Read Item - Epidermolysis Bullosa

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Abstract: Doctor's resource on Epidermolysis Bullosa

Epidermolysis Bullosa (EB)

EB comprises a group of hereditary mechanobullous disorders characterised by traumatic blistering of the skin and mucosa. It is sub-classified according to the pattern of inheritance and the level of the split in the skin. The recessive dystrophic type is the one most commonly associate with cicatricial alopecia, however permanent alopecia may also occur in cicatricial junctional EB and very rarely in EB simplex. EB acquisita is an acquired autoimmune disorder with circulating antibody to type VII collagen in the anchoring filaments (270 kD antigens), mimicking EB dystrophica.

Dystrophic EB presents with bullae at birth. The defect is within the anchoring fibrils and is due to an alteration of type VII collagen that results in separation of the dermis and epidermis with the split forming in the upper dermis. Blisters heal with atrophic scars and occasionally leave a localised absence of skin. The recessive form of dystrophic EB is a particularly devastating disease producing fusion of the digits, oesophageal strictures, blindness and premature death due to neoplasia (SCC) of the skin, mouth and oesophagus. Cicatricial alopecia occurs and may be associated with hair that is generally fine and sparse. Sclerosis of the skin may develop and contribute to the alopecia.

Cicatricial junctional EB is an autosomal recessive condition. The split occurs in the lamina lucida and generalised blistering begins at birth in response to trauma and heals with atrophy. Patches of cicatricial alopecia of the scalp, eyebrows, axillary and pubic regions may occur. The blistering is sufficiently dramatic to cause few diagnostic difficulties, although electron microscopy is required to distinguish this form of EB from dystrophic EB.

Generalised atrophic benign junctional EB is an autosomal dominant condition that produces widespread blistering at birth that improves with age. Dystrophic nails, dental enamel defects and oral involvement occur. There is a multifocal scarring alopecia. The clinical features are less reliable than electron microscopy in determining the subtype of inherited epidermolysis bullosa.

Epidermolysis bullosa acquisita (EBA) is an acquired autoimmune blistering disease that may mimic bullous pemphigoid (BP). Acral blisters may occur and EBA is notoriously unresponsive to treatment. Scarring alopecia may occur. Immunofluorescence of salt-split skin, or immunoblotting of a circulating antibody help to differentiate EBA from BP.

Treatment of the scarring forms of EB is generally unsatisfactory and revolves around supportive care and genetic counseling for the hereditary forms.