Hereditary Epidermolysis Bullosa - A Case Report and Review of Literature

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INTRODUCTION

Epidermolysis bullosa (EB) is a heterogenous group of hereditary disorders characterized by recurrent blister formation as a result of extreme fragility of the skin and mucous membranes, following minor trauma [1]. As the areas of the body most often affected are sites subject to frequent pressure or friction, these conditions are also called as 'Mechanobullous disorders' [2]. Reported incidence varies from one geographical zone to another, affecting approximately 1 in 17000 live births with an estimated 500000 cases worldwide [3,4]. These disorders can be categorized under 3 major types [6]: 1). Epidermolysis Bullosa simplex (EBS), 2). Junctional Epidermolysis Bullosa (JEB), 3). Dystrophic Epidermolysis Bullosa (DEB). Kindler syndrome which includes poikiloderma and photosensitivity as well as early blistering, is also considered a separate form of EB, which differ phenotypically, genotypically and more importantly by the site of ultrastructural cleavage [1]. Diagnosis is by transmission electron microscopy (TEM), immunofluorescence mapping and often DNA analysis. Treatment is primarily directed at preventing skin trauma, with a combination of wound management, good nursing care, infection control, nutrition support with prevention of complications and surgical management if needed [8].

CASE REPORT

A 7 hours old newborn male baby presented with blistering over left arm and erosion of the skin over left eyelid, nape of the neck and right arm since an hour after birth. He was the first born to 3rd degree consanguinous parents...

ABSTRACT

Hereditary epidermolysis bullosa (EB) is a rare autosomal dominant disorder. It is characterised by blister formation following minor trauma. Here we report a case of 7 hours old newborn male baby presented with bullous lesions (fluid filled cavities or blisters, larger than 0.5 cm) over left arm and erosion of skin over left eyelid, nape of the neck and right arm since an hour after birth. After a thorough family history and complete physical examination, the clinical findings of this baby were consistent with epidermolysis bullosa simplex generalised formerly known as Koebner variant. Baby was admitted in NICU of Chalmeda Anand Rao Institute of Medical Sciences, Karimnagar and treated with strict aseptic measures, given intravenous fluids, antibiotics. Baby was discharged on 6th day of life, with proper counselling for management of blisters and prenatal diagnosis in future pregnancy.

Keywords: Blister, EBS, newborn.
Figure 1: Showing blister over left arm, eroded skin over right subcostal region and both right and left kness and ankles.

Figure 2: Showing oral mucosal lesions.

Figure 3: Fresh bullae appeared over pressure areas like trunk, left gluteal region, right and left elbows.

Figure 4: Fresh blister over the nose and right forearm.
without any family history of major medical or genetic disorders. On examination he was full term, weighted 2.5 kg, head circumference of 34 cm and length of 50 cm. He had blister over left arm which was light coloured and measured approximately 10mm size (Fig 1). After 10 hours of admission, he developed new blisters over lower half of right thigh and knee medially. He was given injection vitamin k, intravenous fluids and prevention of infection with Amoxicillin and Cloxacillin combination at appropriate dose given parenterally. Clinically he was stable with vitals- heart rate: 130/min; respiratory rate: 44/min ; cardiovascular system, respiratory system and neurological examination were normal. Spoon feeds was started. On day 2, former blisters healed without any scarring and fresh bullous lesions developed over pressure areas like trunk, left gluteal region and right and left elbows (Fig 3). On the day 6, oral mucosal lesions healed and accepted spoon feeds. Fresh blister developed over the nose and right forearm (Fig 4). Investigations revealed hemoglobin 15.6g/dl, total leucocyte count 13,500cumm, differential count Neutrophils 51%, Lymphocytes 40%, Eosinophils 4%, Monocytes 5%, Platlet count 1,63,000 cumm. Blood culture and sensitivity report was negative. Though diagnosis is by transmission electron microscopy ±transmission electron microscopy (TEM) is the gold standard for determining the level of blistering in EB subtypes and also ultrastructural entities. Skin biopsy is useful in reducing the bacterial load in the patient skin[5]. Soaking wounds this solution for 20 minutes before dressing is beneficial. A topical antibiotic can be applied and an non adherent dressing such as petroleum jelly guaze and covered with a dry guaze[7]. Common agents which cause secondary infection of blisters are staphylococcus aureus, streptococcus and pseudomonas aeruginosa[5]. A modified DAKIN’S solution (0.025% wt per volume sodium hypochlorite) can be useful in reducing the bacterial load in the patient skin[5]. Soaking wounds this solution for 20 minutes before dressing is beneficial. Unfortunately the child with EB faces a life time risk of infection (cutaneous or systemic). Exposure to heat should be avoided to control blister formation. Prenatal

