Opis choroby *

Definicja

A rare, autosomal recessive, organic aciduria that is characterized by variable clinical presentation ranging from acute neonatal onset of metabolic decompensation to later onset of chronic, non-specific manifestations including failure to thrive and/or developmental delay. All patients are prone to intermittent, acute metabolic decompensation. During metabolic episodes, urine analysis demonstrates elevated isovaleric acid derivatives.

Dane

Klasyfikacja
Choroba

Synonimy
Isovaleric acid CoA dehydrogenase deficiency
Niedobór dehydrogenazy kwasy izowalerianowego CoA

Kod ORPHA
33

Kod OMIM
243500

Kod ICD10
E71.1

Kod ICD11
5C50.E0

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*Źródło

orphanet