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
-

Kod ICD10
E71.1

Kod ICD11
5C50.E0

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*Źródło

orphanet