

Hiper-beta-alaninemia

Kod Orpha: 309147 Kod OMIM: 237400

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

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.