

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

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Kod ICD10

E74.3

Kod ICD11

5C61.1

*Źródło

orphanet