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Epidermolysis Bullosa in Saudi Infant, Case Report

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Abstract **Case Report**

Epidermolysis bullosa are a clinically and genetically heterogeneous group of rare inherited disorders characterized by marked mechanical fragility of epithelial tissues with blistering, erosions, and nonhealing ulcers following minor trauma. We present 7 days old newborn with multiple large erythematous erosions with different sizes, mainly over abdomen and lower extremities.the patient was diagnosed as Epidermolysis bullosa

Keyword: Epidermolysis bullosa, blister, Kindler syndrome.

INTRODUCTION

Epidermolysis bullosa (EB) encompasses a clinically and genetically heterogeneous group of rare inherited disorders characterized by marked mechanical fragility of epithelial tissues with blistering, erosions, and nonhealing ulcers following minor trauma [1].

It is caused by mutations that affect skin structural proteins. So far, mutations in 20 genes have been described as being associated with more than 30 clinical EB subtypes [1, 2] that differ in severity and prognosis, clinical and histologic features, and inheritance patterns, but are all characterized by induction of blisters by trauma and exacerbation by warm weather[3].

CASE REPORT

A 7 days old girl infant, full-term baby, Product of Caesarian section with Low Birth Weight (1.6 kg), is born with multiple large erythematous erosions and blisters on the nose, abdomen and lower extremity.

In the Birth History she was 7 Days old, Girl, full Term product of Caesarian section due to pervious two Caesarian sections with breach presentation. Birth weight: 1.6 kg. Intrauterine growth restriction with APGAR Score 6 and 9 in 1 min and 5 min. Needed only for routine care.

Regarding maternal History, Baby born to 27 years old mother, Gravida 4 Para 2+1, and previous two Caesarian sections. Not Booked pregnancy, and has history of prelabor rupture of membranes 7 hours prior to the delivery .No history of Medical history of significant illness. No history of Diabetes mellitus or Hypertension. No history of Chorioamnionitis. No history of Fever, vaginal yeast or gential herpes. No history of Hospital Admission. No history of Medication Intake. No history of Polyhydramnios.

No family history of similar condition. Parents with first degree of consanguinity. No family history of chronic disease. They live in Riyadh with average socioeconomic status.

On Examination: Baby was active, pink, and stable, in good condition, not jaundiced or cyanosed; no dysmorphic feature had few attacks of irritability and crying.

Vital signs are: Temperature: 36.8 Heart Rate: 123 beat per minute Blood Pressure: 46/27 Respiratory Rate: 52 breath per minute Oxygen Saturation: 99% on Room air. Growth Parameter: Weight: 1.6 Kilogram (below 3rd percentile) Height: 40 centimeter (below 3rd percentile. Head Circumference: 31 centimeter (below 3rd percentile).

Head, Ear, Eye, Nose and Throat are: Normal. Cardiovascular system: Normal, Audible First and second heart sounds + no murmur. Capillary Refill Time: 2 seconds. Respiratory system: Clear, Good air entry, no added sound .Abdomen: Soft and lax, no organomegally. Central Nervous System: Active, Anterior Fontanel flat and open, normal primitive reflexes. Musculoskeletal: moving limb. Lymph nodes: No Lymphadenopathy. Genitalia: Normal. Skin:

Multiple large erythematous erosions with different sizes, mainly over abdomen and lower extremities, lesions show peripheral scaling with areas of skin peeling suggestive of Epidermolysis bullosa (Figure 1 and 2).

Management: Kept No per Os, Umbilical vein catheter Inserted, Paracetamole given, Intra Venous Fluid, Cloxacillin, Gentamycin, Vaseline and Fucidin Ointment Applied, Vital sign Monitoring and Dermatologist consultation.

Lab Results: Cell Blood Count: White Blood Cell = 8 Red Blood Cell = 4.2 Hemoglobin = 17.6 Mean Corpuscular Volume = 137.4 Mean Corpuscular hemoglobin = 43 Mean corpuscular hemoglobin concentration = 31 Platelets = 151 NEUT% = 27 LYMP% = 53 NEUT = 2.17 LYMPH = 4.27 Kidney Function Test: Glucose = 3.8 mmole Blood Urea Nitrogen = 6 mmole Creatinine = 59.7 umole Uric Acid = 109 umole Sodium = 136 mmole Potassium = 4.3 mmole Chloride = 105 mmole Calcium = 2.57 mmole Liver Function test: Aspartate Transaminase = 59.7 U/L Alanine Transaminase = 33.7 U/L Alkaline Phosphatase = 94.6 U/L Total Bilirubin = 196 umole Albumin = 20 Protein = 38.6 g/dL GGT=78.

