DISEASE:
Late-onset junctional epidermolysis bullosa

NAME:	Late-onset junctional epidermolysis bullosa
DESCRIPTION:	A form of junctional epidermolysis bullosa characterized by onset in childhood or young adulthood of blistering that first occurs around nails, accompanied by nail dystrophy and shedding, and then affects the hands and feet and, to a lesser extent, the elbows, and knees. Lesions heal with atrophic scarring. Other manifestations include disappearance of dermatoglyphs and palmoplantar hyperhidrosis. Extracutaneous involvement is restricted to soft tissue abnormalities of the oral cavity and enamel defects with development of caries.
ORPHACODE:	79406
SYNONYMS:	Epidermolysis bullosa progressiva JEB-lo Late-onset JEB
XREF(S):	Orphanet ICD-10
ANALYTE(S):	COL17A1
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