

CASE REPORT

EPIDERMOLYSIS BULLOSA IN THE NEWBORN

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We present the case of a new-born baby girl who was delivered at term to a young primigravida mother. The pregnancy was booked late and had no antenatal medical or surgical complications. The baby was born vaginally and had a normal initial examination. However after three hours of birth the baby started to develop skin blisters after minor trauma/friction. The baby was seen by a Dermatologist who diagnosed the condition to be Epidermolysis Bullosa. This hereditary condition is extremely rare, and even rarer is such an early presentation in a neonate. It is caused by mutations in the genes coding for the structural proteins of the skin and is characterized by development of vesicles and blisters. The mainstay of management is supportive care. The baby was discharged on request of the parents on second day of life without any complications.

Keywords: Neonate, epidermolysis bullosa, vesicle, blister, skin lesion

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INTRODUCTION

Epidermolysis bullosa (EB) is a heterogeneous group of inherited, mechanobullous disorders characterized by extreme fragility of the skin and mucous membranes which gives rise to the formation of blisters and ulcers following minor trauma.¹ The pathophysiology is mutations in various structural proteins in the skin.² Epidermolysis bullosa simplex (EBS), junctional epidermolysis bullosa and dystrophic epidermolysis bullosa are the three major types. EBS is the most common among them.³ 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 present a case of a female neonate with blistering of the skin during the immediate neonatal period.

CASE REPORT

A 2.7 kg term baby girl was born to 21 years old primigravida mother via SVD at Aga Khan Hospital for Women Garden. The mother was booked at 31 weeks of gestation, subsequently she had regular antenatal visits and an apparently uneventful antenatal period. There was no history of skin disorder in either the mother's or father's families. Baby was born with APGAR score of 8 at 1st minute and 9 at 5th minute.

Examination immediately after birth was unremarkable with no cutaneous manifestation. At 3 hours of life, the baby developed spontaneous peeling of skin with sub-conjunctival haemorrhage. Gradually there was formation of blisters which initially involved the neck, ear,

ankle and lips. The baby was hence shifted to the high dependency nursery. The examination in the nursery revealed an irritable crying baby with generalized blisters all over the body containing straw-coloured fluid, the blisters would rupture to expose intensely erythematous underlying skin. Minimal friction over the skin elicited fresh blisters. Small ulcer on the tongue was noted. Nails and hairs were normal. Systemic examination was unremarkable. A differential diagnosis of Epidermolysis bullosa, herpes simplex, bullous impetigo and staphylococcal scalded skin syndrome was made. Complete blood counts (CBC), Blood Culture were sent and a lumbar puncture was done; all of these tests were within normal ranges. Intravenous Cloxacillin was started. The baby was seen by a dermatologist who confirmed the diagnosis of Epidermolysis bullosa and advised nursing care, gentle handling, Nystatin oral drops, Fucidin cream, Cloxacillin, Vaseline and liquid paraffin as an emollient. As the baby had mouth ulcers breast milk was given by cup and spoon. The baby was discharged from the hospital on parental request on 2nd day of life.

DISCUSSION

The differential diagnosis for blisters in a neonate is extensive and includes common acquired aetiologies such as sucking blisters or other birth trauma-induced blisters, infection related blisters such as herpes simplex, bullous impetigo, staphylococcal scalded skin syndrome, neonatal candidiasis, neonatal varicella; maternal autoimmune bullous conditions such as bullous pemphigoid, pemphigoid gestations or pemphigus vulgaris; and genetic disorders including incontinentia pigmenti, ectodermal dysplasia, epidermolytic hyperkeratosis, pachyonychia

congenita, and epidermolysis bullosa (all subtypes).

Epidermolysis bullosa is a very rare hereditary skin condition characterized by the development of vesicles and bullae either spontaneously or as a result of minimal trauma. It is caused by mutations in various structural proteins in the skin², the overall incidence and prevalence of the disease within the United States is approximately 19 per one million live births⁴, however, we do not have any local data from Pakistan on the incidence or prevalence of EB. There are four major types of EB, depending on the location of the target proteins and level of the blisters: EB simplex (epidermolytic), junctional EB, dystrophic EB, and Kindler syndrome (mixed levels of blistering).

A new-born infant may present with localized or widespread blistering at birth and may be associated with a variable range of complications. These range from localized skin fragility to very extensive skin lesions leading to neonatal death. In families with EB or those at risk for having a child with EB, prenatal and preimplantation diagnosis is possible in order to appropriately guide the prospective parents. Prenatal diagnosis using DNA has 98% accuracy.⁵

Patients with EB require a multi-disciplinary team approach to management that includes a dermatologist, paediatrician, EB nurse specialist, nutrition specialist, pain management personnel, physical and occupational therapist, geneticist, psychologist, and dentist. There is presently no definitive cure for EB and the mainstay of management is supportive care. This supportive management includes: Prevention of skin trauma to avoid new blister formation by gentle handling of the infant, use of loose-fitting clothing, padding bony prominences, and avoiding adhesives or direct rubbing of the skin, infant should be maintained in cool, air-conditioned environments as overheating can increase skin fragility. New blisters should be drained with sterile large-bore needles at two sites in order to prevent extension. The blister roof should be left in place as it acts as a natural wound dressing. EB is not a contraindication for any vaccination.⁶

Meticulous wound care with appropriate wound dressings to ensure timely healing of wounds and prevention of infection. Topical antibiotic ointments and antimicrobial dressings should be reserved for those wounds that are colonized with bacteria and fail to heal, because of the risk of emergence of antibiotic resistant bacteria. Frank wound infection (increased erythema, swelling, purulence, odour and pain)

often requires systemic antibiotics. Maintenance of good nutrition to maximize growth and wound healing; for which specialized nipples and caloric supplementation of breast milk and/or formula may be required. Supplementary gastrostomy feeding is sometimes necessary to improve growth and nutrition in EB patients.⁸ Oral and dental care should commence as soon as tooth eruption begins. Surveillance for extra-cutaneous complications; and ongoing psychosocial support for both the child and the family to manage this devastating disease.

Research is underway for discovering curative treatment, including Gene therapy, protein replacement by transfection of the corrected protein into the patient's cultured keratinocytes, and bone marrow transplantation.⁹⁻¹¹



Figure-1 & 2: Typical lesion of EB revealing ruptured blisters with underlying intensely erythematous skin

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