Opis choroby *

Definicja

cblC 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 megaloblastic anemia, lethargy, failure to thrive, developmental delay, intellectual deficit and seizures.

Dane

Klasyfikacja
Podtyp kliniczny

Synonimy
CblC defect
Acyduria metylomalonowa z homocystynurią, typu cblC
Defekt CblC
Defekt kobalaminy C
Złożony defekt syntezy adenozylkobalaminy i metylkobalaminy, typu cblD
Cobalamin C defect
Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblC
Methylmalonic aciduria with homocystinuria, type cblC

Kod ORPHA
79282

Kod OMIM
277400

Kod ICD10
D58.8

Kod ICD11
5C50.E0

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*Źródło

orphanet