

## 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

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#### [\\*Źródło](#)

orphanet