

Opis choroby *

Definicja

Combined malonic and methylmalonic acidemia is a rare inborn error of metabolism characterized by elevation of malonic acid (MA) and methylmalonic acid (MMA) in body fluids, with higher levels of MMA than MA. CMAMMA presents in childhood with metabolic acidosis, developmental delay, dystonia and failure to thrive or in adulthood with seizures, memory loss and cognitive decline.

Dane

Klasyfikacja

Choroba

Synonimy

CMAMMA

CMAMMA

Złożona acyduria malonowa i metylomalonowa

Combined malonic and methylmalonic aciduria

Kod ORPHA

289504

Kod OMIM

614265

Kod ICD10

E71.1

Kod ICD11

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*Źródło

orphanet