

Opis choroby *

Definicja

A rare organic aciduria characterized by impaired isoleucine degradation with increased plasma or whole blood C5 acylcarnitine levels (typically observed in newborn screening) and increased urinary excretion of N-methylbutyrylglycine. The condition is usually clinically asymptomatic, although patients with muscular hypotonia, developmental delay, and seizures (among others) have been reported.

Dane

Klasyfikacja

Choroba

Synonimy

2-methylbutyric aciduria
Acyduria 2-metylobutyrylowa
Niedobór dehydrogenazy
krótkich/rozgałęzionych łańcuchów acylo-CoA
Niedobór SBCAD
Opóźnienie rozwoju z powodu niedoboru
dehydrogenazy 2-metylobutyrylo-CoA
Developmental delay due to 2-methylbutyryl-
CoA dehydrogenase deficiency
SBCAD deficiency
Short/branched-chain acyl-coA dehydrogenase
deficiency

Kod ORPHA

79157

Kod OMIM

610006

Kod ICD10

E71.1

Kod ICD11

5C50.E0

*Źródło

orphanet