

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 (cbIC, cbID, cbIF and cbIJ) that are responsible for methylmalonic acidemia - homocystinuria (methylmalonic acidemia - homocystinuria cbIC, cbID cbIF and cbIJ).

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

*Źródło

orphanet