

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

*Źródło

orphanet