DISEASE:
Epidermolysis bullosa simplex due to exophilin 5 deficiency

NAME:	Epidermolysis bullosa simplex due to exophilin 5 deficiency
DESCRIPTION:	A rare, inherited, epidermolysis bullosa simplex characterized by mild, generalized trauma-induced scale crusts and intermittent blistering, sometimes combined with erosions, recovering with slight scarring and post-inflammatory hyperpigmentation. Clinical symptoms improve with age.
ORPHACODE:	412189
SYNONYMS:	EBS due to exophilin 5 deficiency
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	EXPH5
CREATED:	13 May 2019 - 01:02
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