

## Opis choroby \*

### Definicja

An inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which responds to vitamin B12. There are three types: *cblA*, *cblB* and *cblD*-variant 2 (*cblD*v2).

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Adenosylcobalamin deficiency

Acyduria metylomalonowa reagująca na witaminę B12

Adenosylcobalamin deficiency Niedobór adenosylcobalaminy

Vitamin B12-responsive methylmalonic aciduria

#### Kod ORPHA

28

#### Kod OMIM

277410

#### Kod ICD10

E71.1

#### Kod ICD11

5C50.E0

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#### [\\*Źródło](#)

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