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

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*Źródło

orphanet