1 Epidermolysis Bullosa (EB) – the Condition

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1.1 Definition – 2
1.2 Pathology of EB – 2
1.3 The Most Common Sub-Types and Their Signs and Symptoms – 6
1.4 The Most Common Cutaneous and Extracutaneous Complications – 8
1.5 Diagnosis – 10
1.6 Therapy – 10
1.1 Definition

'Bulla (lat.) f: Blister; primary skin eruption; a cavity filled with liquid raised above the normal skin level; caused by a simple separation between the layers of the skin, usually only one chamber; at least 5 mm; differentiation is made between the localisation and the cause: subcorneal bulla (below the cornea), intra-epidermal bulla (in the epidermis), sub-epidermal bulla (between the epidermis and the corium) and acantholytic bulla (in the epidermis due to the breakup of the cell structures); bulla inflammatoris (inflammatory blister due to toxic noxa, inflammatory, allergic reactions, etc.), bulla mechanica (due to mechanical injury in epidermolysis), bulla actinica (due to exposure to the sun, e.g. hydroa vacciniforme), bulla spontanea (e.g. pemphigoid). Compare with efflorescence' (Pschyrembel 2012, p. 328).

'Epidermolysis (; ; lys-*) f: Separation and blister development in the dermoeipidermal zones (e.g. e. bullosa acquisita, forms of e. bullosa hereditaria and bullous pemphigoid) or intra-epidermal (e.g. pemphigoid vulgaris)' (Pschyrembel 2012, p. 597).

Epidermolysis bullosa (EB): EB is a hereditary skin condition in which the skin of the affected person is liable to blistering. The skin can be as sensitive as the ‘wing of a butterfly’.¹

In this rare congenital condition there is a tendency for the skin to blister on account of minor injury. Sometimes, pressure or mild friction alone is sufficient to cause blistering. Depending on the form of the condition, this blistering can also affect the mucous membranes, especially in the mouth and the oesophagus.

The cause of this fragile skin and mucous membranes is a mutation in certain genes. The mutations are in the genes of the structural proteins of the dermoeipidermal basement membrane zone (subepithelial). Normally, these structural proteins hold the layers of the skin together, but in this case they are deficient or defective in their function. Depending on the form, number and localisation, etc., there can be a very wide spectrum of possible clinical forms, which can vary from single blisters on the soles of the feet or palms of the hand to a generalisation over the whole body and the development of serious systemic complications. The clinical picture, progress, prognosis and course as well as the treatment can vary considerably. A causal treatment does not exist at present (cf. Laimer et al. 2008; Netzwerk EB 2009).

1.2 Pathology of EB

In principal, the classification of the three main forms of hereditary EB is done according to the level of blister formation in the skin.

¹ Colloquially, children with EB are known as ‘butterfly children’.