

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

*Źródło

orphanet