Opis choroby *

Definicja

A rare, genetic, syndromic intestinal disorder, characterized by congenital onset of severe watery diarrhea containing high concentrations of sodium, hyponatremia and metabolic acidosis, and generally, uni- or bilateral choanal atresia, and corneal erosions. Additional congenital malformations may include intestinal atresia, and hexadactyly.

Dane

Klasyfikacja
Choroba

Synonimy
Syndromic congenital tufting enteropathy
Syndromic congenital tufting enteropathy

Kod ORPHA
563708

Kod OMIM
-

Kod ICD10
K90.8

Kod ICD11
-

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*Źródło

orphanet