Oral mucosal lesions were also observed(Fig 2), hence feeding was given through nasogastric tube. Normal saline rinses were used for gentle cleaning of the mucosal surfaces. On the day 3, previous lesions healed without scarring and fresh bullae appeared over pressure areas like trunk, left gluteal region and right and left elbows (Fig 3). On the day 6, oral mucosal lesions healed and accepted spoon feeds. Fresh blister developed over the nose and right forearm (Fig 4). Investigations revealed hemoglobin 15.6g/dl, total leucocyte count 13,500cumm, differential count Neutrophils 51%, Lymphocytes 40%, Eosinophils 4%, Monocytes 5%, Platlet count 1,63,000 cumm. Blood culture and sensitivity report was negative. Though diagnosis is by transmission electron microscopy ±transmission electron microscopy (TEM) is the gold standard for determining the level of blistering in EB subtypes and also ultrastructural entities. Skin biopsy is useful in reducing the bacterial load in the patient skin[5]. Soaking wounds this solution for 20 minutes before dressing is beneficial. A topical antibiotic can be applied and an non adherent dressing such as petroleum jelly guaze and covered with a dry guaze[7]. Common agents which cause secondary infection of blisters are staphylococcus aureus, streptococcus and pseudomonas aeruginosa[5]. A modified DAKIN’S solution (0.025% wt per volume sodium hypochlorite) can be useful in reducing the bacterial load in the patient skin[5]. Soaking wounds this solution for 20 minutes before dressing is beneficial. Unfortunately the child with EB faces a life time risk of infection (cutaneous or systemic). Exposure to heat should be avoided to control blister formation. Prenatal

**DISCUSSION**

Epidermolysis bullosa simplex (EBS) is a non scarring autosomal dominant disorder, characterized by intradermal blistering and is associated with keratin gene mutation k5 or k14, which makes up intermediate filaments of the basal keratinocytes[6]. The three most common EBS types are dominantly inherited and include generalized (koebner), localized (weber-cockayne) and herpetiformis (dowling-mera)[5]. Koebner variant shows onset of blisters at birth or latest during early infancy. Blisters are light in colour, vary in size and are formed in response to friction during labour and trauma and appear more frequently in warm conditions. They heal without scarring. In benign cases blisters affect hands and feet and unusually heal without scarring. It is possible to lead a normal life. In severe cases like our case, numerous blisters distributed all over the body, which can potentially lead to infection, dehydration and other problems which may be life threatening. Sites mostly involved are hands, feet, elbows, knees, legs and scalp[6]. Nails rarely become dystrophic. Oral mucosa sometimes show mild erosion, as seen in our case and dentition is normal. Bullae heal with minimal to no scar or milia formation. Palmar and plantar hyperkeratosis and erosions may present , which are not found in our case. Thickening of the soles is common but often doesnot present until later childhood[5]. Secondary infection is the primary complication. Propensity to blister decreases with age and longterm prognosis is good.

Diagnosis[8] is by thorough family history of blistering diseases or skin fragility and complete physical examination particularly skin, oral, genital and conjunctival mucous membranes. Skin biopsy is transmission electron microscopy (TEM) is the gold standard for determining the level of blistering in EB subtypes and also ultrastructural entities. Immunofluorescence antigen mapping (IAM) antibodies to a hemidesmosomal antigen and an antibody to a lamina densa protein, are antibodies mapped to determine the plane in which tissue separation occurs in blister formation. Differential diagnosis includes insect bites, friction blisters, thermal burns, pemphigus vulgaris, bullous pemphigoid and bullous lupus erythematosus. Treatment is mainly supportive. Blisters can be punctured at the base with a sterile needle and pressure applied with a guaze dressing to drain fluid and prevent extension of blistering[7]. Erosions and ulcers should be treated and dressed daily. A topical antibiotic can be applied and an non adherent dressing such as petroleum jelly guaze and covered with a dry guaze[7]. Common agents which cause secondary infection of blisters are staphylococcus aureus, streptococcus and pseudomonas aeruginosa[5]. A modified DAKIN’S solution (0.025% wt per volume sodium hypochlorite) can be useful in reducing the bacterial load in the patient skin[5]. Soaking wounds this solution for 20 minutes before dressing is beneficial. Unfortunately the child with EB faces a life time risk of infection (cutaneous or systemic). Exposure to heat should be avoided to control blister formation. Prenatal
diagnosis is by amniocentesis and chorionic villous sampling as early as 10 weeks of gestation, which is the advanced technique[7].

CONCLUSION

EB simplex, the first disorder shown to be due to mutations in keratin gene K 5 or K 14. Intensive counselling to parents regarding the wound management, prevention of sepsis and prenatal diagnosis may improve the prognosis. In future gene therapy may be useful for the treatment of Epidermolysis Bullosa.

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CONFLICT OF INTEREST

The authors declared no conflict of interest.

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REFERENCES


