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; cblE, cblG and cblD-variant 1 (cblDv1).

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

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*Źródło

orphanet