Rare EBS subtypes		
Autosomal dominant inheritance		
Type	**Affected gene (protein)**	**Clinical symptoms**
EBS, with mottled pigmentation	KRT5 (keratin 5) KRT14 (keratin 14) EXPH5 (exophilin-5)	Skin blistering starts at birth and is generalised of intermediate severity. Mottled or reticulate pigmentation develops gradually. Focal keratoses of the palms and soles, and dystrophic, thickened nails occur over time.
EBS, migratory circinate erythema	KRT5 (keratin 5)	Multiple vesicles (small, fluid-filled sacs) are present from birth onwards and over time take on a circular acquire over time a typical circinate migratory pattern on an erythematous background; post-inflammatory hyperpigmentation develops gradually and may have a mottled pattern. Nails may be dystrophic (appear damaged, misshapen, discoloured, and curvy).
EBS, intermediate with cardiomyopathy (high rate (50%) of de novo inheritance)	KLHL24 (kelch-like member 24)	Extensive skin defects on the extremities are present at birth and heal with hypo- and hyperpigmentation and skin atrophy, initially resembling burn-like scars (Figure 1g).18 Skin blistering diminishes in adulthood, but fragility persists, with erosions occurring after minimal mechanical trauma. Diffuse or focal plantar keratoderma. Nail thickening and onychogryphosis. Diffuse alopecia has been reported in some adult patients. Dilated cardiomyopathy has been reported in young adulthood.20,21 Laboratory screening (pro-BNP, creatine kinase, creatine kinase MB) should be started as early as the age of 2 years, with yearly follow-ups. If pathologic values are found, cardiologic examination, ECG and cardiac ultrasound should be performed.
EBS, intermediate with PLEC pathogenic variants (previously known as EBS Ogna)	PLEC (plectin)	Skin blistering starts at birth, is mainly acral but may be widespread. The autosomal dominant subtype is characterized by a mild course, mainly acral erosions and postlesional violaceous and hypopigmented macules.Only three cases with the autosomal recessive subtype have been published yet, all of intermediate severity. Plantar keratoderma. Dystrophic thickened nails, sometimes onychogryphosis. No muscular dystrophy.
Autosomal recessive inheritance		
Type	**Affected gene (protein)**	**Clinical symptoms**
Recessive EBS, intermediate or severe	KRT5 (keratin 5) KRT14 (keratin 14)	Skin blistering starts at birth and is generalized and severe in most cases. No improvement of cutaneous fragility is expected with age. Healing of lesions leads to post-inflammatory hyperpigmentation. Absence of keratin 5 leads to widespread blisters and erosions and early lethality.

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 https://doi.org/10.1111/bjd.18921

EBS, localized or intermediate with BP230 deficiency	DST (bullous pemphigoid antigen 230)	Skin blistering starts at birth or in childhood, and is mostly localized to acral extremities. Plantar keratoderma. Nail dystrophy.
EBS, localized or intermediate with exophilin-5 deficiency	EXPH5 (exophilin-5)	Generalized skin blistering starts at birth or in infancy. Blistering tendency may diminish with age, while crusts and scabs reflect the fragility of the skin. Mild mottled pigmentary changes may develop.
EBS, intermediate with PLEC pathogenic variants	PLEC (plectin)	Skin blistering starts at birth, is mainly acral but may be widespread. The autosomal dominant subtype is characterized by a mild course, mainly acral erosions and postlesional violaceous and hypopigmented macules. Only three cases with the autosomal recessive subtype have been published yet, all of intermediate severity. Plantar keratoderma. Dystrophic thickened nails, sometimes onychogryphosis. No muscular dystrophy.
EBS, intermediate with muscular dystrophy	PLEC (plectin)	Generalized skin blistering starts at birth and is of intermediate severity. Blistering tendency diminishes with age. Focal plantar keratoderma. Nail dystrophy and loss. Mucosal involvement including oral, ocular and urethral mucosae is common. Dental anomalies. Muscular dystrophy starts at a variable age, ranging from infancy to adulthood. Cardiomyopathy may be associated. Granulation tissue and stenosis of the upper respiratory tract and hoarseness may occur. Muscular dystrophy is usually life-limiting in childhood or early adulthood. Pyloric atresia may be associated in rare cases.
EBS, severe with pyloric atresia	PLEC (plectin)	Widespread full-thickness congenital absence of skin. Pyloric atresia. Involvement of the oral mucosa. Anaemia and growth retardation. Neonatal lethal course.
EBS, localized with nephropathy with CD151 deficiency	CD151 (CD151 antigen)	Only a few individuals with this subtype have been reported so far in the literature. Skin blistering starts at birth and is widespread primarily in the pretibial area but also scattered on other parts of the body, particularly those exposed to trauma. Facial freckling, poikiloderma and atrophy of the skin, and acrogeria of the backs of the hands on the sun-exposed areas reported in one case. Erosions of the oral mucous membranes. Nail dystrophy. Early-onset alopecia. Nasolacrimal duct stenosis. Oesophageal webbing and strictures. Nephropathy manifesting with proteinuria. The scarcity of reported cases precludes firm screening recommendations but annual urinalysis and urea and electrolytes should probably be undertaken following diagnosis.

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