Rare DEB subtypes		
Autosomal dominant or recessive inheritance		
Type	**Affected gene (protein)**	**Clinical symptoms**
DDEB, pruriginosa RDEB, pruriginosa	COL7A1 (type VII collagen)	Usually presents initially as localized or intermediate DDEB or RDEB in childhood and early adulthood. Characterized by intensely pruritic, excoriated violaceous papules or linear plaques and scars particularly on the lower legs, thighs and arms which can become more progressive from adolescence through adulthood. Nail dystrophy and milia are common. May co-exist with non-pruriginosa DEB within families.
DDEB, self-improving RDEB, self-improving	COL7A1 (type VII collagen)	Skin blistering presents at or shortly after birth, usually on the extremities. Aplasia cutis of the lower limbs may be present. Scarring and milia occur at sites of blistering. Skin fragility improves spontaneously and may resolve completely over the first year or two of life although nail dystrophy, particularly of the toenails, may persist throughout life.
Autosomal dominant and recessive inheritance		
Dominant and recessive compound heterozygous DEB	COL7A1 (type VII collagen)	Severe skin and mucosal fragility presenting from birth indistinguishable from severe RDEB. May have a family history of DDEB in family members.
Autosomal recessive inheritance		
RDEB, inversa	COL7A1 (type VII collagen)	In the neonatal period and childhood skin blistering is usually generalized and of intermediate severity. From adolescence to early adulthood, a predilection for flexural sites develops, specifically in the axillae, groins, perianal area and natal cleft. In women, there may be marked vulvovaginal and inframammary skin blistering. Mucosal disease with blistering and scarring in the mouth and oesophagus is characteristic. Involvement of the external auditory canal may lead to narrowing or complete occlusion. Nail involvement is common.
RDEB, localized	COL7A1 (type VII collagen)	Skin fragility usually presents at or shortly after birth but may rarely be of late onset in adulthood. Blistering is limited in extent; it may affect predominantly the hands and feet, but in others may be restricted to the pretibial area. Nail dystrophy and loss are common. Oral and oesophageal mucosal involvement are usually mild or absent.

The information above has been sourced from the 'Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility' article published in the British Journal of Dermatology
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