

### **Epidermolysis Bullosa- A Rare Case**

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### **Abstract**

Epidermolysis Bullosa (EB) is a group of rare disorders, mainly inherited, characterized by blistering and erosions of the skin and mucous membranes resulting from slight mechanical trauma. The inherited disorders occur due to defects in the genes encoding various proteins like collagen and keratin, which are essential for mediating adherence of the epidermis to the underlying dermis.<sup>[1],[2],[3]</sup>In view of the rarity of this disorder, we report 1 case of EB that presented to the department of dermatology of Tata Main hospital.

**Keywords-** Epidermolysis, Bullosa, Dystrophic, Junctional

### **Introduction**

Epidermolysis bullosa (EB) is a rare genetic disorder characterized by abnormal fragility of skin and mucosal surface. The separation of skin layers occurs after application of friction or shearing forces and results in intradermal fluid accumulation and bullae formation.<sup>[4]</sup> This may lead to scarring and debilitating, life-threatening medical conditions. 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). Incidence of Epidermolysis Bullosa is 19.60 per 1 million live births. Here, we report an uncommon case who had classical clinical features of Epidermolysis Bullosa.

### **Case Report**

A newborn male baby was delivered vaginally in Labour room of Tata Main Hospital Jamshedpur State - Jharkhand with fragile thickened skin that had tendency to blister early present over hand and foot. The lesions were initially vesicobullous which later turned into erosions over the same sites.

On Local cutaneous examination , blisters were also present over mouth, throat and scalp leading to scarring, hair loss and milia formation. Two natal mobile teeth were present which were extracted by dental surgeon under topical anesthesia. Nails were thick and horny.



Figure 1: Bullae leading to erosions.



Figure 2: Haemorrhagic erosions



Figure 3: Erosions and skin exfoliating.



Figure 4: Prominent erosions and skin exfoliation leading to scarring.

### Systemic Examination

General Condition-sick , Capillary Refill Time less than 3 sec, Heart Rate- 156/min, Respiratory Rate-48/min, Spo2- 96% on Room Atmosphere, Extremity: warm, pulses well felt, Chest: Bilateral air entry present, Central Vascular System- S1S2 normal, No murmur appreciated, Per Abdomen- soft, Central Nervous System- AF at level, tone/cry/Activity good, Suckling present, pupils- bilaterally reacting to light.

### Lab findings

Histopathologic examination was done which came to be Epidermolysis Bullosa.

Direct Immunoflorescence was also done. Both the reports are attached.

## Histopathology Report

### SKIN PATHOLOGY REPORT

#### Clinical Features :

Term (37 weeks ) born of non consanguineous marriage, with normal new born transition. Baby had developed skin peeling since birth over perioral, periumbilical region, both legs, hands, left side of neck and back. Dystrophic nails and necrotic changes in b/l great toe. Prenatal molar teeth+

#### Clinical Diagnosis :

Epidermolysis Bullosa- Dystrophic Type

#### Gross Pathology :

Skin biopsy from left thigh

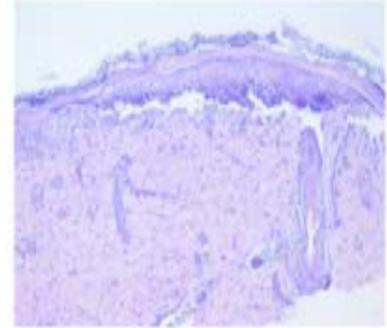
#### Microscopic :

Basket weave stratum corneum overlies thick confluent layer of parakeratosis. There is large suprabasal-subepidermal blister. Epidermal roof is completely separated along the subepidermal portion of the blister which contains detached epithelial fragments mostly of acrotrichia and acrosyringia. No significant inflammation is present. There appears to be some keratinocyte dyscohesion along the basal layer forming the floor of the suprabasal portion and along the lower edge of the epidermis forming the roof along both suprabasal and subepidermal portions.

