

## Opis choroby \*

### Definicja

A severe form of junctional epidermolysis bullosa (JEB) characterized by generalized blistering at birth and congenital atresia of the pylorus and rarely of other portions of the gastrointestinal tract.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Carmi syndrome

Zespół Carmi

JEB with pyloric atresia

JEB-PA

#### Kod ORPHA

79403

#### Kod OMIM

226730

#### Kod ICD10

Q81.8

#### Kod ICD11

EC31

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#### \*Źródło

orphanet