Opis choroby *

Definicja

cblF 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
CblF defect
Acyduria metylomalonowa z homocystynurią, type cblF
Defekt CblF
Defekt kobalaminy F
Niedobór lizosomalnego transportera błonowego kobalaminy
Złożony defekt syntezy adenozylkobalaminy i metylkobalaminy, typu cblF
Cobalamin F defect
Combined defect in adenosylcobalamin and methylcobalamin synthesis, type cblF
Lysosomal membrane cobalamin transporter deficiency
Methylmalonic aciduria with homocystinuria, type cblF

Kod ORPHA
79284

Kod OMIM
277380

Kod ICD10
E72.1

Kod ICD11
5C50.E0

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*Źródło

orphanet