#### Impression :

Histological features in conjunction with Antigen mapping result (path no. 2020-161) rule out dystrophic epidermolysis bullosa, Herlitz type junctional epidermolysis bullosa and severe recessive epidermolysis bullosa simplex involving Cytokeratin14. However, suprabasal clefting with some dyscohesion of keratinocytes raises a possibility of other severe forms of epidermolysis bullosa simplex (EBS) involving Cytokeratin 5, and EB-simplex like disorders (earlier classified as EBS) such as lethal acantholytic EB involving Desmoplakin, severe generalized blistering involving Plakoglobin and skin fragility syndrome involving Plakophilin (see reference). The case requires clinical correlation and genetic analysis for specific diagnosis.

Reference: Bruckner-Tuderman L, McGrath JA, Robinson EC, Uitto J. Progress in Epidermolysis bullosa research: summary of DEBRA International Research Conference 2012. J Invest Dermatol. 2013;133(9):2121–2126.



## Direct Immunofluorescence Report

### ANTIGEN MAPPING TEST

#### Clinical Features :

Term (37 weeks ) born of non consanguineous marriage, with normal new born transition. Baby had developed skin peeling since birth over perioral, periumbilical region, both legs, hands, left side of neck and back. Dystrophic nails and necrotic changes in b/l great toe. Prenatal molar teeth+

#### Clinical Diagnosis :

Epidermolysis Bullosa- Dystrophic Type

#### Gross Pathology :

Skin biopsy from left thigh

#### Microscopic :

Immunofluorescence based antigen mapping performed using normal human skin as positive control. Methylene blue stained frozen sections of lesional skin show a suprabasal blister with disrupted patchy basal layer along the floor.

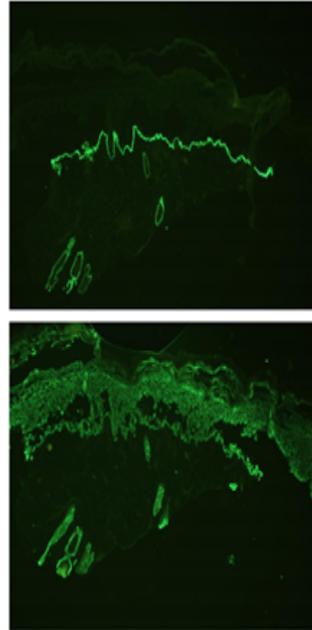
Collagen IV band is mapped to floor of the small blister indicating the level of cleavage to be above lamina densa.

Collagen VII and Laminin 332 bands of intensity comparable to control skin map to the floor of the blister in lesional skin.

Cytokeratin 14 shows a normal pattern of keratinocyte staining in epidermis of lesional skin comparable to control skin. It also highlights the basal layer forming the floor in lesional skin.

#### Impression :

Antigen mapping result in conjunction with histological features (path no. 2020-160) rule out dystrophic epidermolysis bullosa, Herlitz type junctional epidermolysis bullosa and severe recessive epidermolysis bullosa simplex involving Cytokeratin14. However, suprabasal clefting with some dyscohesion of keratinocytes raises a possibility of other severe forms of epidermolysis bullosa simplex (EBS) involving Cytokeratin 5, and EB-simplex like disorders (earlier classified as EBS) such as lethal acantholytic EB involving Desmoplakin, severe generalized blistering involving Plakoglobin and skin fragility syndrome involving Plakophilin (see reference). The case requires clinical correlation and genetic analysis for specific diagnosis.



Reference: Reference: Bruckner-Tuderman L, McGrath JA, Robinson EC, Uitto J. Progress in Epidermolysis bullosa research: summary of DEBRA International Research Conference 2012. J Invest Dermatol. 2013;133(9):2121–2126.

Based on the above clinical, histopathological and DIF findings, diagnosis of Epidermolysis Bullosa was made.

During stay in hospital, iv antibiotics were started, full asepsis maintained and advised wound care with sterile and moist vaselline dressing. Dental referral was done for presence of neonatal grade III molar teeth, extraction under local anaesthesia was done. Plastic surgery consultation was sought who advised conservative management (non operative condition). IV antibiotics were upgraded to Meropenem in view of raised C

Reactive Protein. 2D Echo was done to rule out any congenital heart disease- Scimitar syndrome, Levocardia, Intact Atrial System/Intact Ventricular System, Mild Tricuspid Valve Regurgitation, Trivial Mitral Regurgitation, Ejection Fraction-77% . Baby had 1 episode of desaturation and bradycardia, was kept Nil Per Os, given Normal Saline bolus followed by IV fluids, antibiotics were upgraded to Piptaz, Oxygen by nasal prongs was administered.

