Opis choroby *

Definicja

A rare inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, lethargy, failure to thrive, developmental delay, intellectual deficit and seizures. There are four complementation classes of cobalamin defects (cblC, cblD, cblF and cblJ) that are responsible for methylmalonic acidemia - homocystinuria (methylmalonic acidemia - homocystinuria cblC, cblD cblF and cblJ).

Dane

Klasyfikacja
Choroba

Synonimy
Combined defect in adenosylcobalamin and methylcobalamin synthesis
Acyduria metylomalonowa z homocystynurią
Łączony defekt w syntezie adenozylokobalaminy i metylkobalaminy
Methylmalonic aciduria with homocystinuria

Kod ORPHA
26

Kod OMIM
614857

Kod ICD10
E71.1

Kod ICD11
5C50.E0

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*Źródło

orphanet