

Opis choroby *

Definicja

A rare intestinal disease characterized by impaired absorption of starch and short polymers of glucose due to primary small intestinal glucoamylase deficiency. Patients present in infancy or early childhood with chronic diarrhea, abdominal distention, and bloating. Levels of pancreatic amylase are typically normal, and histopathological analysis shows normal morphology of the intestinal mucosa.

Dane

Klasyfikacja Choroba	Synonimy Maltase-glucoamylase deficiency Niedobór maltazy - glukoamylazy
--------------------------------	---

Kod ORPHA 103907	Kod OMIM -	Kod ICD10 E74.3
----------------------------	----------------------	---------------------------

Kod ICD11
5C61.1

*Źródło

orphanet