

## Opis choroby \*

### Definicja

Vitamin B12-unresponsive methylmalonic acidemia is an inborn error of vitamin B12 (cobalamin) metabolism characterized by recurrent ketoacidotic crises or transient vomiting, dehydration, hypotonia and intellectual deficit, which does not respond to administration of vitamin B12. There are two types of vitamin B12-unresponsive methylmalonic acidemia: *mut0* and *mut-* (see these terms).

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Methylmalonyl-CoA mutase deficiency

Acyduria metylomalonowa niereagująca na witaminę B12

Methylmalonyl-Coenzyme A mutase deficiency

Vitamin B12-unresponsive methylmalonic aciduria

#### Kod ORPHA

27

#### Kod OMIM

251000

#### Kod ICD10

E71.1

#### Kod ICD11

5C50.E0

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### \*Źródło

orphanet