

# A Case of Epidermolysis Bullosa in Bhuth: A Case Report

Gargadi SI<sup>1\*</sup>, Mava Y<sup>2</sup>, Davou K<sup>3</sup>

<sup>1</sup>Plastic Surgeon BHUTH

<sup>2</sup>Pediatrician BHUTH

<sup>3</sup>Nurse in Pediatrics Unit BHUTH

\*Corresponding author: Gargadi SI, 1Plastic Surgeon BHUTH

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## Abstract

Epidermolysis Bullosa (EB) is a group of inherited Bullous disorders characterized by formation of blistering following minor trauma (Nikolsky Sign), healing with scarring. It is caused by one or more mutations in at least one of 20 different genes that synthesize structural proteins that are involve in adherence of epidermal to dermis.

We observed and cared for a 2 day old female neonate with low birth weight, normal antenatal history who presented at birth with spontaneous bullous skin eruptions to parts of skin of the upper and lower limbs which ruptured with minor mechanical trauma. She had conservative care and even though there was lack of finances for genetic test to pinpoint the mutated gene the wounds healed with acceptable scar.

We are glad to share this rare condition with the scientific world.

**Keywords:** Epidermolysis Bullosa (EB) in BHUTH.

## Introduction

The skin lines the outermost part of the body and forms 16% of the body weight with total surface area of 1.8m<sup>2</sup>. It is ectodermal in origin and has three layers: epidermis dermis and subcutis [1]. The dermis has 100 – 200 million papillae in adult skin which is fitted to the papillae of pockets of the epidermis [2]. A multiprotein complexes (hemidesmosomes) facilitates the stable adhesion of epithelial cells to the underlying basement membrane [3].

Epidermolysis Bullosa (EB) is a group of inherited diseases that is characterized by formation of blistering of the skin following minor trauma (Nikolsky sign) and healing with scarring. The incidence in the U.S is 1 in a million [3, 4]. EB is divided into four types: 1. Epidermolysis

Bullosa simple x (weber – cockayne type of EB) – A localized type. It is a genetic disorder that is caused by a dominant – negative mutation in either keratin 5 (KRT5) or the keratin 14 (KRT14) gene. 2. Junctional EB associated with Pyloric atresia – A major type of EB. Moderate to severe, occurs at part. 3. Dystrophic epidermolysis Bullosa is the severest associated with fibrosis and syndactyly. 4. Kindler syndrome: A rare genetic disease affects hands and feet.

EB is caused by one or more mutations in at least one of 20 different genes that synthesize structural proteins that are involve in adherence of epidermal to dermis namely Keratin, Laminin, type VII collagen, integrins, lectin, desmoplakin, plakophilin and plakoglobin.

Generally patients present at birth to infancy and do well with conservative care and grow to adulthood [5].

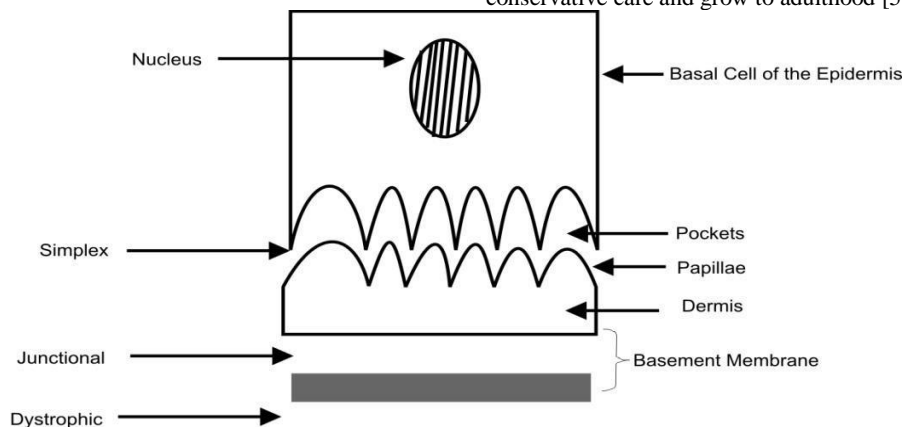


Figure 1. Classification of EB based on the site of the Blister on Electron Microscopy

## Case Report

We present a 2 days old female neonate with low birth weight (2.1kg) born to a 23 years old primiparous Lady following supervised term gestation and spontaneous vertex delivery who was noticed at birth to have localized bullous eruptions affecting parts of the skin of the upper and lower limbs which ruptured with minor mechanical trauma, resulting



**Fig 2: 2 days Female Neonate with EB**

## Discussion

The occurrence of EB in our health facility constitute a challenge since we are dealing with patients with limited funds to do genetic test, however we arrived at a favourable outcome and hope to follow-up our patient to adulthood.

## Conclusion

E.B is a relatively rare skin fragility condition and simple measure of conservative care can give a satisfactory result hence we wish to share with you.

in ulceration. There is no associated family history among first and second degree relations and no history of burns. We could not carry out genetic test due to financial constrain, but clinically we believe is one of the EB type's most likely EB simplex.

Patient was managed conservatively with satisfactory outcome.



**Fig 3: At 9 days old**

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