INHERITED EPIDERMOLYSIS BULLOSA: A CASE REPORT

Shyam Kumar Mahato[1], Susana Lama[2], Nikhil Agarwal[2], Nagendra Chaudhary[1]

ABSTRACT

Epidermolysis bullosa (EB) is a heterogeneous group of genetically determined, mechano-bullous disorders characterized by blister formation in response to mechanical trauma. The blistering of the skin occurs in the varying degrees of severity and can severely incapacitate the life of the afflicted patient. Epidermolysis Bullosa Simplex (EBS), the most commonly occurring type, is dominantly inherited where treatment still remains a major challenge. We report a newborn female with blistering of the skin during the immediate neonatal period.

KEY WORDS: Blistering disorders; Epidermolysis bullosa simplex; Hereditary; Mechanobullous

1. Assistant Professor, Department of Pediatrics, Universal College of Medical Sciences & Teaching Hospital, Bhairahawa, Nepal

2. Junior Resident, Department of Pediatrics, Universal College of Medical Sciences & Teaching Hospital, Bhairahawa, Nepal

For Correspondence
Dr. Nagendra Chaudhary
Department of Pediatrics,
Universal College of Medical Sciences
& Teaching Hospital,
Bhairahawa, Nepal
E-mail: enagendra@hotmail.com

INTRODUCTION

Epidermolysis bullosa (EB) consists of a group of hereditary skin disorders characterized by formation of skin blisters and ulcers following minor skin trauma[1]. As the areas of the body most often affected in EB are sites subjected to frequent pressure or friction, it is also known as mechano-bullous disorder[2].There are mainly 4 major types of EB disorders: Epidermolysis bullosa simplex (EBS), junctional epidermolysis bullosa, dystrophic epidermolysis bullosa and kindler syndrome [3] where EBS is the most commonly occurring type with blisters usually presenting at birth or during the neonatal period [4].

CASE REPORT

A single, term, female baby, small for gestational age, product of non-consanguineous marriage, no antenatal and natal complications, delivered to a multiparous Rh-negative mother by normal vaginal delivery, cried immediately after birth, with birth weight of 2340 g was admitted to neonatal intensive care unit (NICU) of tertiary care hospital (Western Nepal). Baby was put on breast feeding within 30 minutes of birth. Baby had reddish discoloration of limbs (Fig: 1) with skin peeling noticed at birth. During the hospital stay, the baby developed new blisters over limbs and trunk which ruptured spontaneously along with skin peeling (Fig: 2 & 3) and reddish discoloration. Parents gave a history of similar events occurring in three previous siblings, out of which two were females and one male. All the siblings had expired with the first female at 3 months of age, the second female at one and half month and male baby at 1 month of age. On detailed examination of the present case, there was reddish discoloration seen on both legs, more on right than left, extending from knee up to the feet and sole and over the both hand extending up to fingers. Fresh skin lesions in the form of ruptured blisters were seen on the back, buttocks and nape of neck. Oral cavity, conjunctiva, cornea, nails, scalp and genitalia were normal. Systemic examination was within normal limits.

A diagnosis of Epidermolysis bullosa simplex was made clinically. Skin biopsy was planned but could not be done due to parent`s refusal. Baby was managed conservatively and nursed on thick/soft foam pad to prevent trauma inducing new blister formation. Baby was stable and doing well except that there was appearance of new skin lesions mainly at back and buttocks. The parents were explained about the baby's condition. The baby was discharged on day 3 on parents` request who succumbed on day 15 of life at home.

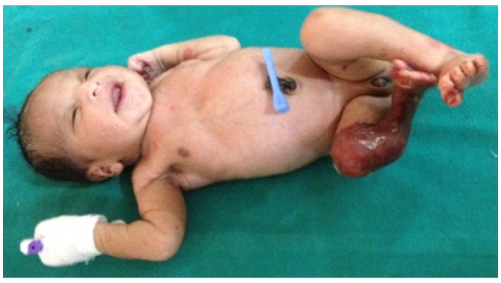

Figure 1: EBS with reddish raw area on right leg

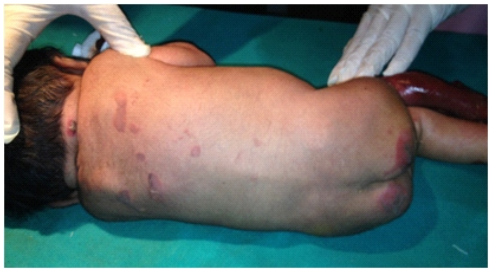

Figure 2: EBS with new lesions on neck, back and buttocks (pressure sites)

