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**EPIDERMOLYSIS BULLOSA SIMPLEX DOWLING-MEARA - A CASE REPORT**Ivelina Yordanova<sup>1</sup>, Snezhina Vassileva<sup>2</sup>, Zdravka Demerjjeva<sup>2</sup>, Dimitar Gospodinov<sup>1</sup>, Nikolai Tsankov<sup>2</sup><sup>1</sup>*Department of Dermatology and Venereology,**Medical University - Pleven, Bulgaria*<sup>2</sup>*Department of Dermatology and Venereology,**Medical University - Sofia, Bulgaria***ABSTRACT**

A case with Epidermolysis bullosa simplex Dowling-Meara is presented. It concerns a 20 days breast-fed girl with severe blistering and erosions on the skin formed at birth after minor mechanical trauma. There are no other affected relatives. A medical examination showed normal somatic and visceral status. There were multiple herpetiform grouped bullous and erosive lesions on the face, trunk, upper and lower limbs, palms and soles. The mucous membranes were not affected. There was also a marked nail dystrophy on the fingers and toenails. The electron microscopic examination revealed cytolysis of the basal keratinocytes with clumping of the perinuclear tonofilaments in the lesional skin. Because of the clinical features and the results of the electron microscopic examination of the skin the case here reported should be considered as an Epidermolysis bullosa simplex Dowling-Meara. The child is followed up to the age of three. The course of the disease was benign with a decreased formation of blisters and erosions. The marked nail dystrophy was persistent. There was palmar and plantar hyperkeratosis. The child's growth, neural and psychic development was normal. The early diagnosis of the bullous epidermolysis is helpful in the prognostic assessment of the disease and in the medical and genetic advice for the parents.

**Key words:** Epidermolysis bullosa simplex Dowling-Meara, tonofilament clumps, electron microscopy.

**INTRODUCTION:**

Epidermolysis bullosa simplex (EBS) is a group of hereditary bullous diseases characterized by intraepidermal blistering due to mild mechanical traumas (3). Patients are presented 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 (6, 7). The prevalence of the EBS has been assessed in several countries (11, 12). The most detailed

investigation has been made in Scotland, where the point prevalence of EBS has been calculated as 28, 6 per million, with the Dowling-Meara subtype accounting for 0, 6 per million (11). The Dowling-Meara subtype of EBS is a rare severe blistering disease inherited in an autosomal-dominant fashion. There is a great range of severity in the various cases. Blistering may be exceptionally severe during the neonatal period and frequently occur on the face, trunk and limbs and tend to be disposed in herpetiform groups. In the most severe cases, blistering may appear spontaneously. Other physical signs include varying degrees of intra-oral blistering, nail dystrophy, minor scarring, palmo-plantar keratoderma, a lack of seasonal variation and improvement during later childhood (3). A case with Epidermolysis bullosa simplex Dowling-Meara is presented.

**Case report:**

A 20 days old breast-fed girl was born from first normal pregnancy, with body weight 2700 g and height 47 cm. The disease started at birth with severe blistering and erosions on the face, trunk and extremities formed after minor mechanical traumas. The blisters also appeared spontaneously. They ruptured and formed extensive superficial erosions. The latter healed in the centre with recurrent blistering at the margins of these areas within a week. There were no other affected relatives. The child was hospitalized in a damaged skin condition.

A medical examination showed normal somatic and visceral status, muscle tone and reflexes with exception of the spontaneous and provoked clonus of the extremities. Dermatological examination revealed multiple herpetiform grouped bullous and erosive lesions, slight scars and milia on the face, trunk, upper and lower limbs (fig.1). There were hemorrhagic blisters on the palms and soles. The mucous membranes were not affected. There was also a marked nail dystrophy on the fingers and toenails. The routine blood count, biochemistry and urine analysis were within the normal limits with the exception of hypoproteinaemia (total protein - 58 g/l; normal values: 60-84 g/l), calcium deficiency (Ca - 1.83 mmol/l, normal values: 2.1 - 2.6 mmol/l) and phosphorus