

Opis choroby *

Definicja

Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency is an extremely rare, autosomal recessive, gastroenterological disorder reported in three families so far that is characterized by meconium ileus without any further stigmata of cystic fibrosis (see this term) including pulmonary or pancreatic manifestations. Two of the reported patients developed chronic diarrhea in infancy. Homozygous mutations in the *GUCY2C* gene (12p12) leading to marked reduction or absence of enzymatic activity of guanylate cyclase 2C were found in the affected patients. The disease was reported to show partial penetrance.

Dane

Klasyfikacja

Choroba

Synonimy

Meconium ileus due to guanylate cyclase 2C deficiency

Niedrożność smółkowa z powodu niedoboru cyklazy guanylowej 2C

Kod ORPHA

314376

Kod OMIM

614665

Kod ICD10

P76.0

Kod ICD11

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*Źródło

orphanet