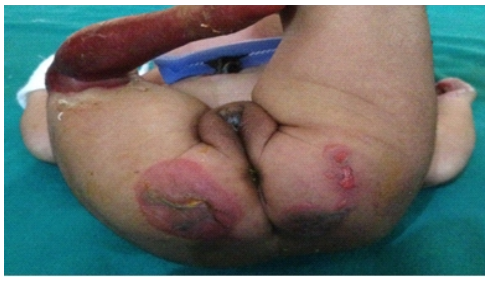

Figure 3: Ruptured blisters on buttocks with raw areas

DISCUSSION

Inherited EB encompasses over 30 phenotypically or geno - typically distinct entities which share a common feature of mechanical fragility of epithelial lined or surfaced tissues, most notably the skin. These disorders represent heterogeneous phenotypes from localized skin fragility to neonatal death. A characteristic feature of all types of EB is the presence of recurrent blistering or erosions on minor traction to these tissues. The reported incidence is approximately 1 in 17 000 live births with an estimated 500 000 cases worldwide[5]. However, in many countries, the actual percentage of children born with EB is unknown[2]. EB has not been reported

from Nepal so far to the best of our knowledge. Incidence is not affected by race or ethnic group[6], and the disease affects both sexes equally.

There are 4 major types of inherited EB, differ not only phenotypically and genotypically but also by the site of ultra-structural disruption or cleavage.[7-8]

Table 1: Classification of EB

Major EB type	Level of Blister formation	Level of skin cleavage	Known targeted antigen/protein(s)
EB Simplex	Intraepidermal	Subcorneal/suprabasal/mid-epidermis("epidermolytic")	Keratins 5 and 14
Junctional EB	Intra–lamina lucida	Intra-lamina lucida("lamina lucidolytic")	Laminin-332, type XVII collagen
Dystrophic EB	Sub–lamina densa	Sub-lamina densa("dermolytic")	Type VII collagen
Kindler syndrome	Multiple levels (Intra-lamina lucida and sub-lamina densa)	Mixed	Kindlin-1

Inherited EB is transmitted as either an autosomal dominant or autosomal recessive disease, depending on EB type and subtype.[9] Mutations and change in targeted proteins/antigens in various types of EB has been mentioned in table 1.[9-11]

The hallmark cutaneous features of inherited EB, in addition to mechanically fragile skin and easy inducibility of blisters or erosions at birth or neonatal period, may be the presence of milia, nail dystrophy, and scarring (usually atrophic). Other findings include exuberant granulation tissue (periorificial, axillary vaults, nape of the neck, lumbosacral spine, periungual and proximal nail folds), localized or confluent keratoderma of the palms and soles, and dyspigmentation (postinflammatory hypo- or hyperpigmentation, mottled or reticulate hyperpigmentation). Rarely, decreased or absent hair, albopapuloid lesions (flesh-colored or hypopigmented papules, usually arising on the lower trunk), and hypo- or hyperhidrosis are seen. The presence or absence of one or more findings may be age-dependent. Common sites of blisters are hands, feet, elbows, knees, legs and scalp.[5] This is similar in this case in which hands and feet were predominantly affected. EBS blisters typically heal with minimal to no scar and do not result in skin atrophy.[5] Secondary infection is the primary complication and the cause of death is resulting septicemia.

The diagnosis of EB is mainly clinical and confirmed by genetic analysis of the patient and the parents to definitively determine the mode of inheritance.[12] Skin biopsy and immunofluorescence mapping is a useful diagnostic tool to determine the plane of separation and to identify the protein affected in each case.[13] The protein affected in a specific case can be identified by analyzing the expression and distribution of antibodies to laminin-332 (formerly known as laminin-5), collagen VII, collagen XVII, plectin, a6b4 integrin, and keratin 14.[14] In present case, biopsy was not consented and facilities for genetic studies were not available with us. Antenatal diagnosis is also possible.[15-17]

There is presently no definitive cure for EB. The objective of treatment is to alleviate symptoms and provide supportive measures. Therapy is therefore focused on the prevention of lesions and complications which requires multidisciplinary approach involving pediatrician, dermatologist, surgeon, nutritionist, dentist, physiotherapist, nurse, psychologist, pain specialist, and geneticist.[18] Psychological support for parents and family members is vital. EB is not a contraindication for any vaccination.[14] The presence of multiple wounds of varying duration and ability to heal makes management of EB difficult and complex. The babies with EB should be given expert nursing care on thick foam pads which protect them from undue trauma induced blister formation. Special precautions need to be taken for older children in the use of adhesive tapes, sphygmomanometer cuffs, tourniquets and other instruments that cause shearing of skin or mucous membranes.[19] The erosions should be cleaned with sterile normal saline and covered with dressing pads or vaseline impregnated gauze. Topical antibiotics should not be applied to prevent development of antibiotic resistant bacteria. Oral and dental care should be kept in consideration as tooth eruption starts. Psychological support for parents and family members is vital .[14] Nutritional support is important for adequate growth and development and to help in optimal wound healing. Parents should be advised about prevention of trauma and occurrences of new lesions.[3]

CONSENT

Verbal consent was taken from the parents.

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