Management: In the second day of life: baby developed mild Respiratory Distress and Hypotension. So baby was started on nasal blender Oxygen, normal saline bolus given, Inotropes, Analgesics and blood re-cultured**.** Antibiotics were change to vancomycin and meropenem**.** Baby was started on Total prenatal nutrition with daily dressing of lesion.

Microbiology Results: Axilla swab culture: No growth, Groin swab culture: No growth, Blood culture: No growth, ABO blood grouping: O+, Direct Comb Test: Negative, RH: Positive. Chest X Ray: Normal (fiure2).

Fig-1: Multiple large erythematous erosions with different sizes, mainly over abdomen and lower extremities, lesions show peripheral scaling with areas of skin peeling upon admission suggestive of Epidermolysis bullosa

Fig-1b: After few days of admission healing of most of skin lesions

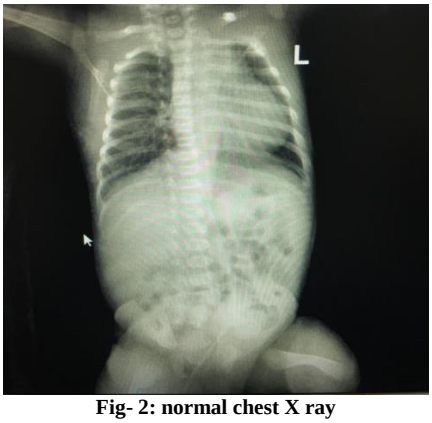

Fig- 2: normal chest X ray

DISCUSSION

Four major types of EB are recognized, based on the ultrastructural split level and blister formation within the epidermal basal membrane zone: (i) epidermolysis bullosa simplex with intraepidermal skin separation[4], (ii) junctional epidermolysis bullosa with

skin separation in lamina lucida, (iii) dystrophic epidermolysis bullosa with sublamina densa separation[4], and (iv) Kindler syndrome, which includes poikiloderma and photosensitivity as well as easy blistering and has various split levels, is considered a separate form of EB[3,4]. The overall incidence and prevalence of inherited EB are 19[5, 6] and 8.2[6] per 1 million live births and per 1 million population, respectively. Similar rates have been reported elsewhere. Onset of epidermolysis bullosa is at birth or shortly after [4]. The clinical presentation of inherited EB varies according to the subtype of the disease [5, 7]. In addition to the blistering and erosions secondary to the mechanical fragility of the skin, inherited EB may lead to the formation of *millium*, nail dystrophy or anonychia, hypo- and hyperpigmentation, and trophic scarring. Common complications include super infection[8], sepsis, dehydration[5], renal failure, upper airway occlusion, failure to thrive, esophageal scarring[4], gastrointestinal obstruction[5], pseudosyndactyly [2, 8], joint contractures, malignant transformation[5] (squamous cell carcinoma[4]) and death.

Diagnosis is clinically based and is confirmed by skin biopsy using immunofluorescence and electron microscopy, and by genetic mutation DNA analysis [4].

The management of patients with EB is largely supportive and requires a multidisciplinary approach [2]. It is based on preventive measures to avoid the development of new lesions, as well as prevention and treatment of complications, both cutaneous and extracutaneous. Implementation of advanced wound care [2] and enhancement of wound healing, control of infection, pain management [7] nutritional support, life style, activity, physiotherapy [4], as well as social support and genetic counselling [4]. The prognosis varies according to type [5], ranging from a normal life span with only minor skin affection, to death in the neonatal period. Dominantly inherited epidermolysis bullosa simplex, dominant dystrophic epidermolysis bullosa, and milder forms of junctional epidermolysis bullosa may not affect a patient's life expectancy adversely [4]. The prognosis for most patients with EB simplex (EBS) is excellent, except the severe generalized form which poses an early infant mortality risk. Patients with the generalized severe form of junctional epidermolysis bullosa have the highest risk during infancy [4], with approximatly 45% dying by age of one year [1].

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