Opis choroby *

Definicja

A rare, genetic disorder of pyrimidine metabolism characterized by increased serum beta-alanine levels and severe phenotype including hypotonia, malaise, seizures, respiratory distress, lethargy and encephalopathy. Urinary excretion of beta-alanine, beta-amino-isobutyric acid, taurine, and gamma-amino-butyric acid is also elevated. There have been no further descriptions in the literature since 1994.

Dane

Klasyfikacja
Choroba

Synonimy
Hyperalaninemia
Hiperalaninemia

Kod ORPHA
309147

Kod OMIM
237400

Kod ICD10
E79.8

Kod ICD11
5C55.1

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*Źródło

orphanet