

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

*Źródło

orphanet