

## SPOT DIAGNOSIS (IMAGE GALLERY)

**BLISTERING SKIN LESIONS - EPIDERMOLYSIS BULLOSA DYSTROPHICA**

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A one day old male newborn was referred to us with a history of skin sloughing and blistering of the skin since birth. He was born of non-consanguineous marriage vaginally at 38 weeks of gestation to a 2<sup>rd</sup> gravida mother. There was no significant antenatal or family history. He cried soon after birth. His birth weight was 2.75 Kg. Few blisters and skin sloughing of palm and foot was present (Fig 1, 2). Nikolky's sign was negative. There were minimal perioral lesions. Nails were dystrophic. Oral cavity, conjunctiva, cornea, scalp and genitalia were normal. Systemic examination was normal. On 3<sup>rd</sup> day of discharge, baby died due to infection.

**What is the diagnosis?**

Epidermolysis bullosa dystrophica (EBD). Epidermolysis bullosa (EB) is a group of inherited disorders characterized by blistering of the skin as a result of minor trauma. Diagnosis is based on clinical symptomatology, histopathology, electron microscopy and genetic studies. (1) Major types of Epidermolysis bullosa previously include epidermolysis bullosa simplex, hemidesmosomal epidermolysis bullosa, junctional epidermolysis bullosa, and dystrophic epidermolysis bullosa, on the basis of the level of tissue separation within the cutaneous basement membrane zone. (2) Epidermolysis bullosa dystrophica (EBD) is a rare inherited skin disease generally presenting in newborns. It is characterized by non-inflammatory bullous lesions which can involve the mucous membranes. The different types of EBD include dominant dystrophic which has onset at birth or early infancy, blistering predominates on dorsa of hands, elbows, knee and lower legs, milia with scarring is present. Nail dystrophy will be seen in 80 percent. Recessive form have widespread blistering, scarring, milia deformity like pseudosyndactyly or joint contracture, severe mucosal and nail involvement. Current treatment for epidermolysis bullosa consists of supportive care for skin and other organ systems and entails a combination of wound management, infection support for chronic wounds, surgical management as needed, nutritional support and prevention of complications. (3) Prognosis varies considerably and is based on both EB subtype and the overall health of the patient. (4) The skin should be protected from trauma, including tapes and adhesives. The blister should be punctured with a sterile needle or a blade by leaving the roof of the blister intact. The ideal dressing is yet to be developed, although there are now a variety of suitable dressings available (5). The affected area should be dressed with white petrolatum-impregnated gauze help optimal healing. Non-adhesive dressing pads or Vaseline-impregnated gauze covered by soft, bulky dressings are ideal. Topical antibiotics, such as mupirocin, may be used prophylactically. (5,6)

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