Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Intestinal obstruction in the newborn due to guanylate cyclase 2C deficiency. ORPHA:314376

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.