

## 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](#)

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