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

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*Źródło

orphanet