

## Recessive Dystrophic Epidermolysis Bullosa-Hallopeau-Siemens Subtype

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### Abstract

*Epidermolysis bullosa dystrophica is a mechanobullous disorder characterized by blister formation in response to minimal trauma. Dystrophic mechanism mainly affects the dermis below the lamina densa at the level of the anchoring fibrils. It is characterized by skin blisters, contractures of digital web spaces, microstomia, and enamel hypoplasia and cementum disorders. The onset is in early childhood and heals with atrophic scars. This case features a 14 year old patient diagnosed with EBD with typical clinical features and histopathological findings.*

**Keywords:** Epidermolysis bullosa, Recessive, Dominant.

### Introduction

Epidermolysis bullosa dystrophica is characterized by blistering and scarring of both skin and mucous membrane in response to mechanical force. The mutation in COL7A1 gene is the main pathogenic mechanism behind the clinical features. Usually patients manifested blistering at birth or shortly thereafter. The other manifestations are blisters and erosions healed with extensive scarring and nail dystrophy. It is a rare skin disorder which comprises of three forms: two autosomal – dominant variants, the cockayne-Touraine and pasini forms and an autosomal recessive form based on the mode of transmission [1]. This paper reports a case of EBD in a 14 year old male with typical clinical features and histopathological findings.

### Case Report

A 14 year old male patient, born of consanguineous marriage reported to our outpatient clinic for routine dental check-up. Patient had a history of spontaneous blister formation when he fell in the school during playing session. Mental and motor milestones of development were normal. There was no history of photosensitivity and family history was noncontributory. Dermatological examination revealed ruptured blister on acral parts (Figure 1), hypohidrosis, hypopigmented areas on the face (Figure 2) and extremities, alopecia (Figures 3a,3b), generalized scarring (Figures 4a,4b) and nail dystrophy (Figure 5). Intraoral findings revealed erythematous gingiva (Figure 6), reduced opening of mouth and extension of tongue. General examination revealed short stature and severe anemia. Routine blood investigations revealed reduced hemoglobin, MCV, MCH and MCHC parameters. The biopsy of leg blisters were carried out. Histopathologic examinations revealed formation of subepidermal blister along with slight perivascular infiltration of lymphocytes

and mononuclear cells in the dermis (Figure 7). In our case, patient was managed by oral prophylaxis, iron supplements and other supportive measures. Apart from this, we advised periodic follow up. For acral lesion, patient was put on systemic antibiotic with neomycin cream.



Figure 1: Ruptured blister on acral parts.



Figure 2: Hypopigmented areas on the face.



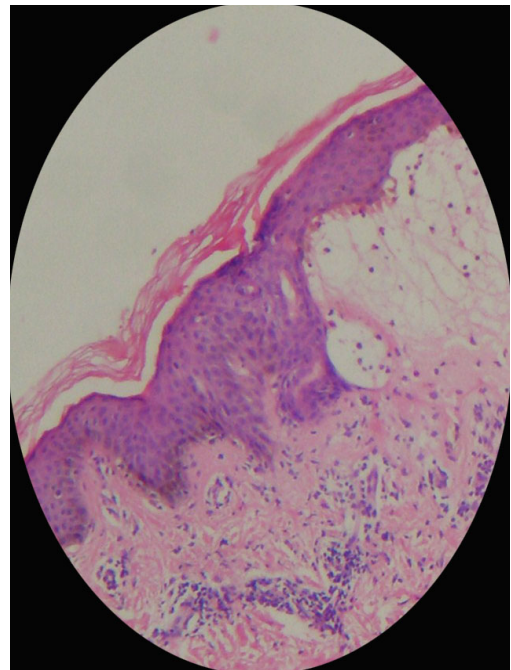
**Figure 3a,3b:** Generalized alopecia.



