Opis choroby *

Definicja

A rare inborn error of metabolism characterized by abnormally high urinary excretion of glutaric acid due to peroxisomal glutaryl-CoA oxidase deficiency. There is no association with a specific clinical phenotype.

Dane

Klasyfikacja
Choroba

Synonimy
Glutaric aciduria type 3
Acyduria glutarowa typu 3
Niedobór oksydazy glutarylo-CoA
Glutaryl-CoA oxidase deficiency

Kod ORPHA
35706

Kod OMIM
231690

Kod ICD10
E72.3

Kod ICD11
5C50.E0

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*Źródło

orphanet