## Opis choroby *

Definicja

3-methylglutaconic aciduria (3-MGA) type I is an inborn error of leucine metabolism with a variable clinical phenotype ranging from mildly delayed speech to psychomotor retardation, coma, failure to thrive, metabolic acidosis and dystonia.

Dane

**Klasyfikacja**
Choroba

Synonimy
3-methylglutaconyl-CoA hydratase deficiency
MGA typu 1
Niedobór hydratazy 3-metyloglutakonylo-CoA
Niedobór hydratazy 3MG-CoA
3MG-CoA hydratase deficiency
MGA1

**Kod ORPHA**
67046

**Kod OMIM**
250950

**Kod ICD10**
E71.1

**Kod ICD11**
5C50.E0

---

*Źródło

orphanet