Opis choroby *

Definicja

A rare inherited disorder of branched-chain amino acid metabolism classically characterized by poor feeding, lethargy, vomiting and a maple syrup odor in the cerumen (and later in urine) noted soon after birth, followed by progressive encephalopathy and central respiratory failure if untreated. The four overlapping phenotypic subtypes are: classic, intermediate, intermittent and thiamine-responsive MSUD.

Dane

Klasyfikacja
Choroba

Synonimy
BCKD deficiency
Ketoaciduria łańcuchów rozgałęzionych
MSUD
Niedobór BCKD
Niedobór BCKDH
Niedobór dehydrogenazy 2-ketokwasów łańcuchów rozgałęzionych
BCKDH deficiency
Branched-chain 2-ketoacid dehydrogenase deficiency
Branched-chain ketoaciduria
MSUD

Kod ORPHA
511

Kod OMIM
248600

Kod ICD10
E71.0

Kod ICD11
5C50.D0

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*Źródło

orphanet