Rare JEB subtypes		
Autosomal recessive inheritance		
Type	**Affected gene (protein)**	**Clinical symptoms**
JEB with pyloric atresia	ITGA6 (integrin α6) ITGB4 (integrin α6β4)	Full thickness skin loss over extensive areas of the head, trunk and limbs at birth. Subsequent severe skin fragility. Skin loss can cause deformity of structures such as the ears and nose. Severe phenotypes can present with rudimentary ears. Nail dystrophy and loss. Pyloric atresia is usually evident within the first days-week of life. May have atresia at other gastrointestinal sites e.g. duodenal or anal. Usually lethal within the first few weeks of life despite surgical correction of pyloric atresia. Milder, non-lethal forms have less severe skin and nail involvement but with frequent genitourinary tract involvement.
JEB localized	COL17A1 (type XVII collagen) LAMA3, LAMB3, LAMC2 (laminin 332) ITGB4 (integrin α6β4) ITGA3 (integrin α3 subunit)	Limited cutaneous fragility and blistering, often only acral. Variable nail dystrophy and mucosal involvement. Variable dental enamel defects. Normal hair.
JEB inversa	LAMA3, LAMB3, LAMC2 (laminin 332)	Onset of blistering from birth in flexural sites. Atrophic scarring. Dental enamel abnormalities. Variable nail loss.
JEB late onset	COL17A1 (type XVII collagen)	Onset of skin fragility in childhood often starting acrally. Progressive fragility with age. Healing with skin atrophy and loss of dermatoglyphs. Scarring leading to flexion contractures of the fingers and reduction of mouth opening may occur with age. Variable dental enamel and nail involvement.
JEB-laryngo-onycho-cutaneous (LOC) syndrome	LAMA3A (laminin α3A)	Onset of skin fragility from birth with blistered areas leaving erosions and granulation tissue (much more than JEB severe). Predilection for the face and neck. Nail dystrophy and loss with granulation tissue of the nail beds. Conjunctival and eyelid granulation tissue leading to symblepharon, scarring and impaired vision. Laryngeal granulation tissue leading to respiratory obstruction which can be lethal. Profound anaemia is a major feature due to bleeding from over granulating wounds.
JEB with interstitial lung disease and nephrotic syndrome	ITGA3 (integrin α3 subunit)	Variable cutaneous features with absence or presence of skin fragility from infancy. Nails may be dystrophic and hair may be sparse. Interstitial lung disease and nephrotic syndrome predominate the phenotype, and can be diagnosed soon after birth. Death in infancy or early childhood is the norm.

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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