Baby was referred to higher centre for further

### Discussion

Epidermolysis Bullosa encompasses an array of autosomal dominant and recessive conditions that may have either localized or generalized dermatological manifestations. The loss or absence of normal intracellular bridges is due to a collagen abnormality, which makes patient susceptible for blister formation by friction/shearing forces and subsequent scarring<sup>[5]</sup>. As there is no specific therapy at present for inherited EB, management mainly revolves around protection, avoidance of provoking factors, prevention and treatment of complications.<sup>[6]</sup> In general, management of patients with EB is one of a "no touch" principle<sup>[7],[8]</sup>. All persons involved in handling these children must be aware of the extreme vulnerability of skin. During transport or mobilization of the patient, the most important task is to maintain the integrity of the skin and avoid friction and trauma. Trolleys and operating tables should be well padded. Patients should be allowed to move themselves on the operating table. Excellent analgesia is important to prevent excessive movements and new skin trauma. A multimodal approach using nonsteroidal analgesics and opioids is the most convenient method.<sup>[9]</sup> In future, gene therapy may become possible for at least some types or subtypes of EB.<sup>[10]</sup>

### Conclusion-

Epidermolysis bullosa, a genetically determined skin fragility disorder can severely incapacitate the life of the afflicted patient. Although the clinical features are multiple and varied, treatment still remains a major challenge. Although there is still a long way to go, good nursing care, and gene therapy could possibly significantly alleviate the suffering of the patients in the future. A high degree of clinical suspicion and awareness will not only shorten the diagnostic delay but also enhance early detection and treatment of these cases.

management.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent form. In the form, the patient's parents have given their consent for the images and other clinical information to be reported in the journal. They understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

### References

1. Fine JD, McGrath J, Eady RA. Inherited epidermolysis bullosa comes into the new millennium: a revised classification system based on current knowledge of pathogenetic mechanisms and the clinical, laboratory, and epidemiologic findings of large, well-defined patient cohorts. *J Am Acad Dermatol* 2000;43:135-7.
2. Fine JD, Eady RA, Bauer EA, Briggaman RA, Bruckner-Tuderman L, Christiano A, *et al.* Revised classification system for inherited epidermolysis bullosa: report of the Second International Consensus Meeting on Diagnosis and Classification of Epidermolysis Bullosa. *J Am Acad Dermatol* 2000;42:1051-66.
3. Mitsuhashi Y, Hashimoto I. Genetic abnormalities and clinical classification of epidermolysis bullosa. *Arch Dermatol Res* 2003;295:S29-33.
4. Thomas J. Epidermolysis bullosa in children: Pathophysiology, anaesthesia and analgesia. *S Afr J Anaesthesiol Analg* 2010;16:13-5.
5. Bruckner AL, Murrell DF. Diagnosis of epidermolysis bullosa. [updated 2012 Mar 6:].
6. Sawamura D, McMillan JR, Akiyama M, Shimizu H. Epidermolysis bullosa: directions for future research and new challenges for treatment. *Arch Dermatol Res* 2003;295:S34-42.

7. 9.Iohom G, Lyons B. Anaesthesia for children with epidermolysis bullosa: A review of 20 years' experience. *Eur J Anaesthesiol* 2001;18:745-54.
8. 10. Thomas J. Epidermolysis bullosa in children: Pathophysiology, anaesthesia and analgesia. *S Afr J Anaesthesiol Analg* 2010;16:13-5.
9. Siddiqui KM, Khan S. Anaesthetic management of an infant with epidermolysis bullosa undergoing inguinal hernia repair. *J Pak Med Assoc* 2010;60:497-8.
10. Fenjves ES. Gene therapy: principles and potential application in inherited epidermolysis bullosa. In: Fine JD, Bauer EA, McGuire J, Moshell A (eds). *Epidermolysis bullosa: Clinical, epidemiologic, and laboratory advances, and the findings of the national Epidermolysis Bullosa - A Series of 12 Patients in Kashmir Valey: Johns Hopkins university press; 1999. p. 407-19.*