

# Kwasica izowalerianowa

**Kod Orpha: 33 Kod OMIM: 243500**

## 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

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.