the blister from extending. If the blister repeatedly refills with fluid, it should be drained several times. Complete and gentle drainage of the fluid, leaving the blister roof intact and covering the affected area with white petrolatum–impregnated gauze help optimal healing. Non-adhesive dressing pads or vaseline-impregnated gauze covered by soft, bulky dressings are ideal. Topical antibiotics, such as mupirocin, may be used prophylactically. Frank crusting or serous drainage is an indication for systemic antibiotics. Secondary infection is the primary complication. In our case, the lesions were secondarily infected with staphylococcus.

Nutritional support is important for adequate growth and development and to promote optimal wound healing. To families of affected children, prenatal diagnosis using molecular techniques offers genetic counseling [1], [5].

Consent

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

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Conflicts of interests: none

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