CASE REPORT

RESPONSE TO PHENYTOIN THERAPY IN EPIDERMOLYSIS BULLOSA SIMPLEX

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ABSTRACT: Report of a 3 year old male child with Epidermolysis Bullosa simplex –Dowling Meara, treated with phenytoin sodium 5mg/kg per day. Patient had multiple fluid filled lesions, erosions, hypopigmented lesions and mild scarring on face, trunk, upper and lower limbs. Dowling-Meara type of EBS is fatal so early diagnosis and prompt treatment is needed.

KEYWORDS: Epidermolysis bullosa disorder, Dowling-Meara syndrome, Phenytoin therapy.

INTRODUCTION: Epidermolysis bullosa simplex (EBS) is a group of hereditary bullous diseases characterized by intraepidermal blistering due to mild mechanical traumas.⁽¹⁾ Patients present with widely varying severity and are classified in three main subtypes: EBS Weber-Cockayne, EBS Kobner and EBS Dowling-Meara, on the basis of the severity and distribution of the blisters, the age of onset, and the ultrastructural investigation of the epidermis and basement membrane zone.

The three major clinical subtypes are all caused by mutations in either the keratin 5 (KRT5) or keratin 14(KRT14) gene,^(2,3)

However various drugs like corticosteroids,^[4] phenytoin,^[4] vit E,^[5] have been used with variable results.^[6] Prevalence of EBS has been calculated as 28.6 per million, with the Dowling-Meara subtype accounting for 0.6 per million.⁽⁷⁾

CASE REPORT: A 3 year old male child presented with multiple fluid filled lesions appearing spontaneously and at sites of trauma from birth. Noticed bulla on 2nd day of life on finger, 10 days later he had generalised blistering after bath.

Since then flaccid blisters of size 0.5cmx0.5cm to 3x4cm continued to appear at sites of friction or trauma healing with hypopigmentation and mild scarring. Marked dystrophy of nails present (Figures 1-4).

Fig. 1: Flaccid Bulla over Left Inguinal Area

Fig. 2: Crusting Over Upper Lip

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Fig. 3: Blisters over Dorsa of feet

Fig. 4: Dystrophic Nails

Hair and teeth are normal. No history of similar illness in family. No history of consanguinity. Other siblings are normal.

INVESTIGATIONS: Hb-10gm%. Total and differential count, Liver function tests, Renal function tests and Complete urine examination were within normal limits.

Histopathology showed intra epidermal bulla. Dermal edema present with perivascular chronic inflammatory cells. Direct Immunofluorescence was not done as patient could not afford it. Based on the clinical and histopathological feature a diagnosis of Dowling –Meara was made.

The patient was started on Syrup phenytoin sodium 5mg/kg per day, divided in 2 doses for 2 months along with supportive treatment. After which patient had healing of old lesions and occurrence of new lesions decreased. Patient is on follow up.

DISCUSSION: Dowling–Meara variant of EBS is characterised clinically by exceptionally severe blistering with an onset in early infancy after minor mechanical trauma. In most severe cases blistering may appear spontaneously particularly in hot environment.

Blisters frequently occur in areas of face, trunk & extremities and are disposed in herpetiform groups with annular or arcuate erythematous borders.^[8,9] Milia and minor scarring may be a transient feature after the blisters have healed.^[1] It is characteristic for blistering on palms and soles to be succeeded by focal keratoderma, usually to a lesser degree.^[10] A rather characteristic thickening of nails is also common in Dowling –Meara EBS.^[11]

In severe cases of EBS-DM, extensive blisters may appear spontaneously particularly in hot environment. Bullae appear more hemorrhagic than in other types of EBS. Laryngeal involvement can also occur resulting in a weak cry. These neonates are more susceptible to recurrent infections and septicemia caused by *S. aureus* and *P. aeruginosa*.^[12]

Death in neonatal period is not infrequent Phenytoin is a commonly used anti-epileptic given in a dose of 5mg /kg per day.

Common side effects of the drug are hirsutism, gum hyperplasia, ataxia, skin rashes, steven-johnson syndrome, megaloblastic anaemia polyneuropathy and rickets.^[13]

Invitro studies have demonstrated that phenytoin inhibits synthesis or secretion of collagenase.

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This ability may be responsible for the beneficial effect of phenytoin in patients with EB.^[14] Our case report shows that phenytoin if used judiciously is safe and effective in reducing blistering and thereby improving outcome.

Here is the image of post treatment (Figures 5).

**Fig. 5: Healed lesions
back view**

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