Scholars Journal of Applied Medical Sciences (SJAMS) ISSN 2320-6691 (Online)
Sch. J. App. Med. Sci., 2014; 2(1B):149-151 ISSN 2347-954X (Print)

(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com DOI: 10.36347/sjams.2014.v02i01.0030

Case Report

Epidermolysis Bullosa in Newborn: A Case Report

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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 and can severely incapacitate the life of the afflicted patient. The severity can range from a mild, localized disease to a generalized, devastating process. Although the clinical features are multiple and varied, treatment still remains a major challenge. 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.
Keywords: Blistering, epidermolysis bullosa, newborn, secondary bacterial infection, treatment

INTRODUCTION

Epidermolysis bullosa (EB) is a heterogeneous group of hereditary disorders characterized by extreme fragility of the skin and mucous membranes, which gives rise to the formation of blisters and ulcers following minor trauma [3]. As the areas of the body most often affected are sites subject to frequent pressure or friction, these conditions are also called mechanobullous disorders. 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.

These 3 subtypes are differentiated according to the level at which the tissue separates and the blisters form, that is, depending on whether this happens above, within, or below the epidermal basement membrane. We describe a male neonate with blistering of the skin during the immediate neonatal period.

CASE REPORT

A newborn male admitted to our hospital with blistering of the skin since birth. The term male baby was born to a 24 year old Primigravida mother with a regularly supervised and apparently uneventful antenatal period.

There was no history of similar disorder in the families of either of the non-consanguineously married parents. He was delivered by caesarean section at term, without any adverse perinatal events. His birth weight was 2850 grams. Baby had blistering of the skin involving both the lower limbs below knee joint including dorsum of foot and in involving the upper limb extending below the elbow joint, dorsum of both the hands (Fig. 1). Baby had blistering involving the lower lip also (Fig. 2). Minimal trauma elicited fresh blisters. Oral cavity, conjunctiva, cornea, nails, scalp and genitalia were normal. Systemic examination was normal. There was no family history of bullous skin lesions. A diagnosis was considered. Dermatologists confirmed the diagnosis and advised skin biopsy. Biopsy was not considered in view of financial constraints.

Fig. 1: Skin sloughing and blisters in a newborn

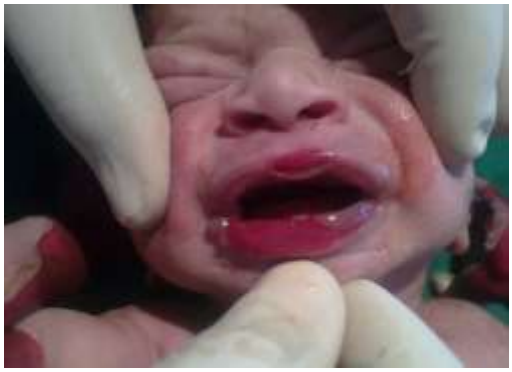

Fig. 2: Lesions on the lips

DISCUSSION

Epidermolysis bullosa (EB) comprises a group of genetically determined skin fragility disorders, which are characterized by blistering of the skin and mucosa, in response to little or no apparent trauma. These disorders represent heterogeneous phenotypes and are associated with a variable range of complications, from localized skin fragility to neonatal death [4]. This complex and heterogeneous group is classified on the basis of the mode of inheritance, clinical, laboratory and epidemiological studies into three major forms: EB simplex (EBS), junctional EB (JEB), and dystrophic EB (DEB).

EB simplex (EBS), is a non scarring, autosomal dominant disorder [1], although the mode of transmission is recessive in some subtypes [5]. With the help of immunohistochemistry, the major epidermolysis bullosa genes identified are those that encode keratins 5 and 14 in EBS, which makes up intermediate filaments of the basal keratinocytes [1, 6]. The intraepidermal bullae result from cytolysis of the basal cells.

In EBS-Koebner, blisters are usually present at birth or during the neonatal period. Sites of predilection are the hands, feet, elbows, knees, legs and scalp [1, 7]. 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 and do not result in skin atrophy [1, 7]. Secondary infection is the primary complication. The propensity to blister decreases with age, and the long term prognosis is good. Blisters should be drained by puncturing, but the blisters top should be left intact to protect the underlying skin [1].

Localized EBS of the hands and feet (Weber-Cockayne type) often presents when a child begins to walk; onset may be delayed, until puberty or early adulthood when heavy shoes are worn or feet are subjected to increased trauma. Bullae are restricted to the hands and feet [1, 5, 8]; rarely, they occur elsewhere such as dorsal aspects of the arms and the shins.

There is presently no definitive cure for EB and the objective of treatment is to alleviate symptoms and provide supportive measures. Therapy is therefore focused on the prevention of lesions and complications. Optimum management of this disease can only be achieved by a multidisciplinary team, which should include the following specialists: dermatologist, surgeon, nutritionist, dentist, physiotherapist, nurse, psychologist, pain specialist, and geneticist. The treatment plan must be individualized and optimal communication among team members is a vital factor in obtaining good results. Psychological support for parents and family members is vital. EB is not a contraindication for any vaccination [9]. A key to successful management is expert nursing care. Nursing the babies on thick foam pads protects them from undue trauma induced blistering. 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 [10]. The erosions should be cleaned with sterile normal saline and covered with non adherent dressings. Topical antibiotics are generally avoided because of the risk of emergence of antibiotic resistant bacteria. Oral and dental care should commence as soon as tooth eruption begins. Non-adhesive dressing pads or Vaseline impregnated gauze covered by soft, bulky dressings are ideal. The treatment plan must be individualized, and optimal communication among team members is a vital factor in obtaining good results. Psychological support for parents and family members is vital [9]. 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, 12].

Consent

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

ACKNOWLEDGEMENTS

I am grateful to the parents for giving permission to publish this image for academic purpose.

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