

Opis choroby *

Definicja

cbID type methylmalonic acidemia with homocystinuria is a form of methylmalonic acidemia with homocystinuria (see this term), an inborn error of vitamin B12 (cobalamin) metabolism characterized by variable biochemical, neurological and hematological manifestations.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

CbID defect

Acyduria metylomalonowa z homocystynurią, typu cbID

Defekt CbID

Defekt kobalaminy D

Złożony defekt syntezy adenozylkobalaminy i metylkobalaminy, typu cbID

Cobalamin D defect

Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cbID

Methylmalonic aciduria with homocystinuria, type cbID

Kod ORPHA

79283

Kod OMIM

277410

Kod ICD10

E72.1

Kod ICD11

5C50.E0

*Źródło

orphanet