Opis choroby *

Definicja

A rare, genetic organic aciduria affecting ketone body metabolism and the catabolism of isoleucine and characterized by intermittent ketoacidotic episodes associated with vomiting, dyspnea, tachypnoea, hypotonia, lethargy and coma, with an onset during infancy and usually ceasing by adolescence.

Dane

Klasyfikacja
Choroba

Synonimy
3-ketothiolase deficiency
Kwasica alfa metyloacetooctowa
Niedobór 3-ketotiolazy
Niedobór 3-oksotiolazy
Niedobór mitochondrialnej tiolazy acetoacetylo-koenzymu A
Niedobór T2
Niedobór tiolazy alfa-metylo-acetoacetylo-CoA
3-oxothiolase deficiency
Alpha methylacetoacetic aciduria
Alpha-methyl-acetoacetyl-CoA thiolase deficiency
Mitochondrial acetoacetyl-coenzyme A thiolase deficiency
T2 deficiency

Kod ORPHA
134

Kod OMIM
203750

Kod ICD10
E71.1

Kod ICD11
5C50.DY

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*Źródło

orphanet