

## 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