Opis choroby *

Definicja

A rare genetic intestinal disease characterized by persistent, potentially life-threatening, watery diarrhea with excessive levels of chloride in stools, hypochloremia, hyponatremia, hypokalemia, and metabolic alkalosis, resulting in chronic dehydration and failure to thrive. Antenatal ultrasound typically reveals polyhydramnios and significant dilatation of the fetal intestinal loops.

Dane

Klasyfikacja
Choroba

Kod ORPHA
53689

Kod OMIM
214700

Kod ICD10
K90.8

Kod ICD11
DA90.1

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*Źródło

orphanet