

## Opis choroby \*

### Definicja

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.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

EBS with pyloric atresia

Postać prosta pęcherzowego oddzielania się naskórka z zarośniętym odźwiernikiem

EBS-PA

#### Kod ORPHA

158684

#### Kod OMIM

612138

#### Kod ICD10

Q81.0

#### Kod ICD11

EC30

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#### \*Źródło

orphanet