DISEASE:
Intermediate generalized junctional epidermolysis bullosa

NAME:	Intermediate generalized junctional epidermolysis bullosa
DESCRIPTION:	A form of junctional epidermolysis bullosa (JEB) characterized by generalized skin blistering, atrophic scarring, nail dystrophy or nail absence, and enamel hypoplasia, with extracutaneous involvement.
ORPHACODE:	79402
SYNONYMS:	Generalized atrophic benign epidermolysis bullosa Generalized junctional epidermolysis bullosa, non-Herlitz type Intermediate generalized JEB Junctional epidermolysis bullosa generalisata mitis Junctional epidermolysis bullosa, Disentis type
XREF(S):	Orphanet OMIM ICD-10 OMIM OMIM OMIM OMIM

ANALYTE(S):	COL17A1 ITGB4 LAMA3 LAMB3 LAMC2
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- Centrum Medische Genetica - UZ Gent
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- integrin subunit beta 4
- laminin subunit alpha 3
- laminin subunit beta 3
- laminin subunit gamma 2

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