Opis choroby *

Definicja

A rare, genetic, intestinal disease characterized by early-onset, chronic diarrhea and intestinal inflammation due to overactivity of guanylate cyclase 2C. Additional manifestations include meteorism, dehydration, metabolic acidosis and electrolyte disturbances. Intestinal dysmotility, small-bowel obstruction and esophagitis (with or without esophageal hernia), as well as irritable bowel syndrome (without severe abdominal pain) and Crohn's disease, are frequently associated.

Dane

Klasyfikacja
Choroba

Kod ORPHA
314373

Kod OMIM
614616

Kod ICD10
P78.3

Kod ICD11
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*Źródło

orphanet