

## Opis choroby \*

### Definicja

*cbIF* type methylmalonic acidemia with homocystinuria is a form of methylmalonic acidemia with homocystinuria (see this term), an inborn error of vitamin B12 (cobalamin) metabolism characterized by megaloblastic anemia, lethargy, failure to thrive, developmental delay, intellectual deficit and seizures.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

CbIF defect

Acyduria metylomalonowa z homocystynurią,  
type cbIF

Defekt CbIF

Defekt kobalaminy F

Niedobór lizosomalnego transportera  
błonowego kobalaminy

Złożony defekt syntezy adenozylkobalaminy i  
metylkobalaminy, typu cbIF

Cobalamin F defect

Combined defect in adenosylcobalamin and  
methylcobalamin synthesis, type cbIF

Lysosomal membrane cobalamin transporter  
deficiency

Methylmalonic aciduria with homocystinuria,  
type cbIF

#### Kod ORPHA

79284

#### Kod OMIM

277380

#### Kod ICD10

E72.1

#### Kod ICD11

5C50.E0

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#### \*Źródło

orphanet

