

Opis choroby *

Definicja

A severe, familial hypobetalipoproteinemia characterized by permanently low levels (below the 5th percentile) of apolipoprotein B and LDL cholesterol, and by growth delay, malabsorption, hepatomegaly, and neurological and neuromuscular manifestations.

Dane

Klasyfikacja

Choroba

Synonimy

Bassen-Kornzweig disease

Choroba Bassena i Kornzweiga

Homozygotyczna rodzinna

hipobetalipoproteinemia

Homozygous familial hypobetalipoproteinemia

Kod ORPHA

14

Kod OMIM

605019

Kod ICD10

E78.6

Kod ICD11

5C81.1

[*Źródło](#)

orphanet