

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

*Źródło

orphanet