

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

*Źródło

orphanet