DISEASE:
Epidermolysis bullosa simplex with pyloric atresia

NAME:	Epidermolysis bullosa simplex with pyloric atresia
DESCRIPTION:	A rare, inherited, epidermolysis bullosa simplex characterized by generalized severe blistering with widespread congenital absence of skin and pyloric atresia that is usually fatal in infancy. Antenatally, pyloric atresia can manifest with polyhydramnios. If patients survive, they experience life-long skin fragility and nail dystrophy. Additional extracutaneous findings include failure to thrive, anemia, sepsis, intraoral blistering, enamel hypoplasia, urethral stenosis and urologic complications.
ORPHACODE:	158684
SYNONYMS:	EBS with pyloric atresia EBS-PA
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	PLEC ITGB4
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