**Figure 6:** Erythematous gingiva.



**Figure 4a,4b:** Generalized scarring.



**Figure 7:** Intra dermal blisters [10X].



**Figure 5:** Dystrophic nails.

### Discussion

More than 20 different subtypes of epidermolysis bullosa have been identified. They are broadly grouped into 3 main categories: epidermolysis bullosa simplex, dystrophic epidermolysis bullosa and functional epidermolysis bullosa. Among these, 46.5% are of the dystrophic type. The gene responsible for dystrophic EB is known as COL7A1 which codes for VII collagen, a protein that anchors the densa within the superficial dermis [2]. Recessive dystrophic EB fibroblasts synthesize very low amounts of type VII collagen. The patients with dominant DEB have mild symptoms, but with recessive DEB have severe debilitating conditions.

### Dominant Dystrophic Epidermolysis Bullosa (DDEB)

The clinical spectrum ranges from localized blistering at sites of maximal trauma to generalized blisters that subsequently heal with scarring. The age of onset is usually from early childhood. Other manifestations are atrophic scarring and abnormal nails of both upper and lower extremities, but no extra cutaneous involvement

in this variety. Pasini and cockayne-touraine are the two variants of DDEB. Among these, pasini DDEB is characterized by albopapuloid lesions, which is a signature feature characterized by small hypo pigmented papules.

### Recessive Dystrophic Epidermolysis Bullosa of Hallopeau-Siemens Subtype

Patients with most severe form of this RDEB have fragile skin which tends to form blisters at birth. Added to this, blisters occur in the mucosa regularly. Microstomia, obliteration of oral vestibule, ankyloglossia, enamel hypoplasia and cementum disorders were seen. The occurrence of squamous cell carcinoma of the skin is the unique complication seen in these patients [3,4].

### Recessive Dystrophic Epidermolysis Bullosa of Non Hallopeau-Siemens Subtype

In this type of recessive form, patient usually lacks the cutaneous and extra cutaneous features. Blisters are present at birth. Also milieu, atrophic scarring, alopecia, nail dystrophy, severe microsomal, ankyloglossia, diffuse scar formation in the esophagus are also present [5].

### Extra cutaneous involvement

The systems such as gastrointestinal tract, upper respiratory tract, genitourinary tract, teeth, eyes and cardiovascular system are the sites most commonly involved. The main manifestations in gastrointestinal tract include dysphagia and chronic constipation [6]. Dental findings are gingival blisters and premature loss of teeth. When it affects the respiratory system, hoarseness and laryngeal stenosis occur. The genitourinary symptoms are urinary retention, glomerulonephritis and nephrotic syndrome. Anemia and growth retardation due to iron deficiency and chronic disease are also manifested [7].

### Management

The approach is multifactorial such as prevention of skin trauma and secondary bacterial infection, maintenance of good nutrition, early dental interventions to avoid premature loss of teeth and topical analgesics [8]. Early dental intervention is required to salvage teeth and gum. As part of oral hygiene, tooth brush with small head, soft bristle is preferred [9]. In the patients with severe microstomia, soft bristles are more desirable. Always a manual tooth brush may be desirable to an electric brush if the mouth is very sore [10]. Cotton buds or clean gauze can be used. Rinsing with water after meals helps in removing food debris or sugar deposits [11]. Adjuvant therapies like the usage of chlorhexidine 0.12%, topical application of fluoride every 3 months and mouth wash with alcohol free formulations are advised [12]. In case of severe generalized RDEB; microstomia is the most common complication. Daily oral exercises can improve mouth opening that in turn favors phonation and swallowing. Liquid-based formulation of drugs is usually advisable to prevent discomfort to the patient, as they are suffering from esophageal stenosis. Every 3-6 months, review appointments are necessary to give dietary advice, topical fluoride application, caries prevention and plaque removal.

### Conclusion

Epidermolysis bullosa has got several complications such as risk of getting squamous cell carcinoma, chronic renal failure, cardiomyopathy and severe anemia. In order to avoid mortality rate, yearly diagnosis and intervention is important.

### Acknowledgement

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