

Opis choroby *

Definicja

<i>cblF</i> 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	Synonimy
Podtyp kliniczny	CblF defect Acyduria metylomalonowa z homocystynurią, type cblF Defekt CblF Defekt kobalaminy F Niedobór lisosomalnego transportera błonowego kobalaminy Złożony defekt syntezy adenozylkobalaminy i metylkomkomaminy, 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

*Źródło

orphanet

