

## Opis choroby \*

### Definicja

Vitamin B12-unresponsive methylmalonic acidemia type mut0 is an inborn error of metabolism characterized by recurrent ketoacidotic comas or transient vomiting, dehydration, hypotonia and intellectual deficit, which does not respond to administration of vitamin B12.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Complete deficiency of methylmalonyl-CoA mutase

Acyduria metylomalonowa typu mut0

niewrażliwa na witaminę B12

Całkowity Niedobór mutazy metylomalonylo-CoA

Vitamin B12-unresponsive methylmalonic aciduria type mut0

#### Kod ORPHA

289916

#### Kod OMIM

251000

#### Kod ICD10

E71.1

#### Kod ICD11

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#### \*Źródło

orphanet