DISEASE:
Localized junctional epidermolysis bullosa

NAME:	Localized junctional epidermolysis bullosa
DESCRIPTION:	A form of junctional epidermolysis bullosa characterized by neonatal onset of localized blistering, and dystrophic or absent nails. Skin blistering is mainly confined to hands, feet, lower legs and face. Additional findings may include dental enamel hypoplasia and an increased incidence of caries.
ORPHACODE:	251393
SYNONYMS:	JEB-nH loc Junctional epidermolysis bullosa, non-Herlitz localized type Localized JEB
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	COL17A1 ITGB4
CREATED:	13 May 2019 - 01:02
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