Opis choroby *

Definicja

A rare organic aciduria characterized by increased urinary excretion of 3-methylglutaconic acid, variably associated with neutropenia (sometimes causing recurrent severe infections and potentially resulting in leukemia) and progressive neurologic manifestations, such as global developmental delay, intellectual disability, hypotonia, movement disorder, and seizures. Microcephaly, cataract, facial dysmorphism, growth retardation, endocrine abnormalities, and cardiomyopathy have also been reported. Brain imaging may show cerebral or cerebellar atrophy, or abnormalities of the basal ganglia.

Dane

Klasyfikacja
Choroba

Synonimy
3-methylglutaconic aciduria-cataract-neurologic involvement-neutropenia syndrome
MGA7
Zespół acydurii 3-metyloglutakonowej, zaćmy, zaburzeń neurologicznych i neutropenii
MGA7

Kod ORPHA
445038

Kod OMIM
616271

Kod ICD10
E71.1

Kod ICD11
5C50.E0

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*Źródło

orphanet