

## 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