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

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*Źródło

orphanet