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Management of Newborn with Epidermolysis Bullosa: Case Report

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Abstract

Epidermolysis Bullosa (EB) is a rare group of inherited disorders that manifests as blistering or erosion of the skin and, in some cases, the epithelial lining of other organs, in response to little or no apparent trauma. However, the term "epidermolysis" is not correct as epidermal disruption is not the primary change in two of the main categories of EB. Currently, there is no definitive and curative treatment for EB. Hence, the mainstay of treatment is symptomatic, supportive and preventive. The major challenge encountered in the care of a neonate with EB is optimum skin care and expert nursing care.

Keywords: Epidermolysis bullosa; Optimum skin care

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].

However, the term "epidermolysis" is not correct as epidermal disruption is not the primary change in two of the main categories of EB [2,3]. According to the recent data, the prevalence of EB has estimated to 8 to 10 million per live birth. The pathophysiology is mutations in various structural proteins in the skin [4]. Epidermolysis Bullosa Simplex (EBS), functional epidermolysis bullosa and dystrophic epidermolysis bullosa are the three major types. EBS is the most common among them [1]. 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 [5].

The disease is always painful, is often pervasive and debilitating. It affects 1 out of every 50,000 live births and those born with it are often called Butterfly Children because as the analogy goes their skin is as fragile as the wings of the butterfly [6]. There is no treatment or cure [6].

Case Presentation

A 2.1 kg, single term male, small for gestation age was born to third degree consanguinity to a second gravida mother. The mother was booked at 31 weeks of gestation; subsequently she had regular antenatal visits. There was history of previous sibling death at day 12 of life due to bullous lesions. The present baby did not require any resuscitation at birth and cried immediately after birth with Apgar score of 8 at 1st minute and 9 at 5th minute. Baby had bullous lesions at legs at birth. He was started on IV antibiotics (Inj Vancomycin) as blood culture grew Methicillin-Susceptible Staphylococcus Aureus (MSSA). Lumbar puncture was not done. Dermatologist opinioned that bullae could be Epidermolysis bullosa (Figure 1). In view of reappearing bullous lesions, baby was sent to higher center on day 9 of life for further management. The baby was referred and admitted in neonatal intensive care unit of our tertiary care facility. There was appearance of new lesions and healing of older one on friction sites. There was no history of poor feeding/recurrent vomiting/ decrease urine output/top milk feeding/umbilical discharge.

Course in NICU

Skin lesions

Epidermolysis Bullosa (Junctional type): In view of waxing and waning course of bullae, skin

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consultation was taken and same diagnosis of EB (Junctional type most likely severe form) was made. Genetic consultation was taken and sample for molecular diagnosis was sent. Parents were counseled regarding skin care, prognosis. Gradually, there was worsening of dermatological status in form of new appearance of bullae leaving denuded skin behind. There was bleeding from denuded skin area and oral mucosa on minimal handling.

Respiratory

On day 14 of life, baby had respiratory distress in the form of increased respiratory rate, new appearance of crepitation in bilateral lung field and frequent desaturation. So, the baby was intubated on day 17 and started on SIMV mode of ventilation.

Renal

On day 14 of life, baby had abdominal distension along with decreased urine output (0.5 ml/kr/hr) with increment in serum urea. NS bolus was given and dehydration correction @75 ml/kg was started. Fluid was continued @150 ml/kg/day. Baby also developed metabolic acidosis with pH of 6.9 and BE-18 for which sodium bicarbonate was given after discussing with Pediatric nephrology team. There was further worsening of renal status in the form of decreased urine output and deranged RFT. Peritoneal dialysis was started.

Hemodynamic

On day 18 of life, baby was having feeble pulse and increased tachycardia. So, NS bolus (10 ml/kg) was given and later dopamine and dobutamine was added. In view of poor pulses, adrenaline was added.

Feeding and nutrition

Baby had abdominal distension on day 14 of life, so baby was kept NPO. Earlier OG feeds and Katori spoon feed was given.

Lethargy/Late onset sepsis

In view of lethargy and abdominal distension, baby was evaluated for sepsis. CBC revealed leucopenia with thrombocytopenia. Inj Piperacillin-Tazobactam, Amikacin and Vancomycin were started in renal modified doses. Child was having persistent thrombocytopenia and required multiple platelets transfusion. Blood culture grew klebsiella (blood culture reported after the demise of baby).

Hyperkalemia

On Day 15, baby had high potassium, so potassium was omitted from IV fluid and NaHCO3 was given. Repeat value was 4.8 mEq/l.

Anemia with thrombocytopenia

Baby was transfused with PRBC and PRP as Hb was 7.7 gm % and Platelets were 13000/mm^3.

Terminal Events

Baby had refractory shock and had sudden cardiac arrest at 2 pm of day. CPR was started and child received 3 doses of adrenaline, but baby could not be revived and expired at 2:15 pm. Parents were counseled for autopsy but consent was not given.

Cause of Death

Immediate cause Septic shock.

Antecedent Cause

Sepsis with Acute renal failure with DIC with Epidermolysis bullosa.

Figure 1: Baby with Epidermolysis Bullosa (EB).

Discussion

Epidermolysis bullosa has no gender, geographical or racial preponderance Epidermolysis bullosa simplex is the most prevalent phenotype whereas autosomal recessive dystrophic and junctional EB are rare [7-10]. 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 [11].

Currently, there is no definitive and curative treatment for EB. Hence, the mainstay of treatment is symptomatic, supportive and preventive [12]. The 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. The major challenge encountered in the care of a neonate with EB is optimum skin care and expert nursing care, more so, with a preterm, low birth weight neonate who undergoes many invasive skin procedures as in the index case [13].

EB is not a contraindication for any vaccination [14]. A key to successful management is expert nursing care. 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 [15]. The erosions should be cleaned with sterile normal saline and covered with non-adherent dressings. 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. 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 [16].

Diagnostically, EB remains a challenge. The definitive diagnosis of inherited EB is made with Transmission Electron Microscopy (TEM), Immunofluorescence antigen mapping (IF), and EB related monoclonal antibody testing as well as mutational analysis. In order to make a correct diagnosis, it is important that the skin biopsy should be performed properly, as described by Intong and Murell [17]. In index case, skin biopsy was not done due to unwillingness of parents.

Conclusion

Epidermolysis bullosa, a genetically determined skin fragility disorder with variable degrees of extracutaneous involvement. The clinical spectrum ranges from localized skin disease to a life-

threatening and disabling disease with extensive extracutaneous involvement. All three major types of EB, namely EB simplex, Junctional EB, Dystrophic EB, can present with blistering and erosions at birth and cannot be distinguished clinically in the newborn period. The diagnosis of EB can be confirmed *via* a skin biopsy for immunofluorescence mapping. Although the clinical features are multiple and varied, treatment still remains a major challenge. There is still a long way to go, meticulous skin care and good nursing care, and gene therapy could possibly significantly alleviate the suffering of the neonates in the future.

Contribution

Dabas Heena has the prime responsibility of data acquisition and draft preparation and review of literature. She is the first author for the paper. Xavier Teenu and Varghese Priyanka did manuscript revision and editing. Teenu Xavier will act as guarantor for the paper.

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