

Opis choroby *

Definicja

Homocystinuria without methylmalonic aciduria is an inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, encephalopathy and, sometimes, developmental delay, and associated with homocystinuria and hyperhomocysteinemia. There are three types of homocystinuria without methylmalonic aciduria; *cbIE*, *cbIG* and *cbID-variant 1* (*cbID*v1).

Dane

Klasyfikacja

Choroba

Synonimy

Functional methionine synthase deficiency
Funkcjonalny Niedobór syntazy metioniny
Niedobór metylokobalaminy
Methylcobalamin deficiency

Kod ORPHA

622

Kod OMIM

236270

Kod ICD10

E72.1

Kod ICD11

5C50.B

[*Źródło](#)

orphanet