Opis choroby *

Definicja

A rare organic aciduria, due to deficiency of 3-hydroxy-3-methylglutaryl-CoA lyase characterized by episodes of metabolic decompensation with hypoketotic hypoglycemia triggered by periods of fasting or infections.

Dane

Klasyfikacja
Choroba

Synonimy
3-hydroxy-3-methylglutaryl-CoA lyase deficiency
Acyduria hydroksymetylglutarowa
Niedobór liazy 3-hydroksy-3-metyloglutaro-CoA
Niedobór liazy HMG-CoA
HMG-CoA lyase deficiency
Hydroxymethylglutaric aciduria

Kod ORPHA
20

Kod OMIM
246450

Kod ICD10
E71.1

Kod ICD11
5C50.E0

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*Źródło

orphanet