Opis choroby *

Definicja

Methylmalonic acidemia due to methylmalonyl-CoA epimerase deficiency is a rare inborn error of metabolism disease characterized by mild to moderate, persistent elevation of methylmalonic acid in plasma, urine and cerebrospinal fluid. Clinical presentation may include acute metabolic decompensation with metabolic acidosis (presenting with vomiting, dehydration, confusion, hallucinations), nonspecific neurological symptoms, or may also be asymptomatic.

Dane

Klasyfikacja
Choroba

Synonimy
MCEE deficiency
Acyduria metylomalonowa z powodu niedoboru epimerazy metylomalonylo-CoA
Acyduria metylomalonowa z powodu niedoboru racemazy metylomalonylo-CoA
Kwasica metylomalonowa z powodu niedoboru racemazy metylomalonylo-CoA
Niedobór MCEE
Methylmalonic acidemia due to methylmalonyl-CoA racemase deficiency
Methylmalonic aciduria due to methylmalonyl-CoA epimerase deficiency
Methylmalonic aciduria due to methylmalonyl-CoA racemase deficiency

Kod ORPHA
308425

Kod OMIM
251120

Kod ICD10
E71.1

Kod ICD11
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*Źródło

orphanet