

Opis choroby *

Definicja

Thiamine-responsive maple syrup urine disease (thiamine-responsive MSUD) is a less severe variant of MSUD (see this term) that manifests with a phenotype similar to intermediate MSUD (see this term) but that responds positively to treatment with thiamine.

Dane

Klasyfikacja

Podtyp kliniczny

Synonimy

Thiamine-responsive BCKD deficiency
MSUD wrażliwa na tiaminę
Niedobór BCKD wrażliwy na tiaminę
Niedobór dehydrogenazy ketokwasów rozgałęzionych wrażliwy na tiaminę
Thiamine-responsive MSUD
Thiamine-responsive branched-chain alpha-ketoacid dehydrogenase deficiency

Kod ORPHA

268184

Kod OMIM

248600

Kod ICD10

E71.0

Kod ICD11

5C50.D0

*Źródło

orphanet