Dystrophic Epidermolysis Bullosa

A 6 year old male child presented with erosion, scarring, closure of the digits of the right foot, absent nails, resulting in mitten appearance (***fig* 1**). The lesion was present since birth with healing and blistering cycle occurring from infancy leading to progressive pseudosyndactyly characteristic of dystrophic epidermolysis bullosa. A few similar lesion were also seen on the other areas of the body. There was loss of upper teeth in our patient.

A differential diagnosis of epidermolysis bullosa and congenital Porphyrias (congenital erythropoietic porphyria, erythropoietic protoporphyria etc), were considered. Absence of photosensitivity, lesions on sun exposed parts, hypertrichosis, and red/pink urine were in favour of diagnosis of epidermolysis bullosa over porphyrias. Based on distinctive clinical feature, a diagnosis of dystrophic epidermolysis bullosa was made.

Dystrophic epidermolysis bullosa (DEB) is characterised by blisters that heal with scarring and milium formation. DEB is derived from defects of ultrastructural entity known as anchoring fibril, which results in sublamina densa separation. DEB can be inherited either in an autosomal recessive or dominant fashion. There are four primary subtypes which include dominant DEB of Cockayne-Touraine, dominant DEB of Albopapuloid or Pasini variant, localised recessive DEB and generalised recessive DEB. Although generalised blistering can take place early in life, blistering usually become localised to repetitively traumatised areas such as knees and acral surfaces. The areas show characteristic scarred, dystrophic appearance. Nail dystrophy or nail loss with atrophic scarring of the digital digits are common. The oropharynx can be severely affected in some cases with scarring that limits the movement of the structures. The teeth may show enamel pitting and caries leading to loss of teeth. Mucosal erosions of the esophagus can also be present, all these features and caloric need for wound healing can lead to malnutrition and growth retardation.

FIG.1 *Erosions, and atrophic scars on feet. Note loss of nails of all digits, pseudosyndactyly and mitten-like deformity of right foot.*

Pseudosyndactyly resulting from closure of the digits in a mitten of skin is extremely common. Skin biopsy with the dermal-epidermal basement membrane zone visualised by electron microscopy or indirect immunofluoroscent microscopy, will show the level of blistering. Management includes treatment of infection and gastrointestinal problems, eye care, supportive skin care, and nutritional management.

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