
DISEASE:
Epidermolysis bullosa simplex due to BP230 deficiency

NAME:	Epidermolysis bullosa simplex due to BP230 deficiency
DESCRIPTION:	A rare, inherited, epidermolysis bullosa simplex characterized by mild, predominantly acral, trauma-induced skin fragility, resulting in blisters. Blisters mostly affect the feet, including the dorsal side.
ORPHACODE:	412181
SYNONYMS:	DST-related epidermolysis bullosa simplex EBS due to BP230 deficiency
XREF(S):	<u>Orphanet</u> <u>OMIM</u> <u>ICD-10</u>
ANALYTE(S):	<u>DST</u>
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