

Niedobór dehydrogenazy izobutyrylo-CoA

Kod Orpha: 79159 Kod OMIM: 611283

Opis choroby *

Definicja

Isobutyryl-CoA dehydrogenase deficiency is an inborn error of valine metabolism. The prevalence is unknown. Only one symptomatic patient (with anaemia, failure to thrive, dilated cardiomyopathy and plasma carnitine deficiency) has been described so far, but several series of patients have been identified through newborn screening programs relying on detection of increased C(4)-carnitine levels by tandem mass spectrometry. The disorder is caused by mutations in the *ACAD8* gene (11q25).

Dane

Klasyfikacja

Choroba

Synonimy

Isobutyric aciduria

Acyduria izobutyrylowa

Kod ORPHA

79159

Kod OMIM

611283

Kod ICD10

E71.1

Kod ICD11

5C50.E0

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.