

Kwasica 3-metyloglutakonowa typ 8

Kod Orpha: 505208 Kod OMIM:

Opis choroby *

Definicja

A rare organic aciduria characterized by neonatal onset of hypotonia, recurrent apneic episodes, lack of psychomotor development, feeding difficulties, extrapyramidal signs, and seizures. Other reported features include microcephaly, sensorineural deafness, bradycardia, and neutropenia. Laboratory studies show increased serum lactate and urinary excretion of 3-methylglutaconic acid. Brain imaging may reveal progressive cerebral atrophy. The disease is lethal in infancy.

Dane

Klasyfikacja

Choroba

Synonimy

MGA8

MGA8

Kod ORPHA

505208

Kod OMIM

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Kod ICD10

E71.1

Kod ICD11

5C50.E0

[*Źródło](#)

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.