key points

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Epidermolysis bullosa (EB) arises from mutations within genes encoding for different proteins which contribute to the structural integrity of the epidermis and basement membrane zone. There are four major EB types: EB simplex (EBS), dystrophic EB (DEB), junctional EB (JEB) and Kindler syndrome.

The main cutaneous features of inherited EB are mechanical fragility of the skin and formation of blisters and erosions with minimal trauma. Clinical manifestations can vary widely and this is dependent on the EB subtype. The initial diagnosis of EB is based on the patient's personal and family history and clinical examination. Skin biopsies are taken from newly induced blisters to identify the level of skin cleavage which helps to determine the subtype.

EBS is the most common form of EB. It is almost exclusively autosomal dominant in inheritance. Patients with EBS localised often present with blisters predominantly on the palms and soles following minimal trauma which are exacerbated by warm weather. Onset of symptoms in EBS is usually at birth or shortly after, although in the localised form it may not present until late childhood or even early adulthood. Patients with severe forms of EBS may have widespread cutaneous blistering sometimes leading to death in infancy.

JEB is inherited in an autosomal recessive pattern. A hallmark feature of JEB is the presence of enamel hypoplasia. In the generalised severe form, extensive generalised cutaneous blistering is present from birth. Anaemia and growth retardation are both common features, and involvement of the gastrointestinal, genitourinary, ocular and respiratory systems are seen. The one-year mortality rate in JEB is high at 40-45%.

In dominant DEB, generalised cutaneous blistering occurs from birth, with subsequent milia, atrophic scarring and dystrophic nails. Recessive DEB (RDEB) presents with extensive blistering from birth and is associated with anaemia, growth retardation and oesophageal strictures. Pseudosyndactyly is a frequent complication. Patients with RDEB are at risk of developing aggressive cutaneous squamous cell carcinomas.

Optimal care of EB patients requires a multidisciplinary approach centred on avoidance of trauma and blistering of the skin, good wound care, prevention of secondary infection, and management of extracutaneous complications. Applying non-adherent dressings to susceptible areas such as the elbows and knees, loose-fitting clothing and cool air conditioning can help prevent formation of new blisters. Wound colonisation is common but infection should be treated to promote healing. Cleansing agents such as normal saline, dilute potassium permanganate, vinegar and diluted bleach have been used. Antibiotics should be used judiciously guided by culture results. There should be close monitoring for cutaneous malignancies and full skin checks by a dermatologist at three-monthly intervals are recommended.